PSYCHOLOGICAL CHARACTERISTICS OF INDIVIDUALS WITH CYTOGENETIC ABNORMALITIES

WITH SPECIAL REFERENCE

TO

TURNER'S SYNDROME

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Ph.D.
University of Edinburgh
1973
ABSTRACT OF THESIS

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February 1st, 1973

Title of Thesis
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The main aim of this study, which presents a general view of individuals with different types of sex chromosome abnormality, is to provide information on a cross-section of characteristics associated with one particular type of sex chromosome aneuploidy - that of Turner's syndrome. A diagnosis of Turner's syndrome is given to female patients who have small stature, primary amenorrhoea and other variable stigmata. Chromosome analysis shows that the majority of such patients have a 45 X0 karyotype, in which one of the pair of sex chromosomes is missing; others have other types of X chromosome abnormality.

Previous psychological research on these comparatively rare individuals (as well as on other types of sex chromosome abnormality) has been seriously criticised on the grounds of poor selection and control methods. This thesis has attempted to provide adequate control data, as well as a survey of all cases of Turner's syndrome registered in the Registry of Abnormal Karyotypes, aimed at delineating more general physical and social characteristics (such as obesity or marital problems) which may be seen to have psychological repercussions.

A group of 24 adult patients was studied in detail, by employing psychological tests - namely Wechsler Adult Intelligence Scale; Benton Visual Retention test; Bender Visual Motor Gestalt test; two experimental tests devised by the author, termed Visual Recognition test and Formboards test; Pickford-Nicolson anomaloscope; Farnsworth Munsell 100-Hue test; auditory screener; and three personality inventories - the Hysteroid-Obsessoid Questionnaire, the Hostility and Direction of Hostility Questionnaire and the Cattell 16 Personality Factor Questionnaire. Results were compared with those obtained from a control group consisting of 60 females (matched for age and social class) on a general practice register.

Results from the group of individuals with Turner's syndrome differed significantly from control group data in the following respects:

(i) There was a shift towards the lower range of Full Scale IQ scores, accounted for by lowered Performance IQ, but not by Verbal IQ.
(ii) Lowered results were achieved on all five Performance sub-tests and the Arithmetic sub-test.
(iii) A greater number of errors were made on the Benton Visual Retention test; this was not considered attributable to a short term memory defect, since no differences on the Digit Span sub-test had been noted.

(iv) A greater number of errors were made on the Bender Visual Motor Gestalt test; greater difficulty was experienced with figures involving contiguity or overlap.

(v) a. A greater number of errors of recognition were made on the experimental Visual Recognition test in specifically defined areas.

b. Greater difficulty with part/whole integration was noted, evidenced by increased completion times on the experimental Formboards test.

(vi) A greater number of errors were made on the IGO-Hue test and wider matching ranges were demonstrated on the Anomaloscope; these are not identifiable in terms of typical forms of colour blindness.

(vii) There was shown to be hearing loss over all frequencies in both ears.

(viii) There were differences in several personality traits, inclined in a typically introverted direction.

The data are discussed in relation to theories of perception and learning. A tentative model is proposed, which attempts to relate the genetic abnormality to the general set of results. The possible consequences of the results on personality functioning is briefly discussed.
I DECLARE that this thesis has been composed by myself and that it contains a record of research carried out by myself. A copy of a paper, already published, based on part of this research is included in the Appendix.
ACKNOWLEDGMENTS

The research project described in this thesis was undertaken whilst I was working as clinical psychologist for the Medical Research Council in the Clinical and Population Cytogenetics Unit, Edinburgh, under the direction of Professor H.J. Evans, whose encouragement was much appreciated. I should like to thank members of the Unit for their assistance; in particular, Dr. W.H. Price, for acting as one of the thesis supervisors; Dr. J.S.S. Milne, Dr. M.Newton and Dr. M.Wright, for providing access to unpublished clinical data; Mrs. A. Frackiewicz and her colleagues, for Registry facilities; and Mr. N. Davidson, for his considerable help with the illustrations.

I am very much indebted to Dr. F.M. McPherson and to Dr. R.J. Wales, my other two supervisors, for their invaluable advice on methodology and constructive criticism of thesis presentation.

I am also grateful to Mr. F. Aspinall, for guidance on the use of the anomaloscope, and to Dr. A.E. Philip, for making available unpublished psychological data.

Finally, I should like to express my gratitude to Dr. D. Williams and Partners, for the provision of surgery facilities, including access to the control sample; and to the patients forming the basis of the study, whose consent was so necessary.
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The main aim of this study, which presents a general view of psychological data on individuals with different types of sex chromosome abnormality, is to provide information on a cross-section of characteristics associated with one particular type of sex chromosome aneuploidy - that of Turner's syndrome. A diagnosis of Turner's syndrome is given to female patients who have small stature, primary amenorrhoea, and other variable stigmata. Chromosome analysis shows that the majority of such patients have a 45 X0 karyotype, in which one of the pair of sex chromosomes is missing; others have other types of X chromosome abnormality.

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The data are discussed in relation to theories of perception and learning. A tentative model is proposed which attempts to relate the genetic abnormality to the general set of results. The possible consequences of the results on personality functioning is briefly discussed.
Negative state - as seen in cell obtained from normal male

Positive state - as seen in cell obtained from normal female

Double positive state - as seen in cell obtained from 47 XXX female

I. Sex chromatin
It is the purpose of this thesis to review and investigate the psychological characteristics of those individuals who have abnormal sex chromosome constitutions. Its particular concern is with describing certain aspects of females with Turner's syndrome.

From this group an attempt has been made to collect a set of parameters from which a profile of psychological characteristics may be constructed. In doing this it was considered advisable to concentrate on those areas concerned with intellectual and cognitive functioning which it could be hypothesised are more likely to be affected by the cytogenetic abnormality involved. A brief study of personality characteristics is also reported, although these should possibly be considered as consequential on the physical abnormalities associated with the syndrome.

It is proposed to introduce the topic by reference to the historical background of the science of cytogenetics, before reviewing sex chromosome abnormalities in general.

**Sex chromatin studies**

An essential preliminary to chromosome studies was the discovery by M.L. Barr and E.G. Bertram in 1949 of a difference existing between nerve cells taken from male and female cats. The nuclei of the cells from the female cats contained a large dark staining body which was not evident in the nuclei of male cat cells. This mass is now commonly referred to as the sex chromatin and is found to be a fairly widespread phenomenon in the animal kingdom, as well as in man. An individual may be classified as chromatin positive or chromatin negative, dependent on whether or not the mass is present. As may be inferred, cells which are chromatin negative have most commonly been taken from a phenotypically male individual, whilst chromatin positive
Clinical picture of female with Turner's syndrome (Adult) showing decreased stature, shield-shaped chest with widely-spaced nipples and increased carrying angle
cells are most characteristically present in an individual having the female phenotype (see Illustration I).

At first skin biopsies were obtained for sex chromatin studies and were often used as diagnostic aids for patients with equivocal external genitalia. This method was soon superseded by the less painful buccal smear technique, which involves the examination of cells obtained by scraping the inside of the cheek.

On extremely rare occasions clinical states were encountered in which the sex, as determined by sex chromatin studies, was at variance with the phenotypic, or physical, sex. (Court Brown, 1961). The states where this occurred were:

(i) The majority of cases with Klinefelter's syndrome
(ii) The majority of cases with Turner's syndrome
(iii) Cases of testicular feminisation

(i) Klinefelter's syndrome: This term was originated by Klinefelter et al. in 1942 in describing a group of men characterised by enlargement of the breasts (medically termed gynaecomastia), very small testes, and absence of sperm in the semen. With the advent of nuclear sexing techniques Plunkett and Barr (1956) found a sex chromatin body to be present in a number of cells taken from males manifesting this syndrome. The idea therefore developed that chromatin positive males with Klinefelter's syndrome were sex-reversed females - that they had been conceived as females, but had developed as males owing to some error in sex differentiation (Court Brown, 1967).

(ii) Turner's syndrome: This syndrome (see Illustration II) was described by the endocrinologist H.H. Turner in 1938. Patients demonstrating it typically presented with the triad of signs of sexual infantilism, webbing of the skin of the neck, and cubitus valgus (increased carrying angle - a deformity of the elbow which prevents the arm being held straight). Whilst it was the combination of these three signs occurring in patients of female phenotype that Turner wished to remark upon specifically, he also commented that the patients all demonstrated retardation in skeletal growth and sexual
Karyotype of normal male with chromosome complement 46XY
Sex chromatin studies on these patients showed the majority (about 80%) to be chromatin negative. Once again sex reversal was considered to have taken place during the development of the foetus, and for a while the patients were referred to in the literature as "genetic males".

(iii) Testicular feminisation syndrome (male pseudohermaphroditism): This condition is characterised by normal female external genitalia and secondary sex characteristics, although at puberty pubic and axillary hair tend to develop only scantily. The first physical sign of abnormality is primary amenorrhoea, although the condition may present earlier with the appearance of hernial sacs, in which testes and male genital ducts can be found. Nuclear sexing procedures indicate the chromatin negative state. This condition, therefore, is the only one of the three under review in which true sex reversal has taken place; as such, it is no further concern of this thesis, since, although the genotype is at variance with the phenotype, it is not in itself abnormal.

Chromosome studies

The techniques of examining chromosomes had long been unreliable and differing estimates had been advanced of the number of chromosomes to be found in normal cells. In 1956 Tjio and Levan established that there are 46 chromosomes present in cells of normal human tissue, and they also provided a reliable classification of these chromosomes. Whilst 22 pairs of them (the autosomes) are identical in males and females, the remaining two chromosomes (the sex chromosomes) are different. In the female these two chromosomes are identical, and termed X chromosomes; but in the male the two sex chromosomes are non-identical, and comprise one (an X chromosome), as in the female pair, and a smaller one, termed Y. The chromosome complement (termed karyotype) of normal males is expressed as 46 XY and that of
IV. Karyotype of female with 45 XO Turner’s syndrome
normal females as 46 XX (see Illustration III).

The development of Tjio and Levan's technique of examining the chromosomal complement of cells meant that the three types of apparent sex reversal could be investigated in more detail, and, indeed, it demonstrated that patients with Klinefelter's syndrome and Turner's syndrome, unlike those with testicular feminisation, were not examples of sex reversal.

Jacobs and Strong (1959) published their findings on a chromatin positive male with Klinefelter's syndrome, who was shown to have a 47 XXY chromosome complement. This report was followed by one (Ford et al., 1959), in which a chromatin negative female having the typical Turner's syndrome phenotype was demonstrated to have only 45 chromosomes, the missing one being one of the X chromosome pair (see Illustration IV). Also in 1959 Jacobs et al. described the first known example of a female with a 47 XXX sex chromosome complement.

Thus, within one year, a new discipline had been established, and three of the four main types of sex chromosome abnormality had been described, namely:

the triple XXX female, karyotype 47 XXX
the Klinefelter male, " 47 XXY
and the Turner female, " 45 XO

The technique described by Tjio and Levan (1956) involves the culturing of human cells to a stage in their development when they are just about to divide. If the process is arrested at this point the chromosomes are sufficiently distinct to be easily counted and classified. Clearly this technique is more time-consuming and laborious than the buccal smear technique described earlier. For this reason nuclear sexing surveys were undertaken, in which abnormalities assessed by taking buccal smears were further analysed by the culturing technique. It should be emphasised that this method isolated only those individuals who had numerical abnormalities of their X chromosomes; it could not be used to identify the 47 XXY karyotype. That
(i) Cell obtained from female

(ii) Cell obtained from normal male, showing one fluorescent body

(iii) Cell obtained from 47 XYY male, showing two fluorescent bodies

V. Fluorescent Y bodies
the double Y karyotype could exist had already been established (Maclean et al., 1962) from a nuclear sexing survey, when a male with 48 XXY chromosome complement had been found. Recently it has been possible to test for the existence of the Y chromosome by employing special staining procedures which affect that chromosome alone (Pearson et al., 1970). By using this technique the presence of single fluorescent bodies has been demonstrated in interphase cells taken from normal 46 XY males, and of double fluorescent bodies in cells from 47 XXY males (see Illustration V).

Isolated studies of males with the karyotype 47 XXY began in 1961 with a preliminary report by Sandberg et al., who made the chance discovery of this karyotype in the father of a child with Down's syndrome whom they were examining. Jacobs (1965) was impressed by the fact that several 48 XXY individuals had been found in maximum security hospitals, and she suggested that the additional Y chromosome might be adversely influencing behaviour. Her own survey in a Scottish maximum security hospital showed that nine out of the 342 men examined had the 47 XXY sex chromosome complement.

The origin of sex chromosome aneuploidy

Sex chromosome aneuploidy is said to exist where there are an abnormal number of sex chromosomes. From the above discussion it may be seen that individuals with karyotypes 45 XO, 47 XXX, 47 XXY and 47 XYY are examples of sex chromosome aneuploidy, whereas patients with testicular feminisation, who have the karyotype 46 XY, are not.

Sex chromosome aneuploidy may be the result of faulty gamete formation in either of the parents during meiosis. This process involves the reduction of the number of chromosomes in the nucleus by half, so that each gamete receives one of a pair of chromosomes, but never both. This division is necessary so that the zygote, formed from the fusion of the parental gametes, shall contain the normal complement of 46 chromosomes.
On the very rare occasions that non-disjunction of the pair of sex chromosomes takes place during the process of meiosis in either parent zygotes having the karyotypes 45 XO, 47 XXY and 47 XXX may result, after fusion of the abnormal gamete with a normal one from the other parent (see Diagram I).

**Diagram I  Formation of abnormal zygotes, following parental non-disjunction**

<table>
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<tr>
<th>Parent</th>
<th>Gametes</th>
<th>Zygotes</th>
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<tbody>
<tr>
<td>46 XX</td>
<td>24 XX + 23 X or 23 Y</td>
<td>47 XXX or 47 XXY</td>
</tr>
<tr>
<td>46 XX</td>
<td>22 0 + 23 X or 23 Y</td>
<td>45 X0 or 45 Y0 (lethal)</td>
</tr>
<tr>
<td>46 XY</td>
<td>24 XX + 23 X</td>
<td>47 XXY</td>
</tr>
<tr>
<td>46 XY</td>
<td>22 0 + 23 X</td>
<td>45 X0</td>
</tr>
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</table>

The abnormal gametes may be formed from the first or second meiotic divisions, whereas the YY gamete can only result from an error at the second meiotic division in the father (see Diagram II).
Diagram II  
**Formation of the 47 XXY zygote, following paternal non-disjunction at the second meiotic division**

<table>
<thead>
<tr>
<th>Parent: 46 XY</th>
<th>Gametes: 23 X, 23 Y</th>
<th>Zygotes: 46 XX, 47 XXY, 45 XO</th>
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</thead>
<tbody>
<tr>
<td>23 X</td>
<td>+ 23 X</td>
<td>46 XX</td>
</tr>
<tr>
<td>23 X</td>
<td>+ 24 Y</td>
<td>47 XXY</td>
</tr>
<tr>
<td>22 X</td>
<td>+ 23 X</td>
<td>45 XO</td>
</tr>
</tbody>
</table>

A more complex type of sex chromosome aneuploidy is that of mosaicism. Mosaic individuals originate from an error in cell division in a normal zygote in the early stages of embryogenesis. This results in a situation where there are two or more populations of cells within the body (termed cell "lines"). Examples of mosaics with Turner's syndrome are discussed more fully in Chapter II.

**Clinical features of individuals with sex chromosome abnormalities**

So far the discovery of the four main types of sex chromosome aneuploidy has been discussed briefly. A more detailed description of the clinical features of these syndromes will be undertaken in order to demonstrate those similarities and differences which may be found to co-exist with an abnormality in sex chromosome number.

**The 47 XXY male (Klinefelter's syndrome)** (Overzier, 1963; Ferguson-Smith, 1966; Court Brown, 1969).

**(i) Clinical characteristics**

It is necessary to specify the abnormal karyotype in such
patients with Klinefelter's syndrome. At the time when nuclear sexing surveys were carried out it was recognised that chromatin negative as well as chromatin positive males existed, who showed no variation so far as the diagnosis of Klinefelter's syndrome was concerned. Since the only genuine sex chromosome abnormality is that which occurs in the sex chromatin positive male, it is possibly more accurate to refer to patients with this constitution as 47 XXY males.

Usually these males seem to develop normally until puberty. Outside mass surveys it is rare for a 47 XXY male to be determined prior to this time, unless as a result of mental retardation or, possibly, congenital malformations. It is not that there are any recognised congenital malformations associated with the karyotype, as there are, for example, in pre-pubertal patients with Turner's syndrome, but merely that chromosomal abnormalities in general are now well-known concomitants of congenital abnormalities.

In adults the only invariable clinical finding is the presence of very small testes associated with otherwise normal external genitalia. Spermatogenesis is absent, causing sterility. Other secondary sexual characteristics tend to appear abnormal, but these present a much more varied picture. The voice certainly becomes deeper in all cases, but may be high-pitched in comparison with 46 XY males. There may be a complete lack of facial hair growth, or it may be so diminished as barely to necessitate shaving. There may be a corresponding lack of chest and axillary hair, whilst pubic hair may show a female configuration. A number of patients also develop gynaecomastia (enlargement of breast tissue) which can be so extreme as to necessitate a removal operation. Patients tend to be taller than average, because they have longer legs in relation to trunk size than do normal males. There is also a tendency to obesity.

Court Brown (1967) showed that these features vary depending on the manner in which the 47 XXY male is ascertained. Males identified at sub-fertility clinics tend to be more masculine than those
referred to endocrinology clinics. Thus 47 XXY males who attend sub-fertility clinics are less likely to have gynaecomastia or female-distributed pubic hair, and will probably need to shave more often than those 47 XXY males attending endocrinology clinics.

(ii) Prevalence of the 47 XXY abnormality

As indicated in the previous paragraph, the mode of ascertainment plays a major role in the discussion of sex chromosome abnormalities. This fact is well illustrated when one considers the differing figures achieved in examining various sub-populations for the incidence and/or prevalence of the 47 XXY male.

(a) Newborn baby surveys: Maclean et al. (1964) surveyed 10,500 consecutive male births in hospitals in the Edinburgh area, using the nuclear sexing technique. Court Brown (1969), in reviewing these surveys, combined results from other Edinburgh studies to give a total of 13,257 live male births, and reported an incidence of .13% for 47 XXY males. Figures for perinatal mortality did not seem to be any higher than expected (Court Brown, 1969).

(b) Survey of school children: Maclean (unpublished data, reviewed in Court Brown, 1969) also conducted a nuclear sexing survey, of 10,659 school entrants. The prevalence of chromatin positive males in this survey was less than .09%. The discrepancy between this figure and that quoted for the newborn survey (.13%) is not statistically significant. It might, however, suggest a comparatively higher rate of infantile mortality or of admissions to special schools or institutions for mentally retarded children. A survey of new entrants to schools for the educationally subnormal did not support the latter hypothesis. It is interesting to note that more recently Ratcliffe et al. (1970) found a lower incidence (.09%), based on 3,500 male births.

(c) Surveys of mental subnormality hospitals: Prevalence figures from surveys of patients in mental subnormality hospitals tend to lie between .7% and 1.3%. As will be discussed in more detail in the
section dealing with Turner's syndrome, figures obtained from such data are suspect on account of difficulties of definition. However, it would seem evident that the prevalence of 47 XXY males in mental subnormality hospitals is about four to five times that of the incidence figure quoted for newborn surveys.

(d) Surveys of criminal institutions: Prevalence figures from studies involving surveys of criminal institutions are difficult to summarise or interpret. The criteria of admission vary from prison to maximum security hospital and also between countries. This indicates how important it has always been to specify closely the population on whom a cytogenetic survey is based. It is also well known that mental subnormality of a fairly high grade is often associated with behaviour disturbances, and the fact that there is a higher prevalence of 47 XXY males in criminal institutions may be a result of their mental subnormality alone. On the other hand, findings reported from California (Mosier et al., 1960), on 47 XXY males isolated in terms of normal IQ in a State hospital, discovered an association with sexual offences in this group.

(iii) Psychological characteristics

Psychological characteristics associated with the 47 XXY karyotype vary depending on the mode of ascertainment as do the physical features. Individuals with the cytogenetic abnormality have been found amongst psychiatric populations, in institutions for the mentally subnormal, prisons and maximum security hospitals, as well as in the general population.

After the discovery of sex chromatin it was soon noted that mental retardation was associated with males having positive sex chromatin. The first buccal smear surveys were reported in 1958 (Ferguson-Smith). Prevalence studies were confused by being based on poorly defined populations, about whom there were few details regarding the way in which IQ was measured, or, indeed, concerning the criteria used to classify the populations as "mentally retarded". Court Brown (1969), in gathering together details of IQ from surveys,
concluded that positive sex chromatin was associated with high grade mental deficiency (i.e. IQ of 50 +) but not with the lower grades (i.e. IQ of 50 -). It therefore appeared that the extra X chromosome was not an important factor in the aetiology of very severe mental subnormality.

Similar difficulties of definition arose in psychiatric hospital surveys from which it was hypothesised that positive sex chromatin was associated with, in particular, a higher prevalence of schizophrenia. Some of these surveys were based on schizophrenic patients only (e.g. Jagiello, 1961 - 5 chromatin positive amongst 530), whilst others included cases with other diagnoses (e.g. Nielsen, 1964 - 5 chromatin positive patients amongst 450). Moreover, as Hambert (1966) noted, the word "schizophrenia" was used in differing contexts in different countries. Hambert's study of 75 males noted a significantly higher prevalence in his psychiatric hospital series, maladjusted retarded males series, and institutionalised epileptics, than in newborn males. In his case histories he described a large number of behavioural problems - tendency to aggressive outbursts, shyness, nervousness in childhood, weak libido, deviant sexual behaviour, abuse of alcohol and anti-social behaviour - none of these traits being universal.

As has already been mentioned, there is an association between the chromatin positive state and high grade mental subnormality. MacLachlan (1969) concluded that the increased frequency of chromatin positive males in delinquency centres might be a function of their IQ, rather than a direct result of their extra X chromosome. Intelligence test (Wechsler scales) results obtained from 18 Klinefelter patients by the author have shown a significant difference in IQ's, significant at .01 level, between those patients ascertained for medical reasons (mean IQ = 98.6, S.D. = 9.0) and those referred from remand homes or institutions (mean IQ = 82.4, S.D. = 8.0). It should be noted that none of these patients was of subnormal IQ. Several of them
seemed to function better than might have been expected from their IQ results, a discrepancy which possibly reflects a low motivation level. For this reason tests of personality (e.g. the Cattell 16 FF questionnaire; Osgood semantic differential; and the Terman Miles Masculinity/Femininity inventory) were given only to selected co-operative patients, a procedure which failed both to produce sufficient numbers and to satisfy the basic principles of comparative research.

Nielsen et al. (1969) tested a group of hypogonadal patients (34 of whom were 47 XXY males, the remainder 46 XY males). They found a comparatively high level of intelligence in the group of 47 XXY males (mean = 102.8), but remarked upon attention defects and a lower verbal comprehension level demonstrated by this group in comparison with hypogonadal controls of normal karyotype.

The association between the extra X chromosome and lowered IQ remains to be proven by further investigation of 47 XXY males in the general population, using sibs and parents as controls. It might be postulated that their additional chromosome material causes them to function at a lowered IQ level in relation to their families - an IQ level not reduced, however, to the degree of mental subnormality. With reference to this point, which recurs in the following discussion of 47 XXY males, the author had the opportunity to test the five sibs, one a dizygotic twin, of a young Klinefelter patient (who was not mentally retarded), ascertained at a local remand home. Reference to the family tree given in Diagram III below will indicate that the 47 XXY male had an IQ of 16 points (i.e. > 1 S.D.) lower than that of his dizygous twin, and that all the other sibs had even higher IQ's (measured using Wechsler scales). The hazards associated with twin births could account for this difference, although there were no obvious organic signs on psychological testing.
Diagram III  Pedigree of family, giving IQ scores for propositus (47 XXY male) and sibs

The 47 XXY male  (Court Brown, 1968)

(i) Clinical characteristics

Reports on phenotype are varied, and no clear picture emerges as to the physical characteristics associated with the 47 XXY chromosome constitution. Whilst the first case was identified from karyotyping the father of a child with Down's syndrome, other cases reported earlier had been referred because of genital abnormalities. More recently a few individuals have been ascertained because of behavioural problems and height.

Increased stature, together with the behavioural trait of aggression, is most commonly, if erroneously, associated with the
47 XYY syndrome. In order to minimise the work-load involved in
karyotyping (rather than buccal smear screening) a population, there
has been a tendency for only those males over a certain height
(often 180 cms.) to be karyotyped. Although there is some indication
from family studies that the 47 XYY males, selected by their height,
are taller than their parents and/or siblings (Court Brown, 1968),
there are not enough data on 47 XYY males ascertained on a purely
random basis to support this association.

Other case studies, often concerned with one patient only, have
cited bone, skin, cardiac and neurological disorders, and dental
irregularities, to mention only a few.

(ii) Prevalence of the 47 XYY abnormality

Mass surveys for 47 XYY males in maximum security hospitals
originated from the observation by Jacobs (1965) that previous buccal
smear surveys (for chromatin positive males) in such institutions had
thrown up several individuals having 48 XXYY constitution. Since an
earlier survey of mentally subnormal males had observed a lower
prevalence of 48 XXYY males (Maclean et al., 1962), she suggested
that the additional Y chromosome in the 48 XXYY males could be
adversely influencing their behaviour, and, if this were so, the
47 XYY males, too, would be more common in maximum security hospitals.

A survey of Carstairs maximum security hospital appeared to support
this hypothesis (Jacobs et al., 1968), in that nine men out of 315
tested had the karyotype 47 XYY, a prevalence rate of 2.9%.

Replication studies carried out in three other maximum security
hospitals (Casey et al., 1966) confirmed the finding of Jacobs et al.
but it should be noted that Casey et al. selected for karyotyping
only those males of 183 cms. in height, and over. Lacking in these
studies was the statistic concerning the frequency of the 47 XYY
abnormality at birth. Court Brown (1968) argued that the frequency
of 47 XYY males at birth must certainly be lower than that of 47 XXY
males, since, whilst the error leading to a 47 XYY conception could
occur only at the second meiotic division in the male, that leading to a 47 XXY conception could arise from errors at first or second meiotic divisions in the female, or at the first meiotic division in the male. Clearly this reasoning has to be substantiated by newborn surveys, and in this connection it is interesting to note the findings of Ratcliffe et al. (1970) of five 47 XXY males and only three 47 XXY males amongst 3,500 male births. If this finding is replicated, it might be postulated that there are selection factors which cause the 47 XXY foetus to be at greater risk of aborting than the 47 XXY foetus.

Not only are data on a larger number of newborn males required, but also mass unbiased screening of adults, so that incidence and prevalence figures may be checked and compared at various stages of development.

(iii) Psychological characteristics

Observations on behavioural disorders and patterns of crime of the nine 47 XXY males found in the original survey of Carstairs maximum security hospital were compared with controls and reported by Price and Whatmore (1967). Amongst their observations it is interesting to note that they found the control patients to be more openly hostile, with more frequent violently aggressive outbursts, than the 47 XXY males. The latter adjusted well to the hospital environment, so much so that their behaviour merited their recent removal from maximum security surroundings, and there are now only two of the original nine remaining. (Of those discharged four are in mental subnormality hospitals and three in the general population). In their criminal behaviour they had displayed less violence against persons than had the control group. They had begun their criminal activities at a much earlier age, and often came from good families who had no criminal or unstable characteristics.

Other reports have produced evidence which conflicts with these findings; e.g. Griffiths et al. (1970), who could find no difference
in mean ages at first conviction. On the other hand, most surveys of this kind support the view that 47 XYY males are more prone to committing petty crimes against property than violent attacks against persons. It is frequently the single case studies which describe aggression in the form of sexual crimes or assault, and which tend to perpetuate the image of the aggressive 47 XYY male. There is, in fact, a noticeable lack of substantiating evidence either to support or refute the theory of the existence of an association between aggression and the 47 XYY syndrome. Instead there has gathered a considerable volume of single case studies, together with surveys on institutionalised groups in which a certain amount of hostility is a recognised feature. Ferguson-Smith (1971), in summarising the papers given on this topic at a symposium, concluded that "the association (of the 47 XYY karyotype) with criminality had been much over-emphasised in the past." The few psychological studies that exist are reviewed below.

Griffiths (1971) administered the Wechsler Adult Intelligence Scale, the Eysenck PEN Questionnaire, and the Foulds Caine Hostility and Direction of Hostility Questionnaire to 12 subjects with 47 XYY constitution who were detained in one London prison, and to 12 controls selected for age and similar length of time in prison. The 47 XYY males scored significantly lower on Full Scale IQ, Performance Scale IQ and Verbal Scale IQ. Sub-test scores which differentiated significantly between the two groups were Comprehension, Similarities, Digit Span and Vocabulary. The 47 XYY males also scored significantly lower on the Extraversion scale of the Eysenck PEN Questionnaire. It is interesting that no other significant differences were noted; in particular, none on the Hostility Questionnaire. The finding of low extraversion scores confirmed previous research carried out by Hope et al. (1967) on seven of the nine 47 XYY males (two refusals) found in the original survey at Carstairs maximum security hospital. Besides this difference the authors also noted that the 47 XYY males
were more defensive in their replies to questionnaires, and it was suggested that this response set might be adversely affecting their replies on all the questionnaires administered, to the detriment of an effective comparison being made between them and the control group. Hope et al. assessed IQ's in the patients, using Mill Hill Vocabulary Scales and Raven's Progressive Matrices, and concluded that the 47 XYY males reflected the distribution of IQ of all patients in Carstairs maximum security hospital.

McKerracher (1971) used the Wechsler Adult Intelligence Scale to assess the IQ's of

(i) A group of 20 47 XYY males
(ii) " " 12 47 XXY "

and

(iii) A random sample of 147 males, being those patients interviewed during the course of one year.

He found that the 47 XXY males were significantly less intelligent than the other two groups, but that there was no difference between the 47 XYY males and the control group. All three groups scored higher on Performance items than on Verbal ones, and there was a trend for the two genetically abnormal groups to have greater discrepancies in this direction than did the control group. On Eysenck's Personality Inventory the 47 XXY males gained higher lie scale scores than did the other two groups, a difference which McKerracher felt might be connected with their lowered IQ's.

It is appropriate at this point to mention an address given by Money (1971) in which three salient points were remarked upon. He suggested that 47 XXY males might not be aggressive so much as impulsive. He postulated that it is the triggering off of reactions, aggression amongst them, which is uncontrolled to a greater extent in 47 XXY males than in normal persons. It has certainly occurred to the author that the 47 XYY males whose records are known appeared to be accident-prone. One of them, employed as a builder's labourer, was killed in a fall from scaffolding, and another frequently requires
hospital admission for accidentally self-inflicted injury. Impulsivity could possibly be experimentally investigated, using the Porteous Maze test (Porteous, 1952), already much employed for assessing delinquent groups.

Money went on to mention the extent to which 47 XYY males appear to be loners. This observation is to a certain degree supported by the research of Hope et al. (1967) and Griffiths (1971), which suggested that 47 XYY males are more introverted. Finally Money commented on the defective attention span of which he found evidence so frequently in school reports. Again, in substantiation, it is interesting to note that in Griffith's research the Digit Span sub-test acted as a significant discriminator between the 47 XYY males and the controls, the former's lower scores possibly resulting from lack of persistence of attention.

The author has tested three 47 XYY males who were ascertained from a random survey of the general population, as well as eight others from criminal and mental subnormality institutions. Whilst there is nothing of note to remark upon in this latter group, beyond the fact that they are, of course, considerably lower in IQ than the general population 47 XYY males, there are three points that were of interest regarding the three ascertained in an unbiased way, which might be more fully investigated when a larger number of 47 XYY males has been similarly ascertained:

(i) IQ results were well within normal limits, but without exception the scores of all three on the Digit Symbol sub-test (often used as a test of psychomotor speed) were the lowest. It is unlikely that this finding has any relevance, since the performance sub-test scores, in which psychomotor speed plays a large part, were not depressed below the verbal test scores. This point will be discussed in further sections, dealing with Turner's syndrome.

(ii) One 47 XYY male demonstrated colour-blindness (protanopia), and it was possible to test his younger brother, both for
colour-blindness and intellectual ability. Whilst the 47 XXY male had an IQ result of 108 on the W.A.I.S., his younger brother achieved a rather higher IQ score of 124, and was not colour-blind. This type of comparative approach, using sibs as controls, has already been mentioned in connection with the 47 XXY males, and would seem to warrant further investigation, in order to examine any intellectual deficit which may have occurred.

(iii) All three 47 XXY males appeared to be exceptionally well-adjusted men; all had held stable employment since leaving school; two had satisfactorily served apprenticeships, and the occupation of the third did not require this. Two were married, and one of these had a child; the other was being investigated for sub-fertility. They related well in interviews and were entirely co-operative in completing all the psychological tests.

The 47 XXX female

(i) Clinical characteristics

The overall evidence on these females is that they are physically normally developed, that they menstruate normally, and are fertile (Court Brown, 1969). However, the first case of a triple X female was that reported by Jacobs et al. (1959), investigated because of secondary amenorrhoea, and there have been other cases reported ascertained after referral for primary amenorrhoea. In these patients there have been no other distinctive clinical features present.

This fact has meant that ascertainment of these females has been open to the usual biases. Buccal smear tests from them show two sex chromatin masses instead of the single one typical of the normal female cell. Most of the triple X females studied have therefore been ascertained from surveys of psychiatric and mental subnormality hospitals.
(ii) **Prevalence of the 47 XXX abnormality**

Hamerton (1971) quotes surveys of newborn female populations as giving an incidence of nine in 23,229 births, which gives an incidence figure of .65 per 1,000 live female births. This is a considerably lower figure than that given for the frequency of chromatin positive male births (2.13 per 1,000), but it is higher than that for chromatin negative females (i.e. individuals with Turner's syndrome), which Hamerton gives as .45 per 1,000.

(iii) **Psychological characteristics**

The prevalence of triple X females in psychiatric hospitals is given by Hamerton (1971) as 3.58 per 1,000, the majority of them being diagnosed as schizophrenic. Surveys of mental subnormality hospitals give a prevalence figure of 3.95 per 1,000. Both these results indicate that the extra X chromosome may pre-dispose the patient towards psychiatric illness or mental subnormality. Again family studies, together with random population surveys, would help to verify the association.

A psychiatric investigation of 22 triple X females (Kidd et al., 1963) classed 11 as mentally subnormal or severely subnormal. The remainder had normal IQ's but were psychotic. Prominent features were psychomotor retardation, poverty of speech and persecutory ideas. Hamerton commented on the similarity between this description and that of the 47 XXY males, but suggested that the illnesses of the latter group are more understandable as being a reaction to the feminisation of physical characteristics. It should be remembered that several other cases have been reported in which there is no evidence of psychiatric illness or mental subnormality.

It is clear that considerable research is required on randomly selected groups in order to form more definitive pictures of the syndromes common to each type of aneuploidy, before it is possible to begin to compare them (intra group). That this should be done is imperative, since it is only by this comparison that the genetic effects
of the extra genes may be determined. Finally it should be emphasised that the foregoing review is in no way intended to constitute an exhaustive survey of the physical and behavioural characteristics of the 47 XXY males, the 47 XYY males, or the 47 XXX females. Inclusion of single case reports was deemed impractical; reference to the works mentioned will provide further details. Earlier researches were often reported more than once, which militates against clear reviewing. There is a strong case for allotting each patient an identification number, as well as for stating clearly that patients have been reviewed or researched previously, where this is so.

This concludes the brief summary of the three main types of sex chromosome aneuploidy which have an extra sex chromosome. It should be noted that rarer forms exist, e.g. 48 XXY males, and females with 48 XXXX. It now remains to describe the fourth main group of individuals with sex chromosome aneuploidy - those females demonstrating Turner's syndrome, in which one sex chromosome is wholly or partially missing. This will be done in the next Chapter, before the data obtained during the study undertaken for this thesis is presented.
So far the phenotypic effects of the sex chromosome trisomies have been introduced and discussed. It is generally accepted that the only viable type of sex chromosome aneuploidy in which a chromosome is missing is that of 45 XO (i.e. 45 chromosomes with one X chromosome absent), and that the condition of 45 YO has never been detected and is assumed to be lethal. As already mentioned, the description of the syndrome associated with the 45 XO abnormality is ascribed to H.H. Turner. Prior to his article in 1938 there had been several reports of females with infantile external genitalia who lacked normal ovarian function (in that they had amenorrhoea and were infertile). Perusal of the literature reveals great confusion in the terminology employed to distinguish the various entities. General terms were invented and abandoned in favour of more specific ones. For example, "ovarian agenesis" was dismissed by Rössle and Wallart (1930), who proved that, although the ovarian follicles were deficient, there was clear evidence of some ovarian tissue in the form of streaks. These writers put forward the term, "sexogenic dwarfism".

A complete discussion of the bewildering range of nomenclature and history of the syndrome under study is deemed to be outside the purpose of this thesis. Moreover, it is essential that the group of individuals under study should be seen to be a clinical entity by psychologists and medical practitioners alike. For this reason it is necessary to begin by describing the clinical aspects of those individuals, and the ways in which these aspects differentiate the subjects as exhibiting a syndrome which is separate from other associated entities.

Clinical Aspects of the Syndrome

An attempt will be made to define the clinical features of individuals with Turner's syndrome by reference to Diagram IV.
Diagram IV  Classification of gonadal dysgenesis, demonstrating distinction between Turner's syndrome and other syndromes (adapted from Hauser, 1963).

The term "gonadal dysgenesis" was suggested in 1955 by Grumbach et al. to describe patients with a condition characterised by the absence of ovarian germ cells, indicative of undifferentiated gonads. Other terms employed are gonadal agenesis or aplasia. External and internal genital organs were usually female in appearance, but remained infantile throughout development. The gonads were present as remains of connective tissue, lacking follicles and ova. It should be noted that the term "gonadal dysgenesis" may also be applied to males; for example, Stewart (1961) distinguishes between "cortical gonadal dysgenesis" as it occurs in females, and "medullary gonadal dysgenesis" as it occurs in males with Klinefelter's syndrome.

The condition of gonadal dysgenesis occurring in a patient of female phenotype may be sub-classified under three headings:

(i) Pure gonadal dysgenesis (Swyder's syndrome)

This condition occurs in females of normal appearance and stature, and is characterised only by the absence of germ cells, resulting in primary amenorrhoea. Cytogenetically they appear to possess very varying karyotypes (Brøgger, 1969). He listed karyotypes 46 XX,
VI. Clinical picture of leg and foot of female baby with Turner's syndrome showing pitting oedema

VII. Clinical picture of hands of female with Turner's syndrome showing bilateral shortening of fourth and fifth metacarpals
VIII. Clinical pictures of female child with Turner's syndrome, showing neck webbing and low hairline
46 XY and mosaics 45 X0/46 XX, 45 X0/46 XY as all having been found at some time associated with the same phenotype (see following section on numerical and structural chromosome abnormalities, pp. 28, 29).

(ii) Gonadal dysgenesis with decreased stature

This condition occurs in females with small stature and primary amenorrhoea. A differential diagnosis between various types of dwarfism and gonadal dysgenesis must be made in these cases, and can usually be determined by hormonal assay methods.

(iii) Gonadal dysgenesis with abnormalities

This condition is frequently associated with the Bonnevie-Ullrich syndrome, which comprises neck webbing, small stature, retarded mental development, skeletal deformities, as well as many other variable signs. A number of these may be seen to resemble closely those listed for Turner's syndrome, but individuals in this category have a normal female karyotype.

(iv) Gonadal dysgenesis with decreased stature and abnormalities

This condition is commonly termed Turner's syndrome. The phrase "decreased stature" as used in this context does not denote a type of dwarfism; there is a generalised and equal retardation of development, caused by inadequate growth of the whole body. Other abnormalities cover a wide range of anomalies, some of which will be described in terms of the region they affect (see Illustrations II, VI, VII and VIII).

**Face**
Prominent ears and small jaw are common features.

**Neck**
The hairline is often low, and the effect may be accentuated by shortness of the neck. Neck webbing (in which a distinct fold of skin extends from below the ears to the shoulder blades) may be present.

**Chest**
This tends to be broad and shield-shaped, and the nipples widely spaced. The latter are often inverted and non-pigmented. There tends to be very little breast development until oestrogen therapy is introduced.
Cardiovascular System The most frequent anomaly in this system is coarctation of the aorta, a condition in which part of the main blood vessel is abnormally narrow. Cardiovascular malformations in general are probably a frequent cause of shortening of life span.

Extremities There may be oedema of ankles and feet and possibly hands in the adult, probably due to lymphoedema (a malformation of the lymphatic system). The fingers of the hand tend to be short; if a fist is made the knuckles of the fourth and/or fifth metacarpal(s) are practically non-existent. The elbow may be deformed, preventing the arm from being held straight (cubitus valgus).

Skin Pigmented spots (or naevi) frequently occur in greater numbers than is considered normal. It is interesting to note that Lindsten and Fraccaro (1969) commented that such naevi are not commonly present in infants.

In the following dissertation the term Turner's syndrome will be used (irrespective of its original definition) in place of indiscriminate labels. It is recognised that the syndrome under discussion is not identical with that originally described by Turner in 1938. However, as a result of the progression of knowledge (which has shown in particular the association of the karyotype 45 X0 with the syndrome) there is now a well-defined entity associated with the syndrome which incorporates those features which Turner defined.

Prevalence of the 45 X0 abnormality

Court Brown et al. (1964) estimated the incidence of chromatin negative females to be 0.37 per 1,000 live female births, a figure which is considerably lower than that for the other types of sex chromosome aneuploidy, and certainly much lower than would be expected on theoretical grounds.

Studies on chromosome abnormalities detected in abortuses indicated that the 45 X0 group contributed 20% of all chromosomally
abnormal foetuses, or about 5% of all spontaneous abortions (Hamerton, 1971). Carr (1965) suggested that only one in 40 zygotes with the 45 XO constitution survive to term (i.e. fewer than 3%). Although at fertilisation the 45 XO anomaly is more common than the 47 XXY, the latter is almost five times more common in newborn infants. It is interesting to speculate what makes possible the survival of the 3%, when so many are so obviously lethally affected by their chromosome abnormality. It would seem imperative to compare physiological features of the mothers of aborted 45 XO foetuses with those of the mothers of the 45 XO individuals who survive.

From the discussion on mortality (see p. 32) it will be clear that a prevalence figure for patients with Turner's syndrome in the general population may well be even lower than the frequency at birth. On the other hand, the studies quoted above, based on buccal smear surveys, disregarded mosaicism and other forms of X chromosome abnormality which are not detected by this method. A reliable estimate of prevalence is therefore difficult to achieve, and a similar problem arises in considering whether or not mental subnormality occurs concurrently with this type of abnormality. This is discussed in detail in Chapter III.

Survey of cases of Turner's syndrome recorded within the Registry of Abnormal Karyotypes, Edinburgh

Individuals with Turner's syndrome are so rare that it was considered advisable to survey the general characteristics of as large a group of patients as possible before concentrating on a smaller group for the purpose of full-scale psychological investigation. The Medical Research Council's Registry of Abnormal Karyotypes was initiated by the late Professor Court Brown in 1959. Its purpose
was to facilitate the study of the pattern of morbidity and mortality in persons with different forms of chromosome abnormality. In the early days of chromosome analysis the Unit was amongst the first to provide the diagnostic aid of karyotyping those individuals whose phenotypes gave medical practitioners cause to question their genotype. However, as a result of the advance in knowledge concerning the techniques involved in the culturing and karyotyping of chromosomes, all major medical centres are now able to provide their own services. For this reason there have tended to be fewer referrals to the Edinburgh Unit during recent years, a fact which limits the effectiveness of obtaining therefrom a representative sample for the purpose of research.

Once the karyotype of such a referral is established as abnormal a proforma is completed, either by the clinician concerned in the case or by a clinician from the Unit. It covers reason for referral, clinical signs and symptoms, measurements of height, weight, hormonal assays, etc. as well as social details on marital status, occupation and rank in sibship. The data gathered in this way vary between the different sources, and methodical surveys are often hampered by lack of information and by imprecise details. The data on each patient are up-dated annually by contact with the general practitioners, or occasionally the consultants, in charge of the cases. They fill in a short questionnaire, involving items of change of address, mortality, illnesses, and referrals to hospital during the past year.

From this data background a total of 128 records were selected of patients who demonstrated Turner’s syndrome, the majority of whom (N = 76) had the karyotype 45 XO. To account for the residual cases it is necessary to digress and to discuss other forms of X chromosome abnormality which give rise to the phenotype having the common characteristics of Turner’s syndrome.

Sex chromosome abnormalities may be sub-divided into numerical and structural types. It is the numerical abnormality characterised
by the karyotype 45 XO which accounts for most cases of Turner's syndrome. Other numerical abnormalities are those represented by the term mosaicism. Structural abnormalities of the X chromosome occur in cells with a seemingly normal complement of 46 chromosomes where one of the X chromosomes is abnormal in its structure. As the numerical abnormalities may include chromosomes with structural defects, the latter will be described first.

Structural defects - There are certain structural abnormalities of the X chromosome which are found to give rise to the same clinical picture as that of the pure 45 XO karyotype. Thus an individual may have a seemingly normal complement of 46 chromosomes, but still manifest the common features of Turner's syndrome.

Isochromosome X (denoted Xqi)
This is a large abnormal chromosome which resembles some of the larger autosomes in size and shape. It is generally assumed that this chromosome is made up of two sets of long arm material from an X chromosome, the short arms being absent. Individuals with the karyotype 46 XXqi are extremely rare, but, generally speaking, it seems that their phenotype resembles that of females with the 45 XO chromosome complement.

Ring X chromosome (denoted Xr)
This chromosome is presumed to be formed by deletion of parts of both the short and long arms of the X chromosome, followed by fusion of the ends. It seems that the Xr chromosome is unstable, since it is unknown for it to exist on its own, being found only in the 45 XO/46 XXr mosaic karyotype. Individuals possessing this demonstrate the clinical features of females with the 45 XO chromosome complement.

Numerical defects (Mosaics) - Whilst the largest group of cases of Turner's syndrome is composed of the 45 XO individuals, it is relevant to list those mosaics who demonstrate the phenotypic features of the syndrome, and who will be included in this study.
45 X0/46 XX mosaicism

This is the most common mosaic variant, and has two cell populations, one 45 X0 and one normal 46 XX. Moore (1966) suggested it was not usual for both populations to occur throughout all the tissues of the body, and consequently it is important that samples be taken from different tissues to ascertain a correct karyotype. The sex chromatin pattern may be positive or negative, and may also differ between various tissues.

Phenotypically these mosaics are less severely affected than the 45 X0 individuals. They tend to demonstrate fewer of the somatic anomalies, and some may menstruate spontaneously and/or grow to normal height. Their phenotypic description is very variable, however, as may be seen, for example, by referring to Leao et al. (1966), who reported on nine girls with the 45 X0/46 XX karyotype, who all had short stature as the presenting complaint. None had neck webbing or lymphoedema. It is generally assumed that these differences in clinical picture may be related to the time of zygote development (embryogenesis) when the X chromosome was lost, and to the distribution of the normal and abnormal cells.

45 X0/46 XY mosaicism

There seems to be no typical clinical picture associated with this karyotype. It has been described in phenotypic females (Jacobs et al., 1961), in phenotypic males (de la Chapelle and Hortling, 1963) and in inter-sexes (Yunis 1965). The XY population of cells tends to produce some masculine development, but this is not always the case. Jacobs et al. (1961) stated that the condition is rarely associated with Turner's syndrome.

Other mosaics represented in the sample studied are 45 X0/46 XXq and 45 X0/46 XXr.

Review of literature on comparison of karyotypes

Since it may be argued that individuals with mosaic or structural karyotypes possess more X chromosome material than those with the karyotype 45 X0, it is tempting to conclude that they will demonstrate
fewer of the symptoms associated with the syndrome. For example, Miroshina and Grumbach (1968) claimed that lymphoedema, neck webbing and coarctation of the aorta are three symptoms which appear less regularly than in individuals with the 45 XO Turner's syndrome. Indeed, Ferguson-Smith (1965) noted that some patients with the mosaic karyotype 45 XO/46 XX had heights within normal ranges, and normal menstruation also occurred. He noted that neck webbing and lymphoedema at birth were infrequent. It seems, therefore, as though the manifestation of Turner phenotype is modified by the normal 46 XX line, or, indeed, by any additional X chromosome material.

Of relevance to this discussion is the 45 XO/46 XY mosaic, in which, again, associated somatic defects seem to be fewer (Miroshina and Grumbach, 1968). There was no evidence of lymphoedema or coarctation of the aorta, but there was extreme variation in external genital development, which might result in individuals with this karyotype being reared either as male or female.

It is from these and other considerations, all of which seemed to be in general agreement, that Ferguson-Smith (1965) and Miroshina and Grumbach (1968) postulated a continuum ranging between normal male phenotype and normal female phenotype, with Turner's syndrome occurring at an intermediate point. Mosaics and structural abnormalities vary in phenotypic expression between the mid-point and either of the two poles (see Diagram V).

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**Diagram V**  
Postulated continuum involving the relationship between normal and abnormal karyotypes  
(Adapted from Ferguson-Smith, 1965)

<table>
<thead>
<tr>
<th>Normal Female</th>
<th>Turner's Syndrome</th>
<th>Normal Male</th>
</tr>
</thead>
<tbody>
<tr>
<td>45 XO/46 XX</td>
<td>46 XX</td>
<td>45 XO</td>
</tr>
<tr>
<td></td>
<td>or</td>
<td>45 XO/46 XY</td>
</tr>
<tr>
<td></td>
<td></td>
<td>46 XXqi etc.</td>
</tr>
</tbody>
</table>
This discussion is of particular interest when the concept of the phenotypic continuum is applied to intelligence. Lindsten (1963) reported that patients of 45 XO/46 XX and 46 XXq1 karyotypes generally had intelligence quotients of 100 or more, a finding which was not supported by Goldberg et al. (1968) or Leão et al. (1966). This clearly merits further investigation (see Chapter III).

Details of survey
Method of Selection

As is clear from the foregoing discussion, the karyotypes described do not all give rise to the Turner's syndrome phenotype. In selecting cases for this survey those patients who showed no evidence of amenorrhoea, or who were over 152 cms. in height, were excluded. Thus, of those patients who had been allotted the karyotype 45 XO/46 XX only three out of seven were involved in the survey.

Survey Method

A Cope chat card (Form 20) was filled out with the following information taken from the case files: details of identification data; karyotype; present age, if alive, or age at death; marital state; occupation; age at ascertainment and reason for referral; details of physical symptoms; height and weight; type of treatment and responsiveness; IQ rating (subjective); and details of any mental illness. A note was also made on whether or not the patient was available for research - a number (N = 47; 37%) were not accessible on account of death, emigration, un-notified change of address or unwillingness to partake in further research.

Survey Results

Relevant data given in terms of the above headings is considered below:

1. Karyotype

Of the total of 128 patients surveyed 76 (59.4%) were 45 XO. The remainder were made up of the numerical and structural types of X chromosome abnormality already listed:
Graph I. Age Distribution

Graph II. Age Distribution at Death
2. Age

Information on 128 individuals indicated that age ranged from birth to 67 years. The mean age of the total group was 25.86 years (S.D. 11.49). It is interesting to note that the oldest patient was a mosaic 45 X0/46 XY, whilst the oldest 45 X0 patient was 49. In the interests of accuracy it was necessary to eliminate those individuals (N = 15) on whom there existed no recent follow-up information because of emigration, loss of contact with G.P. etc. Graph I shows the distribution of ages of patients who were known to be alive at the 1971 follow-up.

The distribution of ages at death (Graph II) is not considered typical, in that not a sufficient number of individuals had been followed through their life-span. The relatively large number of deaths (8 out of 13) occurring before the age of 20 is a typical finding and will be discussed further in the following section.

3. Mortality

From the figures quoted in the previous section it appears that there is an uneven distribution of age at death, skewed towards the lower age groups. Two reasons may be advanced for this, one - an artefact - being that the records are more heavily weighted with younger cases; the other is a tendency for the patients to succumb to the congenital malformations of heart and gut, particularly during the neo-natal period. All those patients who died before the age of 20 had cardiac conditions or disorders of kidney and bowels which proved lethal. Causes of death in the older age group (> 21 years) were more varied. One, a mosaic, died from carcinoma of the ovary, a fact which suggests the advisability of removing the
organs made useless by the cytogenetic abnormality present, on account of a tendency for them to become malignant. Another patient died from a cerebral embolism, and a third committed suicide.

The increased mortality rate skewed towards the lower age group affects the frequency figure quoted for the prevalence of the abnormality (pp. 25, 26). It also affects the number of adult individuals who have to be surveyed in mental deficiency institutions before it may be stated categorically that the cytogenetic abnormality is not associated with mental retardation.

4. Marital Status

Information on the marital status of those patients who were of marriageable age (16+) was noted for 98 cases. Of these only 23 (23.5%) were married. The data are not particularly reliable, in that in most cases the up-dating of records is dependent on the G.P. concerned with the patient informing the Registry at the annual follow-up of the patient's marriage. More important from the psychological aspect is the number of those marriages which might be considered to be "successful". Of the three local married patients who were known personally to the Unit, one had been separated from her husband for some years, after a marriage lasting two years, and another's infertility had resulted in marital disharmony. In a fourth case it is recorded that the husband deserted his wife for another woman, and the patient subsequently committed suicide. This kind of information is not usually available from the G.P., and its elicitation depends more on mutual trust being established personally with the patients.

Of the 60 members of a control group used in the study of the psychological aspects of individuals with Turner's syndrome (see p. only 11 (18.3%) were unmarried. It may be concluded that the individuals with Turner's syndrome are less likely to marry \( \chi^2 = 48.49; p < .001 \), largely on account of their social withdrawal, arising from self-consciousness, lack of sexual attractiveness, and infertility. If it were possible in a long-term study to establish
more personal contact with patients this would seem an important area to study, so that pointers might be obtained towards therapeutic counselling, both before and after marriage. For example, it seems possible for individuals with Turner's syndrome to adapt to their infertility by adopting children. Of the four couples who were known to have done this, three were not available for further research. In itself this wish to sever connections with the Unit may be regarded as a further measure of acceptance of, and adjustment to, their condition.

5. Occupation

Out of 79 patients for whom occupations were noted, 26 (33%) had been employed in skilled work which required some further training after leaving school. Examples of posts in which members of this group were employed included nursing, typing and teaching; one patient was a university student. The remaining 53 (67%) were in unskilled posts, e.g. working in factories or as shop assistants. Closer scrutiny of occupational status was precluded by lack of updated information on the patients, as well as by loosely-defined work-descriptions, e.g. tracer, cashier.

Comparison with control data was impracticable since the majority of the control group married as soon as they left school and had never taken up employment.

6. Age at Ascertainment

From 120 records of patients in which age of ascertainment was available it was seen that 20 (16.7%) were ascertained between birth and 10 years; 64 (53.3%) between the ages of 11 and 20 years; 23 (19.2%) between the ages of 21 and 30 years; 7 (5.8%) between the ages of 31 and 40 years; 5 (4.2%) between the ages of 41 and 50 years; and one at the age of 56 years.

These figures indicated that the peak age of ascertainment was in the pre- and post-pubertal age range, 11 - 20 years.

7. Mode of Ascertainment

Information on this was available for 115 cases. Nineteen (16.5%) were referred on medical grounds other than those normally
associated with the syndrome. Such reasons included injury in
a road accident; complaints of obesity and tiredness; cardiac
abnormality found during routine medical examination; investigation
of chronic cough. Sixty-four (55.7%) patients were referred for
investigation of primary amenorrhoea or menstrual disorders. This
latter category was difficult to define, particularly as in some
cases the notes say 'irregular periods'. One girl had had only one
period, which occurred at the age of 20; another had 'infrequent
and painful' ones. It is interesting to note that five of the
seven cases so categorised were either mosaics (four) or structural
X chromosome abnormalities (one), which might suggest that sufficient
of the second X chromosome material was present to permit of a degree
of normal ovarian functioning. One patient referred for investigation
of primary amenorrhoea had undergone separate operations for
neck webbing and oedema before the diagnosis of Turner's syndrome was
given on the grounds of primary amenorrhoea.

Twenty-one patients (18.3%) were ascertained on the grounds of
failure to grow or to thrive at birth. It seems strange that 17
of these were not referred on these grounds until after the age of
ten. Diminished stature is certainly to be noted earlier than at
this age (see Graph IV); growth seems to be at the same proportional
rate, and there is no evidence that the child with Turner's syndrome
is ever the same height as her peers.

The remaining 11 patients (9.6%) were found from surveys. These
consisted of newborn surveys, both buccal smear and chromosome
types, and surveys of mental subnormality hospitals.

If the age at, and mode of, ascertainment are considered together
the interaction between them emerges as would be expected, i.e.
younger patients were ascertained from birth surveys and referrals
for retardation of growth, and the adult group were ascertained
from referrals to endocrinological and gynaecological clinics for
investigation of primary amenorrhoea. Patients with Turner's
syndrome seem far more aware of, and upset by, their short stature
(a finding which is endorsed by Sabbath et al. 1961) than by their
lack of menstruation and secondary sexual characteristics. What is striking is the length of time which frequently elapses before patients and their families seek help. This has already been noted with respect to retardation of growth. So far as primary amenorrhoea is concerned, 22 patients, or nearly 20% of the total sample, were not investigated until after the age of 21. Female patients in general, and patients with Turner's syndrome in particular, seem hesitant about approaching the medical services with complaints of a gynaecological nature. Moreover, it is usually at the age of 20+ that marriage is contemplated, and the absence of secondary sexual characteristics in individuals with Turner's syndrome becomes more meaningful. At this time realisation of their infertility can be very traumatic, and it is therefore very necessary that these patients should be ascertained as early as possible and appropriate treatment and advice given.

8. Physical characteristics

In this section only the physical characteristics of height, weight and those abnormalities which could affect performance on the psychological tests applied will be considered. It is not suggested that this list is exhaustive. Abnormalities affecting vision and hearing are, respectively, strabismus (squint) and otitis media.

Whilst no specific queries were made on the proforma about strabismus, eight patients were specifically noted as having had the condition at some stage of development. This tendency to strabismus may be a result of the retardation in growth affecting the development of the eyeball. Newton (personal communication) has measured the refractive power of eyes in patients with Turner's syndrome and found a general tendency to hypermetropia (long sight). This error in refraction is likely to be due to the eyeball being smaller than normal, and is known to be associated with an increased incidence of strabismus. This finding has to be borne in mind when considering the results obtained from visual perception testing.
Graph III. Age/Weight Chart of measurements from 59 patients
Graph IV. Age/Height Chart of measurements from 59 patients
So far as otitis media is concerned, nine patients were specifically noted as having the condition - an infection of the middle ear, which may lead to impairment of auditory acuity. Personal experience showed that direct inquiry revealed a much greater incidence than had been indicated in the proformas, which in itself suggests the importance of precision in formulating questions concerned with abnormalities.

With reference to height and weight, details of these were noted for 101 cases, for height alone for 12 cases, and for weight alone, two cases. Unfortunately it was not always clear from the notes at what age these measurements had been made, which makes suspect any attempt at comparative studies using standard norms for the general female population. Where the age was not supplied it was taken as that at ascertainment, since it is at that time that the maximum information on the patient is gathered and a physical examination conducted, which is likely to include measurement of height and weight.

Stunted growth being one of the invariable symptoms of Turner's syndrome, it is interesting to consider whether the abnormality in growth is present at birth and continues until the patient reaches skeletal maturity, or whether it is particularly involved at one stage in development. Graph IV shows the heights and ages of 59 patients with Turner's syndrome between birth and 19 years, plotted on a height standard chart (Tanner et al., 1966). The impression gained from this is that patients with Turner's syndrome are always shorter than their peers.

It seems that there may be an increased pre-disposition towards obesity in patients with Turner's syndrome, although objective impressions are confounded by lack of comparative standards. In the case records there is some mention of obesity as being one of the clinical features noted; one girl was actually ascertained from referral to an endocrinological clinic on account of her excessive weight. It should be mentioned that the annual follow-up procedure
through the G.P.'s is particularly useful in this connection, since the indication is that, once established, obesity is a continuing problem.

Polani (1961) stated that birth weight of individuals with Turner's syndrome is often very low, and it may be seen from Graph III that the few such individuals shown in the Registry records as having been weighed at birth were lighter than the average. It is impossible to use this graph for comparative purposes because of the inter-relationship between weight and height. It is, however, worth noting that, although only three out of 59 patients were within normal limits (lower third percentile) for height (see Graph IV), the statistically significantly greater number of 24 out of 59 were within normal limits for weight \( x^2 = 49.86, \ p < .001 \) and three were actually above the mean. From this it may be inferred that several, although by no means all, of the patients were overweight at the time of measurement.

It will be observed that the Tanner Growth and Development records contain standards up to the age of 19 only. It was, however, possible to compare the adult population of patients with Turner's syndrome with a control group. This, provided by Dr. J.S. Milne, was composed of 119 female blood donors between the ages of 18 and 49. It is recognised that such a group does not adequately control the stature variable, nor is it necessarily a representative sample of adult females, on account of the social and medical characteristics of volunteer groups such as blood donors.

However, using these data it was possible to regress weight upon height for the two groups and to obtain constants for the lines of regression resulting. An arbitrary height of 150 cms. was then adopted for both groups and the mean individual weights calculated for each group equivalent to this hypothetical height, using the formula (Benn, 1971):

\[
W_s = \bar{W} + b (H - \bar{H})
\]

where

- \( \bar{W} \) is the mean weight of the group
- \( H \) " " adopted height (150 cms.)
- \( \bar{H} \) " " mean height

with \( b \) the regression coefficient.
In this way the standard weights of 53.06 Kgms. for the patients with Turner's syndrome and 54.65 Kgms. for the control group were obtained - two figures which are not sufficiently different from each other to support the hypothesis that patients with Turner's syndrome are more pre-disposed towards obesity. For this type of predictive statistical analysis large numbers are necessary, and it is suggested that this line of research should be pursued, using measurements from a larger group of individuals with Turner's syndrome, and a more representative control group.

Another possible statistical approach is to use the "bulk index" which does not require the utilisation of a standard. This method involves the calculation of the exponential n in the expression
\[
\frac{W}{H^n}
\]
where W = weight, and H = height (Benn, 1971).

From this formula it is apparent that if log W is regressed on log H the coefficient of regression is the exponent n, and, hence, the standard error of the coefficient is the standard error of n.

From unpublished data for 900 members of the Edinburgh general population Milne (personal communication) has found that n = 1 for females and n = 2 for males, a result which replicates other studies in this field.

Calculation of the index for the group of blood donors referred to above gave n = 1.19 (S.E. = 0.32), and for the patients with Turner's syndrome n = 1.83 (S.E. = 0.64). This indicates that, whilst the blood donors are fairly representative of female subjects, in that they fit the index \[
\frac{W}{H^n}
\]
patients with Turner's syndrome more closely approximate the index \[
\frac{W}{H^2}
\].

The large standard errors of these exponents preclude any definite conclusions being arrived at, and reinforce the need to investigate larger numbers (particularly of individuals with Turner's syndrome).
It would seem that all that may be concluded from these two calculations is that the individuals with Turner's syndrome were "over-weight", in that their bulk index was higher than expected; but this does not necessarily indicate that their body tissues are composed of excessive fats; it is possible that they may be made up of a greater proportion of muscular tissue or bone.

9. Treatment

It seems common medical practice to prescribe oestrogens in the treatment of patients with Turner's syndrome. Whilst having no effect on the underlying cause of the syndrome, they do promote secondary sexual characteristics, i.e. some breast development, but with no nipple changes; axillary and pubic hair growth; and artificial menses, produced by the withdrawal of the oestrogens for a few days each month. Cyclical oestrogens in the form of the Pill are frequently prescribed.

Reference was made to treatment with oestrogens in 48 cases. Of these 27 contained some statement about the physical effects of such treatment, most of them mentioning slight to moderate breast development and withdrawal bleeding. The lack of standardised instructions on the proforma resulted in a paucity of detail, and in the case of 21 patients no results at all were given. It is quite usual for oestrogen therapy to be discontinued after a few years, often at the request of the patient, since the physical changes created are no longer dependent on continuing therapy, and little seems to be gained from the apparently normal menses. It has already been mentioned that patients with Turner's syndrome appear more upset by their lack of normal stature than by their failure to menstruate.

10. Intellectual status

If the section of the proforma concerned with intellectual status was completed, subjective descriptions of 'Average', 'Normal' or 'Average working-class' were frequently given. It might be
suggested that the halo effect operated here, as the clinicians concerned are accustomed to interviewing individuals with other types of chromosome abnormality in which mental subnormality plays a large part, and with these, individuals with Turner's syndrome would compare very favourably.

This type of record was available for 86 cases. Of these, nine (10.5%) patients were assessed as being mentally subnormal. It is of interest to note that of this group five had the karyotype 45 X0/46 X. This represents nearly half the group of this type of mosaic included in the sample and might indicate that such individuals are pre-disposed towards mental retardation. For the three patients with the 45 X0 karyotype falling into this category no formal IQ test results were available, and it would be unwise to draw any firm conclusions from the existing evidence. Altogether 25 (30%) were described as 'below average', a number which is rather larger than might be expected. This would appear to indicate that the clinicians were able to pick up the low average IQ pattern to be discussed further in Chapter III. It was not practicable to correlate subjective ratings with IQ's obtained from testing by the author because numbers were limited by the fact that subjective impressions were not given if numerical estimates of IQ's were available.

11. Psychiatric illness

Some form of illness was mentioned in connection with eight cases. Two patients had been treated by their G.P. for anxiety states, another for depression. One patient had attempted to commit suicide and was being treated for recurrent bouts of depression; another had committed suicide. A patient admitted during this study was referred for psychiatric assessment and was diagnosed as having "personality disorders with depressive illness". Finally, two patients were reported as having anorexia nervosa. The aetiology of such illnesses was not always suggested, although depressive illnesses seemed to be associated with feelings about infertility.
The proportion of the survey sample (6%) who are known to have had some form of psychiatric disturbance seemed to be smaller than that of the general population - 14% in one year (Shepherd et al., 1964). It is presumed that an increased proportion might have been detected if specific inquiry into mental state had been made.

In conclusion, the survey described demonstrates the difficulty of obtaining consistent data unless a very detailed proforma is employed, and completed in a standard manner. Nevertheless, several points of psychological relevance have emerged, among which may be mentioned those concerned with obesity, marital status and counselling, and treatment.

In the first two Chapters of this thesis the four main types of sex chromosome aneuploidy have been introduced. A distinction has been drawn between the class of aneuploidy involving extra genetic material (i.e. 47 XXY, 47 XYY and 47 XXX), and that involving a lack of such material (e.g. 45 XO).

The remainder of the thesis is concerned with the last-mentioned class, and, in particular, with describing the data obtained from psychological assessment of a group of 24 adult females who demonstrated the physical features of Turner's syndrome, as well as a lack of X chromosome material. A survey of case records from a larger number of such individuals has provided a background of social and physical details against which the group under study may be examined.
CHAPTER III

INTELLECTUAL ABILITY

INTRODUCTION

Incidence of mental retardation

(i) Population Surveys

When Turner's syndrome was first documented as a cytogenetic abnormality, interest focused on the incidence of mental retardation in the individuals concerned. As already discussed, reports on other patients with sex chromosome aneuploidy frequently noted a higher incidence of mental subnormality. With specific reference to the question of mental retardation in Turner's syndrome may be considered the survey carried out by MacLean et al. (1962) on 4,514 patients in mental subnormality hospitals. Only one female with Turner's syndrome was found, giving rise to an incidence of .4 per 1,000 females. This frequency may be compared with the figure quoted by MacLean et al. (1964) of .3 per 1,000 females with Turner's syndrome found in a newborn population. There being no significant difference between these figures Court Brown (1969) concluded that the cytogenetic abnormalities present in Turner's syndrome did not predispose the individuals concerned to mental subnormality. However, it should be noted that there is evidence of a high infant mortality rate for these females, which might suggest a possible increased frequency in hospitals for the mentally subnormal.

Conversely, Bekker and van Gemund (1968) cited the surveys reviewed by Harms (1967). From five population surveys covering 15,938 female neonates he reported an incidence of .4 per 1,000 of 45 XO Turner's syndrome individuals in the general population. This figure he compared with as high an incidence as 1.2 per 1,000 found in three surveys of females in institutions for the mentally retarded, and in E.S.N. schools.
The most recent tabulation of eight general population studies (Moor, 1969) indicated an incidence of 11 chromatin negative subjects in a population of 28,241 females. (As no further information was specified it may be inferred that these 11 subjects had 45 XO karyotypes). This gave a figure of .53 chromatin negative individuals per 1,000 females. Moor compared this figure with that of 1.04 per 1,000 females, taken from five studies of 5,748 mentally retarded females. It should be noted that specific reference to chromatin negative individuals excludes from the surveys those chromatin positive patients who would have been diagnosed as phenotypic examples of Turner's syndrome. Since Moor's survey contained statistics from all the other surveys mentioned, it may be tentatively concluded that the existence of the chromosome abnormality associated with Turner's syndrome may result in predisposition to some degree of mental retardation.

In this context Turner's syndrome may be compared with the other types of sex chromosome aneuploidy. It has been suggested that intellectual functioning remains relatively less impaired in patients with the former, although severe physical abnormality is common. Conversely, patients having other types of sex chromosome aneuploidy demonstrate varying degrees of intellectual impairment, with few somatic abnormalities (Polani, 1970). Similarly, the four sex chromosome groups mentioned differ from other forms of aneuploidy not involving the sex chromosomes (e.g. trisomy 21, Down's syndrome, and trisomy 8) in which physical abnormalities and mental subnormality co-exist.

There is, as Garron and Vander Stoep (1969) indicated, a criticism applicable to all surveys directed at investigating the possibility of the existence of an association between Turner's syndrome and mental retardation - that the definition of the latter is not clear. For example, different phrases such as "educational subnormality" and "severe mental defect" are used without any
indication of the method of determination being given. Whilst it might be argued that the criterion of mental subnormality hospital admission indicates a fairly severe degree of mental retardation present in the patients admitted, it is not valid to compare these patients as a separate entity with those individuals who manage to exist in the general population, and are therefore regarded by authors of surveys as being of normal intelligence.

(ii) Psychological Studies
(a) General intellectual level

Studies of non-institutionalised persons with Turner's syndrome seem also to suggest an increased incidence of mild mental retardation.

Bekker and van Gemund (1968) collected from existing literature reports on 76 cases of 45 XO Turner's syndrome, for whom there were Intelligence Quotient (IQ) results, measured by the Wechsler scales, or, as the authors stated, "adapted versions of them". They quoted a mean Full Scale IQ of 95.6, which differs significantly from the normal distribution mean of 100 (p < .05).

<table>
<thead>
<tr>
<th>IQ</th>
<th>Mean percentage</th>
<th>Normal distribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt; 70</td>
<td>9%</td>
<td>2.5%</td>
</tr>
<tr>
<td>70 - 90</td>
<td>32%</td>
<td>25%</td>
</tr>
<tr>
<td>&gt; 90</td>
<td>59%</td>
<td>72.5%</td>
</tr>
</tbody>
</table>

From the figures shown in Table I Bekker and van Gemund tentatively concluded that mild and severe mental retardation was more frequently found in individuals with 45 XO Turner's syndrome than in persons having a normal chromosome complement.
It is interesting to note that Ferguson-Smith (1965), reviewing cases of 86 patients with 45 X0 Turner's syndrome, claimed that "the incidence of mental retardation is comparatively low". These two reviews overlap considerably in the data called upon, and illustrate well the comment concerning the need for careful definition of terms and criteria used. However, it must be said in fairness that Ferguson-Smith, in discussing the point, was comparing the high incidence of mental retardation in individuals with extra chromosome material (i.e. 47.XXX, 47.XXY, 47.XYY) with those showing Turner's syndrome, lacking such material. Presumably he would have discussed the effect of genetic aspects of this lack of material on the level of IQ if he had indeed found any significant results indicative of an increased incidence of mental retardation.

Money (1964), working along the same lines, compared the IQ distributions of 38 patients with Turner's syndrome with 22 patients with Klinefelter's syndrome (47.XXY), using Wechsler intelligence scales. He found that whilst a high IQ was not incompatible with either of the chromosomal anomalies, there seemed to be an accumulation of defective IQ's in the 47.XXY group. He also pointed out that whilst six members of this group had been found in a mental deficiency hospital, there were only two patients with Turner's syndrome there.

Goldberg et al. (1968) concluded from their review of 53 cases for which cytogenetic results were available that mental deficiency was more common in their cytogenetically heterogeneous group (namely isochromosomes and mosaics) than in their 45.X0 group. Below is presented an extract from their results. Perusal of this will serve to illustrate several criticisms of the whole of the literature in this field.
Table II  Extract of Results, Goldberg et al. (1968)
(Taken from written text but displayed in tabulated form)

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Karyotype</th>
<th>Number</th>
<th>IQ description</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>45 XO</td>
<td>2/27</td>
<td>&quot;mentally deficient&quot;</td>
</tr>
<tr>
<td>2</td>
<td>45 XO</td>
<td>1/27</td>
<td>&quot;dull normal&quot;</td>
</tr>
<tr>
<td>3</td>
<td>45 XO</td>
<td>1/27</td>
<td>IQ = 73</td>
</tr>
<tr>
<td>4</td>
<td>46 XXqi</td>
<td>1/3</td>
<td>&quot;mentally dull&quot;</td>
</tr>
<tr>
<td>5</td>
<td>46 XXqi</td>
<td>1/3</td>
<td>At 13 yrs. &quot;functioned at 10 year level&quot;</td>
</tr>
<tr>
<td>6</td>
<td>45 XO/46 XX</td>
<td>1/8</td>
<td>VIQ = 96; PIQ = 76</td>
</tr>
<tr>
<td>7</td>
<td>45 XO/46 XX</td>
<td>1/8</td>
<td>&quot;rated mentally retarded&quot;</td>
</tr>
<tr>
<td>8</td>
<td>45 XO/46 XXqi</td>
<td>1/3</td>
<td>&quot;dull normal&quot;</td>
</tr>
<tr>
<td>9</td>
<td>45 XO/46 XXqi</td>
<td>1/3</td>
<td>F.S.I.Q. = 77</td>
</tr>
<tr>
<td>10</td>
<td>45 XO/46 XXD</td>
<td>1/1</td>
<td>Stanford Binet IQ = 87</td>
</tr>
</tbody>
</table>

Although Goldberg et al. attempted to draw valid conclusions from these data, such conclusions are of doubtful value for several reasons:

1. By modern standards a person is judged mentally retarded when his IQ fails to attain the level of 70. By this criterion four individuals (Nos. 3, 6, 9 and 10 in Table II) should not have been included. In fact, in the light of more recent developments in the field, Case No. 6 would be classified as being of average intelligence but typical of the syndrome in exhibiting the pattern of Visual IQ (VIQ) being significantly greater than Performance IQ (PIQ).

2. It is usually considered that a person described as "dull normal" has an IQ falling in the range of 70+. By definition (see 1 above) Cases Nos. 2 and 8 should not have been included either.

3. The vagueness of the terms "dull normal", "mentally dull", etc. is characteristic of a large number of papers. For comparative purposes material involving such terms is of little value.
4. With the exception of Case No.10 there is no mention of the IQ scale used; it is therefore impossible for any useful comparison to be made.

This last criticism applies also to the paper by Leão et al. (1968), in which were given detailed descriptions of nine girls having the karyotype 45 XO/46 XX. These descriptions were of value in themselves, since individuals with this karyotype are comparatively rare, and are often omitted from ascertainment studies because they are chromatin positive. However, four of the girls in the sample, who were described as "obviously mentally retarded", included three who had specified IQ's of 45, 53 and 86 respectively. No scales were given, and it is difficult to accept that an IQ of 86 was, in fact, indicative of mental retardation, unless there were other crippling factors present as well.

In conclusion it might be pertinent to note that Lemli and Smith (1963) stated that, although mental deficiency was an unusual finding in the 45 XO syndrome, slight intellectual impairment, judged on the basis of "personal observations and school impairment" seemed quite common. They also stated that of the 24 patients beyond the age of two years whose IQ's they obtained, only one was seriously mentally deficient (IQ = 45, Stanford Binet L-M form). In fact, IQ testing was carried out on only seven other patients, whose IQ's were reported to range from 85 - 119, with a mean value of 95. It is unfortunate that, having specified the karyotype, the authors did not give more data on both the detailed IQ scores of the other seven patients, and the specific tests involved.

(b) Pattern of intellectual abilities

Early reports on intelligence levels tended to be subjective impressions, based on vocational interests and school ratings. It was only as a characteristic pattern of sub-test scores began to emerge that interest was shown in detailed IQ data. Again it is unfortunate that more attention was not paid to specifying the test
given and to documenting the individual results relative to other factors, such as karyotype of patient, age, social class and physical concomitants of the syndrome.

Typical of the earlier type of reports on IQ levels were those by Hampson et al. (1955) and Cohen (1962). The former concluded that their IQ ratings did not support the claim that ovarian agenesis was associated with congenital impairment of intelligence. The method of presentation of their results and the inadequacy of their IQ "ratings" will be commented on in a later section on bimodal IQ distribution.

Cohen (1962) was the first to draw attention to the now generally recognised characteristic IQ pattern of patients with ovarian dysgenesis. His results obtained from a group of ten adolescent girls (who must be presumed, but were not stated, to have been chromatin negative) indicated that the range of IQ's was between 86 and 119, the mean being 96. He noted that all but one of the girls scored higher on the Verbal than on the Performance scale, but there is a lack of data on detailed sub-test scores, and on the precise ages of the patients. Cohen made the observation that the Performance scales were heavily weighted with manual tasks and suggested that the lowered Performance scores were somehow related to the inability of the patients to use their hands. He specifically commented, from an interpretative viewpoint, on the Picture Completion sub-test, in which he found that patients failed to specify the parts of human or animal bodies which were obviously missing. With reference to the verbal items he found that none of the patients gave over-estimates as answers to the question: "How tall is the average American woman?". They had difficulty in answering the question: "How are an egg and a seed alike?", which Cohen suggested was not typical of the responses given by normal girls. His conclusion was that "the intellectual processes of these girls (i.e. those in the sample) seem to be influenced by their physical disorder."
Shaffer (1962) studied a group of 20 cases seen at the paediatric endocrine clinic at the Johns Hopkins Hospital, over a two-year period. In this group 15 cases were reported to be chromatin negative, the remaining five being chromatin positive. Ages ranged from 5.8 to 30.9 years. The tests used were the Wechsler Intelligence scales and the Benton Visual Retention test. Data from the tests were analysed in terms of the two different nuclear sex groups separately, and of the whole group combined, the measures being those of Full Scale IQ, Verbal IQ and Performance IQ. In addition, the factor scores obtained from Cohen's (1957, 1959) factor analysis of the Wechsler Intelligence scales for Adults and Children (W.A.I.S. and W.I.S.C.) were computed for all the subjects over 13 years of age. No reason was given for choosing this age cut-off point. Since the W.I.S.C. is a test appropriate for all ages from five to 16, and Cohen accordingly factor-analysed the subtest scores in terms of these ages, it seems unnecessary to have adopted this cut-off point.

The results indicated no statistically significant difference between the chromatin positive and negative groups. The Full Scale IQ of the combined group was not significantly different from the standardisation mean of 100. Further analysis indicated that the mean Verbal IQ was statistically significantly higher than the mean Performance IQ. Consideration of Cohen's factors also reflected this difference. The combined group had a mean Verbal Comprehension score (loading on Information, Comprehension, Similarities and Vocabulary sub-tests) which was significantly higher than the normative mean, whilst the mean factor scores on Freedom from Distractibility (- or "Memory" - loading on Arithmetic and Digit Span) and Perceptual Organisation (loading on Block Design and Object Assembly sub-tests) were significantly below the normative means. (All findings achieved at least the .05 level of significance.)
(Since Digit Span is not a compulsory sub-test of the W.I.S.C. it may not have been possible to calculate the contribution to the factor of Freedom from Distractability for all the children. Nevertheless, Shaffer's selection of the 13-year cut-off point is still puzzling, and useful information could have been gained by considering the other two factors of Verbal Comprehension and Perceptual Organisation.)

Shaffer's work was confirmed and extended by Money (1963). From testing 36 cases he reported a similar discrepancy between Verbal and Performance IQ's, significant at the .01 level. There was a similar discrepancy between the factor scores on Verbal Comprehension and Perceptual Organisation. From this he concluded that there was a specific space form perception deficit associated with the X chromosome anomaly of Turner's syndrome. Although research has proceeded from the assumption of this association, it should be noted that Cohen (1957) specifically stated that it was incorrect to consider the Perceptual Organisation factor as a spatial factor, since it correlated so highly with other sub-tests (namely Picture Arrangement and Digit Symbol) which contain no spatial component.

Buckley (1971) replicated Money's research by testing 12 adult individuals with Turner's syndrome who had the 45 XO karyotype, using the W.A.I.S. Results indicated that the group had a mean Full Scale IQ significantly lower than the mean of a normally distributed sample (p < .05). Whilst this difference was not true of mean Verbal IQ scores, mean Performance IQ scores certainly differed significantly from those of a normally distributed sample (p < .001). The author concluded that this indicated that it was the Performance (or non-verbal) items of the intelligence test which were contributing to the lowered distribution of Full Scale scores. The Perceptual Organisation factor was also significantly lower than the Verbal Comprehension factor (p < .01). Attention was drawn
to the fact that if the scores on the two sub-tests concerned in the Perceptual Organisation factor were compared, Object Assembly scores were certainly the lowest of the Performance sub-test scores, whilst those on Block Design were marginally the highest. Two comments arise from this observation. Firstly, as Lockyer and Rutter (1970) suggested, in considering IQ scores in a population of autistic children, it is questionable whether Cohen's factors may be applied to an abnormal group of individuals. Secondly, it reinforces the suggestion made by Garron and Vander Stoep (1969) that persons with Turner's syndrome may be poor at all tasks which are primarily non-verbal, and that the label of "space-form blindness" is misleading. An analysis of variance carried out on the Performance sub-test scores showed a significant between-sub-test scores variance (p < .01). It was clear that this was due to the poor performance on Object Assembly in particular. However, this deficiency still does not wholly account for the Verbal/Performance discrepancy, and the author suggested that the basic cognitive deficits giving rise to poor performance on all the non-verbal items should be investigated more fully.

Money (1964) continued his research on Turner's syndrome by contrasting a group of individuals with the chromosome abnormality 45 X0 with 19 individuals having additional X chromosome material, namely persons with Klinefelter's syndrome (47 XXY). He found that there was no similar pattern of scores associated with the latter syndrome.

Bekker and van Gemund (1968) studied a group of 14 girls with Turner's syndrome. Using the Wechsler Intelligence scales they found that the mean Full Scale IQ was not significantly different from the normal distribution. When they compared the mean Verbal IQ with the mean Performance IQ they were unable to replicate the finding of a statistically significant difference between the mean scores. They made the interesting suggestion that this discrepancy could be age dependent, in that they noted a statistically non-significant tendency for the discrepancy to appear in patients older than 13½ years. They
were also only able to demonstrate a significant difference between the Verbal Comprehension and Perceptual Organisation factor scores for their patients over 13 years of age. In this part of the study they used a control group of nine normal girls, matched for IQ and age. Their groups differed significantly on Perceptual Organisation factor scores (p = .05).

The data presented in this paper by Bekker and van Gemund seem to be largely repeated in a chapter on Turner's syndrome in Bekker's book (1969) on the psychological characteristics of individuals with stunted stature and/or sexual infantilism. It is most unfortunate that the comparative data in this book are presented in Dutch only, with an extremely inadequate English summary of the conclusions. The sample of patients with Turner's syndrome is considerably extended to contain 15 patients, karyotype 45 XO, and a further 11 patients with variable mosaic karyotypes. Bekker's approach is of particular interest in that comparisons were drawn between patients with Turner's syndrome and other groups of individuals with small stature, who could therefore be considered as controls for the reduced height variable.

In applying these comparisons some interesting results emerge. Patients with Turner's syndrome were the only group to have an overall mean IQ below normal. Once again the mean Verbal IQ was greater, but not significantly so, than the mean Performance IQ. For other groups, however, this difference was significant, i.e. the IQ profile associated with Turner's syndrome by American researchers was shown to be typical of groups of patients with stunted growth and delayed adolescence, chondrodystrophia and hypopituitarism - to a greater, and indeed statistically significant, extent.

Similar studies on dwarfed children (Pollitt and Money, 1964; Money et al. 1967) failed to endorse the results obtained by Bekker, however. The studies agreed on the point that children with hypopituitary dwarfism were not intellectually retarded, but the American researches failed to produce any significant differences between
sub-scales or Cohen's factors. These studies differ from those of Bekker in involving children only, and it might be postulated that the stage of intellectual development at which the test results were obtained is important.

Another feature of the Dutch study which certainly merits more consideration than can be given in the circumstances, is the follow-up and re-testing of a number of the patients. Seven of the original 15 45 XO patients were re-tested on three occasions, and whilst the Full Scale IQ totals of six of these increased - as might be expected from practice effects - no further pattern emerged. However, Bekker concluded that the relation between Verbal and Performance IQ's was not stable, but changed with age.

In spite of the limitations of utilising the data presented by Bekker, there emerge several important points which have not been raised previously. It appears that the pattern of cognitive abilities formerly associated with Turner's syndrome may not necessarily be specific to that one diagnostic entity. Whether or not it proves to be the case in all studies, there is clearly considerable information to be gained by following up the patients over the years of psychological development.

In his extensive study of several aspects of patients with Turner's syndrome Lindsten (1963) employed two intelligence tests. One, the CVB, was a Swedish modification of the Wechsler Bellevue, which omitted the sub-tests of Digit Symbol, Block Design and Object Assembly (see Garron and Vander Steep, 1969). The other, the SRB, contained three sub-tests, a multiple-choice synonym test, a reasoning test, and a block design test based on Koh's Blocks. As may be seen from reference to the previous papers discussed, the composition of these two tests may well preclude comparison with other studies.

Lindsten reported the correlation between the two tests for results from this sample to be + 0.87, but he did not report on what the figure was when the tests were administered to a normal population.
One might hypothesize that the correlation coefficient would be higher in the latter case where the profile of sub-tests might be expected to be more regular. It is interesting to note for the purpose of comparison with data presented in this thesis that those patients with isochromosomes or 45 XO/46 XX constitution were reported as generally having an IQ of 100 or more. This would seem to fit the model proposed by Ferguson-Smith (1965) already referred to briefly in Chapter II.

It might be valid to postulate that the IQ's of mosaics and other variants of the 45 XO karyotype would be expected to appear on the continuum of phenotypes ranging from normal 46 XX female to the 45 XO female. However, both Leão et al. (1966) and Goldberg et al. (1968) failed to endorse Lindsten's finding. Goldberg et al. claimed that the incidence of mental deficiency in their heterogeneous group of mosaics and structural variants "far exceeded" that in the 45 XO cases. This discrepancy might well be reduced by applying the criticisms already made of this paper (see p. 47). The data from Leão et al. have already been criticised also on much the same grounds, but it is interesting to note that there is so much negative evidence against Ferguson-Smith's formulation. On the other hand, it might also be suggested that IQ is not a true measure of phenotypic expression, and therefore should not be included with other variable characteristics such as neck webbing or cardiac malformations.

Of relevance to this discussion is a paper by Money and Granoff (1965), who attempted to relate IQ to three specific somatic anomalies associated with Turner's syndrome. This was suggested by Polani's conclusion (1960) that, when mental deficiency occurred associated with Turner's syndrome, it was more likely to do so in patients with neck webbing. Whilst these authors found a tendency for cardiac anomalies and neck webbing to occur together, there was no evidence that lowered IQ was associated with neck webbing, cardiac anomalies of sex chromatin type. There was also no evidence to support
Polani's claim that a lowered IQ was associated more often with neck webbing than was a higher score.

(c) Bimodal Distribution of IQ

It has been suggested by several authors that their IQ results obtained from patients with Turner's syndrome fall into a bimodal distribution. The first reference to this was made by Hampson et al. (1955). They presented data on 16 patients, whose karyotype one must infer to have been 45 X0, since the paper was written at the time when the term "male chromosomal pattern" was current. Whilst supplying one Wechsler scale result in full for a 12-year-old individual, the authors merely gave the rest of the patients an IQ "rating" of unspecified derivation. Their results were presented graphically, and examination of these graphs showed that results between IQ's of 91 and 109 had been grouped together, while the rest had been considered in units of ten IQ points. This could well give an artificial peak to the data.

This practice of grouping results was repeated by Haddad and Wilkins (1959). They listed scores obtained from administering Wechsler IQ scales to 20 patients, as follows:

<table>
<thead>
<tr>
<th>IQ Range</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt; 69</td>
<td></td>
</tr>
<tr>
<td>70 - 79</td>
<td>borderline</td>
</tr>
<tr>
<td>80 - 119</td>
<td>average</td>
</tr>
<tr>
<td>&gt; 120</td>
<td>superior</td>
</tr>
</tbody>
</table>

These results might have shown bimodality on account of the large number included in the mid-range.

Lindsten (1965) commented on the tendency for there to be a bimodal distribution of IQ with a peak at a lower intelligence level accounted for by the 45 X0 group of patients. This tendency reached significance level (p = .02) for the SRB scale only. The result was not found to be characteristic of other patients not having the 45 X0 karyotype.
Bekker and van Gemund (1968) were unable to demonstrate bimodality in their group of 14 patients. They also ordered some 76 IQ's culled from various other cases reported in the literature in the same way as did Hampson et al. (1955), but failed once more to demonstrate bimodality. Bekker and van Gemund drew attention to the fact that the sample employed by Hampson et al. was a heterogeneous one, containing chromatin positive cases as well. Finally they also noted the groupings of IQ's between 90 and 109, and divided them into the two more logical groups of 90 - 99 and 100 - 109, as a result of which there appeared a tendency for the scores to be distributed bimodally. This tendency proved non-significant.

Moor (1969) produced two figures to illustrate the bimodal distribution of IQ scores reported for the groups studied by Lindsten and Money, which he claimed reinforced the previous findings. In fact, lack of axis labels and information on derivation of data has made it impossible to reconstruct such figures.

None of the authors who have presented their data in this form gave reasons for considering them in such a way, and they failed to draw conclusions from their results. To prove bimodality would seem relevant only if an attempt were being made to divide individuals with Turner's syndrome into two groups on the basis of IQ.

A review of previous psychiatric studies of IQ in individuals with Turner's syndrome has been given, and areas which are considered to require further investigation have been indicated. In particular, the deficiency of control data has been commented on, and the interpretation of results queried. It is the aim of this study to attempt to remedy the deficiency by employing a control group, thus providing data which make possible inter-group rather than intra-group comparison.
METHOD

Subjects

(a) Experimental subjects

This group consisted of 24 adult female individuals with Turner's syndrome. To be included in the sample the subjects had to satisfy the following criteria:

- Diminished stature
- Primary amenorrhoea
- Abnormal karyotype

For the purpose of analysis the sample may be considered in two groups:

- Group A (N = 16)
  These individuals satisfied the first two criteria listed above, and have the abnormal karyotype 45 X0.

- Group B (N = 8)
  The individuals within this group satisfied the first two criteria listed above, and have varying karyotypes. These may be detailed as:
  - 4 individuals with 46 XXq1 karyotype
  - 1 individual with the mosaic karyotype 45 X0/46 XXq1
  - 2 individuals with mosaic karyotype 45 X0/46 X
  - 1 individual with the mosaic karyotype 45 X0/46 XY

It should be stressed that in terms of the two phenotypic criteria of diminished stature and primary amenorrhoea these individuals presented an unvaried picture.

These patients were selected from those registered in the Registry of Abnormal Karyotypes, of the M.R.C. Human and Population Cytogenetics Research Unit in Edinburgh. Factors limiting selection, besides the criteria already listed, were geographical location of the patient's home; willingness of the referring physician or general practitioner for the patient to be approached for research purposes; the permission
of the patient's parents, if she was still in their care; and, finally, the willingness and continued co-operation of the patient herself. Since the condition from which these patients suffer is so rare and there is little hope of their being offered a corrective remedy, it is quite understandable that they may well feel they are being over-researched with seemingly very little advantage to themselves. In fact, three patients with 45 XO karyotype and one with isochromosome X, who were approached, expressed reluctance to take part, and two others withdrew after the initial testing session. The original group was further depleted by removals to new un-notified addresses and emigration.

Mean Age

The mean age of the whole experimental group was 26.2 years (S.D. = 7.55). The mean age of the 16 individuals included in Group A was 27.6 years (S.D. = 8.67), and that of the eight individuals in Group B was 23.5 years (S.D. = 3.12). For comparative purposes it was necessary to ascertain that these two groups did not differ significantly in age (t = 1.69 N.S.). The age range of the total group was 16 years to 46 years, with all but one patient being under the age of 40. This is not an unexpected finding, since the cardiac anomalies associated with the syndrome considerably decrease the life expectancy of the patients.

Social Class

The experimental group were distributed evenly over the lower and middle classes.

Testing Arrangements

A letter requesting permission to approach a patient was first despatched to her G.P. None refused access. A letter and appointment card were then sent to the patient, explaining the research project and requesting her co-operation. Depending upon circumstances, such as distance from Research Unit, work timetable, etc., the patient either came to the Unit or was visited at her home. Whilst test administration under Unit conditions remained fairly
standard, this could certainly not be said of home visits. Conditions there varied widely. Some arrangements were completely adequate, but a minority were not, a situation which has to be borne in mind when considering results obtained in such conditions.

(b) Control group

A total of 60 females selected from the lists of a local general practice were tested to gather data which could be used for control purposes.

Selection

The control group was selected by employing a computer listing of all the females on the general practice list, arranged in chronological order. It was thus possible to select a random group of controls made up of those females who had approximately the same dates of birth as those of the experimental group. A letter was prepared on an automatic typewriter (which produces a letter which is more personal in appearance than that produced by a stencil). The name, address and time of appointment were inserted by the operator in the appropriate places, by interrupting the tape. With the letter was despatched an appointment card, to be returned in a S.A.E. provided. On this card the recipient could indicate acceptance or refusal of appointment, or suggest an alternative time. 120 persons were contacted in this way, the method of selection being exactly the same for all.

Mean Age

As a result of the selection method adopted, the mean age of the control group corresponded closely to that of the experimental group (mean age = 27.2 years; S.D. = 7.93).

Social Class

This was ascertained roughly by enquiring about the husband's occupation, own occupation before marriage, or father's occupation. It was found that the controls were fairly evenly distributed between the middle and lower social classes.
Testing Arrangements

One of the doctor's surgeries was made available for all testing sessions. These were one and a half hours in duration. As the battery of tests involved required approximately treble this length of time for completion, three control subjects were contacted to every one individual with Turner's syndrome. This was considered more efficient a process than attempting to get the same control to return for two subsequent testing sessions. Since not all the individuals with Turner's syndrome completed the whole battery of tests fewer than the expected 72 controls were required.

The controls were divided into three groups:

Group 1 - Acted as controls for Personality and Colour Vision tests

Group 2 - " " " " the intelligence test and one of the experimental tests

Group 3 - " " " " all the experimental tests

Further details of test order will be supplied in the appropriate chapters.

Measuring Instrument:

Wechsler Adult Intelligence Scale (Wechsler 1955)

This comprises eleven sub-tests, of which six are termed Verbal and the remaining five Performance, sub-tests. They are administered in the following order:

1. Information sub-test. This involves questions about subjects of general knowledge,
   e.g. "What is the population of the United Kingdom?"
   "At what temperature does water boil?"

2. Comprehension sub-test. This involves questions on matters which demand a certain measure of understanding and common sense,
   e.g. "What is the thing to do if, while in the cinema, you are
the first person to see smoke and fire?"
"Why do people have to pay taxes?"

3. Arithmetic sub-test. This consists of questions involving arithmetic. The majority of the problems are concerned with money. Testing of some of the individuals with Turner's syndrome began prior to the introduction of decimal currency, whilst others were tested after this date. To retain as uniform a situation as possible, the test was introduced as sums involving some money calculations (a procedure which is not advised by the manual), and the subject was asked whether she could remember the old currency. Since all stated that they could, subjects of the control group were treated in exactly the same way. Examples of the Arithmetic sub-test questions are:

"How many books can I buy for 36/- if one book costs 6/-?"
"How many inches are there in 2 1/2 feet?"

4. Similarities sub-test. This involves pairs of items; the subject is asked to explain in what way they are alike.

  e.g. "In what way are an axe and a saw alike?"
  "In what way are a fly and a tree alike?"

5. Digit Span sub-test. The subject is asked to repeat a list of digits in the first part of the sub-test; in the second part the subject has to repeat them backwards.

6. Vocabulary sub-test. This involves explaining the meaning of words;

  e.g. "What is the meaning of 'regulate'?"

7. Digit Symbol sub-test. This involves paired numbers and symbols; the subject has to continue writing the correct symbols against each number until the time limit of 90 seconds expires.

8. Picture Completion sub-test. A series of pictures is presented, from each of which a part is missing; the subject has to name the part before the time limit of 20 seconds expires. This test included one picture of an "old" penny from which the flag on the
shield is omitted. In the "decimalised version" of the sub-test this picture has been replaced by one of a 50 p. piece, but this was not available at the time of testing.

9. Block Design sub-test. This involves the copying of a small template, using four red and white blocks for the easier items and nine blocks for the more difficult ones. Time limits are applied and it is possible to gain bonus points if the task is completed exceptionally quickly.

10. Picture Arrangement sub-test. Sets of cards are presented, each set in a fixed random order. The subject has to rearrange the cards so that the pictures on them tell a story. Time limits and bonuses again apply.

11. Object Assembly sub-test. The pieces are put out in a fixed order behind a screen, which is then removed. The subject has to place the pieces together in the right positions to make an object. Time limits and bonuses apply, and scores are also given for part-completion of the task.

The first six of the sub-tests described above contribute to the Verbal IQ and the remaining five to the Performance IQ. By combining these scores the Full Scale IQ is obtained.

Scoring Procedure
This followed the instructions in the manual. For each subject the following scores were noted:
(i) Verbal Scale IQ (VIQ); Performance Scale IQ (PIQ); Full Scale IQ (FSIQ); calculated in the manner laid down in the manual, with values expressed in terms of American norms.
(ii) Cohen's factors of

(a) **Verbal Comprehension**, given by the mean of the age adjusted scores on the four sub-tests, Information, Comprehension, Similarities and Vocabulary.

(b) **Perceptual Organisation**, given by the mean of the age adjusted scores on the two sub-tests, Block Design and Object Assembly.

(c) "**Memory**", given by the mean of the age adjusted scores on the two sub-tests, Arithmetic and Digit Span.

**Procedure**

**Experimental Group**

Individuals within this group were seen for a minimum of one testing session and a maximum of three. The intelligence scale was always administered during the first session, usually preceded by a colour discrimination test (100 Hue). It was given to the 24 subjects within the experimental group.

Administration followed the instructions laid down in the manual, except for the minor adjustments which had to be made for the items which had not been decimalised.

**Control Group**

The test was administered to 24 controls of Group 2. It was always administered before the experimental task.

The testing sessions with members of the control group were always prefaced by a brief description of the research project; it was explained that a set of results had been obtained from a group of individuals having a rare medical abnormality, and that before conclusions could be validly drawn from their results on psychological tests it was necessary for comparative purposes to collect similar results from persons not suffering from this complaint. It was soon
found that this procedure helped considerably in establishing rapport, and in reducing anxiety amongst the more timid members of the group.

The test was scored as specified in the manual, and the three scores mentioned previously were noted for each control on each sub-test.

**Statistical Analysis of Results**

**For Hypothesis III/1**

Full Scale IQ scores of Group A (all karyotype 45 XO) were compared with those of Group B (heterogeneous karyotypes), using the Mann-Whitney U test (Siegel, 1956).

**For Hypothesis III/2**

(a) Full Scale IQ scores,
(b) Verbal Scale IQ scores,
(c) Performance Scale IQ scores, of the total experimental group and the control group were compared, using t-tests (Guilford, 1956).

**For Hypothesis III/3**

Cohen's factors (1957) of
(a) Verbal Comprehension,
(b) Perceptual Organisation,
(c) "Memory" (Freedom from Distractability)
for the total experimental and the control group were compared, using t-tests.

**For Hypothesis III/4**

Raw scores on all 11 sub-tests for the experimental and control groups were compared, using t-tests.

**For Hypothesis III/5**

The experimental group's scores on the five Performance sub-scales were examined, using an analysis of variance technique for single cells (Guilford, 1956).
HYPOTHESES

On the basis of the above discussion the following hypotheses were formulated:

III/1 That there will be no significant difference between the Full Scale IQ scores of Group A when compared with those of Group B.

III/2 That there will be no significant difference between

(a) Full Scale IQ scores,
(b) Verbal Scale scores,
for the experimental and control groups, but
(c) Performance Scale IQ scores for the experimental group will be significantly lower than those for the control group.

III/3 That there will be no significant difference between the experimental and control groups on Cohen's factor of Verbal Comprehension; but that experimental group will have significantly lower scores than the control group on the Cohen factors of Perceptual Organisation and "Memory".

III/4 (a) That there will be no significant difference between the experimental and control groups on raw scores on the six Verbal sub-tests, but
(b) That the experimental group will score significantly lower on the five Performance sub-tests.

III/5 That there will be no significant difference between the scores on the five Performance sub-tests for the experimental group.
RESULTS

Hypothesis III/1

The results supported this hypothesis. Table III shows the mean Full Scale IQ scores for Groups A and B.

Table III  Mean Full Scale IQ scores of Group A and Group B

<table>
<thead>
<tr>
<th></th>
<th>Group A</th>
<th>Group B</th>
<th>U</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>16</td>
<td>8</td>
<td>60.5</td>
<td>N.S.</td>
</tr>
<tr>
<td>Full Scale IQ (mean)</td>
<td>88.75</td>
<td>91.0</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

The Mann-Whitney U test showed that the two groups did not differ significantly in terms of their Full Scale IQ scores. Thus, in terms of intelligence test scores, the two groups may be considered together.

Hypothesis III/2

This hypothesis was partially supported by the results. As may be seen from Table IV the first part of the hypothesis was not supported by the Full Scale IQ score results, but the other two comparisons supported the rest of the hypothesis.

Table IV  Mean Full Scale IQ, Verbal Scale IQ and Performance Scale IQ scores for the experimental group and control group

<table>
<thead>
<tr>
<th></th>
<th>Experimental Group</th>
<th>Control Group</th>
<th>t</th>
<th>P</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean 24 S.D.</td>
<td>Mean 24 S.D.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>N</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Full Scale IQ</td>
<td>89.50 11.87</td>
<td>103.13 11.94</td>
<td>3.88</td>
<td>&lt;.001**</td>
</tr>
<tr>
<td>Verbal Scale IQ</td>
<td>95.54 12.99</td>
<td>102.08 13.66</td>
<td>1.70</td>
<td>N.S.</td>
</tr>
<tr>
<td>Performance Scale IQ</td>
<td>82.79 10.53</td>
<td>103.92 9.87</td>
<td>7.02</td>
<td>&lt;.001*</td>
</tr>
</tbody>
</table>

** indicates 2-tailed test
* indicates 1-tailed test

These results indicated that the experimental group had significantly lower Full Scale IQ's and Performance Scale IQ's than the control group. The two groups did not differ significantly on Verbal Scale IQ scores.
Hypothesis III/3

The results supported this hypothesis. The mean factor scores for Verbal Comprehension, Perceptual Organisation and "Memory" are given in Table V.

Table V  Mean Cohen factor scores for the experiment and control groups

<table>
<thead>
<tr>
<th></th>
<th>Experimental Group</th>
<th>Control Group</th>
<th>t</th>
<th>p*</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N=24</td>
<td>N=24</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Verbal Comprehension</td>
<td>9.35 2.21</td>
<td>10.11 2.71</td>
<td>1.06</td>
<td>N.S.</td>
</tr>
<tr>
<td>Perceptual Organisat.</td>
<td>7.08 2.12</td>
<td>10.81 2.61</td>
<td>5.44</td>
<td>&lt;.001</td>
</tr>
<tr>
<td>&quot;Memory&quot;</td>
<td>8.94 2.54</td>
<td>10.52 2.10</td>
<td>2.35</td>
<td>&lt;.05</td>
</tr>
</tbody>
</table>

* indicates 1-tailed test

These results indicated that whilst the experimental subjects were lower on Perceptual Organisation and marginally so on "Memory" factors than the control subjects, there is no significant difference on the Verbal Comprehension factor.

Hypothesis III/4

(a) The results partially supported this hypothesis, and
(b) wholly supported this. Mean raw scores for the eleven W.A.I.S. sub-tests are given in Table VI.
Graph V.
Wechsler Adult Intelligence Scale.

Mean sub-test scores for experimental and control groups.

- EXPERIMENTAL Ss. (n=24)
- CONTROL Ss. (n=24)
Table VI  Mean raw scores of the 11 sub-tests of the W.A.I.S. for experimental and control groups (Mean scale scores shown graphically on opposite page)

<table>
<thead>
<tr>
<th></th>
<th>Experimental group</th>
<th>Control group</th>
<th>t</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean</td>
<td>S.D.</td>
<td>Mean</td>
<td>S.D.</td>
</tr>
<tr>
<td>Information</td>
<td>14.41</td>
<td>4.54</td>
<td>15.63</td>
<td>5.86</td>
</tr>
<tr>
<td>Comprehension</td>
<td>15.38</td>
<td>3.97</td>
<td>17.33</td>
<td>5.46</td>
</tr>
<tr>
<td>Arithmetic</td>
<td>9.04</td>
<td>3.24</td>
<td>11.50</td>
<td>3.73</td>
</tr>
<tr>
<td>Similarities</td>
<td>11.67</td>
<td>4.71</td>
<td>12.92</td>
<td>5.07</td>
</tr>
<tr>
<td>Digit Span</td>
<td>10.50</td>
<td>2.21</td>
<td>11.42</td>
<td>1.72</td>
</tr>
<tr>
<td>Vocabulary</td>
<td>40.63</td>
<td>15.01</td>
<td>44.71</td>
<td>16.40</td>
</tr>
<tr>
<td>Digit Symbol</td>
<td>39.42</td>
<td>11.37</td>
<td>57.50</td>
<td>11.11</td>
</tr>
<tr>
<td>Picture Completion</td>
<td>8.88</td>
<td>2.95</td>
<td>12.83</td>
<td>2.75</td>
</tr>
<tr>
<td>Block Design</td>
<td>25.58</td>
<td>8.76</td>
<td>33.83</td>
<td>7.90</td>
</tr>
<tr>
<td>Picture Arrangt.</td>
<td>16.63</td>
<td>6.31</td>
<td>24.50</td>
<td>5.10</td>
</tr>
<tr>
<td>Object Assembly</td>
<td>19.75</td>
<td>7.86</td>
<td>32.33</td>
<td>6.16</td>
</tr>
</tbody>
</table>

* 1-tailed test
** 2-tailed test

This table indicates that whilst the experimental group scored significantly lower than the control group on all the Performance sub-tests, they also scored significantly lower on the Arithmetic sub-test.
Hypothesis III/5

This hypothesis was not supported by the results. Table VII gives the results of an analysis of variance of the five Performance sub-test scores.

Table VII  Analysis of variance of the Performance sub-test scores for the experimental group

<table>
<thead>
<tr>
<th>Source</th>
<th>Sum of squares</th>
<th>df</th>
<th>v</th>
<th>F</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Between Ss.</td>
<td>358.19</td>
<td>23</td>
<td>15.57</td>
<td>6.11</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>Between tests</td>
<td>41.47</td>
<td>4</td>
<td>10.38</td>
<td>4.07</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>Remainder</td>
<td>234.93</td>
<td>92</td>
<td>2.55</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>634.59</td>
<td>119</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

This table indicates that as well as the expected significant between subjects variance, there is also a significant between sub-test scores variance.
DISCUSSION

The results from the data presented reflect closely the findings previously described by Shaffer (1962) and Money (1964). This thesis differs from their research in providing control group data for comparative purposes; and utilisation of these produces some results which may prove helpful in extending knowledge of the condition of Turner's syndrome.

Individuals with Turner's syndrome tested for this sample have significantly lower Full Scale IQ scores than have the control group, and investigation of the component Verbal and Performance IQ scores indicated that it was the Performance items and not those of the Verbal scale which were depressing the final Full Scale score.

That this difference was accounted for by the Turner results and not by abnormal control group IQ scores was checked, and indeed the mean FSIQ (103.13), mean VIQ (102.08) and mean PIQ (103.92) were not significantly different from those of a normally distributed sample (mean = 100, S.D. = 15).

Comparison of the two groups within the experimental sample of individuals with Turner's syndrome showed no significant differences between those with the 45 XO karyotype (Group A) and those with heterogeneous karyotypes (Group B). In contrast with Lindsten's results (1963) all of the individuals with the isochromosome karyotype had IQ's well below 100 (range 76-84), and, moreover, none of the remaining group of mosaic individuals achieved a Full Scale IQ of 100. Thus it was not possible to demonstrate any evidence to suggest that Ferguson-Smith's postulated model (p. 30) holds for the "IQ phenotype" - in that isochromosome and mosaic individuals failed to show higher IQ's than those with the 45 XO karyotype.

As has already been suggested (p. 55) this negative result may not affect the validity of the model, since it is difficult to postulate how something as complex as IQ may be affected by genetic imbalance in the same way, as, for example, stature.
It must be remembered, too, that rigid selection criteria ensured a consistent phenotypic expression over the 24 cases, and this in itself may have eliminated those individuals who would have proved exceptions to the finding and might therefore have supported the model.

The fact that Cohen's "Memory" factor (which is made up of Arithmetic and Digit Span mean scores) was found to be significantly lower than the Verbal Comprehension factor had previously led Honey (1964) to suggest the presence of mild dyscalculia associated with Turner's syndrome. The results presented in this thesis replicate this finding, but the difference seems to be attributable more to the Arithmetic sub-test results than to those on the Digit Span sub-test. Comparison of other verbal sub-test scores shows no significant differences. With reference to this discussion, it is worth noting that an analysis of variance carried out on the experimental group's Verbal sub-test mean scores indicated no significant between sub-tests variance ($F = 1.47, \text{N.S.}$). This result suggests that the performance of the experimental group on the Arithmetic sub-test is no worse (or better) than that on any of the other Verbal sub-tests.

By rank ordering the Verbal sub-tests for experimental and control groups it may be seen that, whilst the control group gained higher mean scores on the Arithmetic sub-test than on the remaining five Verbal sub-tests, for the experimental group the Arithmetic mean score was the lowest of the Verbal sub-tests.

<table>
<thead>
<tr>
<th>Rank Order</th>
<th>Experimental group</th>
<th>Control group</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Vocabulary</td>
<td>Arithmetic</td>
</tr>
<tr>
<td>2</td>
<td>Information</td>
<td>Comprehension</td>
</tr>
<tr>
<td>3</td>
<td>Digit Span</td>
<td>Digit Span }</td>
</tr>
<tr>
<td>4</td>
<td>Comprehension</td>
<td>Vocabulary }</td>
</tr>
<tr>
<td>5</td>
<td>Similarities</td>
<td>Information</td>
</tr>
<tr>
<td>6</td>
<td>Arithmetic</td>
<td>Similarities</td>
</tr>
</tbody>
</table>
To name the discrepancy between the two groups' performances "dyscalculia" on such limited evidence seems, therefore, not wholly justified, but clearly the phenomenon warrants further study. In passing it may be noted that two of the younger subjects included in the sample under study had gained passes in Arithmetic 'O' level, and are at present employed as bank clerks.

A Verbal/Performance IQ discrepancy, in which the Verbal IQ exceeds the Performance IQ, (previously described by Cohen, 1962, and Shaffer, 1962), was found to be a characteristic of this sample as well. That this is a significant feature of individuals with Turner's syndrome was confirmed by comparison with control group data. Whilst 12 of the 24 experimental subjects had Verbal/Performance discrepancies of 14+, only two of the control subjects had ($\chi^2 = 8.17; \ p < .01$).

The evidence so far presented indicates that it is those abilities which are tapped by the Performance sub-tests which should be investigated further. The analysis of variance of these tests for the experimental group shows that there exists a significant between sub-tests variance. If differences between individual sub-test scores are investigated, Buckley's finding (1971) on a smaller group ($N = 12$) of individuals with Turner's syndrome is replicated when only Group A subjects are considered. These individuals performed at a significantly lower level on the Object Assembly sub-test than on any of the other sub-tests ($t = 2.19; \ p < .05$). This comparison shows only a non-significant trend when results from all 24 experimental subjects are considered ($t = 1.71; \ p < .1$), but Object Assembly mean scores are certainly significantly lower than those for Block Design ($t = 2.57; \ p < .02$). It is this finding which has led the author to query the wisdom of combining the scores on these two sub-tests into Cohen's Perceptual Organisation factor when considering W.A.I.S. results obtained from individuals with Turner's syndrome. Whilst no conclusions can be drawn as to the validity of considering the two tests combined as a factor, it does
seem possible that information may be lost by not considering separately the tests upon which the factor is based. It could be hypothesised that there is a separate ability particularly concerned with Object Assembly skill which is deficient in patients who have Turner's syndrome.

From the above discussion emerge two factors involved in the deficient performance on non-verbal items on the W.A.I.S. One of these seems to be specific to the Object Assembly sub-test, whilst the other is non-specific and appears to influence performance on all the non-verbal items.

The factor specific to the Object Assembly test may be examined by contrasting the procedures involved in Block Design performance with those involved in Object Assembly. Block Design requires the subject to break down a visually presented geometric pattern involving figure/ground relationships into its component parts, and then to reconstruct it, using the block material. Object Assembly is presented in its component parts, without further clues as to shape and identity, so that the subject is required to develop her own "visual schema" before fitting the parts together to make a whole, helped by the black line cues which are present (except in the case of the Hand item), and which give some impression of substance.

The main differences between the Block Design and Object Assembly tests seem to be four in number:

<table>
<thead>
<tr>
<th>Block Design</th>
<th>Object Assembly</th>
</tr>
</thead>
<tbody>
<tr>
<td>(i) format to copy</td>
<td>no format (except for internalised one)</td>
</tr>
<tr>
<td>(ii) geometric design</td>
<td>concrete objects (albeit very diagrammatically represented)</td>
</tr>
<tr>
<td>(iii) Breakdown and synthesis</td>
<td>synthesis alone</td>
</tr>
<tr>
<td>(iv) surface design alone</td>
<td>interposition of features</td>
</tr>
</tbody>
</table>
As far as (iv) is concerned, there seems to be no evidence to suggest that the experimental group were any less able than the control group to cope with the black line cues of the Object Assembly test pieces, since comparison of performance scores on each of the form objects, Manikin, Profile, Hand and Elephant, failed to show any significant differences for the two groups ($x^2 = 2.66$, $p < .2$).

So far as (ii) is concerned, it could be suggested that the objects are so poorly representational that the experimental group experienced difficulty in abstracting any meaning corresponding to their own normal perceptions of the common objects concerned. On the other hand, it is interesting to note that previous research has commented specifically on difficulty with angles, which might indicate that the geometric patterns involved in the Block Design test would be found especially difficult.

However, one might postulate that a combination of (i) and (iii) could form a theoretical basis for an explanation of the test specific factor affecting deficient Object Assembly performance, namely, an inability to synthesise the given parts into a whole, representative of an internalised schema.

As far as the identity of the non-specific factor affecting all performance sub-tests is concerned, two possibilities may be suggested. One, put forward by Garron and Vander Stoep (1969), is that persons with Turner's syndrome are poor at all tasks which are primarily non-verbal. The second is that, since all the performance tests are heavily biased towards speed, persons with Turner's syndrome are demonstrating some form of motor retardation. Certainly they performed much more slowly on the Digit Symbol sub-test, which is commonly used as a test of speed. If this is true of individuals with Turner's syndrome, it is interesting to speculate whether the motor retardation is unvarying at all ages, or whether this finding represents a "slowing down" or ageing effect in adult persons with Turner's syndrome, which is of a greater severity and manifested at an earlier age than is the case in normal persons.
The latter suggestion is supported by an inverse positive correlation between speed, measured by the Digit Symbol sub-test, and age (Spearman rho = 0.39; \( p < 0.05 \)), for individuals with Turner's syndrome; whereas for the control group there was no evidence of any such relationship (Spearman rho = -0.01, N.S.). This finding would seem to support the view that the motor retardation is related to ageing effects, abnormal in that they occur in an age group far younger than that in which a similar correlation usually begins to appear in the normal population.

In this context reference may be made to results obtained from two girls with Turner's syndrome who were not included in the experimental group as they were below the age of 14 years. One, age eight years at the time of testing with the Wechsler Intelligence Scale for Children (W.I.S.C.), gained a score of 10 for Coding (an equivalent test to Digit Symbol in the W.A.I.S.) - a result which was certainly not the lowest of her sub-test scores. There was also no evidence of a Verbal/Performance discrepancy (VIQ = 96; PIQ = 100). The other girl, aged 12 years, showed a large Verbal/Performance discrepancy (VIQ = 115; PIQ = 99), and gained a score of 8 on Coding, which was the lowest of all her sub-test scores. Clearly these findings warrant further study, particularly of the longitudinal kind.

If it were indeed the case that the non-specific factor negatively influencing performance sub-tests was motor retardation, then it could be argued that those individuals demonstrating the bigger discrepancies between Verbal and Performance Scale IQ's would also be those who were slowest on the Digit Symbol sub-test. In fact, the converse proved to be the case, i.e. it can be statistically significantly proven from these data that persons with a Verbal/Performance discrepancy of 14 + perform more quickly on the Digit Symbol test than do those with discrepancies of 13 - (Fisher Exact Probability test: \( p < 0.025 \)). This paradoxical finding could be due to the fact that those individuals with Turner's syndrome with the
largest Verbal/Performance discrepancies also have higher Verbal IQ's (Spearman rho = - 0.12, N.S.). Thus, although a characteristic common to all individuals with Turner's syndrome is a deficient performance on non-verbal items because of motor retardation, those who have a potentially higher IQ reflect this in their verbal abilities alone.

Further support for this suggestion may be drawn from the high positive correlation between speed as measured by the Digit Symbol sub-test and Verbal IQ (Spearman rho = 0.76; \( p < .01 \)). This would seem to imply that it is the quicker and more efficient individuals with Turner's syndrome who overcome their potentially deficient Full Scale IQ by concentrating on verbal skills, whilst those who are slower and less motivated, do not; accordingly their Verbal IQ stays on a par with their Performance IQ and no significant discrepancy is observed. This could be a reflection of a spurious correlation caused by both being measures of IQ, but the control group showed no evidence of a similar correlation between speed and Verbal IQ (Spearman rho = 0.22, N.S.). This again seems to point to some kind of adaptation on the part of the individuals with Turner's syndrome.

To summarise - this discussion of intelligence test results from individuals with Turner's syndrome postulates two factors adversely affecting Performance sub-tests. One is peculiar to the Object Assembly sub-test alone, depressing the results on it below those on other Performance sub-tests, and involving an inability to perceive part/whole relationships. The second is non-specific, and involves a general inadequacy, possibly related to a premature ageing effect, in all tasks which require speed; and which, therefore, affects the other Performance sub-tests as well as Object Assembly.
CHAPTER IV

EXPERIMENTAL STUDIES INVOLVING SPECIALISED TESTS

INTRODUCTION

From intelligence test results have arisen various studies which have attempted to clarify the unusual pattern of sub-test scores associated with Turner's syndrome. Whilst the majority of these originated from the Johns Hopkins research team there are other isolated studies which are relevant. The tests cover a wide range of assessment and it is difficult to categorise them in any meaningful way or to relate them to the experimental studies undertaken in this thesis. However, the literature may be considered under two general headings:

(a) Tests of defective drawing
(b) Associated tests

(a) Tests of defective drawing

(1) Benton Visual Retention Test

(For a description of the test, see p. 89)

Shaffer (1962) administered this test to 17 of the 20 subjects previously reported (p. 50). He compared their results with Benton's standardised norms, the Full Scale IQ being taken as an index of intelligence level. There were, once again, no differences between the chromatin positive and chromatin negative groups. Whilst only two subjects obtained expected scores, the remainder scored below expectancy, there being a mean difference of two points between obtained and expected scores over the whole group.

This study was followed up by Alexander, Erhardt and Money (1966), who administered the Benton Visual Retention test to 18 patients (neither karyotype nor nuclear sex specified). These authors
gave more details about the form of administration than did Shaffer, in that they stated that both Forms C and D were given, each design being exposed for ten seconds before the patient was requested to draw it from memory. Their method of considering the results was unorthodox and unwieldy. Instead of employing the customary numerical scoring procedure they allotted IQ ratings (very superior, superior, high average, average, low average, borderline and defective) to each patient on the basis of

(i) intelligence test results (given separately for Verbal, Performance and Full Scale scores)
e.g. for Patient A.V. (aged 12 years)
Verbal: "Superior"; Performance: "Average";
Full Scale: "High Average"

and (ii) number of designs reproduced correctly on the Benton Visual Retention test
e.g. for the same patient
Form C: "Low average"; Form D: "Borderline";
Combined C and D: "Low Average/Borderline".

This method still required recourse to Benton's norms, since a control group was not used.

It is difficult to see any advantage in this method over the conventional one. Considerable valuable detail is lost by not enumerating the IQ levels involved. Even so, the results clearly indicated that the patients experienced difficulty in reproducing the designs correctly from memory, and that this difficulty was not a consequence of lowered or deficient IQ.

Alexander et al. also analysed the errors in terms of the classification given in the manual (Benton, 1955). There was a preponderance of errors of distortion (Form C - 49%; Form D - 67%) and of rotation (Form C - 19%; Form D - 15%); whilst errors of omission, perseveration, misplacement and size were presumably (not stated) less common.

Wahler (1956) found that for normal adults over 14 years of age
distortions accounted for 41% of errors and rotation for 13%. According to Alexander et al., using Benton's criteria for diagnosis of a specific organic impairment, ten patients on Form C and 11 patients on Form D would have received this diagnosis. Finally, they stated that figures involving angles proved particularly difficult.

Bekker and van Gemund (1968) compared their group of Benton results on nine patients (karyotype 45 X0) with a control group of nine normal girls, matched for IQ and age. They reported that the patients made significantly more errors than the control group (p = .05), and that the number of correct answers was also lower (p = 0.1). A problem arises from this study which is particularly relevant when matched controls are being chosen for the Benton test. Assuming that a Wechsler scale has been employed to estimate the patient's IQ, and it is desired to match controls in terms of IQ, which form of the IQ (Verbal, Performance, or Full Scale) is the most valid? The Full Scale IQ is the most commonly used, but in the case of patients with Turner's syndrome this will have been depressed by the frequently occurring deficient performance on non-verbal items. For this reason it might be suggested that the Verbal IQ level should be used as the matching criterion, but this, too, may be depressed by further deficiencies as yet possibly unspecified; e.g. Money (1964) claimed that there was evidence of dyscalculia affecting Arithmetic sub-test scores.

(2) Bender Visual Motor Gestalt test (Bender 1930)

(For a description of the test, see p. 90).

This test was used by Alexander et al. (1966) to assess further the defective drawing demonstrated by individuals with Turner's syndrome. This test involves copying simple designs from nine cards, and is therefore a test of visuomotor function rather than of visual memory - the latter is covered by the Benton Visual Retentio
test. The figures were scored in accordance with the criteria laid out in the Monograph by Bender (1938), and had to be accurate enough to maintain the gestalt (or interaction of perceived patterns) of the original stimulus. The test was originally devised to assess the development of visual perception in children from the age of three years, and attempted to gauge the maturation processes involved before the child reaches the adequacy plateau equivalent to adult performance at about the age of 11 or 12 years. The scoring procedure is therefore tied to the various developmental levels associated with each figure, and is of little value in assessing the performance of adults.

Since the group of 18 subjects with Turner's syndrome studied by Alexander et al. did not succeed in copying all nine designs correctly the authors concluded that the performance was so poor as to suggest a deficit of visual perception. This could then certainly account for the difficulties experienced in the Benton test and might indicate that visual memory itself was not grossly impaired. The authors also commented on particular difficulties experienced with angled figures, but whilst some patients' errors were restricted to these alone, other patients found all parts of the test difficult.

(3) Draw-a-Person test (Goodenough, 1926).

This third section, relating to defective drawing ability in Turner's syndrome, concerns Goodenough's test, in which the subject is asked to draw a person. As will be reported more extensively in Chapter VII, Cohen (1962) used this test primarily in its interpretative role to examine characteristics of femininity and rigidity in his group of ten individuals with Turner's syndrome. He drew attention to the poor quality of the drawings, and suggested that this was related to the patients' own body images and their physical deficiencies.

Alexander et al. (1966) used the same test to assess drawing
inadequacies. The test differs from the previous two in that it involves spontaneous drawing. The subject is merely asked to draw a person, with no specification as to sex. The drawings by 18 patients with Turner's syndrome were graded by two judges in accordance with the Quality-scale scoring system introduced by Harris (1963). Fourteen patients had scores below the 50th percentile, a result which indicated that similar difficulties were experienced in producing good spontaneous drawing as in copying geometric figures. With reference to this last point, more data on the inter-relationship between the three test performances would be useful. For example, were the four patients who obtained scores above the 50th percentile those who did better on the Benton and Bender Gestalt tests? It would be interesting, too, to relate the performances on these tests to the IQ results; (IQ details were indeed given alongside the selected samples of drawings, but no further use of the data was referred to).

Finally Bekker (1968) replicated this finding for nine 45 XO patients. He also compared his results with those for nine normal girls matched for age and IQ, and found a similar result.

(b) Associated tests

(i) Road-Map Test of Direction Sense (Money 1965)

The study by Alexander et al. (1964) on right-left directional sense in individuals with Turner's syndrome arose from previous findings on the lowered Perceptual Organisation factor in these patients (Shaffer, 1962; Money, 1964), and attempted to elucidate the space-form dysgnosia postulated by Money (1964). For the purpose a road-map test was evolved (Money et al., 1965), in which the subject was asked to report whether a left or right turn was involved in taking a route on a specially designed map laid in front of him, which he was not allowed to manipulate in any way. This test was given to a group of 13 patients with Turner's syndrome
(karyotype unspecified, cases under 13 years of age and two mental defectives having been excluded), and to a control group of 40 female nursing students. The mean difference in the number of errors made by the two groups was highly statistically significant \( (p < .001) \), the higher number of errors being taken by the authors to support earlier reports of space form dysgnosia. However, they stated that the particular disability was not always present in the syndrome, in that three patients had relatively few errors.

The Road-Map test was also reported to have been administered to groups of children, and the findings from that study indicated that right-left discrimination does not become well established until between the ages of 11 and 13 years. It was for this reason that subjects under the age of 13 years were excluded from their study.

Results on the Road-Map test were replicated by Bekker and van Gemund (1968). They found that the mean number of errors made by 11 45 X0 patients with Turner's syndrome was significantly greater than the mean number made by a control group of seven normal girls.

(2) Other relevant research

The result from the previous experiment, taken together with evidence of space form dysgnosia and dyscalculia from other studies (Money 1964) led Alexander and Honey (1966) to compare the neuro-psychological characteristics of Turner's syndrome with those of Gerstmann's syndrome. Both postulated entities arose from consideration of Cohen's factor analysis of the Wechsler scales. The author of this thesis has already questioned the validity of using these factors in investigating the scores from individuals with Turner's syndrome; in addition, it should be noted that the validity of the Gerstmann's syndrome is by no means established. The Perceptual Organisation factor represents a clustering of abilities required for the successful completion of the tests of Block Design
and Object Assembly and therefore might appear less precise than the label "space form dysgnosia" would warrant.

The evidence of the existence of dyscalculia, on the other hand, is derived from low results on the Cohen factor Freedom from Distractability, which loads on Arithmetic and Digit Span sub-test scores. It is not clear why the defect in this factor was attributed only to inability to do the Arithmetic sub-test successfully, and why the contribution from the Digit Span sub-test was disregarded. Indeed, Alexander and Money (1966) showed from 28 patients that the mean Digit Span sub-test scores were significantly lower than the mean Arithmetic sub-test scores (p = .02).

The symptoms comprising Gerstmann's syndrome are

(i) finger agnosia - inability to name, recognise or differentiate between the individual fingers;
(ii) right-left disorientation - Gerstmann asserted that this symptom was restricted to body relationships, but Alexander and Money cited other studies which claimed that there might be right-left disorientation with respect to objects outwith the body;
(iii) dyscalculia - defined as "an inability to perform simple or complex arithmetical operations and discriminations for the sequence of digits within a figure."
(iv) dysgraphia - defined as "a complex inability to form letters or words."

Alexander and Money concluded that the typical Gerstmann syndrome was not present in patients with Turner's syndrome, in that dysgraphia and finger agnosia did not appear at all. Dyscalculia was described as mild. Right-left discrimination was limited to extra-personal space, and did not include body image. Some of the associated symptoms of Gerstmann's syndrome were found, namely constructional apraxia (disturbance in formative activity), demonstrated by poor scores on the Perceptual Organisation factor, and defective geometric and figure drawing ability; and visual-spatial
disorientation (definition given as "closely related to constructional apraxia") which appeared to involve the ability to abstract the properties of objects placed away from the subject. The patients with Turner's syndrome showed themselves deficient in this ability by being significantly worse at recognising a figure after it had been rotated in space (on the Space sub-test of the Science Research Associates (SRA) Primary Mental Abilities test), and also in not being able to draw the floor plan of the room in which they were sitting.

It seems unlikely, therefore, that the cognitive difficulties associated with Turner's syndrome are consequent on cerebral dysfunction in the parieto-occipital area, as is tentatively postulated for Gerstmann's syndrome. The authors suggested instead that the neurological defect associated with Turner's syndrome might be in the parietal lobes, the area that Critchley (1955) had suggested was primarily concerned with the perception of form and spatial relationships.

Alexander and Money (1965) argued that dysfunction in this area could interfere with reading ability in the differentiation between shapes and directions of letters (e.g. c and e, p, b, q and d) and words (e.g. pot and top). To investigate this hypothesis a reading test was administered to a group of 17 girls with Turner's syndrome (karyotype unspecified). The measures of reading speed, vocabulary and comprehension of the group corresponded closely to the standardised scores of normal individuals of similar chronological age. It therefore seems that the postulated space form and directional sense deficits associated by the authors with Turner's syndrome are specific to drawing and do not affect reading or writing.

Finally, a study by Money and Alexander (1966) should be mentioned. They used the SRA Primary Mental Abilities test to follow up their previous studies using the Wechsler Intelligence scales and the Benton Visual Retention test. Since the SRA gives separate measures
for verbal-meaning, space, reasoning, number and word fluency, they hypothesised that certain of these would be deficient in patients with Turner's syndrome. Indeed, scores from 16 patients (14 being 45 XO and two 45 XO/46 XX) on sub-tests of space, number and word fluency were significantly lower. This was expected so far as the first two results were concerned, although the performance on the number sub-test was barely low enough to achieve statistical significance, a finding which casts further doubt on the surmise of dyscalculia occurring in Turner's syndrome. The low performance score on the word fluency sub-test (described as a controlled association test, in which the subject has to write down as many words as possible beginning with 's' in five minutes) was associated by Money and Alexander with the Cohen factor of Freedom from Distractability. It may be recalled that there is evidence that this factor (loading on the Wechsler sub-tests of Arithmetic and Digit Span) is also lower than in the Verbal Comprehension factor in individuals with Turner's syndrome (Money, 1964), a finding not supported by Bekker's results (1968).

This chapter and the previous one have attempted to review the research done by Money and his co-workers at the Johns Hopkins University, and the follow-up studies thereon, which tend to endorse the original findings. On the other hand, Bekker often failed to find similar results with his smaller sample of patients with Turner's syndrome.

As in the intelligence testing, the need for selected controls was often neglected and standardised norms were used in their place. When control groups were used, they tended as Garron and Vander Stoep (1969) indicated to be poorly selected in terms of age and social variables. This problem becomes even more acute when Bekker's findings [summarised in the English translation of his Dutch monograph (1969)] are recalled. They indicated that the patterns delineated by Money et al., which Bekker was not always able to find
associated with his group of patients with Turner's syndrome, showed tendencies to be associated with other syndromes involving dwarfed stature.

One of the primary reasons for studying this group of chromosomally abnormal individuals may be seen to be the provision of data which may be generalised to individuals having normal chromosomes. Garron (1970) discussed this concept in relation to the hypothesised sex linked recessive inheritance of spatial and numerical abilities. Such tasks have been claimed by authors (e.g. Macfarlane Smith, 1964) to be performed better by males than by females. Stafford (1961), however, hypothesised that these abilities could be sex-linked. From results on a test of spatial visualisation he found that correlations between mother and son, and between father and daughter, were equal to, or higher than, the correlation between mother and daughter.

These correlations may be explained in terms of X chromosome linkage. The correlation between mother and son arises from the fact that males receive their X chromosome from their mothers. Females receive one X chromosome from each parent. The correlation between father and daughter is higher than that between mother and daughter because only the X chromosome (which is assumed to carry the superior "spatial" gene) is involved.

McClearn (1967) suggested that instead of considering patients with Turner's syndrome as having a deficient Performance IQ, it was of more value to consider specific impaired abilities - namely, the spatial and numerical ones. There is a paradox here; why are patients with Turner's syndrome and only one X chromosome not similar to normal males with only one X chromosome in possessing spatial and numerical abilities superior to those of normal females with two X chromosomes? In the case of another X linked recessive trait - that of red/green colour-blindness - it has been established that the incidence is higher in individuals with Turner's syndrome than in
normal females; this makes them similar to normal males in this respect (Polani et al., 1956). So far as spatial and numerical abilities are concerned, however, individuals with Turner's syndrome have been proved to be inferior to normal females, who, in turn, are inferior to normal males on tests involving these abilities.

It is difficult to postulate an explanation for this paradox. Garron (1970) suggested that the heterogeneity of karyotypes in patients with the Turner phenotype might be confusing the issue, but Money and Granoff (1965) and Shaffer (1962) were unable to find any differences between their groups of chromatin positive and negative individuals.

So the paradox remains. It may be due to lack of definition and confusion of terms. The concepts of "spatial" and "numerical" abilities are rather imprecise. Stafford's original experiment (1961) consisted of a spatial visualisation task. The abilities involved in completing this task adequately may be far removed from those thought by Money et al. to be deficient in patients with Turner's syndrome; in these patients several inter-related abilities, having some relation to spatial perception, may be seen to be involved, but may not necessarily be equated with each other.

As already discussed, the existence of a deficient numerical ability in individuals with Turner's syndrome is open to doubt, and therefore should not be used as a factor in attempting to solve the paradox.

Finally, since the loss of chromosome material clearly has such an adverse effect on other phenotypic features (e.g., height, structure of reproductive organs, etc.) it could be hypothesised that non-verbal characteristics (in particular those associated with spatial perception) might be similarly affected. Whilst the potential "high performance" gene might be present in the remaining chromosome, the effect of the missing chromosomal material (in the form of either an entire chromosome or pieces) may be so pathological as to nullify any other more favourable effects.
IX. Benton Visual Retention Test
METHOD

Subjects

The experimental group consisted of the majority of those subjects already described in Chapter III, with the exception of those who withdrew. A corresponding number of control subjects were tested. Precise details of the numbers involved are given in the descriptions of the testing procedures.

Measuring Instruments

(i) Benton Visual Retention Test (Benton, 1955)

The standard test material consists of ten cards with geometric designs of increasing complexity drawn on them (see Illustration IX). The first two in the series show only one geometric figure; the remainder are composed of three geometric figures, two main larger ones and one smaller peripheral one.

The cards are presented singly, and the subject is asked to study the design for ten seconds before drawing it from memory on to a single sheet of paper, corresponding roughly in size to the stimulus card. This procedure is classed Administration A by the manual. Other types of administration involve only five seconds' exposure of each design with immediate reproduction from memory (Administration B); copying of designs (Administration C); and ten seconds' exposure of each design followed by 15 seconds' delay before the subject is allowed to reproduce it from memory (Administration D).

Scoring Procedure

There are two scoring systems provided by the manual: Number of correct reproductions: Each drawing is scored on an "All or none" basis out of a maximum of ten. Thus it is necessary for all three figures of the more complex cards to be correct before a score of one can be allotted.
Error score: This score is made up of errors incurred on all the separate figures making up each design. It is particularly useful because it is possible to classify the errors into six major categories, namely:

1. Omissions  
2. Distortions  
3. Perseverations  
4. Rotations  
5. Misplacements  
6. Size errors

This allows for a qualitative as well as a quantitative analysis to be made of a subject's performance on the test. Details of scoring, together with examples, are given in the manual (Benton, 1955).

(ii) Bender Visual Motor Gestalt Test (Bender, 1938)

This test consists of nine cards with geometric patterns on them. The cards are presented one at a time, and the subject has to copy them on to a large sheet of paper.

Scoring procedure

The test was originally evolved for assessment of perceptual maturation to be used as a gauge in child development studies. For this reason the scoring procedure suggested by the Monograph (Bender, 1938) was not particularly suitable for use with the experimental group under study who had all achieved chronologically adult status. Also, whilst some of their errors could be scored in terms of the "levels" described by Bender in Chapter XI of her Monograph, it was felt that valuable qualitative information was being lost.

A new scheme was therefore devised. This attempted to evaluate in as objective a manner as possible the errors made by the experimental subjects, and to compare them with test performance by the control group. It seemed possible to categorize the nine designs as follows:

Category 1, containing four cards (identification A, 4, 7 and 8); the main feature of these is that the two figures shown are either contiguous or overlapping.
Category 2, containing the remaining five cards of the series (identification 1, 2, 3, 5 and 6); in these the main feature is one of orientation, either of the total figure or of parts within the figure.

The details specific to scoring each card within these categories are given below:
Bender Visual Motor Gestalt Test

Category 1: Contiguity or Overlap
Category 1: Contiguity or Overlap

Card A

Scoring criterion: Marked correct if:

Figures contact correctly without overlapping

Card 4

Scoring criteria: Marked correct if:

(a) Figures contact correctly without overlapping, and
(b) Point of contact between figures is correct

Card 7

Scoring criterion: Marked correct if:

Angles A and B are drawn within upright figure

Card 8

Scoring criterion: Marked correct if:

Both points X and Y of diamond touch larger figure
1. 

2. 

3. 

4. 

5. 

6.

XI. *Bender Visual Motor Gestalt Test*  
*Category 2: Orientation*
Category 2: Orientation (see Illustration XI)

Card 1
Scoring criterion: Marked correct if:
Orientation of line of dots is parallel to horizontal axis of paper (to within ± .5 cms. deflection)

Card 2
Scoring criterion: Marked correct if:
(a) Orientation of figure is parallel to horizontal axis of paper, and
(b) Orientation of groups of three dots is at an angle (and not vertical)

Card 3
Scoring criterion: Marked correct if:
Central transverse line of dots is parallel to horizontal axis of paper

Card 5
Scoring criterion: Marked correct if:
(a) Main part of figure is semi-circular in shape, and
(b) Correct contact is made between line and main figure

Card 6
Scoring criterion: Marked correct if:
Angle X is within the limits of $70^\circ \pm 10^\circ$
XII. Experimental Visual Recognition Test
(iii) Experimental Visual Recognition test (see Illustration XII)

This test was made up of eight sets of cards, 3" x 3" in dimension. Each set contained from six to eight cards, each card having one geometric design drawn on it in Indian ink. To protect the card, the surface was covered with a matt opaque self-adhesive cover.

The figures in the first six sets of cards were adapted from Benton designs. Each set was constructed to differ in specific ways from the other sets, e.g. closure of figure v. non-closure; internal detail v. no internal detail; upright orientation v. rotation.

Sets 7 and 8 were the same as those used by Taylor and Wales (1970) to study form discrimination in pre-school children. Subjects had to pick out from the set the card which corresponded to the standard they had been very briefly shown. They then had to pick out two more cards which were as similar as possible to the standard. The procedure was repeated for each card in each set (a total of 55 cards). The subjects' choices were noted on a standard sheet.
Experimental Formboards Test

XIII. Practice Board (above) As presented to subject
     (below) Completed
(iv) Experimental Formboards test

The apparatus consisted of one practice and four experimental wooden formboards. The former (see Illustration XIII) was of fairly simple design and contained pieces having both curved and straight edges.
XIV. Experimental Formboards Test

Series 1  (above)  Large & Small Formboards  (below)  Small Formboard showing alternative placements.  (N.B. Letters A, B, C and D superimposed in photograph only for identification purposes)
The experimental formboards constituted two series. Series I (see Illustration XIV) involved two formboards which differed in size alone. They had similar circular outlines and similarly shaped pieces. These pieces contained no straight line angles but only angles formed by intersecting arcs. As may be seen from the illustration, there were three possible placements for the pieces.
XV. Experimental Formboards Test

Series 2  (above) Cross
         (below) Diamond
Series 2 (see Illustration XV) involved two formboards of different outlines. The pieces to be fitted into the boards were all made up of acute and/or right angles.
The subject was introduced to the test using the practice formboard. She had first to fit the pieces into the board visually; she was then blindfolded and asked to repeat the task using touch alone (haptically). Series 1 was then presented, with the first board always presented visually. The order of the subsequent three boards was randomised, in an attempt to cancel out learning effects across the group. Series 2 was similarly presented, except that the first encounter with the boards in this series could be haptic.

The order in which the boards were administered and the time taken (in seconds) to complete each board were noted on the standard sheet. For Series 1 the placement of the pieces was noted as well. From this an approximate measure of "placement consistency" across the four boards was achieved by assigning a rating to each individual in line with those given below (Table VIII).
Table VIII  Rating Scale devised to assess "placement consistency"

(Where X and 0 represent different sets of similar placements;
Blank indicates non-X; non-0.)

<table>
<thead>
<tr>
<th>FORMBOARD 1</th>
<th>FORMBOARD 2</th>
<th>FORMBOARD 3</th>
<th>FORMBOARD 4</th>
<th>RATING</th>
</tr>
</thead>
<tbody>
<tr>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>6</td>
</tr>
<tr>
<td>X</td>
<td>X</td>
<td>X</td>
<td></td>
<td>5</td>
</tr>
<tr>
<td>X</td>
<td>X</td>
<td>X</td>
<td>X</td>
<td>5</td>
</tr>
<tr>
<td>X</td>
<td></td>
<td>X</td>
<td>X</td>
<td>4</td>
</tr>
<tr>
<td>X</td>
<td></td>
<td></td>
<td>X</td>
<td>4</td>
</tr>
<tr>
<td>X</td>
<td>X</td>
<td></td>
<td>0</td>
<td>3</td>
</tr>
<tr>
<td>0</td>
<td>0</td>
<td>X</td>
<td>X</td>
<td>3</td>
</tr>
<tr>
<td>0</td>
<td>X</td>
<td>X</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>X</td>
<td>X</td>
<td></td>
<td></td>
<td>1</td>
</tr>
</tbody>
</table>
Procedure

Experimental group

The Benton Visual Retention test was administered to 24 subjects during the first testing session. It always succeeded the intelligence test. The remaining three tests were administered during the second, or often the third, testing session, and the order tended to be rather more random. The Bender Visual Motor Gestalt test was administered to 20 subjects; the experimental Visual Recognition test to 15 subjects; and the experimental Formboards test to 13 subjects. The discrepancy in the numbers of subjects to whom the tests were administered was accounted for by loss of subjects during the later stages of testing, as already mentioned.

Control group

The four tests were administered to controls corresponding in number to the subjects in the experimental group, except in the case of the controls taken for the experimental Formboards test, where 29 were employed. (It was considered advisable to obtain results from a greater number of individuals in view of the complex design of the experiment).

When all four tests were administered to a control subject (i.e. Group 3 control), it was necessary to take into account the similarity of the experimental Visual Recognition Figures to those used in the Benton test, and the resultant practice effects. For this reason the administration of the Benton Visual Retention test preceded that of the experimental Visual Recognition test to half the control group, and succeeded it for the other half.

(i) The Benton Visual Retention test

The subject was given blank sheets of paper, dimensions roughly 11 cms. x 20 cms., a pencil and a rubber. The following instructions (termed by the manual 'Administration A') were given:
"I am going to show you some cards with patterns on them. I want you to study them for ten seconds and then to draw them from memory when I take the card away." The first card was then presented for ten seconds; it was then withdrawn, and the further request made: "Now, please draw what you saw on that card." The subject drew one pattern per sheet of paper, and each attempt was taken away and turned face downwards before the next card was presented.

(ii) The Bender Visual Motor Gestalt test

The subject was given one large sheet of blank paper, dimensions roughly 32 cm x 20 cm, a pencil and a rubber. The following instructions were given:

"I am going to show you some figures which I should like you to copy on to that sheet of paper." No time limit was placed on the test.

(iii) The experimental Visual Recognition test

The subject was asked to close her eyes whilst the set of 6, 7 or 8 cards were put out in a fixed circular order (the pre-determined order was specified by letters of the alphabet). A screen was placed over this display whilst the subject was given the following instructions:

"Under this screen I have laid out a set of cards with a different pattern on each one. I shall show you a pattern on one of these cards" (indicating the pack of standard cards retained by the experimenter) "very quickly and then take the screen away. I should like you to show me the card which has the same pattern as the one you have just seen - I shall turn this card face downwards. Then I should like you to show me another which is similar. I shall turn this one face downwards too, and ask you to choose yet another which is similar to the one I showed you. Now, there are eight sets of
cards, and to prevent the test becoming too boring we shall stop after the first four sets and do some other tests, before continuing with the last four sets."

The standard cards were presented in a pre-determined fixed order denoted by serial digits on the back of the cards. Thus each card had an identification made up of a letter plus a number, e.g. E 4. The identification letters of each card chosen were noted on a prepared sheet.

(iv) The experimental Formboards test

The subject was presented with the practice formboard and its pieces separately. She was given the following instructions:

"Please will you fit these pieces into this board as quickly as you can. This is only a practice run to show you what the test involves." After the practice board had been satisfactorily completed when it was presented visually, the pieces were removed from the board and the subject was blindfolded. She was then asked to fit the pieces into the board again.

The Series 1 experimental formboards were then presented, in one of the sequences given in the schedule below.
Schedule of Formboard sequences (Series 1)

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>$V_L$</td>
<td>$V_L$</td>
<td>$V_L$</td>
<td>$V_L$</td>
</tr>
<tr>
<td>2</td>
<td>$H_S$</td>
<td>$H_L$</td>
<td>$H_L$</td>
<td>$V_S$</td>
</tr>
<tr>
<td>3</td>
<td>$H_L$</td>
<td>$H_S$</td>
<td>$V_S$</td>
<td>$H_S$</td>
</tr>
<tr>
<td>4</td>
<td>$V_S$</td>
<td>$V_S$</td>
<td>$H_S$</td>
<td>$H_L$</td>
</tr>
</tbody>
</table>

| 5 | $V_S$ | $V_S$ | $V_S$ | $V_S$ |
| 6 | $H_S$ | $H_L$ | $H_L$ | $V_L$ |
| 7 | $H_L$ | $H_S$ | $V_L$ | $H_S$ |
| 8 | $V_L$ | $V_L$ | $H_S$ | $H_L$ |

Where $V$ = visual
$H$ = haptic
$L$ = large
$S$ = small

Thus the subject to whom Sequence 3 had been allotted would complete the large circular formboard visually first; then the large one haptically (blindfolded, by touch); then the small one visually, and, lastly, the small one haptically. It will be noted that the
first presentation was always visual, since it was considered inadvisable to present the circular boards haptically at the beginning of the experiment, because they were very difficult and tended to influence negatively the subject's performance on the boards which followed.

The time taken to complete each board, and the layout of the pieces on completion of the board, were noted on a standard form.

The formboards in Series 2 were then administered, again following a schedule (see below). In this series it was possible that the first encounter with a board could be haptic. The time taken to complete each board was noted on a standard form.

<table>
<thead>
<tr>
<th></th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
</tr>
</thead>
<tbody>
<tr>
<td>H&lt;sub&gt;C&lt;/sub&gt;</td>
<td>V&lt;sub&gt;D&lt;/sub&gt;</td>
<td>H&lt;sub&gt;D&lt;/sub&gt;</td>
<td>V&lt;sub&gt;C&lt;/sub&gt;</td>
<td></td>
</tr>
<tr>
<td>V&lt;sub&gt;C&lt;/sub&gt;</td>
<td>H&lt;sub&gt;D&lt;/sub&gt;</td>
<td>V&lt;sub&gt;D&lt;/sub&gt;</td>
<td>H&lt;sub&gt;D&lt;/sub&gt;</td>
<td>C</td>
</tr>
<tr>
<td>V&lt;sub&gt;D&lt;/sub&gt;</td>
<td>H&lt;sub&gt;C&lt;/sub&gt;</td>
<td>V&lt;sub&gt;C&lt;/sub&gt;</td>
<td>H&lt;sub&gt;D&lt;/sub&gt;</td>
<td></td>
</tr>
<tr>
<td>H&lt;sub&gt;D&lt;/sub&gt;</td>
<td>V&lt;sub&gt;C&lt;/sub&gt;</td>
<td>H&lt;sub&gt;C&lt;/sub&gt;</td>
<td>V&lt;sub&gt;D&lt;/sub&gt;</td>
<td></td>
</tr>
</tbody>
</table>

Where V = visual

H = haptic

D = diamond

C = cross
Statistical Analysis of Results

The following statistics were then applied to the results:

For Hypothesis IV/1

(a) "Number correct" scores and (b) "Error" scores of the experimental and control groups on the Benton Visual Retention test were compared, using the Mann-Whitney U test.

For Hypothesis IV/2

The types of errors were expressed as percentages of the total error score for the experimental and control groups, and compared, using the $X^2$ test for K independent samples (Siegel, 1956).

For Hypothesis IV/3

To test this hypothesis comparison was made between the scores obtained on each of the (a) four cards contained in Category 1 and (b) five cards contained in Category 2, by the experimental and control groups, using the $X^2$ test for two independent samples (Siegel, 1956).

For Hypothesis IV/4

The number of recognition errors scored on the first choice experimental Visual Recognition test were compared for the experimental and control groups, using the Mann-Whitney U test.

For Hypothesis IV/5

Size (i.e. large v. small) and modality (i.e. visual v. haptic) performances on the Series 1 experimental Formboards test were compared for the experimental and control groups, using a $2^3$ factorial design analysis of variance (Hays, 1965).

For Hypothesis IV/6

Modality (i.e. visual v. haptic) performances on the Series 2 experimental Formboards [(i) the diamond, and (ii) the cross] were compared for the experimental and control groups, using a $2^2$ factorial design analysis of variance (Hays, 1965).
HYPOTHESES

On the basis of the above discussion the following hypotheses were formulated:

For the Benton Visual Retention test

IV/1 That the experimental group will have
(a) significantly lower "number correct" scores than the control group,
(b) significantly greater "error" scores than the control group.

IV/2 That the experimental group will differ significantly on the types of qualitative errors they commit, when the errors are classified in the manner described on p. 89.

For the Bender Visual Motor Gestalt test

IV/3 That the null hypothesis stands, in that there will be no significant differences between the experimental and control groups on any of the measures obtained from the scoring procedure previously described (p. 90).

For the experimental Visual Recognition test

IV/4 That the experimental group will commit a significantly greater number of errors of recognition than will the control group.

For the experimental Formboards test

IV/5 That for Series 1 the experimental group will not differ significantly from the control group in the amount of time
they require to complete the two formboards across the four administrations.

That for Series 2 the experimental group will not differ significantly from the control group in the amount of time required to complete

(a) the Cross Formboard, and
(b) the Diamond Formboard,
   (i) visually, and
   (ii) haptically.
RESULTS

Hypothesis IV/1

The results supported both parts of this hypothesis. Table IX shows the mean "number correct" and "error" scores for the two groups.

Table IX  Mean "number correct" and "error" scores of the experimental and control groups on the Benton Visual Retention test

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss</th>
<th>Control Ss</th>
<th>U</th>
<th>p*</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>24</td>
<td>24</td>
<td></td>
<td></td>
</tr>
<tr>
<td>&quot;Number correct&quot; (mean)</td>
<td>5.0</td>
<td>7.6</td>
<td>4.0</td>
<td>&lt;.00003</td>
</tr>
<tr>
<td>&quot;Errors&quot; (mean)</td>
<td>8.5</td>
<td>3.3</td>
<td>4.12</td>
<td>&lt;.00003</td>
</tr>
</tbody>
</table>

* 1-tailed test

These results indicate that the experimental group obtained correct scores on significantly fewer designs on the Benton Visual Retention test than did the control group, and as a consequence, gained a significantly greater number of errors.

Hypothesis IV/2

The results did not support this hypothesis. Table X shows the percentage error scores split up in terms of the six qualitative types of error (as given in the manual) for the two groups.

Table X  Percentage error scores for six qualitative categories of the Benton Visual Retention test for experimental and control groups

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss</th>
<th>Control Ss</th>
<th>x^2</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>24</td>
<td>24</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Omissions</td>
<td>5.9%</td>
<td>8.7%</td>
<td>8.118</td>
<td>&lt;.2</td>
</tr>
<tr>
<td>Distortions</td>
<td>47.5%</td>
<td>47.5%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Perseverations</td>
<td>10.8%</td>
<td>16.3%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Rotations</td>
<td>19.1%</td>
<td>17.5%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Misplacements</td>
<td>10.3%</td>
<td>10.0%</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Size Errors</td>
<td>6.4%</td>
<td>0%</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
This result indicates that the experimental group had no greater tendency towards committing any one particular type of error than had the control group.

Hypothesis IV/3
(a) The null hypothesis was not upheld for results on any of the four cards included in Category 1. Table XI shows the total number of subjects within the experimental and control groups who scored correctly on the criteria previously tested for the four cards.

Table XI  Number of experimental and control subjects who scored correctly on Category 1 designs (Contiguity and Overlap) of the Bender Visual Motor Gestalt test

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>$x^2$</th>
<th>$p^{**}$</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>20</td>
<td>20</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Card A</td>
<td>10</td>
<td>18</td>
<td>5.83</td>
<td>&lt;.02</td>
</tr>
<tr>
<td>Card 4</td>
<td>9</td>
<td>16</td>
<td>3.84</td>
<td>&lt;.05</td>
</tr>
<tr>
<td>Card 7</td>
<td>4</td>
<td>13</td>
<td>6.55</td>
<td>&lt;.02</td>
</tr>
<tr>
<td>Card 8</td>
<td>7</td>
<td>16</td>
<td>6.55</td>
<td>&lt;.02</td>
</tr>
</tbody>
</table>

** 2-tailed test

These figures indicate that the experimental group made a significantly greater number of errors when copying designs which involved the overlapping or contiguity of two figures than did the control group.

(b) The null hypothesis was generally supported from the results gained from analysis of the five designs included in Category 2. Table XII shows the total number of subjects within the experimental and control groups who scored correctly on the criteria previously listed for the five cards.
Table XII  Number of experimental and control subjects who scored
correctly on Category 2 designs (Orientation)
of the Bender Visual Motor Gestalt test

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>X²</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Card 1</td>
<td>20</td>
<td>20</td>
<td>.28</td>
<td>N.S.</td>
</tr>
<tr>
<td>Card 2</td>
<td>17</td>
<td>19</td>
<td>9.80</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>Card 3</td>
<td>6</td>
<td>17</td>
<td>2.75</td>
<td>N.S.</td>
</tr>
<tr>
<td>Card 5</td>
<td>10</td>
<td>16</td>
<td>~0</td>
<td>N.S.</td>
</tr>
<tr>
<td>Card 6</td>
<td>18</td>
<td>19</td>
<td>~0</td>
<td>N.S.</td>
</tr>
</tbody>
</table>

** 2-tailed test

These results indicate that, on the whole, the experimental
group was not significantly different from the control group in
copying figures involving orientation. An exception to this finding
was the result on Card 2, where the experimental group failed
significantly more frequently than the control group to copy the
design correctly.

Hypothesis IV/4

The results confirmed this hypothesis. Table XIII shows the
mean number of first choice errors of recognition gained by the two
groups on the experimental Visual Recognition test.

Table XIII  Mean number of errors scored by the experimental and
control groups on the experimental Visual Recognition test

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>U</th>
<th>p*</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>15</td>
<td>15</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Number of errors (mean)</td>
<td>11.3</td>
<td>5.7</td>
<td>60.5</td>
<td>&lt;.025</td>
</tr>
</tbody>
</table>

* 1-tailed test
Graph VI. *Series 1 Formboards*

*Graphical Representation of Means with 5% confidence limits*
This result indicates that the experimental group committed significantly more errors of recognition than did the control group on the experimental Visual Recognition test.

**Hypothesis IV/5**

The results did not support this hypothesis. Table XIV gives the mean completion times for the four presentations of the formboards (together with their standard deviations, S.D.) for the experimental and control groups. From these is obtained Graph VI, which demonstrates the means and confidence limits of the four measures for the two groups.

**Table XIV**  
Mean completion times for the four presentations of the formboards for the experimental and control groups

<table>
<thead>
<tr>
<th></th>
<th>Experimental S.</th>
<th>Control Ss.</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean</td>
<td>S.D.</td>
</tr>
<tr>
<td>Visual large (V_L)</td>
<td>27.7</td>
<td>20.06</td>
</tr>
<tr>
<td>&quot; small (V_S)</td>
<td>35.8</td>
<td>36.59</td>
</tr>
<tr>
<td>Haptic large (H_L)</td>
<td>94.6</td>
<td>79.12</td>
</tr>
<tr>
<td>&quot; small (H_S)</td>
<td>114.3</td>
<td>85.74</td>
</tr>
</tbody>
</table>

From these results the $2^3$ factorial design analysis of variance (shown below in Table XV) was set up, with the treatments of size (large v. small) and modality (visual v haptic) applied to the experimental and control groups.
Table XV  
$2^3$ factorial design analysis of variance applied to Series 1 experimental Formboards

<table>
<thead>
<tr>
<th>Source</th>
<th>Sum of Squares</th>
<th>df.</th>
<th>$V$</th>
<th>$F$</th>
<th>$p$</th>
</tr>
</thead>
<tbody>
<tr>
<td>Between size</td>
<td>5351.24</td>
<td>1</td>
<td>5351.24</td>
<td>2.681</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; groups</td>
<td>20935.43</td>
<td>1</td>
<td>20935.43</td>
<td>10.487</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>&quot; modality</td>
<td>98546.49</td>
<td>1</td>
<td>98546.49</td>
<td>49.364</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>Size x group</td>
<td>122.1</td>
<td>1</td>
<td>122.1</td>
<td>.061</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; x modality</td>
<td>3487.94</td>
<td>1</td>
<td>3487.94</td>
<td>1.747</td>
<td>N.S.</td>
</tr>
<tr>
<td>Group x modality</td>
<td>11679.85</td>
<td>1</td>
<td>11679.85</td>
<td>5.851</td>
<td>&lt;.05</td>
</tr>
<tr>
<td>Size x gp.x mdlty.</td>
<td>324.6</td>
<td>1</td>
<td>324.6</td>
<td>.162</td>
<td>N.S.</td>
</tr>
<tr>
<td>Between cells</td>
<td>139798.45</td>
<td>7</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Within cells</td>
<td>319085.73</td>
<td>159</td>
<td>2006.83</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Total  458884.18  166

(Note: It having been established that the second order interaction was not statistically significant, a new error term of 319410.33, i.e. 319085.73 + 324.6, with 160 degrees of freedom was adopted for testing against first order and main effects).

This table shows that whilst the size effect (i.e. large v. small comparison) is not significant, the other two main effects of group (i.e. experimental v. control comparison) and modality (i.e. visual v. haptic comparison) are statistically significant. There is also a statistically significant first order interaction between group and modality effects.

For Hypothesis IV/6

The results supported this hypothesis in part but not entirely. Table XVI shows the mean completion times for the Series 2 Formboards (the diamond and the cross) performed visually and haptically for the experimental and control groups.
Table XVI  Mean completion times for the Series 2 Formboards for the experimental and control groups under different modality treatments

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>13</td>
<td>29</td>
</tr>
<tr>
<td>Mean</td>
<td>S.D.</td>
<td>Mean</td>
</tr>
<tr>
<td>Diamond (visual)</td>
<td>32.9 31.71</td>
<td>15.9 16.24</td>
</tr>
<tr>
<td>&quot; (haptic)</td>
<td>97.8 113.98</td>
<td>45.6 35.97</td>
</tr>
<tr>
<td>Cross (visual)</td>
<td>9.2 2.83</td>
<td>8.5 3.31</td>
</tr>
<tr>
<td>&quot; (haptic)</td>
<td>42.1 50.57</td>
<td>37.7 61.37</td>
</tr>
</tbody>
</table>

From these results two 2 x 2 design analyses of variance were set up, one each for the diamond and cross Formboards, with the treatment of modality (visual v. haptic) applied to the experimental and control groups in both cases (see Tables XVII and XVIII).

Table XVII  2 x 2 analysis of variance applied to Series 2 Formboards (i) diamond

<table>
<thead>
<tr>
<th>Source</th>
<th>Sum of squares</th>
<th>df.</th>
<th>V</th>
<th>F</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Between groups</td>
<td>21476.39</td>
<td>1</td>
<td>21476.39</td>
<td>71.28</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>&quot; modality</td>
<td>34526.29</td>
<td>1</td>
<td>34526.29</td>
<td>11.70</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>Group x modality</td>
<td>5558.08</td>
<td>1</td>
<td>5558.08</td>
<td>1.88</td>
<td>N.S.</td>
</tr>
<tr>
<td>Between cells</td>
<td>61560.76</td>
<td>3</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Within cells</td>
<td>227138.13</td>
<td>77</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>288698.89</td>
<td>83</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
These tables show that, whilst the significant difference between the modality treatments persists over the total group of subjects, a significant difference exists between the experimental and control groups on the diamond Formboard only. There are no significant interaction effects.
DISCUSSION

The Benton Visual Retention Test

Results confirmed previous studies which had used this test on individuals with Turner's syndrome. Very significant differences between the experimental and control groups were demonstrated on both the measures involved. Thus individuals with Turner's syndrome were found to score fewer correct (out of a maximum of ten), and, correspondingly, made a far greater number of errors in reproducing geometric designs from memory. Clinically this result indicated that individuals with Turner's syndrome demonstrate some sort of organic impairment which particularly affects visual short-term memory processes. Of relevance in this connection it may be recalled that the experimental group also scored significantly lower than the control group on Cohen's third factor of "Memory". However, it is doubtful whether this result reflects a specific short-term memory defect, since the difference seems attributable to the Arithmetic sub-test scores, and not to those in the Digit Span sub-test. The latter is commonly used in assessing short-term memory, and it may be noted that there existed no significant difference between the control and experimental groups on this measure. It would seem to be implied, therefore, that it is a defect in the visual perceptive system, and not of short-term memory, which is concerned in this context.

It should be noted that not all the individuals with Turner's syndrome experienced this difficulty with the test; one person in the sample completed the test without any errors, and four others scored errors lower in number than the mean total of the control group.

An unexpected negative result was that obtained when percentage error scores on the six qualitative categories were compared. In particular it might have been hypothesized that the Distortion sub-category would be over-subscribed to by the experimental group. This proved not to be the case, although it could be suggested that this
category in itself covers too broad a definition to differentiate between the two groups of subjects. For example, a simple case is that of the second design, a hexagon. As will be seen by reference to the illustrations contained in the Appendix, errors by the experimental group included the typical one of failure to reproduce the obtuse angles correctly, producing instead a line forming an acute angle to the horizontal one already drawn. Controls, on the other hand, if they failed to reproduce the figure correctly, tended to include two extra sides, making an octagon. Both these errors are included within the Distortion sub-category, sub-classified as Substitution errors, and, indeed, it is difficult to see how any other method could be adopted.

Other attempts to differentiate between the performances of experimental and control subjects also proved fruitless, e.g. ordering the ten designs in terms of error scores on each indicated a similar order of difficulty for both groups (i.e. design 10 proved the most difficult; design 7 was found to be the next most difficult; then, in order, design 9, etc.). There was no difference between the two groups when the percentage difficulty of each of the three single figures making up the complex design was compared, except in the cases of designs 8 and 9, where the figures with "half-moon" internal details were particularly poorly reproduced by the experimental group, who seemed unable to coincide the points in the correct position (see Appendix for examples). This suggests some sort of difficulty in achieving contiguity or positioning, and might bear some relationship to the difficulties experienced with the Object Assembly sub-test.

It has already been noted that some of the experimental subjects were well able to perform adequately on the Benton Visual Retention test. In this context it is interesting to observe a high positive correlation for the experimental group (Spearman rho = 0.98, p < .01) between verbal skills and ability to reproduce the Benton Visual
Retention test designs correctly. This might indicate, in terms of the model suggested in the previous chapter, that some individuals with Turner's syndrome, with a potentially high IQ, develop adaptive processes to overcome characteristic difficulties experienced in performance tests. Such processes might involve some kind of verbal encoding of visual material, a suggestion which is supported by significant correlations between VIQ and other tests involving visual material, namely the Bender Visual Motor Gestalt test (Spearman rho = 0.38, p < .05) and the experimental Visual Recognition test (Spearman rho = 0.63, p < .01).

The Bender Visual Motor Gestalt test

The utilisation of this test, together with the modified method of scoring, produced interesting qualitative results.

The fact that the experimental group made nearly three times the number of copying errors of the control group indicated that perceptual errors occurred at the input and reproduction stages rather than in any system of visual short-term memory (as could be suggested from the results on the Benton test). Whilst the author does not claim that the modified scoring procedure evolved for the purpose of this thesis discriminates between "wrong" and "right" copies, it does highlight the very obvious difficulties in copying experienced by individuals with Turner's syndrome. These are particularly marked in connection with designs involving two separate figures bearing some kind of contiguous or overlapping relationship with each other. There is no evidence to suggest that the 'gestalt' itself is disrupted; the designs are broken up into their component figures in the same way as are those drawn by the controls, but individuals with Turner's syndrome fail to integrate the two figures into a correct reproduction of the original design (see Appendix, for examples). Whilst this task appears to involve part/whole relationships (as does Object Assembly), it differs from the W.A.I.S. sub-test in that it provides the subject with an externalised visual schema which can always be checked against the final reproduction.
Performance on the Bender Visual Motor Gestalt test seemed to correlate with that on the Benton Visual Retention test (Spearman rho = 0.40, p < .05), which could indicate that a similar perceptual ability or disability is involved in performance on the two tests.

The experimental Visual Recognition test

Results indicated that the experimental group made a significantly larger number of errors of recognition than did the control group, thus demonstrating that the difficulty seems to be one of input (perception) rather than of output (motor).

For the purpose of singling out the Sets and single items within the Sets which caused most difficulty for the experimental group, it was considered valid to exclude results from three subjects included within the control group. These contributed over 70% of the control error score, and it seemed that circumstances outwith the testing situation (a severe attack of influenza; an unwanted pregnancy; and suspicion that the author had something to do with the child-fostering services) were influencing performances in such a way as to confound any comparison made with the experimental group. It should be remembered that the significant result that the experimental group gained a greater number of recognition errors was obtained from comparison with a control group of 15 subjects. Further comparisons considered in the following discussion are therefore based on 12 subjects (and 15 of the experimental group).

The designs constituting Set III were by far the best discriminant between experimental and control groups. The Fisher Exact Probability test indicated differences (significant at .025 level) for Cards A2, B4, C3 and Gl. They were most commonly confused with their mirror images, i.e. C3 for Gl, A2 for B4 - which was also true of those controls who made recognition errors on this Set. As far as the specific Set differences were concerned, Set III differed from
other Sets in consisting of closed figures, five of which had complete internal details (which, viewed another way, resulted in two sub-figures, bearing a contiguous integrated relationship to each other, in forming a larger whole figure). On the other hand, Card D7, although constructed on the same principle, failed to discriminate between the experimental and control groups.

Sets I, IV, V, VII and VIII did not discriminate between the groups. One card, Fl (in Set II) was particularly poorly recognised ($p < .001$), as was Card D6 ($p < .05$). In Set VI Card C4 discriminated between experimental and control subjects ($p < .01$).

Without exception the designs which caused recognition errors for the experimental group tended to be confused with those designs selected as second choice (or "most similar") by the control group. As a corollary, the correct design was consequently the most often selected as second choice (or "most similar") by the experimental group. This seems to indicate that the internal categorisation methods employed by the experimental group are similar to those of control subjects. It is possible that they employ similar coding methods for perceptual and storage processing, and that confusion and error arise from pre- and post-coding procedures.

Further insight is gained by comparing individual performances on those Benton figures in Designs 7 and 10 which relate to Set III of the Visual Recognition test. Nine of the experimental group both reproduced and recalled the designs incorrectly. What is more interesting is that five others were able to reproduce the Benton designs correctly, but failed on the Visual Recognition figures, and not one subject made mistakes on the Benton test whilst performing correctly on the Visual Recognition task. It seems strange that what would seem to constitute a more difficult task in terms of complexity of the initial design and the method of recall required (i.e. reproduction, as in the Benton test) was, in fact, performed better by the experimental group than was the less complicated Visual
Recognition task. The only feature of test administration which might explain this differentiation is that of exposure time of the test material. The Visual Recognition cards were shown for a very brief period of time only, whilst the Benton designs had the customary ten seconds’ exposure. It could be hypothesised that, had the Visual Recognition test not been administered manually, but tachistoscopically (thus cutting down the exposure time within controlled limits), this difference would have been even more apparent. It would seem possible that individuals with Turner’s syndrome require a longer time to assimilate a geometric design, and this might be related to the psychomotor retardation postulated in the previous chapter. Indeed, performance on the Visual Recognition test correlates significantly with that on the Digit Symbol sub-test (Spearman rho = .53, p < .05), indicating that the more retarded of the experimental group make a greater number of recognition errors. Evidence to support this idea could be taken from the significant correlation which exists for the control group between the Benton Visual Retention test errors and Visual Recognition test errors (Spearman rho = .68, p < .01), but which does not exist for the experimental group (Spearman rho = .40, N.S.). This would seem to indicate two facts:

(i) that the Visual Recognition task, in correlating with the Benton Visual Retention test for controls, has validity, in that it measures what it was devised to do;

(ii) that, since this relationship is not replicated by the experimental group, it could be postulated that some factor, namely the deficient registration time, is interfering with their ability to act in the same way as the control group on the recognition task.

Finally, it would be interesting to repeat the same type of test, with the introduction of certain modifications, e.g. tachistoscopic presentation; inclusion of further sets which emphasised the
"parts integrated into a whole" theme. The administration of the test might also be altered to introduce a ranking procedure of "most like" (i.e. correct match for standard) graded through 6, 7 or 8 cards comprising the Set, to "least like", in place of the present method of noting choices 1, 2 and 3. This would greatly facilitate statistical analysis, but would require the number of Sets in the test to be reduced in order to be acceptable.

The experimental Formboards Test

The results given by the $2^3$ analysis of variance of the Series 1 circular formboards, taken together with the graphical representation of the results, indicated that performance differed between the two groups of subjects, in that the experimental group required more time to complete the series of formboards (i.e. the total of four presentations) than did the controls, and that both groups required more time to complete the formboards haptically than visually. The significant interaction effect indicates that the two main effect differences were not independent of each other. Whilst interaction effects are difficult to interpret in terms of practical parameters, it could be suggested that this interaction indicates that over the sequence $V_L - V_S - H_L - H_S$ the time taken to complete the tasks became progressively longer for experimental and control groups, but that this progression was at a greater rate for individuals with Turner's syndrome than it was for controls.

Before proceeding further it should be noted that the customary procedures of investigation of the suitability of the Formboards data for the use of the analysis of variance technique (using Bartlett's test of homogeneity of variances) indicated that the assumption of homogeneity was not valid. For the experimental group the sampling statistic was $B = 26.55$; for the control group $B = 19.50$. However, transforming the raw data to log normal scores achieved suitable non-significant Bartlett's tests. Repeating the $2^3$ analysis of
variance procedure on log normal scores produced similar main effects differences, significant for between groups and between modalities variances, but not for between sizes. The interaction effect was also not significant, a consequence which was not unexpected, since the result of employing log normal scores is to reduce the variance available for calculation of interaction effects.

Whilst accepting these limitations, it may be recalled that the F ratio has been shown to be a very robust measure (Norton, cited in Lindquist, 1953), and it would seem inappropriate to dismiss what appears to be an interesting result on the basis of analysis of transformed data, a statistical technique which in itself has its severe critics.

In retaining the interaction effect as valid it might be postulated that, whilst the increase in difficulty over the sequence of formboards demonstrated by the control group was the natural concomitant of performing an unaccustomed task using relatively novel abilities, this was also the case for the experimental group, but that another variable, specific to them, played a part. It might be suggested that this latter variable involves an inability to learn, or retardation in learning, from practice. Further evidence for this hypothesis is provided by considering the "placement consistency" measure adopted (see p. 98). It may be recalled that this rating depended on the consistency with which the subject employed the same manner of placement (out of three choices) across the four presentations of the formboards. It was noted that there was a non-significant trend for the experimental group to be less consistent than the control group ($X^2 = 1.69; p < .1$). The impression gained during testing was that fitting the pieces together using either modality was generally far more haphazard in individuals with Turner's syndrome than it was in control subjects, although on the whole the former were more motivated to complete the task. A further index of this haphazardness is obtained by comparing the choices of placement most frequently made
by control and experimental subjects. It was demonstrated that a significant difference existed between the two groups' choices ($X^2 = 7.19, p < .025$). Whilst the controls selected the balanced placement (see p. 96, top Illustration), the experimental subjects most often used the unbalanced one (p. 96, lower right-hand Illustration). Once again it might be postulated that there exists a poor internalised scheme employed by the experimental group to integrate the pieces of the formboard into the "whole" outline. In this way it might be expected to correlate with performance on Object Assembly, a correlation which is found to be significant only for the formboards performed visually (visual formboards: Spearman rho = .89, $p < .01$; haptic formboards: Spearman rho = .19, N.S.). This would indicate that it is the visual component of the test which is important.

Finally, it should be noted that the experimental group's deficient performance on the formboards is not related to the psychomotor retardation previously postulated, since there are non-significant correlations between visual/haptic times and Digit Symbol scores for both experimental and control groups.

Consideration of the results obtained from the Series 2 Formboards has also to be limited by the factor of non-homogeneity already discussed with reference to the Series 1 Formboards. The results achieved by analysing the Series 2 Formboards raw data reflected those obtained from Series 1, in showing a statistically significant difference between visual and haptic performances on both the diamond and cross formboards. A statistically significant difference existed between the scores of the experimental and control groups on the diamond formboard only, and not on the cross formboard. From scrutiny of the times taken to complete the latter, it seems fairly clear that, as a test, it was a much easier shape than the other formboards in the two series. The fact that there was no difference between the two groups' performances on it would indicate that the experimental group...
were able to emulate the control group in their performance, and it was only as the complexity of the task increased that they performed at a lower level than normal individuals.

With reference to the diamond formboard, it is interesting to note that non-parametric comparison of the completion times of the experimental and control groups for the boards performed visually and haptically indicated a significant difference for the visual presentation alone (Mann-Whitney $U = 72; z = 3.17; p < .001$). This would seem to indicate that there is a particular disability associated with the visual performance of the test but not with the haptic.
CHAPTER V

COLOUR VISION

INTRODUCTION

Theories of Colour Vision

The mechanisms involved in colour vision have been variously described on a considerable scale, but there still does not exist a single proven physiological explanation which is sufficient to cover all colour vision phenomena.

Theories of colour vision as described by Lakowski (1969) are presented in a simplified form which will serve to introduce the terms and concepts which may then be employed in considering the results obtained from the individuals under discussion.

Historically speaking, there are two theories, which at one time were set up in opposition to each other, but which now may well prove of greater value if amalgamated into a more complex stage model of colour vision.

The trichromatic theory [after Young, (1807), and Helmholtz, (1892)] postulates the existence of three sets of response mechanisms in the retina, each set responding independently to the primary colours of red, green and blue. Any other colours experienced are theoretically produced by summing two or more of the primaries, e.g. yellow is produced by the sum of red and green responses, and white by the sum of equal amounts of red, green and blue. It is known, however, that colour matchings with the three primaries cannot reproduce all colours, so the model can clearly be an approximation only. In more recent research on the physiology of the eye, and in particular, the retina, it has been possible to isolate structures (cones) which could well correspond to the receptors postulated in this theory. This suggestion has been supported as a consequence
of measuring the absorption peaks of the cones, and obtaining results which roughly, but not entirely, correspond to the three colour response characteristics postulated by Young and Helmholtz (reviewed by Dartnall, 1970).

The second approach, formulated by Hering (1878), puts forward the idea of a visual mechanism involving three opponent-pairs of colour sensations (rather than stimuli) - black/white, red/green, and yellow/blue. Equal stimulation of the two members of a pair gives rise to a neutral or achromatic response. The theory assumes that the light is absorbed by photopigments in the receptors, and this absorption then triggers off activity in the rest of the visual system, it being this activity which is responsible for the colours we see.

More recently stage models using both these basic theories of colour vision have been adopted. Thus, whilst the Young-Helmholtz three components model may be taken as valid at the receptor stage, the sensations from these are deemed to be organised in the manner of Hering's theory. There still persists controversy regarding the precise stage of the visual process at which the organisation occurs; different exponents postulate different structures, e.g. the retina, the optic nerve, and occipital lobe. Further details on the different theories of stage models may be seen in Judd (1966).

**Colour defects**

The normal observer (the trichromat) is able to make light/dark, red/green and yellow/blue discriminations without difficulty. On the other hand, the colour-blind observer (the dichromat) requires only two primaries to match with other colours. The two most common types of colour-blindness occur in the red/green confusion category and are termed protanopic and deutanopic. People with protanopia lack red-absorbing pigmented cones in the central area of the retina (Kalmus, 1965). Their
sensitivity to light is also affected in that they require the red colour to be steadily darkened towards the red end of the spectrum before matching it with yellow. Deuteranopic colour-blindness cannot be explained by a lack of a particular set of receptors of any type, but it is suggested that in this condition there is only one common class of cones, which therefore fails to distinguish between red and green stimuli. This explanation covers the fact that, in addition, deuteranopes do not experience brightness loss, as the protans do. Since the conditions described above are postulated to affect the red and green cones, it seems likely that there may be a similar defect associated with the blue cones. This condition, known as tritanopia, has been found difficult to isolate.

If the three primaries are required for colour matching, but the intensity ratios are different from those of the standard observer, anomalous colour vision is said to be present. Anomalous trichromats can be divided into:

(i) protanomalous - those who require more red stimulation
(ii) deuteranomalous - " " " green "
(iii) tritanomalous - " " " blue "

Colour defects may be either congenital or acquired.

**Congenital defects**

It has long been established that colour-blindness of the congenital type is sex-linked. This means that the transmission of defective colour vision is effected by an abnormal gene on the X chromosome. In addition, this is said to be a recessive characteristic, in that it becomes manifest in the female only if the same abnormal gene occurs on both X chromosomes. Women who possess only one abnormal gene are called heterozygotes, and have a 50:50 chance of passing the characteristic on to their offspring, but do not always manifest colour vision deficiency themselves.
Pickford (1951) stated that colour defects were incompletely recessive, since heterozygotes for the abnormal gene usually had small red/green defects (termed Schmidt's sign) or might actually manifest the major defects themselves. Kalmus (1965) also described the occurrence of Schmidt's sign in women, heterozygous for protanopia, who required to dim orange and red colours in order to achieve a match with yellow. This, he explained, was taken to indicate that the brightness component of the red-cone path was impaired, rather than that any defect existed in the number or structure of the red cones themselves.

**Acquired defects**

This type of defect differs from the congenital type in that the assumption is made that at some time the whole of the colour vision apparatus was normal, from the genes concerned with colour vision to the retinal receptors and more central structures. In the acquisition of colour vision defects some form of pathology is therefore involved.

Ageing effects have been suggested as one of the pathological variables. Some of the minor alterations in colour vision which occur with age may be attributed to lens changes. Lakowski (1962) found that there was a gradual loss of fine colour discrimination and an increase in the number of people whose performances resembled that of major defectives. Yellow filters used to mimic lens changes produced some similar alterations in colour vision of normal young subjects, but these alterations could not account for the total amount of deterioration noted. Lakowski (1969) suggested, therefore, that there might be additional pathological changes which affect some people after, say, about 30 years of age, which accelerate the senile degenerative processes.

In acquired defects in general, i.e. those having some pathological basis, the results of colour vision testing are extremely variable and unpredictable, which is not, of course, true of congenital defects.
There is other pathology listed by Kalmus (1965) besides lens discolouration; lack or mal-distribution of one or several cone pigments, or of pigmented cones or rods themselves; faulty neural connections in the retina or the primary and secondary visual centres.

Some 500 eyes affected by different diseases were examined by Verriest (1963). He isolated four types of colour discrimination deficiency and associated them with pathology at different levels of the visual system. There is some suggestion that lesions in the outer layer of the retina lead to yellow/blue defects, whereas lesions in the inner layers, or in the optic nerve, lead to red/green defects (Lakowski, 1969).

Colour vision studies in individuals with Turner's syndrome

From the previous discussion it may be seen that the incidence of congenital patterns of colour-blindness in males (7-8%) will be much higher than that in females (.6%). This discrepancy between sexes arises from the fact that, whilst males will manifest red/green defects if the abnormal gene is present on their one and only X chromosome, the abnormal gene has to be present on both X chromosomes for such defects to be manifested in females. On the rare occasions that this does occur, the father of the propositus should be colour-blind, whilst the mother should be heterozygous for the colour-blindness gene. So, when Walls (1959) investigated, in 1952, the case of a colour-blind girl with both parents having normal colour vision, he assumed that the girl could only be termed a "manifesting heterozygote". This, as has already been stressed, was unlikely, since the recessive colour-blindness gene tends not to be manifest in women carrying the normal gene as well. Walls discussed this case along with those of other manifesting heterozygotes and remarked upon the fact that the defect produced seemed to be irregular, in not being the same as that
produced in a colour-blind male, adding that the action of the colour-blindness gene in the former case appeared to affect only the colour pathways and not the receptors.

In 1958, after the publication of the paper to be reviewed next in this Introduction (Polani et al., 1956) it occurred to Walls that the girl in question might in fact be a chromosomal male, and blood smears indicated that she was indeed chromatin negative. This led Walls to recommend that the presence of an irregular colour defect in a female should be investigated further to ascertain whether she had a 45 XO karyotype, and also that checks should be made for this irregularity in colour-blind females with a known 45 XO karyotype.

Polani et al. (1956), working with only the evidence of nuclear sexing procedures, postulated that females with ovarian agenesis and "male chromosomal sex" might be expected to carry those genetically determined characteristics more commonly found in males than in females. To investigate this hypothesis they chose to examine the incidence of red/green colour-blindness in these individuals. Using the Ishihara tables they tested 25 patients. They divided this group into two - those considered to have pure ovarian agenesis (N = 12), and those with Turner's syndrome (N = 11 + two "probables"), i.e. those having ovarian agenesis together with webbing of the neck, cubitus valgus and other variable malformations. Nuclear sexing techniques indicated that 10/12 of Group 1 and 10/11 of Group 2 were chromatin negative. Of these 20 chromatin negative individuals four were found to give such responses to the Ishihara plates as to classify them as abnormal. This figure gave the incidence of colour-blindness in Turner's syndrome as 16%, which is clearly closer to that of normal males than to that of normal females.

These results supported the popular ideas of that time concerning the genetic maleness of "females" with ovarian agenesis. However, with considerable acumen, Polani et al. also suggested "that we may be dealing with persons who have an XO pattern of sex-chromosomes".
Once the karyotype associated with Turner's syndrome had been established by Ford (1959), further studies of the incidence of colour-blindness in those individuals began. By testing for the presence of colour-blindness in the parents of a colour-blind individual with Turner's syndrome inferences about parental meiotic non-disjunction could be made. Thus Lennox (1961) reported four cases of colour-blind patients with Turner's syndrome. Since the fathers of these cases all had normal colour vision it was suggested that the mothers were heterozygous for the colour-blindness gene. It could therefore be concluded that non-disjunction had occurred in the father in all four cases. This could be represented thus:

\[
\begin{align*}
\text{(Heterozygous mother)} & \quad \text{(Normal father)} \\
\text{\(X\)} & \quad \text{\(X\)} \\
\text{\(X_C\)} & \quad \text{\(XY\)} \\
\text{\(X_C\)} & \quad \text{\(X\)} \\
\text{\(X_C\)} & \quad \text{\(XY\)}
\end{align*}
\]

\((C = \text{colour-blind})\)
Lennox also reported one case in which the father, as well as the daughter with Turner's syndrome, was colour-blind, indicating that non-disjunction had occurred in the mother, thus:

\[
\begin{array}{c}
\text{(Normal mother)} \\
XX \\
\text{OX} \\
XX
\end{array}
\quad
\begin{array}{c}
\text{(Colour-blind father)} \\
XCY \\
\text{OX} \\
XcY
\end{array}
\]

\[XcO\]

\[(C = \text{colour-blind})\]

Lennox concluded that non-disjunction can occur in either parent. Polani (1961) tentatively supported this view but remarked that the evidence for maternal non-disjunction was not clear-cut. Until it is possible to differentiate between females who are heterozygous for the colour-blindness gene and those who are normal, Lennox's conclusion is not proven, since non-disjunction could still have occurred in the colour-blind father, with the mother (now postulated to be a heterozygote) supplying the abnormal gene.

It is also important to establish as far as possible that the putative father with normal colour vision is actually the father of the patient. In just such a case Stewart (1960) in fact tested several other branches of the family (see Diagram VI) and established beyond doubt that the mother (as well as one of her
sisters) was heterozygous for the gene governing deuteranomaly. It was also claimed that minor colour discrimination defects in these individuals were established from anomaloscope testing.

Diagram VI Pedigree of family having colour-blind propositus with Turner's syndrome

(C = colour-blind)
There followed reports of colour-blindness occurring in the relatives of individuals with Turner's syndrome who had an isochromosome X for the long arms of the X chromosome. From these studies it was fairly certainly concluded that the locus for the colour-blindness gene (of deuteranopia, at least) was on the short arm of the X chromosome. For example, Lindsten et al. (1963) published the pedigree of an individual with Turner's syndrome, lacking the short arm material of one X chromosome, who manifested deuteranopia (see Diagram VII).

**Diagram VII** Pedigree of family having propositus with Turner's syndrome (karyotype 46 XXqi) demonstrating mode of inheritance of deuteranopia

(C = colour-blind)
Since the father had normal colour vision it must have been the mother's abnormal gene that the girl had inherited. The fact that the effect of this abnormal gene was not cancelled by the normal X chromosome material supplied by the father indicated that the missing part of the paternal chromosome must have carried the cancelling agent.

Other conclusions may be drawn (Stewart, 1961) from the study of individuals with isochromosome Turner's syndrome, relative to those with 45 X0 karyotypes:
(i) that a factor for sex chromatin manifestation is present in the long arm of the X chromosome (i.e., these patients are chromatin positive, unlike the patients with 45 X0 Turner's syndrome);
(ii) that a factor for normal stature development is present in the short arm of the X chromosome;
(iii) that a factor for normal gonadal development is present in the short arm of the X chromosome.

As part of the survey described in Chapter II six cases of Turner's syndrome associated with colour-blindness in themselves or in their relations were noted.

(i) A/388/67 (Registry identification number) - karyotype 45 X0. Colour-blindness was demonstrated in both the propositus and her father, which indicated maternal non-disjunction as having given rise to the abnormal karyotype.
(ii) A/24/60 - karyotype 45 X0 - being the case already cited, and reported by Stewart (1960).
(iii) Family K 40 - karyotype 45 X0 - propositus having normal colour vision, but two brothers were shown to be colour-blind. This indicated that the mother was a heterozygote, but gave no positive indication of parental non-disjunction.
(iv) A/38/60 - karyotype 46 XXqu. Father and propositus both demonstrated deutan colour-blindness. This case provided further
support for the theory that the deutan locus is on the short arm of the X chromosome, since the double amount of long arm material received from the mother (as a result of a fault in maternal gametogenesis) failed to counteract the effects of the X chromosome bearing the abnormal gene contributed by the father.

(v) \( \text{A/5/63} \) - karyotype 46 XXqi. Both mother and patient were reported colour-blind on the Ishihara plates. This case provided similar evidence for the suggestion regarding the colour-blindness gene locus, but differs from the previous case in that the mother was colour-blind, and it was inferred that she was homozygous for the colour-blindness gene. Faulty paternal gametogenesis must therefore have resulted in this case.

(vi) \( \text{A/62/63} \) - karyotype 45 X0/46 XXqi/47XXqi Xqi. The patient, her parents and her four sibs were assessed by Dr. R. Lakowski, of the Department of Psychology, Edinburgh. He concluded that, whilst the patient's colour discrimination was perfect on the red/green equation, she demonstrated "an almost tritan type of defect" on the blue/green equation. He found similar defects in other members of the family, but seemed not to postulate an inherited type of defect. This finding is of particular interest, in the light of the results presented in the following pages, since it could be suggested that Dr. Lakowski was describing a pattern of colour discrimination scores similar to those recorded there.
METHOD

Subjects

The experimental group consisted of the majority of those subjects already described in Chapter III, with the exception of those who withdrew. A corresponding number of control subjects were tested. Precise details of the numbers involved are given in the descriptions of the testing procedures.

Measuring Instruments

(i) Ishihara Plates (Ishihara 1968 Edition)

The test material consists of 24 plates which display multi-coloured figures picked out in dots against a dotted multi-coloured background. The colours used in the plates are chosen so that both normal and defective observers can see a figure, but this differs for each of the two classes. The test is usually employed to distinguish between normals and defectives, and not to study variations within these two categories. However, Lekowski (1964, 1965) found that very young and elderly subjects, whilst having essentially normal colour discrimination, experienced difficulty in reading some of the plates "correctly".

The subject is asked to read off the number or numbers she sees on each plate, without too much hesitation. These numbers are noted.

(ii) The Farnsworth-Munsell 100-Hue Test (Farnsworth 1957).

As well as discriminating between normal and congenital colour defectives, this test provides data on very small differences in colour discrimination.

The test material consists of four wooden boxes which altogether hold 85 movable caps, in each of which Munsell colours are mounted. Each box is made up of two trays hinged together. When a box is
XVI. Farnsworth-Munsell 100-Hue Test

(above) Box 85-21 ready for presentation to subject

(below) The four completed boxes
open and ready for presentation the tray nearest the subject is inclined at 45 degrees and has two extra repeated caps, one at each end of the tray, which act as fixed reference points. The movable caps are arranged in a random order in the other tray, which lies flat farther away from the subject (see Illustration XVI). The colours in the first box range from pink to yellow-green; those in the second from yellow-green to blue-green; those in the third from blue-green to blue; and those in the fourth from blue to pink.

The subject has to rearrange the randomly ordered caps, moving them from one tray to the other, to range between the fixed reference caps, into an order which resembles a continuous colour series with the two reference caps at the extreme poles of the series.

The manual recommends that standard lighting should be employed, and suggests that the reliability of results is lessened if it is not. Practical considerations in the testing of both experimental and control groups militated against the attainment of standard conditions, and the compromise of placing the subject near the best source of daylight available was employed. Since such conditions were therefore essentially randomized for both experimental and control groups it was considered that the results obtained from the two groups could still be validly compared.

**Scoring procedure**

The order of caps produced by the subject was recorded on the scoring sheets provided. Error scores were then calculated in the manner laid out in the manual. If the order is correct, the numbers on the reverse of the caps should run consecutively; if it is incorrect, the order is noted and an error score is calculated for each cap by adding the differences between the number of that cap and the number of the caps placed on either side of it. Because this
XVII. Pickford-Nicolson Anomaloscope

(above) Diagram of apparatus

(below) Photograph
method gives an error score of two for a cap in its correct position, a corrected error score is calculated by subtracting two from each error score:

<table>
<thead>
<tr>
<th>Cap Order</th>
<th>8 9 10 13 14 12 11 15 17</th>
</tr>
</thead>
<tbody>
<tr>
<td>Error Score</td>
<td>2 2 4 4 3 3 5 6</td>
</tr>
<tr>
<td>Corrected Error Score</td>
<td>0 0 2 2 1 1 3 4</td>
</tr>
</tbody>
</table>

The corrected error scores may be summed to give total error scores. This was done for all subjects. The error scores may be plotted on the circular graph provided on the score sheet. This, too, was done for all subjects, to ascertain whether there was any particular area of confusion or whether there was just a general lack of discrimination.

(iii) The Pickford-Nicolson Anomaloscope (1960)

The anomaloscope resembles the 100-Hue test in that it measures individual differences in colour discrimination, as well as differentiating between normal and abnormal colour vision.

The apparatus consists of a light source (A) from which light passes to two integrating boxes (B). (See Illustration XVII). These boxes illuminate a circular viewing aperture (D), covered with lightly frosted perspex. The viewing aperture is divided vertically in such a way that light from each integrating box illuminates a separate half of the aperture. The light from the light source passes through specified colour filters held in slides (E), moving in vertical slots (F). These slides can hold up to four filters, and the position of the slide in its slot determines the colour of the corresponding side of the viewing aperture. Two additional shutters control the intensity of the illumination of the halves of the viewing aperture, without affecting the colour. Controls for the two sides and two shutters are placed on top of the apparatus and
are fitted into graded dials. These are marked off in 82 arbitrary divisions.

For the purpose of this experiment the large viewing aperture, measuring 2.5 cms., was employed. One side of the aperture was maintained constant in both colour and illumination for all subjects, whilst the other half was varied by manipulation of the appropriate controls by the experimenter.

The arrangement of the filters and the dial settings for the particular anomaloscope used (Instrument No. 50; manufacturers, Rayne and Keeler) are given below. The settings of the controls necessarily vary from one apparatus to another.
Settings for the Filters, Slides and Shutters

for the three tests

<table>
<thead>
<tr>
<th>Test 1: Red &amp; Green</th>
<th>L.Slide</th>
<th>R.Slide</th>
</tr>
</thead>
<tbody>
<tr>
<td>OY3</td>
<td>OGr 1</td>
<td></td>
</tr>
<tr>
<td>Above</td>
<td>Above</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>ON 32</td>
<td>OR 1</td>
<td></td>
</tr>
<tr>
<td>Below</td>
<td>Below</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Right Slide:</td>
<td>Normal Setting about 31</td>
<td></td>
</tr>
<tr>
<td>Right Shutter:</td>
<td>Open at setting 82 ) kept</td>
<td></td>
</tr>
<tr>
<td>Left Slide:</td>
<td>Set at 54 ) constant</td>
<td></td>
</tr>
<tr>
<td>Left Shutter:</td>
<td>About 21</td>
<td></td>
</tr>
</tbody>
</table>

Test 2: Green & Blue

<table>
<thead>
<tr>
<th>L.Slide</th>
<th>R.Slide</th>
</tr>
</thead>
<tbody>
<tr>
<td>OB 2</td>
<td>OB 10</td>
</tr>
<tr>
<td>Above</td>
<td>Above</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>ON 32</td>
<td>OGr 1</td>
</tr>
<tr>
<td>Below</td>
<td>Below</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Right Slide:</td>
<td>Normal Setting about 35</td>
</tr>
<tr>
<td>Right Shutter:</td>
<td>Open at setting 82 ) kept</td>
</tr>
<tr>
<td>Left Slide:</td>
<td>Set at 66 ) constant</td>
</tr>
<tr>
<td>Left Shutter:</td>
<td>About 39</td>
</tr>
</tbody>
</table>

Test 3: Yellow & Blue

<table>
<thead>
<tr>
<th>L.Slide</th>
<th>R.Slide</th>
</tr>
</thead>
<tbody>
<tr>
<td>Nil</td>
<td>OB 10</td>
</tr>
<tr>
<td>Above</td>
<td>Above</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>OR 1</td>
<td>ON 32 +</td>
</tr>
<tr>
<td></td>
<td>OY 3</td>
</tr>
<tr>
<td>Below</td>
<td>Below</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Right Slide:</td>
<td>Normal Setting about 31</td>
</tr>
<tr>
<td>Right Shutter:</td>
<td>Open at setting 82 ) kept</td>
</tr>
<tr>
<td>Left Slide:</td>
<td>Set at 29 ) constant</td>
</tr>
<tr>
<td>Left Shutter:</td>
<td>About 51</td>
</tr>
</tbody>
</table>

Key to Colour Filters

- OY 3 = yellow
- ON 32 = neutral
- OGr = green
- OB 2 = blue-green
- OB 10 = dark blue
- OR 1 = red
Given the standard the subject has to say whether the two halves match at various settings of the variable shutter. Some subjects may have a small matching range (MR), accepting only one setting, whereas others may accept many settings as matching the standard and thus have a larger MR. For the latter subjects a mid-matching point (MNP) may be calculated.

Three standards were presented, those which involved matching mixtures of:

(i) red and green
(ii) green and deep blue
(iii) deep blue and yellow

Once again it was impossible to achieve standard external lighting conditions. Light sources were dimmed as much as possible; lights switched off, curtains or blinds drawn, etc.; and the subject was placed sideways to this dimmed source. If the light still seemed to interfere with testing conditions a screen was used alongside the apparatus to block the light even further. This was necessary only infrequently, i.e. when there was direct sunlight. The experimenter donned a white coat to prevent interference from coloured clothes.

Finally it might be noted that the administration and interpretation of the anomaloscope are more complex than those associated with the two colour tests mentioned previously. It is necessary to test in such a way that the subject gains no clues, and to establish the end points as reliably as possible. It is also necessary to be able to recognise the types of profile common to the colour vision abnormalities. To achieve as competent a testing procedure as possible the author

(a) observed an experienced tester (P.A.) during several administrations of the anomaloscope;
(b) administered the anomaloscope to some 30 persons (colleagues and general medical patients);
(c) tested five males with congenital forms of colour-blindness, namely, three deutans, one protan and one deuteranomalous subject. In addition, the experienced tester (P.A.) re-tested two members of the experimental group at the Eye Pavilion under standard conditions and found abnormal results which corresponded closely to those obtained by the author.

Whilst these measures may not be considered completely sufficient, inclusion of a control group must help to counteract the effects of unskilful administration.

**Procedure**

The battery of colour vision tests was administered in different testing sessions to the experimental and control subjects.

**Experimental group**

The Ishihara and 100-Hue tests were administered to all 24 subjects during the first testing session. The 100-Hue test was found to be a very acceptable test for establishing rapport and interest, and this was accordingly always administered before the intelligence scale. The Ishihara plates were interposed between the 100-Hue and intelligence scale. The anomaloscope was administered during a later testing session to the 19 subjects, out of the original experimental group of 24, who were available.

**Control group**

All three tests were administered to controls corresponding in number to the subjects in the experimental group (Group 1 controls). The tests were ordered (i) 100-Hue

(ii) Ishihara plates

(iii) Anomaloscope

These colour tests always preceded the administration of the 16 Personality Factor Questionnaire (see Chapter VII).
(i) The Ishihara plates

The book of plates was laid open flat in front of the subject, in such a position as to attain maximum daylight. The following instructions were given:

(Indicating practice plate) "What number do you see here? Now, please read off the numbers on the other pages." If the subject hesitated over any of the plates she was encouraged to make a guess at it, and to pass on to the next plate. The responses were noted. The Ishihara plates were used as a screening test only.

(ii) 100-Hue test

The first box was laid open in front of the subject, so that the tray with the fixed reference caps was nearest the subject (see Illustration XVI). Maximum lighting was obtained by seating the subject facing, and as near as possible to, a source of daylight. Verbal instructions did not follow verbatim those given in the manual, since it was considered that they were not easy to understand. Instead the subject was given the following instructions:

"This is a test of colour vision. These two caps" (indicating reference caps) "are fixed, but these" (indicating tray of random movable caps) "you can move around. Please will you pick out the one that is closest in colour to this one" (indicating left-hand reference cap) "and lay it alongside it." This was usually correctly done, but no help was given. "Now, please re-arrange the other colours along here, so that they start with these colours" (indicating left-hand reference cap and the one placed there by subject) "and finish with this one" (indicating right-hand reference cap). "You can change the order if you are not satisfied with it when you have finished." No time limit was set for either experimental or control subjects, to avoid flustering the former. However, the two-minute limit advised by the manual was seldom exceeded, and it was never considered necessary to hasten anyone.

The order of the completed boxes was noted on the scoring sheet before the caps were re-arranged in random order ready for use with the next subject.
(iii) Pickford-Nicolson anomaloscope

The room was darkened as much as possible by switching off lights and drawing curtains and blinds. The subject was placed sideways to any source of light, at a distance of about one metre in front of the anomaloscope. The height of the anomaloscope was adjusted until the aperture was at eye level. The preliminary setting of the shutters on the anomaloscope was as follows: the standard on the left-hand side of the viewing aperture (as viewed by the subject) and the colour shutter set at the rough 6 MP for the right-hand side. Thus the colours were roughly equated. The shutter governing the brightness of the variable half was shut off so that it appeared black.

The following verbal instructions were given:
"This is another colour test. I want you to look at the two halves of this circle and tell me whether they are the same or different. At the moment I think you will agree that they are different... Right; that is because this side" (indicating R.H.S.) "is much darker than this side" (indicating L.H.S.). "Now, I can make it much lighter" (manipulating brightness shutter). "Now they are still different because this side" (indicating R.H.S.) "is too light now. Please will you tell me when they are about the same." (Brightness shutter manipulated until subject indicated they were the same). "Now I shall just cover the aperture" (with hand over viewing aperture) "because you tend to get used to the two sides, rather as you get used to seeing in the dark, and you may be telling me they are the same, when in actual fact they are different." (Uncovering viewing aperture) "Now, are they still the same, or different?" When the subject agreed that they were the same, the question was asked, "Are the colours the same?" The statement was added, "The colours can change, just as you have seen that the brightness and darkness can, but I am not going to show you how - I want you to tell me when the colours are different."
The method of testing followed was then to increase the variable colour shutter reading by five graduated steps as marked on the circular graded dials, asking, "Are they the same or different?" If the answer was "Different" the subject was asked to describe in what way they were different — often an adjustment of the brightness shutter would be sufficient for S to decide they were the same. (Any adjustments were always made with the hand covering the viewing aperture; this both introduced a break in adaptation processes for the subject and prevented her from seeing the effects of the adjustment). A response to the effect that the colours were the same having been obtained, the colour shutter reading was increased by further steps of five graduations until an end point was reached at which the subject insisted the colours were different and was able to describe this difference adequately. It was then necessary to check the individual graduated steps in the last five steps to see at exactly what end point the subject first perceived a difference in the colours.

The process was repeated in steps below the rough mid-matching point to obtain the end point at the other end of the matching range. The light source was then switched off before the settings for the next colour comparisons were made.

Statistical Analysis of results

The following statistics were then applied to the results. It should be recalled that the Ishihara test was given only as a screening device, and no statistical procedures were therefore involved in its consideration.

For Hypothesis V/1

Total error scores on the 100-Hue test were compared for Groups A and B of the experimental group, using the Mann-Whitney U test.

For Hypothesis V/2

Total error scores on the 100-Hue test for the experimental and control groups were compared, using the Mann-Whitney U test.
For Hypothesis V/3

Error scores for the four boxes of the 100-Hue test (i.e. ranges pink-green: Caps 85-21; green-green/blue: Caps 22-42; green/blue-blue: Caps 43-63; and blue-pink: Caps 64-84) were compared for the experimental and control groups using the Mann-Whitney U test.

For Hypothesis V/4

Regarding the anomaloscope results:

(a) The mid-matching points (MMF's) and
(b) the matching ranges (MR's)
for the experimental and control groups were compared, using Mann-Whitney U tests.
HYPOTHESES

The following hypotheses were formulated:

V/1 That there will be no significant difference between the experimental Groups A and B in total error scores on the 100-Hue test.

V/2 That there will be no significant difference in total error scores on the 100-Hue test for the experimental and control groups.

V/3 That there will be no significant differences in the four colour ranges on the 100-Hue test for the experimental and control groups.

V/4 (a) That there will be no significant differences in the mid-matching points for the three colour equations on the anomaloscope for the experimental and control groups.
   (b) That there will be no significant differences in the matching ranges for the three colour equations on the anomaloscope for the experimental and control groups.
RESULTS

Results obtained from screening all subjects with the Ishihara test indicated that none of either the experimental or control subjects demonstrated congenital red/green colour-blindness.

Hypothesis V/1

The results supported this hypothesis. Table XIX shows the mean total error scores on the 100-Hue test for the experimental Groups A and B.

Table XIX  Mean total error scores on the 100-Hue test for experimental Groups A and B.

<table>
<thead>
<tr>
<th>Group</th>
<th>N</th>
<th>Total error score (mean)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Group A</td>
<td>16</td>
<td>158.4</td>
</tr>
<tr>
<td>Group B</td>
<td>8</td>
<td>136.8</td>
</tr>
</tbody>
</table>

U = 42.5 N.S.

This table indicates that there was no significant difference in the total number of errors gained by the two sub-groups of the experimental subjects. In future analysis they are therefore considered in toto.

Hypothesis V/2

The results did not support this hypothesis. Table XX shows the mean total error scores on the 100-Hue test for the experimental and control groups.

Table XX  Mean total error scores on the 100-Hue test for experimental and control groups

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>U</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>24</td>
<td>24</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total error score (mean)</td>
<td>151.2</td>
<td>73.9</td>
<td>133 (z=3.19)</td>
<td>&lt;.001</td>
</tr>
</tbody>
</table>

** 2-tailed test
These results indicate that the experimental group gained a significantly higher total number of errors on the 100-Hue test than did the control group.

Hypothesis V/3

The results did not support this hypothesis. This finding is therefore in line with the results for Hypothesis V/2. Table XXI shows the mean error scores on the four boxes of the 100-Hue test for the experimental and control groups.

<table>
<thead>
<tr>
<th>Box</th>
<th>Mean error score</th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>U</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td>85-21</td>
<td>22.0</td>
<td>11.3</td>
<td>178.5</td>
<td>(z=2.26)</td>
<td>&lt;.02</td>
</tr>
<tr>
<td>22-42</td>
<td>43.2</td>
<td>21.9</td>
<td>132.0</td>
<td>(z=3.22)</td>
<td>&lt;.001</td>
</tr>
<tr>
<td>43-63</td>
<td>47.0</td>
<td>21.7</td>
<td>137.5</td>
<td>(z=3.10)</td>
<td>&lt;.002</td>
</tr>
<tr>
<td>63-84</td>
<td>38.9</td>
<td>19.0</td>
<td>160.5</td>
<td>(z=2.63)</td>
<td>&lt;.01</td>
</tr>
</tbody>
</table>

** 2-tailed test

These results indicate that the experimental group gained a significantly greater number of errors on all four colour ranges than did the control group.

Hypothesis V/4

(a) The results did not wholly support this hypothesis. Table XXII gives the mean mid-matching points (MMP's) for the three equations of the anomaloscope for the experimental and control groups.
These results indicate that, whilst there were no differences in the mid-matching points chosen by the experimental and control groups for the red/green and blue/green equations, there was a significant difference in the MMP's chosen for the yellow/blue equation.

(b) The results did not support this hypothesis. Table XXIII gives the matching ranges (MR's) for the three equations of the anomaloscope for the experimental and control groups.

** Table XXII **  Mean MMP’s for the three equations of the anomaloscope for the experimental and control groups

<table>
<thead>
<tr>
<th>Equation</th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>U</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Red/green equatn.</td>
<td>30.4</td>
<td>31.0</td>
<td>139 (z=1.25)</td>
<td>N.S.</td>
</tr>
<tr>
<td>Blue/green</td>
<td>31.4</td>
<td>34.2</td>
<td>136 (z=1.33)</td>
<td>N.S.</td>
</tr>
<tr>
<td>Yellow/blue</td>
<td>27.0</td>
<td>30.2</td>
<td>109.5 (z=2.10)</td>
<td>&lt;.05</td>
</tr>
</tbody>
</table>

** 2-tailed test

** Table XXIII **  Mean MR's for the three equations of the anomaloscope for the experimental and control groups

<table>
<thead>
<tr>
<th>Equation</th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>U</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td>Red/green equatn.</td>
<td>8.7</td>
<td>2.6</td>
<td>43.5</td>
<td>&lt;.002</td>
</tr>
<tr>
<td>Blue/green</td>
<td>22.5</td>
<td>4.9</td>
<td>24.0</td>
<td>&lt;.002</td>
</tr>
<tr>
<td>Yellow/blue</td>
<td>25.9</td>
<td>7.4</td>
<td>90.0</td>
<td>&lt;.02</td>
</tr>
</tbody>
</table>

** 2-tailed test
These results indicate that there was a significant difference in the matching ranges on all three equations for the experimental group, who accepted a larger range of colours as matching all three standards than did the control group.
EXPERIMENTAL GROUP.

TOTAL ERROR SCORE = 20

CONTROL GROUP.

TOTAL ERROR SCORE = 28

TOTAL ERROR SCORE = 154

TOTAL ERROR SCORE = 72

TOTAL ERROR SCORE = 428

TOTAL ERROR SCORE = 163

Farnsworth-Munsell 100-Hue Test

Error Distribution Charts (for further explanation see text)
DISCUSSION

From the results presented in this chapter has emerged an indication that, in patients with Turner's syndrome, there was evidence of a deficiency in colour perception and/or discrimination. Previous studies of colour vision in these individuals have involved research into the congenital types of colour-blindness only, and the subject of colour discrimination has not been explored. By using the Ishihara plates as a screening device for congenital types of colour-blindness it was established that all the individuals in the sample had "normal" colour vision. This finding was confirmed by further testing with the 100-Hue test. The results indicated that the experimental group experienced difficulty in discriminating colours, a deficiency not identifiable with any of the types of colour-blindness usually isolated by this test. Although the experimental group made a significantly greater number of errors on the whole test, it was not possible, by splitting up the error scores in terms of the four sections of the colour spectrum represented in each of the four boxes, to isolate one particular area of confusion. Indeed, the charts used to illustrate the variation in error scores for the experimental and control groups (see Illustration XVIII) tend to demonstrate a typically "anarchic pattern" (Lakowski, 1969). This type of pattern has two main characteristics:

(i) a large total error score;
(ii) errors distributed randomly.

It may be seen from these charts that the first characteristic is not always true of the experimental subjects, since the lowest error score overall was achieved by one of the patients with Turner's syndrome (see Charts A and B of Illustration XVIII). Other variations are difficult to compare, since any cut-off points which exist for the 100-Hue test are, necessarily, arbitrary ones.
The manual states that about 16% of the population has been found to make total error scores of more than 100, a figure which is almost replicated by the control group tested here (17%), but not by the experimental group, 70% of which scored over 100 errors. This point is illustrated by Charts C and D of Illustration XVIII, from which a comparison can be made of the results of an experimental and control subject who score at the mean for their respective groups. Charts E and F contrast examples of maximum error scores for individuals from each group.
Graph VII. Farnsworth-Munsell 100-Hue Test
Mean corrected error scores
In order to examine and contrast the patterns of colour confusion more closely the mean corrected error score for each of the 85 caps was calculated for the experimental and control groups (see Graph VII). Examination of the graphs thus obtained shows a remarkable correlation between the two lines representing the experimental and control groups' performances, and indicates that that of the experimental group mimics, at a higher level of error score, that of the control group. Since the test may be seen as involving skills dependent on an interaction between ordering and colour discrimination abilities, this observation would rule out any suggestion that the experimental group were in some way deficient in ability to order material into a sequence; it would rather indicate a difficulty in their colour discrimination which was an exaggeration of normal.

These results made it necessary to check for the anomalous types of colour discrimination defects in the experimental group, which the 100-Hue test is not designed to do. Anomaloscope testing provided good replication results, indicating no significant anomalous shifts of the mid-matching points (MMP's) on the red/green and blue/green equations, only an elongation of the matching ranges (MR's) on all three equations when compared with controls.
Red-Green test

Control group (N=19)

Experimental group (N=19)

Graph VIII. Pickford-Nicolson Anomaloscope
Graphical representation of results
Blue-Green test

Graph IX. Pickford-Nicolson Anomaloscope
Graphical representation of results
Yellow-Blue test

Control group (N=19)

Experimental group (N=19)

±3S.D.

Dial Reading

Graph X. Pickford-Nicolson Anomaloscope

Graphical representation of results
Results from anomaloscope testing are presented as graphical illustrations (Graphs VIII, IX and X). The lines ± 3 S.D.'s relate to the mean MMP and were calculated from the control group's results, so that the abnormal readings taken from the experimental group could be related to the central tendencies within the normal population. This method was introduced by Pickford (1951), who considered that the limits of ± 3 S.D.'s marked the boundaries of normal distribution, and that outside these limits lay "MMP's of genetically determined anomalous trichromats." (Lakowski, 1969). From the three graphs it may be seen that 6/19; 5/19; 11/19 of the experimental group fell outside these limits, on, respectively, the red/green, blue/green and yellow/blue tests. This result, taken on its own, would indicate the existence of anomalous colour vision defects, although it is noted that it is possible for all three types of defect to occur in one individual - an uncommon finding in persons having genetically-determined colour vision defects. The statistical analysis of the results of the group indicates a significant shift in MMP on the yellow/blue equation alone.

Statistical analysis of the matching range (MR) data indicates a significant difference between experimental and control subjects on all three colour equations, and the graphs show that the experimental group accepted a larger number of matches to the standard than did the control group. This again would seem to suggest that the experimental group had poor colour discrimination. With the exception of the significant shift of the MMP noted on the yellow/blue equation, these results appear to support those obtained from the 100-Hue test, in indicating a basically "normal" colour vision apparatus, with particular deficiencies which cause a poor level, rather than a complete lack, of colour discrimination. Thus it could be suggested that individuals with Turner's syndrome possess all the necessary basic colour vision mechanisms (in that
they do not lack cones relevant to particular colours in the same way as congenitally colour-blind persons do) but that their particular genetic abnormality has given rise to defects in the perceptual processing of the colours they see.

In this connection it would seem necessary to discuss very briefly some of the experimental evidence on the physiological structures involved in colour vision. Sheppard (1968) presented a comprehensive review of the research carried out in this area, and discussed in turn the retina, lateral geniculate nuclei and striate cortex, which he claimed are all essentially involved, although little was known as yet of the detailed structure and interconnections of the system. He concluded that evidence for the existence of photopigments in the cones was not conclusive. He also demonstrated that the neuronal connections with the cones increased from single interconnections in the periphery of the eye to three per cone around the central part, or fovea, and suggested that these ratios corresponded to the areas concerned with colour vision, in that the ratio 3:1 gives full colour vision, whilst 1:1 gives achromacy. Together with other cone physical characteristics this would seem to imply that colour perception at the retinal level depends on a variation of all these characteristics, and cannot be simplified to photopigment absorption alone. It is appropriate to mention a study by Jesberg (1968), who found a high incidence of retinal degeneration in a group of patients with Turner's syndrome. Such degeneration will interfere with all aspects of vision, at the retinal level.

Sheppard also reviewed research on the sub-cortical synapses involved in the lateral geniculate nuclei, which are interconnected with the cones in varied and complex ways. In the monkey the various layers of the nuclei have been shown to react differently to visual stimuli (Wiesel and Hubel, 1966), and their firing activity appears to be governed by the spectral wavelengths involved. Finally
Sheppard considered evidence on the terminating area of afferent impulses - the occipital lobes, or, more closely, the striate area. Here it had been shown that each area of the retina had its corresponding projection area, although he emphasised that functionally the visual cortex is not rigid, and, indeed, is very much under the control of portions of the cortex adjacent to it.

It is impossible to hypothesise which areas are particularly affected in individuals with Turner's syndrome, but it may be recalled that it has been suggested that all the areas are probably relatively intact, in that the patients concerned merely showed exaggerated profiles of normal colour discrimination errors. It is also impossible to categorise the defect described here as congenital or acquired within the accepted definitions of the terms. A detailed comparative longitudinal study is called for, since it cannot be assumed that the deficiencies are present from birth, or that they always develop in the same way, or, indeed, that ageing effects are not responsible for them.

In conclusion, it should be borne in mind that the physiological deficiency of a patient with Turner's syndrome may be expressed at the cellular level, so that some or all of the areas itemised above, as well as others which are not, may be affected - to result in the picture described in this thesis. It is questionable what is to be achieved by continuing to draw comparisons with normal processes; in any case, theories regarding these are often derived from experiments on lower organisms and are, therefore, suspect in their application to man.

In the context of the established relationship between the X chromosomes and colour vision it is relevant to introduce the Lyon hypothesis, (1962). This postulates that the chromatin body normally seen in female cells is the inactivated second X chromosome from the female sex chromosome pair. Thus, males lack the chromatin body because they have a Y chromosome in the sex chromosome pair and
not a second X chromosome. On the other hand, individuals with Turner's syndrome who are chromatin negative and have 45 chromosomes (karyotype 45 XO) lack the chromatin body because they have no second X chromosome, and not because they are genetically male. The further suggestion is made that there may well be suppression of the activity of the genes located on the X chromosome which forms the chromatin body in normal females. This inactivation cannot be complete, however, or individuals having the 45 XO karyotype would be no different from those with the 46 XX chromosome complement.

Evidence set out in this chapter suggests that the missing X chromosome material has resulted in some type of deficit, be it in terms of absent genes or organic abnormality, which has interfered with colour perception ability. It could be that during the embryonic stage the missing X chromosome did not provide information that the second temporarily active X chromosome in the normal female does, or that the X chromosome is not totally inactivated, and that ongoing colour discrimination is dependent on an interaction between the two X chromosomes. In this context might be mentioned an article in the Lancet (1970), based on X blood grouping, which indicates that genetic inactivation of one of the X chromosomes in human females is not total. In order to investigate these points more closely it would certainly be necessary to contrast patients having Turner's syndrome with the 45 XO karyotype with large numbers of mosaic patients having the 45 XO/46 XX karyotype, in order to observe variations and their corresponding characteristics.
CHAPTER VI

AUDIOMETRY

INTRODUCTION

In an auditory screening programme it is customary to obtain a "profile" of auditory acuity from both ears over several frequencies. Any hearing loss that is noted is measured in decibels, and if such loss exceeds about 20 decibels it is considered to constitute deafness. The origins of this deafness may be sub-classified into:

(i) **conductive defects**: these are caused by disease or deformity of the outer, or, more usually, the middle, ear. There is a reduction in the conduction of sound by air, as well as by the bony chain of the middle ear (i.e. the malleus, incus, stapes link).

(ii) **perceptive defects**: these usually imply a deficiency of the inner ear and/or its neural connections. It is claimed that abnormalities of the cochlear (part of the inner ear) may be distinguished from nerve deafness by the use of recruitment tests.

One of the associated symptoms of Turner's syndrome which may become apparent any time from birth onwards is deafness. In his review of gonadal dysgenesis Hauser (1963) commented that partial or total deafness had occasionally been reported. In their review of 55 cases Haddad and Wilkins (1959) commented on a frequent abnormality of the ear setting with protrusion and deformity (55%). Apart from a table indicating that "deafness" occurred in three patients, no further comment was made in the discussion of the anomalies. There were consequently no details concerning the degree of manifestation of deafness and no suggestion as to what the cause was thought to be.

Lemli and Smith (1963) described the pattern of various anomalies associated with the 45 XO syndrome and, whilst they mentioned the
existence of prominent ears as one of the frequently occurring anomalies (in 84% of the 25 patients studied), no comment was made on the auditory acuity of their patients. In a short case report written with specific reference to hearing impairment Stratton (1965) reported on the occurrence of deafness in a patient with Turner's syndrome. At the first referral she was found to have bilateral conduction deafness due to the adherence of the drums to promontories. An operation for removal of adenoids was carried out to remedy this. Five years later, when the same case was reviewed, the girl was found to have bilateral chronic otitis media and an average hearing loss of 60 decibels in each ear. In considering the report Stratton reviewed some 234 cases of gonadal dysgenesis in which only eight (3%) were found to have some form of deafness. Of these the causes were given as: congenital - two; otitis media - two; otosclerosis - nil; cause not stated - four.

By far the most detailed study in this field arose from Lindsten's monograph (1963), which covered a large number of aspects of Turner's syndrome. In a paper on hearing impairment in these patients (Anderson et al., 1969) it became apparent for the first time how necessary auditory screening is when this syndrome is being investigated. A survey of 79 patients with varying but specified karyotypes was presented. Each of these patients was questioned about previous history of middle ear infections, and an audiometric analysis made. To distinguish between nerve and conduction defects acoustic recording of the intra-aural muscular activity ("stapedius reflex") was employed.

Of 76 patients 68% had had a middle ear infection for which they had received medical attention. All the 17 patients who were found to have a conductive type of hearing impairment had a previous history of ear infections, whilst 24 of the 34 patients with nerve deafness also had a similar history. A more detailed but technical account of the findings is given in the paper, but certain results are relevant to this thesis. In the more common type of impairment, nerve
deafness, the area chiefly affected was found to be that around the frequency of 1,000 c.p.s. This is the same type of impairment as that which occurs in labourers using mechanical road drills, but in their cases the area of impairment is at a higher frequency. Since human speech is in the range about 1,000 c.p.s. it is clearly important for educational and social reasons to discover and assess this impairment in patients with Turner's syndrome. In analysing the results with respect to age, the authors reported that hearing impairment seemed to appear around or after ten years of age, and then remained stationary or increased very slowly.

Measurements of the skulls indicated that the middle ear and Eustachian tube were abnormally orientated, which the authors suggested might predispose the patients to middle ear infections. However, they stressed that middle ear infections can be linked only with hearing impairment of the conduction type, which occurred less often in their sample than nerve deafness. With respect to this latter impairment, results from specialised tests suggested that the impairment was associated with a defect in the outer hair cells of the organ of Corti. However, in radiographic examination of the cochlea in ten cases, no abnormality could be observed. An attempt was also made to study (by electronmicroscopy) cochlear specimens from two patients who died, but this failed on account of autolytic changes having taken place. The authors concluded by hypothesising that the cochlear defect might be associated (along with other observations of abnormalities in morphology and function of other organs) with the genetic imbalance associated with Turner's syndrome.
Audio-testing Instrument
Record Chart for Audio-Tester


METHOD

Subjects

The auditory acuity of all the members of the experimental group was assessed, along with that of a corresponding number of control subjects.

Measuring Instrument

The instrument used was the Keeler Audio-Tester Mk III. This instrument is a very simple and quick screening device by means of which it is possible to ascertain whether a person's hearing acuity is within normal limits, or, if it is not, what amount of loss there is.

The instrument consists of a battery-operated power source which emits three frequencies of sound through an earphone, which the subject holds close to her ear. These frequencies (250, 1000 and 4000 cycles per second) are tested for both ears at normal threshold level, and if the subject cannot hear them, hearing loss may be assessed in ten decibel steps up to 30 decibels plus a practice range of 60 decibels. The results on both ears are plotted on a chart (see Illustration XIX).

Procedure

The subject was asked to place the earphone over her right ear, and a check was made to ensure that the centre of the earphone was correctly aligned with the external auditory canal. The output level switch (see Illustration) was then turned to 60 decibels (db) and the frequency switch to 1000 cycles per second (cps). The test switch was depressed and the subject was asked to listen for a sound. It having been established that she understood the instruction, she was asked to indicate when she heard a sound by tapping on the table.
Lower levels of loudness were then tested, starting with the 30 db. level, and the lowest point at which the subject could still hear the sound was registered on the chart. The point was always checked by depressing the test switch a couple of times. The process was repeated for frequencies of 250 and 4000 cps. and then the whole testing procedure was carried out on the left ear.

Two precautions were observed. The audio-screener was so orientated that the subject could not see when the test switch was depressed; and the interval between changing the sound level and depressing the key was varied so that the subject did not learn to respond to the interval alone.

Statistical analysis of results

Results were split into two groups for each frequency in terms of decibel loss. One group was considered within normal range (0 - 20 db.) and the second as demonstrating hearing loss (30 + db.).

The experimental and control groups were compared on this measure, using $x^2$ for two independent samples.

HYPOTHESIS

That the experimental group will differ significantly from the control group in demonstrating greater decibel loss in both ears, on all frequencies.
RESULTS

The results supported the hypothesis. Tables XXIV and XXV give details of the number of subjects in the experimental and control groups who demonstrated severe hearing loss over the three frequencies.

<table>
<thead>
<tr>
<th>Table XXIV</th>
<th>Number of experimental and control subjects demonstrating hearing loss in the RIGHT ear</th>
</tr>
</thead>
<tbody>
<tr>
<td>Experimental Ss.</td>
<td>Control Ss.</td>
</tr>
<tr>
<td>N</td>
<td>24</td>
</tr>
<tr>
<td>Frequency 250 cps.</td>
<td>15</td>
</tr>
<tr>
<td>&quot; 1000 &quot;</td>
<td>9</td>
</tr>
<tr>
<td>&quot; 4000 &quot;</td>
<td>8</td>
</tr>
</tbody>
</table>

* 1-tailed test

<table>
<thead>
<tr>
<th>Table XXV</th>
<th>Number of experimental and control subjects demonstrating hearing loss in the LEFT ear</th>
</tr>
</thead>
<tbody>
<tr>
<td>Experimental Ss.</td>
<td>Control Ss.</td>
</tr>
<tr>
<td>N</td>
<td>24</td>
</tr>
<tr>
<td>Frequency 250 cps.</td>
<td>10</td>
</tr>
<tr>
<td>&quot; 1000 &quot;</td>
<td>10</td>
</tr>
<tr>
<td>&quot; 4000 &quot;</td>
<td>11</td>
</tr>
</tbody>
</table>

* 1-tailed test

These results indicate that the experimental group demonstrated a highly significant greater degree of auditory acuity loss than did the control group. This finding was common to both ears and over all three frequencies.
DISCUSSION

That a defect in hearing is associated with Turner's syndrome is clearly shown from the results presented in this Chapter. It should be remembered that hearing acuity and loss are represented as a continuum and not as the dichotomy Deaf/Not Deaf. Most previous research has reported hearing difficulties only when they have been so apparent as to require remedial attention. In comparing the group of patients with a control group it was possible to demonstrate some degree of hearing acuity loss on all frequencies in both ears as being a characteristic of patients with Turner's syndrome. The pattern of impairment noted by Anderson et al. (1969) of a bilaterally symmetrical dip in the middle frequencies (around 1000 cps.) was not replicated, although it could be suggested that an audiometer with a wider range of sensitivity might have to be used to isolate this pattern.

Six of the patients studied reported episodes of otitis media, when asked specifically about illnesses involving the ears. It was not possible to prove that these particular individuals demonstrated an even greater loss of auditory acuity than the other patients included in the sample. There also seemed to be no difference between the 16 patients with the 45 XO karyotype and the remainder of those having varied karyotypes.

As far as the cause of the hearing loss is concerned, it was considered outwith the limits of a psychological thesis to proceed further to establish whether the origins of hearing loss were conductive or perceptive in nature - or, indeed, whether the two types co-existed within the group of patients tested. It must be assumed that the six patients already mentioned who had previous histories of middle ear disease were of the conductive hearing loss category.

Using the simple Rinne and Weber techniques (Best and Taylor, 1966) the author established the presence of perceptive as well as
conductive hearing losses in a few members of the group. (The techniques involve the use of a vibrating tuning fork applied to various positions on the bones of the skull).

These results, in general, reflect the findings of Anderson et al. in indicating at least two areas involved in the auditory perceptual process, which are detrimentally affected in patients with Turner's syndrome. Thus the hearing loss cannot be associated with deficiency at any particular level because it also seems possible that the chromosome abnormality may affect the development of the physical structure of the ear itself, as well as the intermediary areas of the middle ear and the sensory-neural end organs.

The application of these findings to the other results described in this thesis is important. The VIQ/PIQ discrepancy is not affected in the expected direction, i.e. when deafness is a factor performance on verbal items is generally affected to reduce the VIQ below the PIQ level. It could be hypothesised that poor auditory acuity would contribute to the picture of social withdrawal and introversion, described in the following chapter - a consideration which emphasises the necessity for early auditory screening of individuals with Turner's syndrome.
CHAPTER VII

PERSONALITY

INTRODUCTION

There exist few data on the personality characteristics of individuals with Turner's syndrome. Earlier studies (e.g., Hampson et al., 1955) contained the incorrect concept that the individuals under study were "genetic males", having a female phenotype. The fact that the authors concluded that all their subjects fitted unequivocally into the female role reflects credit on their research methods, since more recent findings on patients with established karyotype have endorsed this conclusion.

The literature may be reviewed under two headings:

(i) General personality characteristics
(ii) Incidence of mental illness

(i) General personality characteristics

The study by Hampson et al. (1955) on 11 patients with "male chromosomes" has already been mentioned briefly. In comparing their patients with a group of 11 normal girls the authors were unable to find any differences in gender orientation which could be attributed to the "male chromosomes".

Cohen (1962) used the Draw-a-Person test (Goodenough 1926), amongst a battery of other tests, to assess a group of nine patients with ovarian dysgenesis (no cytogenetic details were given). Results were compared with those from a group of nine controls, and were marked by three raters who were asked to judge the tests in terms of the sexual maturity revealed by the drawings. Whilst "femininity" was expressed in all the drawings obtained from the control group, this
could be said to be true of only three of the patients with gonadal dysgenesis. Cohen suggested that this finding was related to the patients' physical deficiencies. The ratings were in fact so much higher for the control subjects that Cohen postulated that there might indeed be two populations involved, but no statistical evidence was put forward to support this idea.

Cohen's finding might be seen as contradicting the previously reviewed report by Hampson et al. on the association between "unequivocal femininity" and Turner's syndrome. In order to explain this apparent contradiction the important concept of psycho-sexual maturity must be introduced. Whilst patients with Turner's syndrome fit into the feminine role, their maturational development in this role may well be delayed or even retarded when the sex hormones fail to produce secondary sexual characteristics, thus not only resulting in physical immaturity, but also influencing other fields of behaviour as well. It is therefore possible that those of Cohen's patients who did not express "femininity" in their drawings were affected in this way.

Alexander et al. (1966) also included the Draw-a-Person test amongst their battery of tests to assess the form-perception disability associated with the syndrome. They commented on the tendency to draw a female figure first, from which they inferred feminine gender orientation. They also found that both human and geometric figures were poorly drawn, suggesting that the deficiency is visuo-constructional and not a reflection of physical deficiencies, as Cohen suggested.

By far the largest study of the personality characteristics associated with Turner's syndrome was made by Shaffer (1963). He administered two questionnaires to a group of 13 patients with gonadal dysgenesis, cytogenetically described as eight cases chromatin negative and five chromatin positive. The first questionnaire, the Minnesota Multiphasic Personality Inventory (MMPI), was originally designed primarily to assess psychological maladjustment. Results on this
test were compared with data from ninth grade girls (modal age 15 years) and with the standardised norms given for adult women. Only one scale, "Hypomania", differentiated significantly between the controls (both adult women and ninth-grade girls) and the patients with Turner's syndrome. This, by definition, indicated that the latter lacked enthusiasm, tending to be passive in their approach to everyday problems. Shaffer also commented on findings from the Masculinity/Femininity scale, which indicated that his group of individuals with Turner's syndrome tended to be more feminine than either the adult women or ninth-grade girls, a trend which did not reach a significant level. All other scales showed no differences, a finding which in itself points to adequate psychological adjustment having taken place.

A second questionnaire, the Guilford Zimmerman Temperament Survey (GZTS), was also administered, employing for comparative purposes the standardised norms for adult women alone. The patients with gonadal dysgenesis scored significantly lower on the scale measuring general activity and energy, and on the Masculinity/Femininity scale, reinforcing the previous finding of a definite tendency to be more feminine. They scored significantly higher on the scale measuring general co-operation.

At this juncture it is relevant to introduce a reference to the work done by Bekker (1969) on the psychological characteristics of persons demonstrating various syndromes involving retarded growth and sexual infantilism. Included in the study were 15 patients with karyotype 45 XO and a further 11 with mosaic cell lines, resulting in phenotypes resembling Turner's syndrome. As already mentioned (p.53) only a summary of conclusions is given in English and consultation of tabulated data has been difficult. Personality assessment involved the use of projective tests - the Rorschach and the Thematic Apperception test (TAT). Bekker noted that his patients' outlook on life and their future needs and prospects tended to be naive, but their
personality development was not found to be as delayed as that of other patients having stunted growth, whose abnormality tended to become obvious at an earlier age. Bekker also commented specifically on the marked lack of vitality, which links well with Shaffer's results.

(ii) Incidence of mental illness

In general patients with Turner's syndrome demonstrate little psychopathology. The evidence from Shaffer's study using the MMPI emphasised this fact, in showing very little difference in the personality profile obtained for patients with Turner's syndrome compared with those for control groups. Hampson et al. (1955) rated their 11 patients on a 4-point scale of personality healthiness ranging from healthy to severely (morbidly) non-healthy. None of the patients fell into this last category; ten were regarded as "healthy"; the remaining one was described as "moderately unhealthy" on account of what the authors diagnosed as a mild adolescent adjustment problem.

Money et al. (1956), in their review of nearly 300 persons with varying types of hermaphroditism, found that patients with Turner's syndrome were scarcely more inclined towards mental disorder than were other types of hermaphrodites. Isolated case reports of patients with Turner's syndrome having some form of mental illness may be found in the literature. Since these are usually written for the purpose of describing the combination of illness with a rare syndrome, it is difficult to form a valid overall estimate of the incidence of mental illness amongst individuals with Turner's syndrome.

Michaux et al. (1967) collected reports on three surveys covering 402 schizophrenics. Whilst there was a raised incidence of 47 XXX females with schizophrenia, none was found with a 45 XO karyotype. However, the authors completed their paper by reporting the case of a girl who "after sexual relations and the revelation of her ovarian agenesis, had a confusional bout with deliriant ideas centred upon
her absence of ovaries." Follow-up on the girl indicated that the episode was an isolated one, and was possibly a result of the two trauma occurring concurrently. A similar episode was recorded by Sabbath et al. (1961); one of the seven patients they studied responded to receiving an engagement ring by demonstrating overt psychotic symptoms of both auditory and olfactory hallucinations. Hoffenburg et al. (1957) reviewed 27 cases, two of whom demonstrated psychotic illnesses. One had a manic depressive psychosis; the other developed an endogenous depression later on in her life. Since these details are given in tabulated form only there is no further information available.

Most authors suggest that the defences of patients with Turner's syndrome may already be generally weakened and strained by their stature and infantilism problems. For this reason it is particularly necessary that a sympathetic explanation should be made to them of their retarded sexual development and resulting infertility.

Electroencephalographic evidence from patients with Turner's syndrome has in some cases indicated the existence of some form of neurological abnormality. Mellbin (1965) described four cases of chromatin negative patients with both psychiatric illnesses and abnormal EEG recordings. The form taken by the psychiatric illnesses varied considerably. One case had a long-standing psychosis for which she had had to be hospitalised. A second, with epilepsy, had had one acute psychotic episode from which she completely recovered. The third had had a very clinging immature personality and at the age of 49 had had to be admitted to an old people's home. The fourth was reported briefly, and is mentioned more extensively in the section on anorexia nervosa. Mellbin suggested that the EEG abnormalities in the cases he reported were indicative of cerebral dysfunction, and that this might be responsible for the wide variety of psychiatric disorders observed in these patients.
Forssman et al. (1970) drew attention to reports on the coincidence of Turner's syndrome with anorexia nervosa. The authors suggested that the two syndromes were so rare that their concurrent appearance in the same person must have some significance. The first case was reported by Pitts and Guze (1963). Their patient was put on a restricted diet at the age of 13, because of obesity. She refused to abandon this diet when told that her weight was normal, and continued to lose weight by restricting her intake of food and exercising excessively. She also resorted to regurgitation of the infrequent meals she took, becoming increasingly depressed and self-critical, and having to be hospitalised on several occasions to stabilise her weight loss. Lindsten (1963) also mentioned a case of anorexia nervosa occurring in a patient with 45 XO Turner's syndrome. The patient reported by Forssman et al. (1970) first came to the authors' notice when she developed anorexia nervosa because she did not want to "grow fat like Mummy". The anorexia lasted about two years and then gradually disappeared after the patient was discharged and began to work. It was only ten years later, when she returned to the same department complaining of primary amenorrhoea, that the diagnosis of Turner's syndrome was made. A further case was reported by Dickens (1970), of a patient (one of a pair of dizygotic twins) who started to diet following her sister's example, but failed to stop when her sister did.

It is interesting to note that a common factor which might link Turner's syndrome and anorexia nervosa is EEG abnormality, although it is recognised that the last-mentioned, in particular, is difficult of definition. Crisp et al. (1968) proved that cerebral dysfunction, as assessed by the EEG, is fairly common in anorexia nervosa. Mellbin (1966), in his study of four patients with karyotypes 45 XO, found that they all had abnormal EEG's. Forssman et al. (1970) commented on the case of a patient suffering from both of these clinical entities, who had an abnormal EEG, as had the patient reported by Dickens above. These reports are of interest since this thesis includes in its sample of patients with 45 XO Turner's syndrome one girl who was just recovering from an episode of anorexia nervosa.
In conclusion, there is clearly a need for controlled data on personality characteristics, as well as for standardised methods of enquiry into psychiatric illness, as already suggested in connection with the survey reported in Chapter II.
METHOD

Subjects

The experimental group consisted of the majority of those subjects already described in Chapter III, with the exception of those who, for one reason or another, withdrew. A corresponding number of control subjects were tested with the Cattell 16 Personality Factor Questionnaire, but time limitations precluded control subjects from completing the remaining two questionnaires within the personality assessment battery.

Measuring Instruments

(i) The Hysteroid:Obsessoid Questionnaire (Caine and Hope, 1967). (termed the Self-Description Questionnaire)

The Hysteroid:Obsessoid Questionnaire (or HOQ) is a personality questionnaire which was devised to measure personality traits and attitudes as opposed to psychiatric signs and symptoms. Its origin is closely associated with Foulds's theory of personality dysfunction (Foulds 1965) and both this questionnaire and the following one (the Hostility and Direction of Hostility Questionnaire) represent an attempt to quantify and differentiate between personality traits and psychiatric symptoms. Foulds considers that they are differentiated in three ways:

(a) personality traits are common (universal), whilst psychiatric symptoms are not

(b) personality traits are relatively acceptable to both the patient and his associates; psychiatric symptoms are extremely distressful to the patient (on most occasions) and to his associates

(c) personality traits are relatively enduring, whilst psychiatric symptoms are fairly transient.
The HOQ is found to correlate highly with the Eysenck Personality Inventory (EPI, Eysenck and Eysenck, 1964) and is considered to correspond closely to the extraversion:introversion continuum, in which a hysteroid person is typically an extravert (outgoing and careless) and, conversely, the obsessoid type of person is an introvert (quiet and meticulous).

The Manual of the Hysteroid:Obsessoid Questionnaire (Caine and Hope, 1967) provides details on the validity and reliability of the Questionnaire, as well as normative data for various normal and psychiatric populations.

The Questionnaire of 48 items was given to the subject to fill in by herself, and she had to respond by indicating whether statements were "true" or "false".

**Scoring Procedure**

Scoring was achieved with the aid of a perspex stencil. Points were allotted for those items endorsed in a hysteroid direction. The manual suggests that patients scoring 24 or more on the HOQ are placed in the hysteroid category, the rest being regarded as obsessoid.

(ii) **The Hostility and Direction of Hostility Questionnaire** (Caine, Foulds and Hope, 1967)

This questionnaire (HDHQ) is also an attempt to elucidate personality traits and not psychiatric symptoms. It is designed to measure "a wide range of possible manifestations of aggression, hostility or punitiveness." (Caine, Foulds and Hope, 1967). It is suggested (Foulds, 1965) that the ability of a person to mature depends on establishing satisfactory mutual relationships. The immature person, experiencing difficulty in this, becomes egocentric in his thinking and behaving. Foulds suggested that "general punitiveness" could provide an indirect measure of egocentricity, since a feature of an immature personality is the need to apportion blame to other people or to self.
The questionnaire comprises items taken from the Minnesota Multiphasic Personality Inventory. These sub-divide to give five scales:

- acting out hostility (AH);
- criticism of others (CO);
- projected delusional hostility (PH);
- self-criticism (SC);
- delusional guilt (DG).

The first three scales are considered measures of hostility directed outwards towards other people (extrapunitiveness), the last two as measures of hostility directed inwards self (intropunitiveness).

The Manual of Hostility and Direction of Hostility Questionnaire (Caine, Foulds and Hope, 1967) gives details of validity and reliability, as well as scores relating to normal and psychiatric populations. The questionnaire is of the same "true/false" type as the HOQ.

**Scoring Procedure**

Scoring was again facilitated by using a perspex stencil. This indicated the categories which might be allotted to an item if it was endorsed in a particular direction.

(i) The total hostility score was obtained by summing all the categories, i.e. $AH + CO + PH + SC + DG$

(ii) The direction of hostility by

$2(SC + DG) - (AH + CO + PH)$

(iii) Philip (to be published) has collected data which support the view that extrapunitiveness and intropunitiveness should be considered as separate entities, where

- extrapunitiveness $= AH + CO + PH$
- intropunitiveness $= SC + DG$

It is those scores obtained from the first and last categories, as well as individual sub-scale scores, which have been used for comparative purposes.
(iii) **The Sixteen Personality Factor Questionnaire** (Cattell and Eber, 1965).

This questionnaire (16PF) assesses personality in terms of 16 first order factors and four second order factors. Cattell has always insisted on the multivariate analysis of personality (Cattell, 1965), and these factors arise from his factor analytic research.

Below are given brief descriptions of the bi-polar first order factors as set out in Philip's Ph.D. thesis (1968). It facilitates discussion and interpretation of the results to adopt these simpler terms rather than those used by Cattell, e.g. Parmia v. Threctia (Factor H).

<table>
<thead>
<tr>
<th>Factor</th>
<th>Low Score</th>
<th>High Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>Aloof</td>
<td>Warm, outgoing</td>
</tr>
<tr>
<td>B</td>
<td>Unintelligent</td>
<td>Intelligent</td>
</tr>
<tr>
<td>C</td>
<td>Emotionally unstable</td>
<td>Mature, stable</td>
</tr>
<tr>
<td>E</td>
<td>Submissive</td>
<td>Dominant</td>
</tr>
<tr>
<td>F</td>
<td>Reticent</td>
<td>Enthusiastic</td>
</tr>
<tr>
<td>G</td>
<td>Expedient</td>
<td>Conscientious</td>
</tr>
<tr>
<td>H</td>
<td>Shy</td>
<td>Venturesome</td>
</tr>
<tr>
<td>I</td>
<td>Tough-minded</td>
<td>Sensitive</td>
</tr>
<tr>
<td>L</td>
<td>Trustful</td>
<td>Suspecting</td>
</tr>
<tr>
<td>M</td>
<td>Practical</td>
<td>Self-absorbed</td>
</tr>
<tr>
<td>N</td>
<td>Simple</td>
<td>Sophisticated</td>
</tr>
<tr>
<td>O</td>
<td>Confident</td>
<td>Apprehensive</td>
</tr>
<tr>
<td>Q1</td>
<td>Conservative</td>
<td>Radical</td>
</tr>
<tr>
<td>Q2</td>
<td>Group dependent</td>
<td>Self-sufficient</td>
</tr>
<tr>
<td>Q3</td>
<td>Uncontrolled</td>
<td>Self-controlled</td>
</tr>
<tr>
<td>Q4</td>
<td>Relaxed</td>
<td>Tense</td>
</tr>
</tbody>
</table>

Second order factors are calculated from first order factors as follows: (Cattell and Eber, 1965)

**Anxiety**  
$$3.7 - 0.2C - 0.2H + 0.2L + 0.30 - 0.2Q_3 + 0.4Q_4$$

**Introversion:Extraversion**  
$$0.2A + 0.2E + 0.4F + 0.5H - 0.2Q_2 - 1.1$$
In line with Philip (1968) the remaining two second order factors were omitted, since their validity and description are as yet uncertain.

In order to compare one individual's scores on several factors it is possible to convert them to sten scores, which represent scores out of a total of ten. The raw score mean is fixed to give a sten score of 5.5 and raw scores which are one half of a standard deviation below or above the mean give stens of 5 and 6 respectively.

Form A (1962 edition) was used; the subject completed a separate answer sheet to respond to the 187 statements.

Scoring procedure

A cardboard stencil laid over the completed answer sheet indicated whether an endorsed item contributed a score of 2 or 1 to the total. The stencil was also so marked that the total raw score for each of the 16 first order factors might be entered on the answer sheet. These raw scores could then be converted to sten scores by referral to standardised tables (based on American populations). British norms, recently compiled by Saville (1972) were not available at the time of the study.

Procedure

The battery of personality tests was administered to the experimental and control groups respectively in the following manner:

Experimental group

The HOQ and HDHQ were administered to 19 subjects of this group and the 16 PF to 20. If the experimental subject was being tested at her home, she was given all three questionnaires after the first testing session, with a careful explanation of how to complete them, and asked to return them by post to the Unit in the stamped, addressed envelope provided for this purpose. Only one subject failed to return the forms.
Control group

As has already been mentioned time limitations did not allow for all the personality questionnaires to be completed by the control group. It was therefore decided that control 16 PF data should be collected in order to avoid having to use American norms for comparison of sten scores. The HOQ and HDHQ, on the other hand, have been standardised on British populations (albeit on much smaller numbers). Twenty members of the control group (Group 1) filled in the 16 PF at the surgery (taking on average at least one hour), after they had completed the three colour vision tests.

Administration of all three questionnaires was as stated in the manuals; the subjects were asked not to miss any items (either by intention or in error). It was also suggested that the best method to be adopted in judging the items was to respond with the initial reaction immediately on reading the question, and not to waste time in consideration. Finally, it was pointed out that items on the 16 PF could be answered in one of three ways, as opposed to the "true/false" format of the HOQ and HDHQ.

Statistical Analysis of results

For Hypothesis VII/1

The mean total score on the HOQ for the experimental group was compared with that of the largest group of normal female subjects (N=69) given in the manual (i.e. mean = 24.01; S.D. = 5.48) using a t-test.

For Hypothesis VII/2

(i) The mean total hostility score on the HDHQ for the experimental group was compared with

(a) that of the largest group of normal female subjects (N=31) given in the manual (i.e. mean = 12.1; S.D. = 5.1);

and

(b) that of a much larger group of normal females (N=330)
(Philip unpublished data) (i.e. mean = 14.42; S.D. = 6.28)

using t-tests.
(ii) Mean extrapunitiveness and intropunitiveness scores on the HDHQ for the experimental group were compared with those of Philip's group for normal females (N=330).

- Extrapunitiveness mean = 7.92; S.D. = 3.87
- Intropunitiveness mean = 6.50; S.D. = 3.42

using t-tests.

(iii) The mean sub-scale scores on the HDHQ for the experimental group were compared with those of the largest group of normal female subjects (N=31) given in the manual.

(i.e.)
- AH mean 3.4 S.D. 1.8;
- CO " 3.3 S.D. 2.0;
- PH " 0.5 S.D. 0.6;
- SG " 3.7 S.D. 2.1;
- DG " 1.2 S.D. 1.2

using t-tests.

For Hypothesis VII/3

(i) Mean raw scores on the 16 first order factors of the 16 PF were compared for the experimental and control groups, using t-tests.

(ii) Mean sten scores on the 16 first order factors of the 16 PF for the experimental group were compared with Philip's data on normal females (N=179), using t-tests.

(iii) Mean sten scores on the two second order factors of Anxiety and Extraversion:Introversion on the 16 PF were compared for the experimental and control groups, using t-tests.
HYPOTHESES

On the basis of the above discussion the following hypotheses were formulated:

VII/1 That the experimental group will have significantly lower scores on the HOQ than those given in the manual for normal females.

VII/2 (i) That the experimental group will have significantly higher total hostility scores on the HDHQ than (a) those given in the manual, and (b) those obtained from Philip data.  
(ii) That the experimental group will not differ significantly from the Philip group on extrapunitiveness, but that they will score significantly higher than the Philip group on intropunitiveness.  
(iii) That there will be no significant differences between the scores of the experimental group and the Manual norms on the HDHQ sub-scales, with the exception of SC (self-criticism), where it is hypothesised that the experimental group will score significantly higher.

VII/3 (i) That there will be no significant differences between the experimental and control groups' raw scores on the 16 first order factors, with the exception of those which contribute to the loadings for the second order factors of Extraversion:Introversion, namely Factors A, E, F, H and Q^2. It is hypothesised that the experimental group will score significantly lower than the control group on Factors A, E, F and H and significantly higher on Factor Q^2.
(ii) That a similar pattern to (i), of significant and non-significant differences, will emerge from comparison of sten scores for the experimental group and Philip data.

(iii) That, whilst there will be no significant difference between the experimental and control groups on the second order factor of Anxiety, the experimental group will have significantly lower scores on the Extraversion:Introduction second order factor.
RESULTS

Hypothesis VII/1
The results supported this hypothesis. Table XXVI shows the mean total HOQ scores for the experimental group and the Manual normal females.

Table XXVI  Mean total HOQ scores of the experimental and normal female groups (Manual norms)

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Manual norms</th>
<th>t</th>
<th>p*</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>19</td>
<td>69</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mean total HOQ score</td>
<td>20.37 (S.D. 4.97)</td>
<td>23.7 (S.D. 5.48)</td>
<td>2.72</td>
<td>&lt;.01</td>
</tr>
</tbody>
</table>

* 1-tailed test

This result indicated that the experimental group scored significantly lower on the HOQ; i.e., that they tended to demonstrate obsessoid, rather than hysteroid, personality characteristics.

Hypothesis VII/2
(i) The results supported both parts of this hypothesis. Table XXVII gives mean total hostility scores on the HDHQ for the experimental group, as well as (a) Manual norms and (b) those obtained from Philip data.

Table XXVII  Mean total hostility scores for the experimental group, as compared with Manual norms and Philip data

<table>
<thead>
<tr>
<th></th>
<th>N</th>
<th>Mean</th>
<th>S.D.</th>
<th>t</th>
<th>p*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Experimental Ss.</td>
<td>19</td>
<td>17.26</td>
<td>5.87</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Manual norms</td>
<td>31</td>
<td>12.1</td>
<td>5.1</td>
<td>3.17</td>
<td>&lt;.005</td>
</tr>
<tr>
<td>Philip data</td>
<td>330</td>
<td>14.42</td>
<td>6.28</td>
<td>2.04</td>
<td>&lt;.025</td>
</tr>
</tbody>
</table>

* 1-tailed test

These results indicated that the experimental group obtained significantly higher hostility scores than either Manual norms or Philip data.
(ii) The results did not support this hypothesis. Table XXVIII shows mean extrapunitive and intropunitive scores for the experimental group and Philip data.

Table XXVIII Mean extrapunitive and intropunitive scores for the experimental group and Philip data

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Philip data</th>
<th>t</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td>19</td>
<td>330</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mean</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mean</td>
<td>9.74</td>
<td>7.92</td>
<td></td>
<td>.1</td>
</tr>
<tr>
<td>S.D.</td>
<td>4.54</td>
<td>3.87</td>
<td>1.71</td>
<td></td>
</tr>
<tr>
<td>Extrapunitive</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Intropunitive</td>
<td>7.53</td>
<td>6.5</td>
<td>1.47</td>
<td>N.S.</td>
</tr>
<tr>
<td>S.D.</td>
<td>2.94</td>
<td>3.42</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

** 2-tailed test

These results indicate that whilst there were no differences in the hypothesised direction on intropunitive scores, there was a trend (which failed to reach significance) which indicated that the experimental group were more extrapunitive than the group of females tested by Philip.

(iii) The results partially supported this hypothesis. Table XXIX shows the mean sub-scale scores for the experimental group and compares them with norms obtained from the HDHQ manual.
Table XXIX  Mean sub-scale scores for the experimental group and Manual norms

<table>
<thead>
<tr>
<th>Sub-scale</th>
<th>Experimental Ss.</th>
<th>Manual norms</th>
<th>t</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N</td>
<td>Mean</td>
<td>S.D.</td>
<td>Mean</td>
</tr>
<tr>
<td>AH</td>
<td>19</td>
<td>4.0</td>
<td>1.78</td>
<td>3.4</td>
</tr>
<tr>
<td>CO</td>
<td>19</td>
<td>4.84</td>
<td>2.91</td>
<td>3.3</td>
</tr>
<tr>
<td>PH</td>
<td>19</td>
<td>0.89</td>
<td>0.83</td>
<td>0.5</td>
</tr>
<tr>
<td>SC</td>
<td>19</td>
<td>5.84</td>
<td>1.95</td>
<td>3.7</td>
</tr>
<tr>
<td>DG</td>
<td>19</td>
<td>1.68</td>
<td>1.30</td>
<td>1.2</td>
</tr>
</tbody>
</table>

* 1-tailed test
** 2-tailed test

These results confirmed the hypothesis that the experimental group would score significantly higher on SC items (self-criticism). The Table also indicates that the experimental group scored significantly higher on the CO sub-scale (criticism of others) and that there was also a non-significant trend for them to score higher on PH items (projected delusional hostility).

Hypothesis VII/3

(i) The results partially supported this hypothesis. Table XXX gives the mean raw scores on the 16 first order factors for the experimental and control groups.
Table XXX  Mean first order factor raw scores on the 16 PF for experimental and control groups

<table>
<thead>
<tr>
<th>Factor</th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>t</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean</td>
<td>S.D.</td>
<td>Mean</td>
<td>S.D.</td>
</tr>
<tr>
<td>Factor A</td>
<td>12.25</td>
<td>2.22</td>
<td>12.25</td>
<td>3.82</td>
</tr>
<tr>
<td>&quot; B</td>
<td>6.65</td>
<td>1.95</td>
<td>7.05</td>
<td>1.70</td>
</tr>
<tr>
<td>&quot; C</td>
<td>14.00</td>
<td>3.77</td>
<td>13.60</td>
<td>3.99</td>
</tr>
<tr>
<td>&quot; E</td>
<td>8.55</td>
<td>3.10</td>
<td>8.40</td>
<td>3.90</td>
</tr>
<tr>
<td>&quot; F</td>
<td>12.00</td>
<td>4.09</td>
<td>14.55</td>
<td>5.63</td>
</tr>
<tr>
<td>&quot; G</td>
<td>13.75</td>
<td>2.55</td>
<td>13.00</td>
<td>3.16</td>
</tr>
<tr>
<td>&quot; H</td>
<td>8.30</td>
<td>3.73</td>
<td>10.75</td>
<td>5.55</td>
</tr>
<tr>
<td>&quot; I</td>
<td>11.15</td>
<td>2.92</td>
<td>12.15</td>
<td>2.39</td>
</tr>
<tr>
<td>&quot; L</td>
<td>6.90</td>
<td>3.54</td>
<td>7.15</td>
<td>2.83</td>
</tr>
<tr>
<td>&quot; M</td>
<td>12.30</td>
<td>2.56</td>
<td>12.90</td>
<td>3.40</td>
</tr>
<tr>
<td>&quot; N</td>
<td>11.45</td>
<td>2.39</td>
<td>11.40</td>
<td>2.62</td>
</tr>
<tr>
<td>&quot; O</td>
<td>11.85</td>
<td>4.66</td>
<td>13.50</td>
<td>4.41</td>
</tr>
<tr>
<td>&quot; Q₁</td>
<td>7.95</td>
<td>2.50</td>
<td>8.55</td>
<td>2.56</td>
</tr>
<tr>
<td>&quot; Q₂</td>
<td>13.10</td>
<td>2.75</td>
<td>11.10</td>
<td>3.34</td>
</tr>
<tr>
<td>&quot; Q₃</td>
<td>13.05</td>
<td>2.89</td>
<td>10.25</td>
<td>3.14</td>
</tr>
<tr>
<td>&quot; Q₄</td>
<td>12.00</td>
<td>4.45</td>
<td>14.05</td>
<td>4.12</td>
</tr>
</tbody>
</table>

* 1-tailed test
** 2-tailed test

These results:
(a) supported the hypothesis in indicating that the experimental subjects scored higher than the control group on Factor Q₂ (i.e. they were more self-sufficient than the control group);
(b) partially supported the hypothesis in indicating non-significant trends in the directions postulated for Factors F and H, indicating that the experimental group seemed more likely to be reticent (\(F\)) and shy (\(H\)) than the control group;

(c) did not support the hypothesis in that they did not indicate any significant difference between the experimental and control groups on Factors A and E;

(d) supported the null hypothesis for the remaining factors, except for Factor \(Q_3\) where the experimental group scored significantly higher than the control group, indicating that they were more self-controlled.

(ii) The results supported the hypothesis more strongly when the Philip data employing sten scores from 179 females were used for comparative purposes. Table XXXI gives the mean sten scores for the two groups.
### Table XXXI

Mean first order factor stem scores on the 16 PF for experimental group and Philip data

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Philip data</th>
<th>t</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mean</td>
<td>S.D.</td>
<td>Mean</td>
<td>S.D.</td>
</tr>
<tr>
<td>N</td>
<td>20</td>
<td>179</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Factor A</td>
<td>5.75</td>
<td>1.41</td>
<td>5.18</td>
<td>2.05</td>
</tr>
<tr>
<td>&quot; B</td>
<td>6.50</td>
<td>1.64</td>
<td>7.59</td>
<td>1.49</td>
</tr>
<tr>
<td>&quot; C</td>
<td>4.80</td>
<td>2.02</td>
<td>5.89</td>
<td>1.59</td>
</tr>
<tr>
<td>&quot; E</td>
<td>4.90</td>
<td>1.62</td>
<td>5.90</td>
<td>2.12</td>
</tr>
<tr>
<td>&quot; F</td>
<td>4.85</td>
<td>1.79</td>
<td>5.85</td>
<td>2.27</td>
</tr>
<tr>
<td>&quot; G</td>
<td>5.75</td>
<td>1.68</td>
<td>5.22</td>
<td>1.83</td>
</tr>
<tr>
<td>&quot; H</td>
<td>3.95</td>
<td>1.54</td>
<td>5.03</td>
<td>2.07</td>
</tr>
<tr>
<td>&quot; I</td>
<td>5.00</td>
<td>1.95</td>
<td>5.72</td>
<td>2.24</td>
</tr>
<tr>
<td>&quot; L</td>
<td>5.25</td>
<td>2.38</td>
<td>5.26</td>
<td>2.01</td>
</tr>
<tr>
<td>&quot; M</td>
<td>5.25</td>
<td>1.52</td>
<td>5.62</td>
<td>1.99</td>
</tr>
<tr>
<td>&quot; N</td>
<td>6.20</td>
<td>1.82</td>
<td>5.31</td>
<td>2.21</td>
</tr>
<tr>
<td>&quot; O</td>
<td>6.20</td>
<td>2.26</td>
<td>5.70</td>
<td>1.72</td>
</tr>
<tr>
<td>&quot; Q₁</td>
<td>5.05</td>
<td>2.14</td>
<td>5.94</td>
<td>1.80</td>
</tr>
<tr>
<td>&quot; Q₂</td>
<td>7.35</td>
<td>1.39</td>
<td>5.85</td>
<td>2.10</td>
</tr>
<tr>
<td>&quot; Q₃</td>
<td>6.40</td>
<td>1.79</td>
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<td>2.08</td>
</tr>
<tr>
<td>&quot; Q₄</td>
<td>5.20</td>
<td>1.77</td>
<td>5.37</td>
<td>1.88</td>
</tr>
</tbody>
</table>

* 1-tailed test

** 2-tailed test
These results:

(a) supported the hypothesis and confirmed the non-significant trends noted for Factors F and H in the previous section, in indicating that the experimental subjects gained significantly lower scores than Philip data on Factors E, F and H, indicating that they were more submissive (E), reticent (F) and shy (H). The experimental group again scored significantly higher on Factor Q2 (indicating that they were more self-sufficient);

(b) endorsed the previous finding on Factor Q3 which indicated that the experimental subjects were more self-controlled;

(c) showed further significant differences between the two groups, namely on Factor B (Philip subjects were more intelligent than the experimental group), Factor C (experimental group were inclined to be more emotionally unstable than that tested by Philip), Factor N (experimental group might be more sophisticated than the Philip group), plus a non-significant trend on Factor Q1, which suggests that the experimental subjects might be more conservative.

Before conclusions are drawn from these data consideration must be given to the intellectual ability and social background of the subjects contained in the Philip survey, and a comparison made between them and those included in the control group (see p. 196).

(iii) The results supported this hypothesis. Table XXXII gives the mean second order factor scores (Anxiety and Extraversion: Introversion) for the experimental and control groups.

Table XXXII  Mean Second order factor scores on the 16 PF for the experimental and control groups

<table>
<thead>
<tr>
<th></th>
<th>Experimental Ss.</th>
<th>Control Ss.</th>
<th>t</th>
<th>p*</th>
</tr>
</thead>
<tbody>
<tr>
<td>N</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>20</td>
<td>20</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mean</td>
<td>5.66</td>
<td>6.42</td>
<td>1.22</td>
<td>N.S.</td>
</tr>
<tr>
<td>S.D.</td>
<td>2.06</td>
<td>1.87</td>
<td>1.58</td>
<td>0.22</td>
</tr>
<tr>
<td>Factor 1</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>(Anxiety)</td>
<td>3.48</td>
<td>4.70</td>
<td>1.88</td>
<td>&lt;.05</td>
</tr>
<tr>
<td>S.D.</td>
<td>1.58</td>
<td>2.43</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* 1-tailed test
This table indicates that, whilst the experimental and control groups did not differ significantly on the second order factor of Anxiety, the experimental group was more introverted than the control group.
DISCUSSION

Testing individuals with Turner's syndrome on three personality inventories demonstrated several areas in which the experimental group were different from normal. It is recognised that the lack of control group data for the Hysteroid:Obsessoid Questionnaire and the Hostility and Direction of Hostility Questionnaire, and the smallness of the numbers of subjects involved in all the personality assessments, limit the conclusions which may be drawn from such a survey. In many instances, however, inter-test similarities gave some idea of the reliability of the results achieved.

Hysteroid:Obsessoid Questionnaire

Only four of the 19 individuals with Turner's syndrome obtained scores on this test which placed them in the hysteroid category. The remainder were classified as obsessoid. That the group tested were predominantly obsessoid was confirmed by comparing their mean scores with those given by the manual. This showed that the experimental group had significantly lower scores than normals, which placed them, as a group, in the obsessoid category. Expressed in terms of means and standard deviations their scores were more characteristic of psychiatric populations, and in particular of neurotic ones.

It may therefore be concluded that individuals with Turner's syndrome are likely to display little emotion, preferring to remain in the background and not to attract attention. They also tend to be slow in making decisions, a finding which could be seen to be relevant to the discussion in Chapter III on psychomotor retardation.

Hostility and Direction of Hostility Questionnaire

The experimental group obtained significantly higher total hostility scores on this questionnaire than did the group of normals upon whom the test was standardised. This was also true of the comparisons made with Philip normative data. It was considered
advisable to utilise the Philip data as well as the norms given by the manual for two reasons:

(i) Philip obtained his results from a larger normal population;
(ii) Philip (1968) suggested that local norms on such tests may vary, and it was therefore expedient to employ Aberdeen norms, which were more appropriate geographically and culturally than those obtained from an English population (i.e. Essex). It should be noted that his normative group contained a high proportion of hospital staff of various grades and showed a bias towards higher socio-economic status. As may be seen from the Sixteen Personality Factor Questionnaire results, this in itself raises problems in the interpretation of significant differences between the experimental and control groups.

Having established that a high degree of hostility existed, it was necessary to investigate the directions in which it was channelled. If the Direction of Hostility was calculated in the manner suggested by the manual, the results showed that 13 of the 19 experimental subjects directed their hostility intropunitively. However, in terms of the Manual normative data for normals there was no indication of abnormality in the amount of Direction of Hostility. On the other hand, if the directions were considered separately (as suggested by Philip: personal communication), in terms of hostility directed inwardly compared with that directed externally, and these measures were compared with Philip data, a non-significant trend could be noted for the experimental group to express hostility towards other people more often than normals did. There was no indication of a difference in the tendencies of the two groups to direct hostility inwardly towards themselves.

These somewhat equivocal results were only resolved by comparing mean sub-scale scores on the test. This comparison was made of necessity with Manual norms, and showed particularly clearly that the experimental group, besides achieving a non-significant trend towards
higher Projected Delusional Hostility scores, achieved particularly significantly higher scores on the Self-criticism and Criticism of Others sub-scales. This indicates that, in addition to being highly critical of themselves, individuals with Turner's syndrome are also critical of other people. Since the former is an intropunitive and the latter an extrapunitive measure, this finding explains the non-significant Direction of Hostility discriminant discussed above.

If these results are extended to include the original theory upon which the test is based, it might be suggested that individuals with Turner's syndrome, in being more hostile than normals, are accordingly less mature. Instead, however, it could be maintained that they have sufficient cause, in terms of their physical stigmata and concurrent reactions of normal people to these, to justify their high level of hostility. This should not be viewed as an indication of a neurotic state (as, for example, it would be in patients in the psychiatric category), but rather as a natural reaction to their condition, and one which a clinician should be aware of when dealing with such patients.

**Sixteen Personality Factor Questionnaire**

This discussion is primarily concerned with those significant differences which emerged from comparison of the experimental group with the smaller group of controls, those tested by the author. It was thus possible to use raw scores, without requiring to convert the scores to stens employing American norms; and the groups were better matched in terms of intellectual and socio-economic status. The Philip data provided a useful source of confirmatory material, particularly in the case of non-significant trends which might be observed in initial comparison. It is noted that t-tests carried out between the experimental group and Philip data had to be viewed with caution, on account of the large difference in the N of the two groups (i.e. 20 and 179).
Two primary factors emerged which discriminated clearly between the experimental and control groups — those of $Q_2^+$ and $Q_3^+$. High scores on Factor $Q_2$ indicated that individuals with Turner's syndrome were more self-sufficient, preferring to make their own decisions, and not being group dependent. It could be suggested that this is an adaptive process brought about partially by rejection by peer groups, and partially by self-consciousness, since a problem which very frequently became apparent during interviews was the lack of social contacts and activities outside work and family. Scoring higher than controls on Factor $Q_3^+$ indicated a tendency to be very controlled and socially exact, and corresponded in many ways to factor G (super-ego). Again, it might be suggested that this result reflects the extent to which the patient is isolated by her syndrome and remains within the family structure to a greater degree than is usual. Mothers of such patients in particular are naturally protective towards them and, as a result of their daughters' symptoms, have a fairly close relationship with them. It could therefore be suggested that the patients acquire many of the parents' values and social mores to a greater extent than is usual.

Two other primary factors emerged as trends. These were factors $F$ and $H$, and the trends were confirmed by comparison with Philip data. Low scores on Factor $F$ indicate characteristics of sobriety and caution, with such a person being beset by cares and possibly worries. It would again seem appropriate for a patient with Turner's syndrome to score in this manner, since her physical condition is such that it cannot be alleviated or cured, and in some cases is not fully explained to the patient.

The experimental group's low scores on Factor $H$ are particularly interesting, since the factor has been shown to have appreciable constitutional and autonomic associations (Cattell et al., 1970). Such individuals are very shy, emotionally cautious, and tend to withdraw from personal contacts. It has been suggested that,
constitutionally, these individuals have an over-responsive sympathetic nervous system, which makes them particularly "threat reactive". It may be seen that this factor, taken together with factor \( Q_2^+ \), whether they precede or succeed the physical conditions associated with the Turner syndrome, produces a rather depressing social prognosis for such patients.

As far as further comparison with Philip data is concerned, it may be seen from Table XXXIII that some of the apparent significant differences arising from such comparison might be dismissed because they seem (from comparison with control data) to be related to intellectual, socio-economic variables, rather than to the medical condition of Turner's syndrome.
Table XXXIII  Mean first order sten scores on the 16 PF for control group and Philip data

<table>
<thead>
<tr>
<th>Factor</th>
<th>Control Ss.</th>
<th>Philip data</th>
<th>t</th>
<th>p**</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>20</td>
<td>179</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Mean S.D.</td>
<td>Mean S.D.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Factor A</td>
<td>5.8 2.29</td>
<td>5.18 2.05</td>
<td>1.16</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; B</td>
<td>6.8 1.32</td>
<td>7.59 1.49</td>
<td>2.50</td>
<td>&lt;.05</td>
</tr>
<tr>
<td>&quot; C</td>
<td>4.65 1.84</td>
<td>5.89 1.59</td>
<td>2.90</td>
<td>&lt;.01</td>
</tr>
<tr>
<td>&quot; E</td>
<td>4.85 1.81</td>
<td>5.90 2.12</td>
<td>2.42</td>
<td>&lt;.05</td>
</tr>
<tr>
<td>&quot; F</td>
<td>6.15 2.58</td>
<td>5.85 2.27</td>
<td>.50</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; G</td>
<td>5.55 1.82</td>
<td>5.22 1.83</td>
<td>.77</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; H</td>
<td>4.90 2.25</td>
<td>5.03 2.07</td>
<td>.25</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; I</td>
<td>5.70 1.53</td>
<td>5.72 2.24</td>
<td>.05</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; L</td>
<td>5.50 1.96</td>
<td>5.26 2.01</td>
<td>.52</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; M</td>
<td>5.55 2.14</td>
<td>5.62 1.99</td>
<td>.14</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; N</td>
<td>6.25 1.80</td>
<td>5.31 2.21</td>
<td>2.16</td>
<td>&lt;.05</td>
</tr>
<tr>
<td>&quot; O</td>
<td>6.90 2.10</td>
<td>5.70 1.72</td>
<td>2.46</td>
<td>&lt;.05</td>
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<tr>
<td>&quot; Q₁</td>
<td>5.55 1.70</td>
<td>5.94 1.80</td>
<td>.97</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; Q₂</td>
<td>6.20 1.85</td>
<td>5.85 2.10</td>
<td>.79</td>
<td>N.S.</td>
</tr>
<tr>
<td>&quot; Q₃</td>
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<td>4.84 2.08</td>
<td>.74</td>
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</tr>
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<td>&quot; Q₄</td>
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<td>5.37 1.88</td>
<td>1.19</td>
<td>N.S.</td>
</tr>
</tbody>
</table>

**2-tailed test

From this table it is clear that differences on Factors B, C, E and N may not be considered as typical of patients with Turner's syndrome; only the non-significant trend for the experimental group to score lower than the Philip group on Factor Q₁ may be mentioned,
as indicating that the former are more likely to be conservative in their thought and behaviour, a finding which is not incompatible with the high loading on Factor $Q_2$ for this group.

Differences on second-order factors emerged as predicted. The experimental group showed no variation from normal on the Anxiety factor, indicating that they were generally as well adjusted as normal females. As would be expected from the profile of primary factors which has been discussed above, together with the result of the Hysteroid:Obsessoid Questionnaire, the experimental group emerged as significantly different on the second-order factor Introversion/Extraversion in an introverted direction.

In summary, the conclusions to be drawn from the results presented indicate that patients with Turner's syndrome are typically an introverted group of individuals with a high level of hostility against themselves and other people, managing to cope with this by abiding strictly by social rules and also by being naturally withdrawn and wary of social interactions. The fact that the second-order factor of Anxiety/Adjustment resembled that of control groups reinforces the hypothesis that individuals with Turner's syndrome are not prone to psychological maladjustment, which, of course, is not the case with the other classes of cytogenetic abnormality mentioned in Chapter I. As may be seen from the case notes contained in the Appendix, two subjects had received psychiatric attention, one for anorexia nervosa, the other for agitated depression. On the personality inventories on which they were assessed, their scores, by falling at the extremes of the measures, placed them within the limits scored by psychiatric populations.
CHAPTER VIII

GENERAL CONCLUSIONS

The general area concerning psychological aspects of individuals with sex chromosome abnormalities has been reviewed. The study undertaken was primarily concerned with the sex chromosome abnormality in which there is a lack of genetic material involving the X chromosome (i.e. 45 XO). There are certain points, however, which may be mentioned in connection with the three types of sex chromosome abnormality in which there is an excess of genetic material (i.e., 47 XXY, 47 XYY, 47 XXX). From clinical experience of interviewing and testing individuals with such abnormalities arose the impression that suggestions of an increased frequency of mental subnormality in these individuals should be investigated more fully. It seems clear that some type of intellectual deficit is present, but in order to define this deficit more closely careful comparative studies employing family members as controls are necessary. It is suggested that such studies might reveal a tendency not towards a direct and increased probability of mental retardation, but towards a comparatively depressed IQ. If the "family IQ" were low, that of the abnormal member might well fall within subnormal limits. In addition, as has been stressed throughout the thesis, it is essential to obtain data from persons with sex chromosome aneuploidy ascertained from random surveys before effective conclusions can be drawn from psychological assessment. Patients with Turner's syndrome, on the other hand, pose fewer ascertainment problems, in that they are clinically recognisable on account of their phenotypic abnormalities. As a group they are genetically unique, since they lack cytogenic material.
A general survey of the case histories of a population of these individuals revealed areas in which further study is required, particularly with reference to marital problems, obesity and treatment procedures. This could be of direct help in the treatment and counselling of such patients.

Against this general background a smaller group were intensively studied and a profile based on the results of psychological assessment was obtained. Previous studies on IQ in patients with Turner's syndrome were confirmed by the findings of the present study. Analysis of the data showed a shift of Full Scale IQ scores towards the lower IQ ranges, with the significantly lower scores on all Performance items being chiefly responsible. Interpretation of the results differed from previous research in which control group data were not utilised. In particular it has been suggested that some form of psychomotor retardation may play a larger part in depressing Performance Scale IQ. In this connection two points may be mentioned and tentative suggestions put forward.

1. There appears to be no strong evidence that the group represented by the karyotypes 47 XXY, 47 XXX, 47 XXX had significantly lower scores on the Performance sub-tests. The scores on the Digit Symbol sub-test, in particular, were at a level which would be expected from the overall IQ. In this context the finding of Wright (personal communication) is interesting. He found that peripheral nerve conduction in the afore-mentioned group was significantly slower than normal. This would not appear to produce a detectable effect on psychomotor speed. In the group with Turner's syndrome no slowing in peripheral nerve conduction was demonstrated, and, indeed, there might even have been an increase. The observed low scores on the Performance sub-tests, and on the Digit Symbol sub-test in particular, of the group with Turner's syndrome would seem to indicate the existence of some central effect, and are not explicable in terms of slowing of peripheral nerve conduction.
2. Certain physical features of adult patients with Turner's syndrome are reminiscent of those of an older person, e.g. dried, crinkly skin on hands, fine and thinning hair, cardiac complications. It may therefore be postulated that the observed psychomotor retardation might have some connection with an ageing effect, possibly therefore related to the absence of the appropriate hormones at puberty, causing a premature onset of ageing effects usually associated with later periods of the life-span. To clarify these points further research is required, both cross-sectional, involving children as well as adults, and longitudinal.

It remains very clear to the author that psychomotor retardation is by no means the principal factor affecting Performance sub-tests. From observation of the difficulties experienced in attempting to complete the Object Assembly sub-test in particular it became obvious that further research should involve an attempt to examine the cognitive processes utilised by patients with Turner's syndrome.

Attention was focused on processes involved in vision, touch and hearing. It is interesting to note that the processes of smell and taste, although not studied in this thesis, have been investigated by Henkin (1967). In nine patients with Turner's syndrome he found raised median detection and recognition thresholds for sour and bitter tastes, as well as raised thresholds for all vapours tested.

Previous research had indicated abnormality within the visual processing systems of patients with Turner's syndrome. Such abnormalities were variously termed "space form dysagnosia", "spatial deficit", "visuospatial defect", such definitions having arisen primarily from intelligence test data, and secondarily from tests considered to measure such disabilities. Labels of this nature contributed little to an understanding of what is very clearly a unique deficiency associated with genetic imbalance of a particular kind. Indeed, confusion arises in the definition of spatial
perception itself, and in its complex inter-relation with form perception. Without attempting such a definition it is at least possible to state that spatial ability is not measured by Cohen's factor of Perceptual Organisation, and that, conversely, a low score on this factor is not invariably associated with poor spatial ability. Indeed, as has already been noted, Cohen (1957) stated specifically that the Perceptual Organisation factor should not be identified as a spatial factor, since it correlated highly with sub-tests which had no involvement with spatial ability tests (e.g. Picture Arrangement).

Attempts to define spatial ability have sometimes involved factor analysis of tests evolved to measure that ability. Such an analysis by Michael et al. (1957) produced three factors, which were identified as

1. Spatial relations and orientation
2. Visualisation
3. Kinaesthetic imagery

The following discussion indicates in what sense it would seem that patients with Turner's syndrome are deficient on all three factors.

1. Spatial relations and orientation: defined as "an ability to comprehend the nature of the arrangement of elements within a visual stimulus pattern with respect to the examiner's body as frame of reference." Geometric relationships of component elements are topological in that movement of one causes no change in the relationships of others. It is also implied that depth perception, in which three-dimensional distinctions are made, is involved. Piaget and Inhelder (1956, as cited in Piaget and Inhelder, 1969) have described the first spatial intuitions of the developing child in topological terms. Some of the errors perpetrated by individuals with Turner's syndrome in a simple copying test (the Bender Visual Motor Gestalt test) suggest difficulties in visual perception typical of this stage, in that errors occur in contiguity or positioning of
points in relation to other parts of composite figures. It is not suggested, however, that such individuals are necessarily arrested at this stage in their spatial ability development, since they must have undergone other types of maturational changes in order to reproduce the more sophisticated figures, albeit incorrectly. It is difficult to separate concepts of topological geometry from those of three-dimensional representation in these results. It could be argued that two figures reproduced correctly in form, but incorrectly in terms of topological relationship with each other, are being viewed as lying in different planes. Gregory (1970) has drawn attention to the ability of the brain to interpret images essentially transmitted as two-dimensional figures as objects having three-dimensional shape. It is interesting to speculate that the errors of contiguity, as isolated by the Bender Visual Motor Gestalt test, may represent some kind of incorrect processing of perceptual images, either in the input stage or in the reproductive stages, which manifests itself as an inability to achieve correct contiguity of figures when the designs are being reproduced. It would seem most likely that errors occur at the input stage, since those patients who made mistakes rarely seemed distressed by their attempts at reproduction, and tended not to correct their initial drawings with the rubber provided. This observation is reinforced by the experimental Visual Recognition test results, in which no actual reproduction in the form of drawing was necessary. Further research in this area could well concentrate on interpretation of ambiguous figures, which it might be postulated would be incorrectly solved by individuals with Turner's syndrome.

2. Visualisation: requires "mental manipulation of visual objects."

In previous discussion it has been suggested that deficient performance on the Object Assembly sub-test could be linked with an inadequate visual schema for the task in hand. Performance on the experimental Formboards test may be facilitated by a good ability to visualise,
both for the boards presented visually, and for them presented haptically when visual memory could play a part if the formboards have already been presented visually.

3. Kinaesthetic imagery: this is termed a "tentative" factor by Michael et al. and is dependent on left-right discriminations. As such it has not been incorporated into this battery of tests, but it will be recalled that it was specifically tested by the Road-map test devised by Money et al. (1965), who showed individuals with Turner's syndrome to be deficient in this area.

From this discussion it may be inferred that the author favours a more 'global' explanation of the psychological phenotype described in connection with the genetic imbalance associated with Turner's syndrome. Money (1968), in enumerating "space form dysgnosia, directional sense dysgnosia and mild dyscalculia" as associated with the syndrome, postulated a developmental right parietal lobe anomaly. Without necessarily negating this suggestion, data presented in this thesis do seem to warrant consideration of the possible involvement of other areas, particularly the striate cortex. It seems unjustified in the present state of knowledge to attempt to localise too closely the area involved. Instead, Luria's concept (1966) of a "complex functional system" seems more appropriate, involving as it does a series of simple functions and interactions. Thus, adequate visual perception depends on retinal cell firing, neuronal conduction within the optic tracts, and cortical function.

In many ways this concept is more acceptable since it is generally assumed that each cell contained within a patient with Turner's syndrome is genetically abnormal, in that it lacks X chromosome material. The effect of such an abnormality on specifically differentiated cells (e.g. rods, taste buds, or neurones) is unknown, but it seems reasonable to postulate that some types of cell may remain unaffected if the genetic material carried on the X chromosome is not
involved in their differentiation, whilst the opposite must be true of other types. In this context it may be recalled that genetic material of chromosomes is composed of biochemical chains (deoxyribose-nucleic acid - D.N.A.) which provide the template for messenger-R.N.A. (ribose-nucleic acid), a chemical substance necessary for the carrying out of cell functions. Experiments have shown that R.N.A. concentration in neuronal cells is reduced by minimising environmental input and increased by complex stimulation (Riesen, 1970). Correlated with the increase in R.N.A. is a growth of fine cell structures. It is interesting to speculate whether the growth of these finer structures suffers as a result of lack of initial D.N.A. as well as of decreased environmental stimulation. The genetic abnormality giving rise to Turner's syndrome features concerns some degree of absent D.N.A. in the form of X chromosome material. Such speculations have to be affected by the fact of the existence of the Y chromosome in males. Whilst being a very small chromosome, with less D.N.A. than an X chromosome, it clearly carries enough material to be equated with an X chromosome, and males are not different from normal females in the same way as patients with Turner's syndrome are.

There may also be considered the molecular model for learning and memory postulated by Hyden (1970), which involves those parts of the gene not normally seen as "active" (about 90%). Hyden has suggested that such areas become activated by the impact of environmental factors and induce synthesis of R.N.A. If this model is applied to Turner's syndrome it implies that, since the normal amount of gene area is never present, the potentiality for such development is reduced.

Of relevance to this discussion is the Lyon hypothesis, previously mentioned on p.158). It suggests that activity of the genes located on the X chromosome forming the chromatin body may be suppressed in females. Recent research has resulted in the hypothesis being modified, in that parts of the supposedly inactive material have been
shown to continue to influence particular immunological reactions of the body. There is also the possibility that the inactive X chromosome was active in the very early stages of cellular development in the foetus. At birth, brain cells have undergone the main differentiation which makes them nerve cells, and the chromatin body is clearly present. Even when Hebb (1949) postulated his theory of primary and secondary learning, he maintained that "primary learning" required initial formation of complex perceptual and motor organisations in the brain, upon which the environment might then act to bring about normal processes of learning and memory. If this initial stage fails to occur because of faulty genetic coding there will be a resultant developmental deficiency.

Such theories clearly have relevance in connection with a cytogenetic thesis, but historically they have evolved with emphasis being placed on the mechanisms involved in memory and learning. Gibson (1966) has pointed out the fallacy of making a distinction between mechanisms of short-term memory, learning and perception, and has suggested instead the use of the word 'apprehension' as a substitute for the active processes involved. Garner (1966), in considering perceptual organisation by means of the study of differential effects of redundancy, has emphasised the cognitive aspects of perception. "Perceiving is a cognitive process involving knowing, understanding, comprehending, organising, even cognising" and "As such, perception is much more closely related to classification, conceptualisation and free-recall learning than to sensory or discriminatory processes."

The model adopted by Neisser (1966) considers that perception involves the active processes of synthesising or constructing visual figures. As far as present knowledge extends, the initial process is the formation of an "icon", a term adopted by Neisser to explain the phenomenon of transient visual memory described by Sperling (1960). Sperling showed that, although a rectangular array of nine letters
(3 x 3) was not read and correctly reported after 50 msec. exposure time, the line indicated by a pre-arranged tone immediately after exposure of the display was repeated with nearly 100% efficiency. The term "icon" was adopted to correspond to the description of a "visual image" reported by the subjects. With regard to the suggestions already put forward to explain data on registration times (with reference to the experimental Visual Recognition test), and visualisation abilities (with reference to the Object Assembly sub-test), it might be relevant to repeat Sperling's experiment with patients with Turner's syndrome. In this type of experiment emphasis has been placed on the role of verbal coding in transferring the stimulus to short-term memory. The high correlation observed between performance on the Benton Visual Retention test and VIQ has already been noted, lending support to the idea that the more verbally adequate patients with Turner's syndrome used their skills to deal with the potentially difficult problems presented by the test. It could be argued from the fact that the experimental group performed adequately on the Similarities sub-test of the W.A.I.S. that there are no concurrent difficulties involving relationships of verbal concepts. Along more subtle lines of approach it could be postulated that investigation of the ability to comprehend and use words carrying spatial relationship meanings, especially those expressed by prepositions and adverbial phrases, might reveal relevant difficulties.

Eye movements have also been found to play an important role in perception. Whilst rapid tachistoscopic presentation does not allow of successive fixations, eye movements after presentation are observed (Crovitz and Davies, 1962). It could be postulated that the subjects under study are generally slower in their eye movements, which might cause scanning problems and inadequate registration. However, it is important to point out that performances on the Benton Visual Retention and Bender Visual Motor Gestalt tests provided evidence of sufficient registration of the configuration of the
figures to be represented. If, therefore, eye movement data are relevant, they may be relevant only to difficulties experienced with reference to detailed relationships. In the absence of evidence to this effect it seems probable that deficient eye movements contribute less to the problem than do abnormalities in central organisation.

Of considerable relevance to this discussion are theories of pattern recognition. It was noted that several of the Benton Visual Retention test designs were incorrectly reproduced because their axes of orientation were distorted, e.g. diamonds were reproduced as squares (see Appendix), and in this way they were reminiscent of results obtained by Gibson et al. (1962) from young children who, whilst being indifferent to rotation, clearly recognised other discriminating features. On the other hand, it could be argued that patients with Turner's syndrome noted the configuration but without those features which would distinguish such a configuration from other similar, but rotated, ones.

Theories of visual recognition classically include two types, template matching and feature analysing. Theories of template matching postulate that a new figure is identified by noting its congruence with a basic model - a theory which fails to account for accurate recognition despite changes of position of image on the retina, and in size and orientation. A connection between the template model and that following - the feature analysing model - is the consideration that, involved in a template theory, is the concept that it generates a set of rules, so that if a new stimulus is to be translated in terms of these rules, certain features have to fit. If the wrong features are initially perceived, problems of discrimination arise.

Feature analysing models postulate a cognitive searching system containing hierarchically arranged analysers which test for the presence of specific features. This type of explanation has gained
in popularity with the discoveries made by Hubel and Wiesel (1962) and other researchers, which have shown specific sets of cells within the visual system which respond to particular edges within a stimulus. Later studies (Wiesel and Hubel, 1966) showed similar structures to be involved in colour discrimination. Individuals with Turner's syndrome show a pattern of colour discrimination errors similar to normals, although at a less accurate level. It would therefore seem that their deficiencies are due to quantitative rather than qualitative differences.

In more general terms, evidence collected in this thesis, dealing with pattern recognition and discrimination of colours and designs, points to a perceptual organisation defect associated with the genetic anomaly of Turner's syndrome. This is an interesting concept, since it indicates that complex processes can be influenced by genes. This might appear to be a similar suggestion to that of Jensen (1969) with respect to IQ. It should, however, be noted that a part of his argument about the heritability of IQ hinged on the research done by Money on patients with Turner's syndrome. Hudson (1970) has already queried the validity of this on methodological grounds (size of sample, age ranges, etc.). It seems, however, that a more fundamental issue is involved - that, in applying IQ tests to an individual with Turner's syndrome, one is dealing with a subject who is basically displaying some form of brain damage. Therefore to involve the evidence obtained from the results of such tests in an argument for the genetic influence on IQ is as inappropriate as applying the same concept to a patient displaying phenylketonuria and severe mental defect - where the lowered IQ is secondary to the effects of the absent enzyme, a condition in itself genetically determined.

The effects discussed so far may be seen as primary consequences of the genetic defect associated with Turner's syndrome. Personality characteristics may be considered as secondary consequences of the
interaction between the physical and psychological phenotypic features of the syndrome. It might be postulated that patients with Turner's syndrome experience configurational problems in their perception of other people similar to those they experience in perception of patterns. By invoking Kelly's Personal Construct theory (Kelly, 1955) it might be suggested that such individuals build up inaccurate systems (as a result of inadequate perception), which, in turn, affect their ability to predict and anticipate other people's behavioural reactions. The situation is additionally influenced by their physical abnormalities. The result could well be a withdrawal by the patient from social contacts. There is no direct evidence in this thesis that this theoretical explanation can be applied to the personality data presented. It is put forward as a parallel to studies by McPherson and Buckley (1970) on thought disorder in schizophrenia, in which a similar breakdown in construct systems has been postulated, resulting in the schizophrenic symptoms of thought disorder and, secondarily, of flattening of affect (McPherson et al., 1971).

In summary it may be stated that, prior to the preparation of this thesis, little methodological consideration had been given to the study of the psychological characteristics of individuals with Turner's syndrome. The lack of control data in particular has been commented on. Methodologically, in this thesis an attempt has been made to remedy the situation by using a control group, and by investigating and extending the behavioural detail (in terms of differential test content), thus dispensing with theories dependent on uncontrolled extrapolation from IQ results.

It may be concluded that patients with Turner's syndrome have, relatively speaking, little difficulty in perceiving simple individual figures, but that problems arise when they attempt to interrelate them. Behaviourally then, they evidence difficulties with complex
figures - complex, that is, because of the number of lines and angles, or a degree of overlap, contained within the figures. It has been argued that the evidence presented suggests that patients with Turner's syndrome have a centrally determined problem of integrating relative information, and this has been discussed with regard to the implications concerning theories of pattern recognition. Taking also into consideration data obtained from testing colour vision and auditory acuity, using biochemical models originally applied to learning, tentative suggestions have been put forward of the way in which the genetic deficiency may influence R.N.A. level, which may, in turn, affect the neural capacity for apprehension of conceptual relationships. Finally, the implication of configurational difficulties has been related to their possible consequences in the development of personality.


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APPENDIX A

Case Notes
Karyotype 45 XO  
Date of birth: 16-3-38  Marital status: Single  
Referred to gynaecologist because of primary amenorrhoea. Noted to have poor breast development, which improved with oestrogen therapy. She had been operated on at the age of 14 for correction of neck webbing. She has strabismus with a left-sided deviation. She has two half-sisters and works very efficiently as a private secretary. She was always in the top three at school and has pursued her career successfully.

Testing was always carried out at the Unit, where she very cheerfully takes part in any research project. Reference to her results indicates that, as a member of the sample having the highest IQ, she is an example of the way in which it is possible to compensate for psychological deficits apparent in other members of the group, with lower IQ's.

Psychological Test Results

Wechsler Adult Intelligence Scale  
FSIQ 112  
VIQ 119  PIQ 101  
Detailed sub-test scores  
Information 13  Digit Symbol 12  
Comprehension 14  Picture Completion 10  
Arithmetic 15  Block Design 9  
Similarities 12  Picture Arrangement 12  
Digit Span 12  Object Assembly 7  
Vocabulary 14  

Benton Visual Retention Test  
No. correct 10  
No. errors 0  

Bender Visual Motor Gestalt Test  
Errors of overlap of contiguity 0  
" orientation 0  

Experimental Visual Recognition Test  
No. errors 0
Experimental Formboards Test

Series 1

V_L (1) 17"  V_S (4) 14"  H_L (2) 46"  H_S (3) 81"

Placement consistency rating 1

Series 2

Diamond V (1) 30"  H (2) 29"
Cross V (2) 8"  H (1) 41"

100-Hue

Total error score 117
Box 85-21: 7  Box 22-42: 32
" 43-63: 35  " 64-84: 43

Anomaloscope

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Hysteroid:Obsessoid Questionnaire

Total 19

Hostility and Direction of Hostility Questionnaire

Total 9

Extrapunitiveness 5  Intropunitiveness 5

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

A  3  L  6
B  7  M  6
C  6  N  6
E  6  O  4
F  6  Q_1 7
G 10  Q_2 7
H  3  Q_3 8
I  4  Q_4 3

Second order factors

Anxiety  3.9  Introversion:Extraversion  3.2
Karyotype  45 XO  
Date of birth: 2-5-42  Marital status: Married

Presented in her late teens with primary amenorrhoea and failure to grow. Noted to have no breast development, marked increased carrying angle and neck webbing, with low hair line, but normal female distribution of pubic hair. She was already receiving cyclical oestrogen therapy. Only significant previous medical history was a congenital squint treated surgically at the age of nine years.

Fourth in a sibship of five (three girls and two boys), she is now a housewife, having previously worked as a machinist of children's clothes. Being aware of her infertility, her main interest was in the adoption of a child, which caused her to be resistant to further investigation of herself. Partial assessment was carried out at her home, with the presence of a small niece causing some distraction.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 96  
VIQ 98  PIQ 93

Detailed sub-test scores

Information 11  Digit Symbol 7  
Comprehension 10  Picture Completion 7  
Arithmetic 7  Block Design 10  
Similarities 9  Picture Arrangement 11  
Digit Span 11  Object Assembly 9  
Vocabulary 11

Benton Visual Retention Test

No. correct 6  
No. errors 5

Bender Visual Motor Gestalt Test

NT

Experimental Visual Recognition Test

NT
A/2/60 (cont.)

Experimental Formboards Test

NT

100-Hue

Total error score 20

Box 85-21: 0
" 43-63: 8
" 64-84: 4

Anomaloscope

NT

Hysteroid: Obsessoid Questionnaire - Not returned
Hostility and Direction of Hostility Questionnaire - Not returned
Sixteen Personality Factor Questionnaire - Not returned
A/169/60  Bender Visual Motor Gestalt Test
1. □
2. ○
3. ○ ○ □
4. □ ○ △
5. ○ □ △
6. □ △
7. △ □ △
8. △ □ △
9. □ □ □
10. □ □ □
**Karyotype** 45 XO  
**Date of birth:** 20-9-33  
**Marital status:** Single  
Presented, aged 18 years, with primary amenorrhoea. At that time noted to have poor breast development, absent axillary and pubic hair, short fourth metacarpal on right hand, but no neck webbing. Demonstrated to have streak ovaries at laparotomy. She was treated for a short time with oestrogen to promote breast development. Only previous medical history was of discharging ears, which were treated surgically.

The second of four girls, she is at present employed in sticking on labels in the bottling room of a distillery. Testing was carried out in the Unit and good rapport was easily established.

**Psychological Test Results**

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**Detailed sub-test scores**

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<td>Object Assembly</td>
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**Benton Visual Retention Test**

| No. correct | 4 |
| No. errors  | 8 |

**Bender Visual Motor Gestalt Test**

| Errors of overlap or contiguity | 3 |
| "  " orientation               | 1 |

**Experimental Visual Recognition Test**

| No. errors | 15 |
A/169/60 (cont.)

Experimental Formboards Test

Series 1

\[ V_L (1) \ 18'' \ V_S (4) \ 7'' \ H_L (2) \ 27'' \ H_S (2) \ 22'' \]

Placement consistency rating 1

Series 2

Diamond V (1) 38'' H (2) 26''
Cross V (2) 7'' H (1) 49''

100-Hue

Total error score 188

Box 85-21: 26 Box 22-42: 46
'' 43-63: 57 '' 64-84: 59

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Hysteroid:Obsessoid Questionnaire

Total 13

Hostility and Direction of Hostility Questionnaire

Total 13

Extrapunitiveness 4
Intropunitiveness 9

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

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Second order factors

Anxiety 5.4
Introversion:Extraversion 3.2
Karyotype  45 XO
Date of birth:  17-12-45    Marital status: Single
Referred by her G.P. after noting swelling of her feet when she was 15 years of age; until that time she had had no major illnesses, but had complained of migraine.

She is the firstborn of a sibship of 11, eight of whom are half-sibs. Other clinical signs common to Turner's syndrome patients are short neck with low occipital hair line and slight webbing, cubitus valgus and short fourth and fifth metacarpals. Oestrogen therapy produced breast development and withdrawal bleeding.

She left school at 15 years of age with no qualifications and now works at a wire factory as a winder. Crowded home conditions made it essential that the testing was carried out in the Unit.

Psychological Test Results
Wechsler Adult Intelligence Scale

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<td>PIQ</td>
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Detailed sub-test scores
Information  9  Digit Symbol  7
Comprehension 9  Picture Completion 6
Arithmetic 10  Block Design 7
Similarities 10  Picture Arrangement 8
Digit Span 10  Object Assembly 7
Vocabulary 9

Benton Visual Retention Test
No. correct 7
No. errors 3

Bender Visual Motor Gestalt Test
Errors of overlap or contiguity 2
  " orientation 1

Experimental Visual Recognition Test
No. errors 22
Experimental Formboards Test

**Series 1**

- $V_L (4)$ 27"  
- $V_S (1)$ 24"  
- $H_L (3)$ 153"  
- $H_S (2)$ 173"

Placement consistency rating 0

**Series 2**

- Diamond V (2) 16"  
- H (1) 60"  
- Cross V (1) 11"  
- H (2) 24"

100-Hue

- Total error score 168
- Box 85-21: 33  
- Box 22-42: 46  
- " 43-63: 49  
- " 64-84: 40

Anomaloscope

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<tr>
<td>Y/B &quot;</td>
<td>34</td>
<td>28-39</td>
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Hysteroid:Obsessoid Questionnaire

- Total 25

Hostility and Direction of Hostility Questionnaire

- Total 20
- Extrapunitiveness 10  
- Intropunitiveness 10

Detailed sub-test scores

<table>
<thead>
<tr>
<th>AH</th>
<th>SC</th>
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<tbody>
<tr>
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<table>
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<table>
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Sixteen Personality Factor Questionnaire

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<th>C</th>
<th>E</th>
<th>F</th>
<th>G</th>
<th>H</th>
<th>I</th>
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<table>
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<td>O</td>
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<td>Q1</td>
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<td>Q2</td>
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<tr>
<td>Q3</td>
<td>3</td>
</tr>
<tr>
<td>Q4</td>
<td>4</td>
</tr>
</tbody>
</table>

Second order factors

- Anxiety 5.6  
- Introversion:Extraversion 1.5
A/169/61  Bender Visual Motor Gestalt Test
Karyotype 45 X0

Date of birth: 21-2-34  Marital status: Single

Referred to gynaecology clinic at the age of 26 years with amenorrhoea which was presumed to be secondary, since the patient claimed to have had her menarche at the age of 12. She has had several psychiatric referrals and has been described as being "seriously depressed with an abnormal personality." She was noted to have slight increased carrying angle, but no neck webbing. Oestrogen implants produced good breast development.

The second of three sisters, she is presently unemployed but has previously worked as a cinema usherette. Testing was initially carried out in the patient's home and completed in the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 75
VIQ 85  PIQ 65

Detailed sub-test scores

Information 7  Digit Symbol 4
Comprehension 9  Picture Completion 5
Arithmetic 4  Block Design 3
Similarities 6  Picture Arrangement 4
Digit Span 10  Object Assembly 3
Vocabulary 9

Benton Visual Retention Test
No. correct 3
No. errors 14

Bender Visual Motor Gestalt Test
Errors of overlap or contiguity 0
" " orientation 2

Experimental Visual Recognition Test
No. errors 14
Experimental Formboards Test

Series 1
\[ V_L (3) \ 60'' \quad V_S (1) \ 85'' \quad H_L (2) \ 60'' \quad H_S (4) \ 94'' \]

Series 2
Diamond V (1) 60'' \quad H (2) 23''
Cross V (2) 15'' \quad H (1) 25''

100-Hue
Total error score 60
Box 85-21: 20 Box 22-42: 20
'' 43-63: 12 '' 64-84: 8

Anomaloscope
<table>
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<tr>
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<td>Y/B</td>
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Hysteroid: Obsesscid Questionnaire
NT

Hostility and Direction of Hostility Questionnaire
NT

Sixteen Personality Factor Questionnaire

<p>| | | |</p>
<table>
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<tbody>
<tr>
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<td>B</td>
<td>7</td>
<td>M</td>
</tr>
<tr>
<td>C</td>
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<td>E</td>
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<td>O</td>
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<td>F</td>
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<td>Q_1</td>
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<td>Q_3</td>
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<tr>
<td>I</td>
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<td>Q_4</td>
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Second order factors

<p>| | |</p>
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</thead>
<tbody>
<tr>
<td>Anxiety 7.9</td>
<td>Introversion: Extraversion 2.7</td>
</tr>
</tbody>
</table>
Karyotype: 45 XO

Date of birth: 5-9-42  Marital status: Married

Ascertained at birth on account of extreme neck webbing which was later excised, leaving heavy scarring. She also has increased carrying angle, low set ears, absent breast development, and lymphoedema of her right ankle.

She lived for two years with her husband before they separated; her husband is now co-habiting with another woman, known to the patient. She herself lives with her mother.

The youngest of three girls, she works as a machinist in a carton manufacturing factory, and is frequently required to be the financial supporter of the remainder of her family. Testing was carried out at the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 85

VIQ 90  PIQ 80

Detailed sub-test scores

Information 8  Digit Symbol 6
Comprehension 11  Picture Completion 8
Arithmetic 7  Block Design 7
Similarities 9  Picture Arrangement 6
Digit Span 7  Object Assembly 7
Vocabulary 9

Benton Visual Retention Test

No. correct 3
No. errors 10

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 4
" " orientation 3

Experimental Visual Recognition Test

No. errors 17
Experimental Formboards Test

Series 1

\[ V_L (1) \quad 14'' \quad V_S (2) \quad 28'' \quad H_L (4) \quad 42'' \quad H_S (3) \quad 62'' \]

Placement consistency rating 5

Series 2

Diamond V (2) \quad 36'' \quad H (1) \quad 65''
Cross V (1) \quad 14'' \quad H (2) \quad 16''

100-Hue

Total error score 111

Box 85-21: 19 \quad Box 22-42: 28
" 43-63: 34 \quad " 64-84: 30

Anomaloscope

\begin{array}{ccc}
\text{H/G equation} & \text{MMP} & \text{MR} \\
29 & 22-34 \\
35 & 30-40 \\
25 & 15-35 \\
\end{array}

Hysteroid: Obsessoid Questionnaire

Total 22

Hostility and Direction of Hostility Questionnaire

Total 22

Extrapunitiveness 12 \quad Intropunitiveness 10

Detailed sub-test scores

\begin{array}{ccc}
\text{AH} & 3 & \text{SC} \\
\text{CO} & 7 & \text{G} \\
\text{PH} & 2 & \\
\end{array}

Sixteen Personality Factor Questionnaire

\begin{array}{ccc}
\text{A} & 6 & \text{L} \\
\text{B} & 8 & \text{M} \\
\text{C} & 4 & \text{N} \\
\text{E} & 6 & \text{O} \\
\text{F} & 7 & \text{Q}_1 \\
\text{G} & 5 & \text{Q}_2 \\
\text{H} & 4 & \text{Q}_3 \\
\text{I} & 6 & \text{Q}_4 \\
\end{array}

Second order factors

Anxiety 6.0 \quad \text{Introversion: Extraversion} 4.7
Karyotype  45 XO
Date of birth:  10-1-36  Marital status:  Single

Referred to gynaecologist complaining of primary amenorrhoea and subsequently ascertained during a survey of such patients. Noted to have broad chest with immature breast development, marked neck webbing and increased carrying angle. A congenital lesion has recently caused the development of congestive cardiac failure. She was known to have otitis media in childhood and this has produced increasing bilateral perceptive deafness necessitating the wearing of a hearing aid in the right ear. She is also under review by a clinic specialising in renal disorders.

The ninth in a sibship of ten, she lives at home with her mother, and works full-time as a cotton mill operator. All testing was carried out in the Unit, where she is a frequent attender for review.

Psychological Test Results

Wechsler Adult Intelligence Scale

<table>
<thead>
<tr>
<th>Scale</th>
<th>FSIQ</th>
<th>VIQ</th>
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Detailed sub-test scores

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<tr>
<td>Comprehension</td>
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<td>Arithmetic</td>
<td>7</td>
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<tr>
<td>Similarities</td>
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<tr>
<td>Digit Span</td>
<td>10</td>
</tr>
<tr>
<td>Vocabulary</td>
<td>8</td>
</tr>
<tr>
<td>Digit Symbol</td>
<td>8</td>
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<tr>
<td>Picture Completion</td>
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</tr>
<tr>
<td>Block Design</td>
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<tr>
<td>Picture Arrangement</td>
<td>4</td>
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<tr>
<td>Object Assembly</td>
<td>4</td>
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Benton Visual Retention Test

<table>
<thead>
<tr>
<th>Test</th>
<th>Score</th>
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<tbody>
<tr>
<td>No. correct</td>
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<tr>
<td>No. errors</td>
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Bender Visual Motor Gestalt Test

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<th>Score</th>
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<tbody>
<tr>
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<td>&quot;  &quot; orientation</td>
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Experimental Visual Recognition Test

<table>
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<tr>
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<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>No. errors</td>
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</tbody>
</table>
Experimental Formboards Test

Series 1
\[ V_L (1) \ 43'' \ V_S (4) \ 23'' \ H_L (3) \ 43'' \ H_S (2) \ 31'' \]

Placement consistency rating 4

Series 2
Diamond V (2) 22''  H (1) 167''
Cross V (1) 6''  H (2) 21''

100-Hue

Total error score 257
Box 85-21: 31  Box 22-42: 80
'' 43-63: 74  '' 64-84: 72

Anomaloscope

<table>
<thead>
<tr>
<th>R/G equation</th>
<th>MMP</th>
<th>MR</th>
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</thead>
<tbody>
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<td>Y/B</td>
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Hysteroid:Obsessoid Questionnaire

Total 29

Hostility and Direction of Hostility

Total 16
Extrapunitiveness 12  Intropunitiveness 4

Detailed sub-test scores

<table>
<thead>
<tr>
<th>AH</th>
<th>SC</th>
<th>CO</th>
<th>G</th>
<th>PH</th>
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<td>3</td>
<td>7</td>
<td>1</td>
<td>2</td>
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</tbody>
</table>

Sixteen Personality Factor Questionnaire

A  6  L  7
B  4  M  5
C  4  N  6
E  7  O  8
F  5  Q_1 7
G  6  Q_2 8
H  6  Q_3 7
I  4  Q_4 3

Second order factors
Anxiety 5.3  Introversion:Extraversion 4.9
1. 

2. 

3. 

4. 

5. 

6. 

7. 

8. 

9. 

10. 

A/61/64  Benton Visual Retention Test
Karyotype 45 X0

Date of birth: 25-1-46  Marital status: Single

Referred at the age of 18 to an endocrinology clinic with primary amenorrhoea. Noted to have sparse axillary and pubic hair, slight neck webbing with low hairline, no breast development and increased carrying angle. Also had marked oedema of both feet, which condition had been present since birth. Strabismus was noted at the age of six months and corrected surgically at the age of 13. More recently she has been put on a low calorie diet for obesity.

An only child, she lives with her mother and works as a box maker. Testing was carried out at the Unit and, although initially reserved, she became progressively more outgoing in subsequent interviews.

Psychological Test Results

Wechsler Adult Intelligence Scale

<table>
<thead>
<tr>
<th></th>
<th>FSIQ</th>
<th>VIQ</th>
<th>PIQ</th>
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<tbody>
<tr>
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Detailed sub-test scores

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<tbody>
<tr>
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<td>Similarities</td>
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<td>Digit Span</td>
<td>10</td>
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<td>Vocabulary</td>
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<tr>
<td>Digit Symbol</td>
<td>6</td>
</tr>
<tr>
<td>Picture Completion</td>
<td>7</td>
</tr>
<tr>
<td>Block Design</td>
<td>10</td>
</tr>
<tr>
<td>Picture Arrangement</td>
<td>9</td>
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<tr>
<td>Object Assembly</td>
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Benton VisualRetention Test

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<th>Score</th>
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Bender Visual MotorGestalt Test

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Experimental Visual Recognition Test

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<th>Score</th>
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</thead>
<tbody>
<tr>
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</tbody>
</table>
Experimental Formboards Test

Series 1

\[ V_L (4) \ 8" \ V_S (1) \ 26" \ H_L (2) \ 95" \ H_S (3) \ 50" \]

Placement consistency rating 0

Series 2

Diamond V (1) 15" H (2) 22"
Cross V (2) 8" H (1) 25"

100-Hue

Total error score 45
Box 85-21: 0 Box 22-42: 21
" 43-63: 16 " 64-84: 8

Anomaloscope

<table>
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<tr>
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Hysteroid:Obsessoid Questionnaire

Total 23

Hostility and Direction of Hostility Questionnaire

Total 14

Extrapunitiveness 7 Intropunitiveness 7

Detailed sub-test scores

<table>
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<tr>
<th></th>
<th>SC</th>
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<tbody>
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Sixteen Personality Factor Questionnaire

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<tr>
<td>I</td>
<td>2</td>
<td>Q_4</td>
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</table>

Second order factors

Anxiety 5.3 Introversion:Extraversion 3.8
Karyotype: 45 XO

Date of birth: 19-10-50  Marital status: Single

Referred to an orthopaedic clinic at the age of 14 on account of retarded growth. Noted to have low hairline, absent breast development and oedema of hands and feet, but no other abnormality. Treatment with oestrogen implant produced slight breast development.

The eldest of eight children, she lived at home with her family in overcrowded conditions and was employed as a petrol pump attendant. Initial testing was carried out at the patient's home. Completion of tests was prevented by her emigration to Australia.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 63
VIQ 63  PIQ 67

Detailed sub-test scores

Information 2  Digit Symbol 5
Comprehension 3  Picture Completion 7
Arithmetic 5  Block Design 4
Similarities 5  Picture Arrangement 5
Digit Span 1  Object Assembly 3
Vocabulary 4

Benton Visual Retention Test

No. correct 3
No. errors 13

Bender Visual Motor Gestalt Test

NT

Experimental Visual Recognition Test

NT
 experimental formboards test

nt

10C-Hue

Total error score 252
Box 85-21: 56  Box 22-42: 44
" 43-63: 94  " 64-84: 58

Anomaloscope

nt

Hysteroid: Obsessoid Questionnaire

Total 23

Hostility and Direction of Hostility Questionnaire

Total 24

Extrapunitiveness 17  Intropunitiveness 7

Detailed sub-test scores

AH 6  SC 6
CO 9  G 1
PH 2

Sixteen Personality Factor Questionnaire

A 7  L 8
B 8  M 7
CC 5  N 6
E 5  O 5
F 7  Q1 1
G 6  Q2 6
H 6  Q3 8
I 3  Q4 6

Second order factors

Anxiety 5.4  Introversion: Extraversion 5.9
Karyotype: 45 XO

Date of birth: 10-1-48 Marital Status: Single

Referral at the age of 17 to endocrinology clinic on account of short stature and primary amenorrhoea. Noted to have short neck, pitting oedema of right ankle, sparse axillary and pubic hair. An oestrogen implant in 1967 and oral oestrogens for two months in 1969 produced good breast development. She is markedly overweight and has recently been admitted to hospital for weight reduction and investigation of ankle oedema.

The first of five sibs, she has two brothers who also have abnormally short stature but a normal chromosomal complement. She left school without qualifications and is now employed as a machinist. Testing was carried out at the patient's home.

Psychological Test Results

Wechsler Adult Intelligence Scale

<table>
<thead>
<tr>
<th>Scale</th>
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<td>57</td>
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Detailed sub-test scores

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<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Information</td>
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<tr>
<td>Comprehension</td>
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<td>Arithmetic</td>
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<td>Similarities</td>
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<td>Digit Span</td>
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<td>Vocabulary</td>
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<td>Object Assembly</td>
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Benton Visual Retention Test

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<th>No. correct</th>
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<tr>
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Bender Visual Motor Gestalt Test

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<tbody>
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<td>&quot; orientation</td>
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Experimental Visual Recognition Test

<table>
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<tr>
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</tr>
</thead>
<tbody>
<tr>
<td>No. errors</td>
<td>14</td>
</tr>
</tbody>
</table>
Experimental Formboards Test

**Series 1**
- $V_L$ (2) 70"
- $V_S$ (1) 43"
- $H_L$ (4) 52"
- $H_S$ (3) 75"

Placement consistency rating 6

**Series 2**
- Diamond V (1) 18"
- H (2) 110"
- Cross V (2) 6"
- H (1) 210"

**100-Hue**

Total error score 120

- Box 85-21: 8
- Box 22-42: 27
- " 43-63: 51
- " 64-84: 34

**Anomaloscope**

<table>
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**Hysteroid:Obsessoid Questionnaire**

NT

**Hostility and Direction of Hostility Questionnaire**

NT

**Sixteen Personality Factor Questionnaire**

NT
Karotype 45 XO

Date of birth: 12-6-33  Marital status: Single

Referred at the age of 35 years to endocrinology clinic with suspected hypothyroidism, complaining of dry skin and thinning of hair. Found to have primary amenorrhoea. Noted to have no breast development and absent axillary and pubic hair.

The second of four sisters, she lives at home with her mother and father, and works in the office of a local school where her father is caretaker. Testing was carried out at her home; at that time she was entirely co-operative, but has subsequently expressed a wish not to take part in any future research.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ  91
VIQ  96  PIQ  86

Detailed sub-test scores

Information  11  Digit Symbol  6
Comprehension  9  Picture Completion  7
Arithmetic  6  Block Design  9
Similarities  10  Picture Arrangement  7
Digit Span  10  Object Assembly  6
Vocabulary  10

Benton Visual Retention Test

No. correct  5
No. errors  9

Bender Visual Motor Gestalt Test

NT

Experimental Visual Recognition Test

NT
Experimental Formboards Test

NT

100-Formboards Test

Total error score 156

Box 85-21: 28  Box 22-42: 43
  " 43-63: 41  " 64-84: 44

Anomaloscope

NT

Hysteroid:Obsessoid Questionnaire

Total 22

Hostility and Direction of Hostility Questionnaire

Total 21

Extrapunitiveness 15  Intropunitiveness 6

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

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Second order factors

Anxiety 7.2  Introversion:Extraversion 2.8
A/95/69  Benton Visual Retention Test
Karyotype  45 X0

Date of birth: 6-9-50  Marital status: Single

Referred to gynaecologist at the age of 19, with primary amenorrhoea. Noted to have slight neck webbing, increased carrying angle and minimal breast development. Previous medical history had included recurrent episodes of otitis media and an episode of acute glomerulonephritis. She was subsequently admitted to a psychiatric hospital, owing to the development of anorexia nervosa. This had begun as a conscious effort to reduce weight by dieting along with her mother.

The eldest of five children, she is employed as a clerk. Testing was undertaken at the patient's house. Whilst she co-operated fully in the completion of the tests there was very little of an effective relationship formed, and it seemed possible that she had not fully recovered from her psychiatric illness.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ  97  
VIQ  107  PIQ  84

Detailed sub-test scores

Information  8  Digit Symbol  10
Comprehension  10  Picture Completion  8
Arithmetic  12  Block Design  5
Similarities  12  Picture Arrangement  8
Digit Span  12  Object Assembly  6
Vocabulary  10

Benton Visual Retention Test

No. correct  7
No. errors  5

Bender Visual Motor Gestalt Test

NT

Experimental Visual Recognition Test

NT
Experimental Formboards Test

100-Hue

Total error score 324
Box 85-21: 58  Box 22-42: 64
" 43-63: 93  " 64-84: 109

Anomaloscope

NT

Hysteroid:Obsessoid Questionnaire

Total 11

Hostility and Direction of Hostility Questionnaire

Total 25

Extrapunitiveness 13  Intropunitiveness 12

Detailed sub-test scores

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Second order factors

| Anxiety: 9.5 | Introversion:Extraversion: 0.6 |
A/97/69 Benton Visual Retention Test
Karyotype  45 XO
Date of birth:  25-8-53        Marital status: Single
Referred to endocrinology clinic at the age of 16, complaining of primary amenorrhoea. On physical examination was found to have slight breast development, absent axillary and pubic hair, bilateral short fourth and fifth metacarpals, low hairline with no neck webbing. She had been troubled by otitis media in the left ear, up to the age of seven, prior to her referral. Her sister suffered with scanty and irregular menstruation (oligomenorrhoea) and a maternal aunt is known to have had secondary amenorrhoea from the age of 21. Both of these relatives are known to have a normal karyotype.

Third in a sibship of three, she lives at home with her mother and stepfather. Her father died from drug-overdosage. She has gained 'O' levels in Arithmetic, French, English and Home Management, and has been employed as a bank clerk since leaving school. She has been most helpful in completing psychological tests, which have always been carried out at the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale
FSIQ  104
VIQ  113        PIQ  91

Detailed sub-test scores
Information  11        Digit Symbol  10
Comprehension  9        Picture Completion  8
Arithmetic  13        Block Design  9
Similarities  11        Picture Arrangement  9
Digit Span  14        Object Assembly  6
Vocabulary  10

Benton Visual Retention Test
No. correct  7
No. errors  4

Bender Visual Motor Gestalt Test
Errors of overlap or contiguity  0
"  " orientation  1

Experimental Visual Recognition Test
No. errors  2
Experimental Formboards Test

Series 1

\( V_L \) (4) 43" \( V_S \) (1) 15" \( H_L \) (2) 185" \( H_S \) (3) 217"

Placement consistency rating 1

Series 2

Diamond V (2) 15" H (1) 80"
Cross V (1) 8" H (2) 12"

100-Hue

Total error score 172
Box 85-21: 24 Box 22-42: 53
" 43-63: 46 " 64-84: 49

Anomaloscope

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Mysteroid:Obsessoid Questionnaire

Total 25

Hostility and Direction of Hostility Questionnaire

Total 12

Extrapunitiveness 3 Intropunitiveness 9

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

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Second order factors

Anxiety 2.4 Introversion:Extraversion 6.0
Karyotype: 45 XO
Date of birth: 20-3-53
Marital status: Single

Referred to an endocrinology clinic at the age of 16 for investigation of short stature. There was no other abnormality noted, and she is now an attractive slim girl with slight breast development (not induced by exogenous oestrogens). In her previous history she was described as failing to thrive shortly after birth and was enuretic up until the age of eight-and-a-half. She is the second of three children; her elder sister has had severe ulcerative colitis, necessitating colectomy.

The patient obtained several 'O' levels, including Arithmetic, and is now employed as a bank clerk. Testing was undertaken at her home.

Psychological Test Results

Wechsler Adult Intelligence Scale

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Detailed sub-test scores

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<td>Similarities</td>
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<td>Digit Span</td>
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<tr>
<td>Vocabulary</td>
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<tr>
<td>Digit Symbol</td>
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<tr>
<td>Picture Arrangement</td>
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<td>Object Assembly</td>
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<td>Benton Visual Retention Test</td>
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<td>No. correct</td>
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<tr>
<td>No. errors</td>
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Bender Visual Motor Gestalt Test

Errors of overlap or contiguity: 3
" " orientation: 2

Experimental Visual Recognition Test

No. errors: 7
Experimental Formboards Test

**Series 1**

V<sub>L</sub> (1) 7"  V<sub>S</sub> (3) 144"  H<sub>L</sub> (2) 29"  H<sub>S</sub> (4) 76"

Placement consistency rating 5

**Series 2**

Diamond V (1) 133"  H (2) 17"
Cross V (2) 7"  H (1) 30"

100-Hue

Total error score 209

Box 85-21: 30  Box 22-42: 59
" 43-63: 55  " 64-84: 65

Anomaloscope

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Hysteroid:Obsessoid Questionnaire

Total 20

Hostility and Direction of Hostility Questionnaire

Total 14

Extrapunitiveness 6  Intropunitiveness 8

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

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Second order factors

Anxiety 4.7  Introversion:Extraversion 3.6
A/188/70  Bender Visual Motor Gestalt Test
A 29.

Karyotype: 45 X0

Date of birth: 4-9-52   Marital status: Single

Referred to an endocrinology clinic at the age of 17 because of primary amenorrhoea. She was noted to have only slight breast development and no neck webbing.

The second of two children, she attended a boarding school between the ages of 10 and 17, whilst her parents were living abroad. She gained one Higher and five Ordinary Level passes, including Arithmetic. She had just left school and had enrolled in a secretarial course, with the possibility of learning drama. Her elder sib obtained 'O' levels at evening classes and entered the Civil Service.

Testing was carried out at the Unit, during a very short admission period for investigation, and it was not practicable to complete all the tests.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 97
VIQ 103   PIQ 89

Detailed sub-test scores

Information 11   Digit Symbol 8
Comprehension 11   Picture Completion 9
Arithmetic 7   Block Design 9
Similarities 10   Picture Arrangement 9
Digit Span 7   Object Assembly 5
Vocabulary 12

Benton Visual Retention Test

No. correct 6
No. errors 7

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 3
" " orientation 2

Experimental Visual Recognition Test

NT
Experimental Formboards Test

NT

100-Hue

Total error score 124

Box 85-21: 17  Box 22-42: 47
" 43-63: 39 " 64-84: 21

Anomaloscope

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Hysteroid: Obsessoid Questionnaire

NT

Hostility and Direction of Hostility Questionnaire

NT

16 Personality Factor Questionnaire

NT
Karyotype: 45 XO

Date of birth: 23-7-23  Marital status: Single

Admitted at the age of 47 to a respiratory unit for investigation of bronchitis. Noted to have features of Turner's syndrome: primary amenorrhoea (which had not previously been brought to medical attention), absent axillary hair and scanty pubic hair, no breast development, neck webbing with low hairline and increased carrying angle.

The elder of two sibs, she lives with her mother and has sheltered employment as an office worker. Testing was undertaken at the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 79

VIQ 85  PIQ 79

Detailed sub-test scores

Information 11  Digit Symbol 4
Comprehension 7  Picture Completion 4
Arithmetic 5  Block Design 4
Similarities 4  Picture Arrangement 6
Digit Span 7  Object Assembly 2
Vocabulary 9

Benton Visual Retention Test

No. correct 2
No. errors 16

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 4
" " orientation 1

Experimental Visual Recognition Test

No. errors 14
Experimental Formboards Test

100-Hue

Total error score 211

Box 85-21: 40  Box 22-42: 59
" 43-63: 69  " 64-84: 43

Anomaloscope

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Hostility and Direction of Hostility Questionnaire

NT

Sixteen Personality Factor Questionnaire

NT
Karyotype: 46 XXq1

Date of birth: 27-2-44 Marital status: Single

Referred to an endocrinology clinic at the age of 16 with obesity. Noted to have primary amenorrhoea, no breast development, no axillary or pubic hair, increased carrying angle, but no neck webbing. Cyclical oestrogens have produced withdrawal bleeding and good breast development. Previous medical history of relevance has been aortic stenosis for which a valvotomy was performed; the extraction of all teeth; and episodes of bronchitis.

The second of two children, she lives at home with her parents and works in sheltered factory employment. As a hobby she trains sheepdogs and has won prizes at sheepdog trials.

Testing was carried out at the patient's home.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 95
VIQ 106 PIQ 81

Detailed sub-test scores

Information 11 Digit Symbol 9
Comprehension 14 Picture Completion 7
Arithmetic 9 Block Design 8
Similarities 11 Picture Arrangement 6
Digit Span 10 Object Assembly 5
Vocabulary 12

Benton Visual Retention Test

No. correct 5
No. errors 8

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 4
" orientation 0

Experimental Visual Recognition Test

No. errors 13
Experimental Formboards Test

Series 1
\[ V_L (1) \ 34'' \ V_S (2) \ 11'' \ H_L (4) \ 249'' \ H_S (3) \ 221'' \]
Placement consistency rating 4

Series 2
Diamond V (1) \ 20'' \ H (2) \ 202''
Cross V (2) \ 10'' \ H (1) \ 47''

100-Hue
Total error score 40
Box 85-21: 4 Box 22-42: 9
43-63: 11 64-84: 16

Anomaloscope

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Hysteroid:Obsessoid Questionnaire
Total 22

Hostility and Direction of Hostility Questionnaire
Total 16
Extrapunitiveness 7 Intropunitiveness 9

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

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Second order factors
Anxiety 6.4 Introversion:Extraversion 3.0
A/34/63  Bender Visual Motor Gestalt Test
Karyotype: 46 XXq1

Date of birth: 29-3-41
Marital status: Married

Referred to General Hospital at the age of 22 with symptoms suggesting laryngeal obstruction. No cause was found but she was noted to have primary amenorrhoea, neck webbing, increased carrying angle, and webbing between second and third toes. Cyclical hormone therapy has produced some breast development and withdrawal bleeding.

She was married prior to her ascertainment and it is known that there is a background of marital disharmony. Following discussion of her infertility episodes episodes of depression have developed, one of which resulted in aspirin overdose.

She now lives with her mother and has had various unskilled jobs, e.g. canteen worker, and potato sorter. Testing was carried out in the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale
FSIQ 78
VIQ 82  PIQ 76

Detailed sub-test scores
Information 9  Digit Symbol 7
Comprehension 6  Picture Completion 7
Arithmetic 6  Block Design 6
Similarities 8  Picture Arrangement 6
Digit Span 6  Object Assembly 5
Vocabulary 8

Benton Visual Retention Test
No. correct 4
No. errors 11

Bender Visual Motor Gestalt Test
Errors of overlap or contiguity 4
" " orientation 1

Experimental Visual Recognition Test
NT
A36.

A/34/63 (cont.)

Experimental Formboards Test

NT

100-Hue

Total error score 154

Box 85-21: 9  Box 22-42: 64
" 43-63: 51 " 64-84: 30

Anomaloscope

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Hysteroid: Obsessoid Questionnaire

Total 27

Hostility and Direction of Hostility Questionnaire

Total 30

Extrapunitiveness 18  Intropunitiveness 12

Detailed sub-test scores

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Sixteen Personality Factor Questionnaire

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<td>3</td>
</tr>
<tr>
<td>C</td>
<td>N</td>
<td>3</td>
</tr>
<tr>
<td>E</td>
<td>O</td>
<td>9</td>
</tr>
<tr>
<td>F</td>
<td>Q₁</td>
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</tr>
<tr>
<td>G</td>
<td>Q₂</td>
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<tr>
<td>H</td>
<td>Q₃</td>
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<tr>
<td>I</td>
<td>Q₄</td>
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</table>

Second order factors

Anxiety 8.4  Introversion: Extraversion 4.3
Benton Visual Retention Test

A/97/63
Karyotype 46 XXq1

Date of birth: 17-3-44  Marital status: Single

Referred to endocrinology clinic at the age of 19 for primary amenorrhoea and growth retardation. Noted to have short fourth and fifth metacarpals, increased carrying angle, but no neck webbing. Cysticaloestrogen therapy produced only one episode of withdrawal bleeding, slight development and scanty growth of pubic hair. She gave a ten-year history of intermittent diarrhoea, ascribed to gastroenteritis.

She suffers from episodes of depression, in which she becomes very tearful, withdraws from her already-limited social contacts and takes to her bed. One such episode has necessitated medical attention. She is the firstborn of seven children. She did not learn to read until the age of 14, and both reading and writing are very retarded. She now works as a sewing machine operator.

Testing was carried out partly at home and partly at the Unit, where she was admitted for investigation of her gastro-enteritis. She has since refused any further contact.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 75
VIQ 72  PIQ 84

Detailed sub-test scores

| Information | 5 | Digit Symbol | 5 |
| Comprehension | 5 | Picture Completion | 7 |
| Arithmetic | 7 | Block Design | 10 |
| Similarities | 3 | Picture Arrangement | 6 |
| Digit Span | 7 | Object Assembly | 9 |
| Vocabulary | 5 |

Benton Visual Retention Test

No. correct 5
No. errors 8

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 1
" " orientation 2

Experimental Visual Recognition Test

NT
Experimental Formboards Test

**100-Hue**

<table>
<thead>
<tr>
<th>Box</th>
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</tr>
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<tbody>
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**Hysteroids-Obsessoid Questionnaire**

Total 22

**Hostility and Direction of Hostility Questionnaire**

Total 17

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**Detailed sub-test scores**

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**Sixteen Personality Factor Questionnaire**

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<th>C</th>
<th>N</th>
<th>E</th>
<th>O</th>
<th>F</th>
<th>Q1</th>
<th>G</th>
<th>Q2</th>
<th>H</th>
<th>Q3</th>
<th>I</th>
<th>Q4</th>
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<td>4</td>
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**Second order factors**

<table>
<thead>
<tr>
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<th>4.1</th>
</tr>
</thead>
<tbody>
<tr>
<td>Introversion:Extraversion</td>
<td>5.7</td>
</tr>
</tbody>
</table>
Benton Visual Retention Test
Karyotype: 46 XX

Date of birth: 9-3-46  Marital status: Single

Referred at the age of 18 for investigation of primary amenorrhoea. No stigmata except absence of breast development, which responded to cyclical oestrogen therapy, which has now stopped.

She is the second of two children; her brother is also infertile and has adopted a child. She left school at the age of 16, without any qualifications, and is now employed as an assembly worker. She lives with her parents. Testing was undertaken at her home.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 88

VIQ 96  PIQ 80

Detailed sub-test scores

<table>
<thead>
<tr>
<th>Sub-test</th>
<th>Score</th>
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<tr>
<td>Information</td>
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<tr>
<td>Comprehension</td>
<td>9</td>
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<tr>
<td>Arithmetic</td>
<td>8</td>
</tr>
<tr>
<td>Similarities</td>
<td>9</td>
</tr>
<tr>
<td>Digit Span</td>
<td>6</td>
</tr>
<tr>
<td>Vocabulary</td>
<td>11</td>
</tr>
<tr>
<td>Digit Symbol</td>
<td>7</td>
</tr>
<tr>
<td>Picture Completion</td>
<td>6</td>
</tr>
<tr>
<td>Block Design</td>
<td>8</td>
</tr>
<tr>
<td>Picture Arrangement</td>
<td>6</td>
</tr>
<tr>
<td>Object Assembly</td>
<td>8</td>
</tr>
</tbody>
</table>

Benton Visual Retention Test

No. correct 4

No. errors 13

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 4

" orientation 4

Experimental Visual Recognition Test

No. errors 14
Experimental Formboards Test

Series 1

\[ V_L (1) \ 12'' \ V_S (4) \ 28'' \ H_L (3) \ 220'' \ H_S (2) \ 67'' \]

Placement consistency rating 1

Series 2

Diamond V (2) \ 14'' \ H (1) \ 441''

Cross V (1) \ 12'' \ H (2) \ 28''

100-Hue

Total error score 428

Box 85-21: 52

Box 22-42: 121

" 43-63: 152

" 64-84: 103

Anomaloscope

<table>
<thead>
<tr>
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<th>MR</th>
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<tbody>
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<tr>
<td>Y/B</td>
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<td>0-50</td>
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Hysteroid:Obsessoid Questionnaire

Total 22

Hostility and Direction of Hostility Questionnaire

Total 12

Extrapunitiveness 7

Intropunitiveness 5

Detailed sub-test scores

<table>
<thead>
<tr>
<th>AH</th>
<th>SC</th>
<th>5</th>
</tr>
</thead>
<tbody>
<tr>
<td>CO</td>
<td>G</td>
<td>0</td>
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<tr>
<td>PH</td>
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Sixteen Personality Factor Questionnaire

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<thead>
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<tr>
<td>B</td>
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<td>6</td>
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<td>C</td>
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<td>8</td>
</tr>
<tr>
<td>E</td>
<td>O</td>
<td>4</td>
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<tr>
<td>F</td>
<td>Q1</td>
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<td>Q2</td>
<td>8</td>
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<td>H</td>
<td>Q3</td>
<td>9</td>
</tr>
<tr>
<td>I</td>
<td>Q4</td>
<td>3</td>
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</table>

Second order factors

Anxiety 2.3

Introversion:Extraversion 2.5
A/282/70  Bender Visual Motor Gestalt Test
1.  

2.  

3.  

4.  

5.  

6.  

7.  

8.  

9.  

10.  

A/282/70  Benton Visual Retention Test
Karyotype  45 X0/46 XXqi (initially assigned 45 X0)

Date of birth: 27-5-51  Marital status: Single

Referred at the age of 19 to a gynaecologist for investigation of primary amenorrhoea. Noted to have low hairline and slightly increased carrying angle, short fourth metacarpal, strabismus and nystagmus. Relevant medical history was of acute nephritis at the age of three; now has a non-functioning kidney; middle ear infection and perforation of eardrum at the age of 12. Oestrogen therapy for two years produced normal breast development.

The surviving twin in a sibship of seven, she now works in a factory making dolls' clothes. She left school at the age of 17 without qualifications. Her school report indicates that she was timid and lacking in self-confidence. Initial psychological testing was carried out in the Unit during her admission for investigation, and subsequent testing was not possible because of home location.

Psychological Test Results

**Wechsler Adult Intelligence Scale**

<table>
<thead>
<tr>
<th>Scale</th>
<th>FSIQ</th>
<th>VIQ</th>
<th>PIQ</th>
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<tbody>
<tr>
<td></td>
<td>90</td>
<td>95</td>
<td>85</td>
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**Detailed sub-test scores**

- Information: 9
- Comprehension: 10
- Arithmetic: 7
- Similarities: 9
- Digit Span: 7
- Vocabulary: 10
- Digit Symbol: 7
- Picture Completion: 9
- Block Design: 7
- Picture Arrangement: 9
- Object Assembly: 6
- Visual Retention Test
  - No. correct: 3
  - No. errors: 14
- Bender Visual Motor Gestalt Test
  - Errors of overlap or contiguity: 3
  - " " orientation: 2
- Experimental Visual Recognition Test
  - No. errors: 7
**Experimental Formboards Test**

**100-Hue**

<table>
<thead>
<tr>
<th>Box 85-21</th>
<th>Box 22-42</th>
<th>Box 43-63</th>
<th>Box 64-84</th>
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</thead>
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**Anomaloscope**

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<th>MR</th>
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**Hysteroid:Obsessoid Questionnaire**

**Hostility and Direction of Hostility Questionnaire**

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<th>Intropunitiveness</th>
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**Sixteen Personality Factor Questionnaire**

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<th>D</th>
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<th>F</th>
<th>G</th>
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<td>2</td>
<td>6</td>
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<td>M</td>
<td>N</td>
<td>O</td>
<td>Q1</td>
<td>Q2</td>
<td>Q3</td>
<td>Q4</td>
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**Second order factors**

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<tr>
<td>6.9</td>
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</table>
Karyotype: 45 X0/46 XXr

Date of birth: 6-5-49  Marital status: Single

Ascertained at cardiology clinic where she was under review, following an operation for coarctation of the aorta at the age of ten. Noted to have increased carrying angle, low hairline with some neck webbing, and sparse scalp hair. Cyclical oestrogen therapy produced some breast development, but was stopped after three years. Her previous medical history included an episode of otitis media, and she is now overweight.

The elder of two sisters, she gained six 'O' levels but failed Highers. She worked for a while in a haematology laboratory but found the tempo too great. She then took a medical secretarial course and now works in her father's office; (he is a solicitor).

Testing was undertaken in the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 156
VIQ 115  PIQ 94

Detailed sub-test scores

Information 12  Digit Symbol 7
Comprehension 10  Picture Completion 9
Arithmetic 13  Block Design 11
Similarities 13  Picture Arrangement 9
Digit Span 15  Object Assembly 10
Vocabulary 12

Benton Visual Retention Test

No. correct 8
No. errors 4

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity 1
" " orientation 2

Experimental Visual Recognition Test

NT
Experimental Formboards Test

**NT**

**100-Hue**

Total error score 76

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<td>64-84</td>
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**Hysteroid: Obsessoid Questionnaire**

Total 11

**Hostility and Direction of Hostility Questionnaire**

Total 26

Extrapunitiveness 14
Intropunitiveness 12

**Detailed sub-test scores**

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<tbody>
<tr>
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**Sixteen Personality Factor Questionnaire**

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<th>B</th>
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<th>E</th>
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<th>G</th>
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**Second order factors**

Anxiety 9.5
Introversion: Extraversion 1.3
A/7/71  Bender Visual Motor Gestalt Test
Karyotype  45 X0/46 XX,

Date of birth: 10-7-49  Marital status: Single

Referred at the age of 22 to a gynaecologist for investigation of amenorrhoea and short stature. She reported having had a single menstrual bleed at the age of 20. Noted to be slightly obese, to have shield-shaped chest with no breast development, sparse ancillary, pubic and scalp hair, but no neck webbing.

She is the third of three girls and works as a copy typist.

Testing was carried out at the Unit.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ  92  VIQ  97  PIQ  87

Detailed sub-test scores

- Information  10  Digit Symbol  7
- Comprehension  9  Picture Completion  8
- Arithmetic  8  Block Design  9
- Similarities  7  Picture Arrangement  8
- Digit Span  12  Object Assembly  9
- Vocabulary  10

Benton Visual Retention Test

No. correct  6
No. errors  6

Bender Visual Motor Gestalt Test

Errors of overlap or contiguity  4
" orientation  2

Experimental Visual Recognition Test

No. errors  4
Experimental Formboards Test

Series 1

\[ V_L (4) \ 7'' \ V_S (1) \ 17'' \ H_L (3) \ 29'' \ H_S (2) \ 317'' \]

Placement consistency rating 1

Series 2

Diamond \ V (2) \ 11'' \ H (1) \ 29''
Cross \ V (1) \ 8'' \ H (2) \ 16''

100-Hue

Total error score 40

Box 85-21: 2 Box 22-42: 9
" 43-63: 15 " 64-83: 14

Anomaloscope

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<tbody>
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<tr>
<td>Y/B &quot;</td>
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Hysteroid: Obsessoid Questionnaire

Total 20

Hostility and Direction of Hostility Questionnaire

Total 16

Extrapunitiveness 11 Intropunitiveness 5

Detailed sub-test scores

<table>
<thead>
<tr>
<th>AH</th>
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<tbody>
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Sixteen Personality Factor Questionnaire

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<td>I</td>
<td>Q_4</td>
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</tr>
</tbody>
</table>

Second order factors

Anxiety 3.0 Introversion: Extraversion 5.4
Karyotype: 45 X0/46 XY

Date of birth: 31-12-46  Marital status: Single

Referred to paediatric clinic for investigation of retarded growth. Noted to be rather obese, but to have no other physical stigmata apart from short stature. On gynaecological investigation demonstrated to have primitive ovarian rather than testicular gonads.

The second in a sibship of two, she gained five 'C' levels and is now employed as a primary school teacher. Her father has impaired red/green discrimination.

Testing was carried out in the Unit where she was admitted for gynaecological investigation. Although she completed the initial series of tests it was considered unlikely that she would wish to co-operate further.

Psychological Test Results

Wechsler Adult Intelligence Scale

FSIQ 104
VIQ 106  PIQ 99

Detailed sub-test scores

Information 11  Digit Symbol 9
Comprehension 9  Picture Completion 9
Arithmetic 10  Block Design 14
Similarities 12  Picture Arrangement 8
Digit Span 9  Object Assembly 10
Vocabulary 15

Benton Visual Retention Test

No. correct 6
No. errors 6

Bender Visual Motor Gestalt Test

NT

Experimental Visual Recognition Test

NT
Experimental Formboards Test

NT

100-Hue

Total error score 88

Box 85-21: 7  Box 22-42: 28

" 43-63: 45 " 64-84: 8

Anomaloscope

NT

Hysteroid: Obsessoid Questionnaire

Total 16

Hostility and Direction of Hostility Questionnaire

Total 8

Extrapunitiveness 6  Intropunitiveness 2

Detailed sub-test scores

AH 3  SC 2
CG 3  G 0
PH 0

Sixteen Personality Factor Questionnaire

A 4  L  1
B 8  N  7
C 6  N  5
E 5  O  3
F 5  Q1 7
G 9  Q2 7
H 4  Q3 6
I 8  Q4 6

Second order factors

Anxiety 4.0  Introversion: Extraversion 3.3
APPENDIX B

Published paper
PRELIMINARY REPORT ON INTELLIGENCE QUOTIENT SCORES OF PATIENTS WITH TURNER'S SYNDROME: A REPLICATION STUDY

BY
FELICITY BUCKLEY

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ROYAL MEDICO-PSYCHOLOGICAL ASSOCIATION

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THE BRITISH JOURNAL OF PSYCHIATRY
Vol. 119, No. 552, November 1971