Thesis for M.D.

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FRIEDRICH'S ATAXIA.

by

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While acting as House Physician to Dr. J. W. Anderson at the Glasgow Royal Infirmary I had the opportunity of observing two cases of Friedrichs Ataxia the subject I have selected as my Thesis. The following notes which were made on admission Dr. Anderson has kindly allowed me to make use of.
CASE I.

Phemie Sungeon Aet.11. admitted to ward VIII on 25th April 1893.

History of present Illness.

Two years ago she had an attack of measles accompanied by Bronchitis. This only confined her to bed for a few days, but shortly afterwards, her mother noticed that her walking had become somewhat impaired, her feet crossing each other causing her to trip and fall. At the same time her arms became affected causing her to drop things out of her hands. About six months afterwards she was sent to school and from that time began to get worse, her speech becoming affected in addition to the above, especially marked when excited. (She was a long time in beginning to speak originally).

About a year ago she had another attack of measles since which she has got worse with intervals when she appeared to be somewhat better. Latterly she has had frequent falls out of bed through the night, and occasionally when attempting to rise has fallen. Her hands and arms have got a good deal worse lately.

Family History.

Parents both alive and healthy. Two sisters
in good health, one brother affected in pretty much the same way as herself, and another affected since infancy with paralysis of one side. No history of nervous disease in Mother's family, in Father's there is a history of Chorea. Personal History. Up to two years ago she enjoyed good health. She has always been rather a nervous child. Has never had Scarlet Fever or Rheumatic Fever.

Physical Examination.

On inspection patient is seen to be a well developed healthy looking child with a good deal of colour in her face. While lying quiet in bed nothing particular is seen, but soon jerking of the head is observed while the lips twitch to one or other side. The eyes keep steady and do not seem to be affected, occasionally their is a contraction of the muscles of the chest. On putting out the tongue nothing abnormal is noticed in the method of doing so, although an occasional tremor may be observed in it. So long as she remains quiet there does not seem to be any movement of the limbs, but on asking her to stretch out her arm, the fingers and then the whole arm begin to show involuntary movements though not to any great
extent. The legs at time of examination showed very little movement, but on attempting to walk the feet are jerked across one another interfering with locomotion.

**Respiratory System.**

On examination nothing abnormal is to be made out, the muscles concerned in respiration appearing to be unaffected, her Mother states that at night her breathing becomes rather noisy as if she had something in her throat.

**Circulatory System.**

P. 104 regular and of good volume, Apex heat in 5th interspace. Cardiac dullness of normal extent, no murmurs to be detected, heart sounds accentuated.

**Alimentary System.**

Tongue moist and clean, bowels regular, appetite good.

**Locomotory System.**

The difficulty of locomotion before alluded to seems to be rather due to spasmodic action of the muscles than to any loss of power.

**Urinary System.**

Urine clear, slight mucus deposit, Sp. Gr. 1026 acid no sugar or albumen.
Nervous System.

Sensibility to pain and touch unimpaired. Superficial and deep reflex almost abolished. Intelligence good, speech slow and deliberate. Sensory functions normal. Muscular sense only slightly impaired, sight good, pupils moderately dilated, react both to light and accommodation, hearing good. Motor functions. Organic reflexes normal, skin reflex diminished; tendon reflex almost gone. Vaso motor functions present no abnormality. Cerebral functions, intelligence fairly good, attention and memory good, speech slow and drawling, sleep good. Cranium large, no pain on percussion of spine or cranium.
CASE II.


History of present illness.

Two years ago suffered from measles and Bronchitis at the same time but in a more aggravated form than his sister. Since then he has never been able to walk more than a few yards unless holding on to something. Shortly after this, while sitting up he would fall backwards. From commencement he has had difficulty in using his hands, always dropping things out of them. Unlike his sister he has never been thrown out of bed, his speech is affected and in a more marked degree than hers.

Personal history.

While a baby he seemed well and strong, but since twelve months of age he has never appeared very strong, though until two years ago had nothing definitely wrong with him.

Physical Examination.

Face pale and rather unhealthy looking. Breathing heavy as if suffering from some nasal obstruction. He is small and rather poorly developed. There is twitching of both eyebrows, nystagmus and squinting of both eyes. On pushing out his tongue it is done
so in a way characteristic of Chorea and trembles when protruded. Movements but not violent occur in all the limbs while at rest. On voluntary movement he seems not to have complete control over them being unable to guide his hand directly to his mouth. He is unable to walk without assistance or even to stand alone. He walks with his feet apart advancing each foot a few inches at a time and staggering very much. While walking he keeps his hands flexed on his forearm and the latter on the arm. His speech is slow and deliberate.

Respiratory System.

Left side of chest more prominent than the right side and seems to expand more freely. Lungs normal.

Circulatory System.

P.124, regular and of good volume. Heart sounds accentuated. No murmurs.

Alimentary System.

Tongue moist and slightly furred. Bowels regular, appetite good.

Locomotory System.

Inability to walk and loss of control over movements of his arms and legs. There is some
wasting of the adductor muscles of the thumb, otherwise nothing noticeable with the exception of slight Talipes Varus in both feet.

**Urinary System.**

Urine pale, no deposit, acid, no albumen or sugar.

**Nervous System.**


Craniun large but otherwise normal. Spine shows lateral curvature to right in dorsal region. There seems to be loss of power in both hands especially the left, but none in legs.

On first listening to the Mother's statement Chorea seemed to be the most likely affection from which the children were suffering, her description of her daughter being thrown out of bed onto the floor
resembling very much what would happen in a marked case of Chorea while the jerky and clumsy movements of the hands seemed also to point to this disease. In the case of the boy the manner in which he put out his tongue, the nystagmus, twitching of the eyebrows and jerkiness of the limbs favoured the diagnosis of Chorea. On going more fully into the examination however, the fact that the jerky movements are bilateral, are little seen in the facial muscles, that the reflexes are almost gone, and that the mental condition remains clear and bright militates against the diagnosis of Chorea. The manner of locomotion also is unlike that of a Choreic patient.

On seeing the children walk one thought of locomotor Ataxia, but against this diagnosis were the age of the patients, the fact of the upper limbs being involved so early in the disease, the absence of the Argyle Robertson symptom and of lightning pains, also the presence of nystagmus and the peculiar form of speech. The gait also was not exactly that of a patient suffering from Tabes Dorsalis, as both cases were quite unable to walk in a straight line but staggered all over the room.

Excluding Chorea and Locomotor Ataxia the age of the patients, history of onset and symptoms in
general seemed to point to the diagnosis of Friedrich's Ataxia as the affection from which they were suffering. A point in favour of this in the case of the boy lay in the existence of Talipes and spinal curvature both of very common occurrence in cases of Friedrich's disease.

Both cases were detained in Hospital for several months but were ultimately dismissed without any sign of improvement having taken place.
FRIEDRICH'S ATAXIA.

Synonyms. Hereditary Ataxia, Friedrich's disease. Family Ataxia, Generic Ataxia and Diffuse Sclerosis of spinal cord and bulb. Of these the term Hereditary Ataxia is rather a misnomer as the disease is except in a few rare cases (1) not directly hereditary from parent to child. The term Family Ataxia seems to me a better one as the disease in the great majority of cases has as its victims several members of the same family.

History. Friedrich's Ataxia was discovered by Friedrich in 1861 in which year he reported 6 cases, later on, other cases were reported by other observers and several more by himself, but by 1880 only 31 cases had been reported in all. In 1885, 57 cases were reported by Dr. Everitt Smith in the Boston Medical and Surgical Journal (Oct. 15th 1885). Since then numerous cases have been reported by different observers in America, Germany, Italy, France, Switzerland and England. Few cases have as yet been reported in Scotland. The greatest number of cases have come from America, Dr. Griffiths reporting in 1889, 143 cases (2) since which others have been added. In 1888, Soea in his These de
Paris published 165 cases but many of these are considered doubtful cases (3).

**Definition.** Friedrichs Ataxia is a disease affecting the posterior and postero lateral regions of the spinal cord and involving ultimately in many cases the Medulla and Pons more especially in the region of the hypoglossal nucleus. It is characterised by slowly developing Ataxia commencing first in the lower then attacking the upper limbs, and in the latter stages the muscles of the back, head, and neck. As a rule there is an entire absence of the knee jerk. There are no sensory abnormalities, nor is there any failure of the mental functions. Nystagmus is present along with a peculiar affection of the speech. There is also very commonly present some curvature of the spine along with some form of club foot. The disease is of prolonged duration and is generally brought to a close by death from exhaustion or from some complication. Hitherto treatment has had very slight effect. It is further characterised by the occurrence of several cases in the same family, isolated cases being rare though such have been reported by Dreschfield (4) McCann (5) and Ewart (6).

**Etiology.** With the exception of a few isolated
cases this disease occurs in families where there is a neurotic history, some members of the family exhibiting some neurosis such as Epilepsy, Insanity, Inebriety or great nervous irritability. Gowers mentions a case where the mother of the patient and her mother again suffered from the disease (7) while Byrom Bramwell also mentions a case of direct inheritance reported by Vizioli (8). Alcoholism in the parents is said to have some influence but this is doubtful though it has been observed in many cases, consanguinity in parents has probably some influence in predisposing towards the disease. Keating mentions Tuberculosis as a cause, while MacCunn reports a case in a family with strong tubercular but no neurotic history (9). Syphilis does not seem to be a factor.

As to the influence of race, it is probably more common in America than elsewhere.

The disease may develop from 2-24 though the case is recorded in which it came on at the age of 65 (10).

Sex has not much if any influence on the disease as the number of cases is about equal in each sex though there is a slight preponderance of
the male sex except in America where it is vice versa. The majority of cases are found in the labouring classes. As far as immediate causes go the disease has been reported as occurring after Measles, Scarlet Fever, Variola and Chorea. It has also followed injury such as a blow on the head.

Symptoms. In all probability the first symptoms noticed will consist in some alteration in the patient's gait or he will be noticed to be getting clumsy in handling things. These symptoms may not be at all marked at first so that little attention may be paid to them but as time goes on they become more noticeable, the feet are jerked up and in some cases put down with a stamp as in Locomotor Ataxia. The difficulty in walking is especially marked in the dark, the patient becomes unable to walk in a straight line and often falls. He may still be able to walk a considerable distance with assistance as there is no failure of strength it may be for many years. The upper limbs eventually begin to show an incoordination in their movements, the patient finding difficulty in performing any act requiring delicacy of manipulation, in fact he may become unable to feed himself. In the addition to the unsteadiness of gait the way the patient holds
himself is peculiar the head and back being bent forwards while the body sways from side to side, this swaying movement is also noticed in many cases while standing at rest. In addition there is in some cases a movement resembling that of Athetosis. The jerky inco-ordinate movements especially when also noticed in head make the resemblance to Chorea a close one. A case has been recorded by W. E. Tresidder in which there was inco-ordination of the muscles in swallowing (11). Another symptom present in the majority of cases is a flexure of the spinal cord, while another very frequent symptom is the presence of some form of Club foot though Nolan says it is a mistake to regard this as Pathognomonic (12). Nystagmus more or less marked is a constant feature, though the movement is said to be slower than the ordinary form of nystagmus, this symptom is however one of the later characteristics of the disease and may not come on for some years. The Argyle Robertson symptom is absent. There is no optic atrophy. Sensory affections as a general rule are absent in this disease. There are no lightning pain nor visceral crisis as in Locomotor Ataxy nor is there any impairment of sensation though exceptions to them have been recorded by various observers. Dr. Bramwell reports three
cases where the sensibility to touch, heat, cold
and pain were all exaggerated, while in two were
lightning pains and visceral crisis (13).
Muscular sense is not impaired. The knee jerk is
in typical cases absent but may be present in one
and absent in other or present in both. Dr. Ewart
(14) mentions a case where they were increased.
On making the patient stand up Romberg's symptom
is absent in the great majority of cases. The
mental functions in this disease show no impairment,
in fact, many of those affected seem to be the
sharpest members of the family. A peculiarity
often observed is a tendency to uncontrolable fits
of laughter. Nolan however mentions three cases
of mental deficiency (15).

The progress of the disease is very slow;
many years 20, 30 or more may elapse before death
occurs and when this does happen it is through
sheer weakness or from some complication arising.
The progress however, if slow is steady, the
symptoms gradually increasing until the patient is
unable to move about at all even with assistance.
In consequence of want of use the muscles atrophy
and contraction occur. The muscular weakness may
be confined to the legs, where it is at all events
always greater, the electrical contractility seems to be slightly decreased.

Speech as the disease progresses becomes affected, the words being drawn out in a slow monotonous manner and as time goes on becoming thick and indistinct. Vertigo is commonly observed, while in some cases headache is noticeable. There are no Vasomotor changes as a rule, though Polyuria has been observed by Dawa (16), while in one of Friedrich's cases flushing oedema, polyuria and salivation were noticed.

Fragility of the bones is mentioned by Dawa as a curious symptom.

**Differential Diagnosis.** There are several diseases which on superficial examination may be confounded with Friedrich's Ataxia, viz. Locomotor Ataxia, Cerebellar disease, Ataxia paraplegia, Chorea and Disseminated Sclerosis.

**V. Locomotor Ataxia.** Between these two diseases while there are considerable points of resemblance there are also other points of marked difference. Friedrich's Ataxia in the first place is a disease commencing during childhood, while Locomotor Ataxia is a disease occurring in adult life, though cases
have been recorded by Remak (17) occurring in childhood. Gowers says that there is a difficulty of diagnosis in "children with slight takes subjects of inherited Syphilis where it is not uncommon to have some weakness of the legs, coexisting with disordered speech in consequence of some early lesion of the brain". On looking into the history a decided difference is observed. Friedrich's Ataxia, except in rare cases occurring in several children of the same generation, while it is exceptional to find this in Locomotor Ataxia. On looking into the symptomatology there is considerable resemblance between the two. In both there is the peculiar unsteadiness in walking and standing, the peculiar stumping character of the former however is more marked in Locomotor Ataxia; the difficulty in walking seems to be greater in Friedrich's Ataxia than in Locomotor Ataxy, the patient staggering about like a drunk man and being unable to walk in a straight line which is not the case in Locomotor Ataxy. Romberg's symptom is present in L. A. not in Friedrich's Ataxia. In well marked cases of Friedrich's there is a jerky movement of head, neck and body (choreiform in character) not present in Locomotor Ataxy. On investigating the sensory symptoms a
decided difference is noticed, the lightning pains and crisis present in Locomotor Ataxy being absent in Friedrich's Ataxy; nor does there appear to be the impairment of the sensory functions in Friedrich's disease. The knee jerk as a rule is found absent in Friedrich as in Locomotor Ataxy. The upper limbs are seldom affected in Locomotor, always (sooner or later) in Friedrich. Nystagmus always present in F. A. is absent in Locomotor Ataxy, while on the other hand it is just vice versa with Argyle Robertson symptom. Optic atrophy is never observed in F. A. but may be in L. Ataxy. Another diagnostic feature is the speech which present no peculiarity in Locomotor Ataxy. It is common to have rectal and bladder affection in Locomotor, rare in Friedrich's Ataxy. While fragility of the bones has been observed in Friedrich's there are no marked bone lesions as in Locomotor Ataxy. Trophic lesions are rare in Friedrich's disease, common in Locomotor Ataxy. Both are diseases of long duration, but the symptoms are not so steadily progressive in L. A. as in Friedrich's Ataxy. The presence of spinal curvature and talipes so often observed in F. A. cannot be looked upon as a diagnostic symptom in Locomotor Ataxy.

V. Insular Sclerosis. There are many points
of difference between these two diseases though in atypical cases where the rhythmical tremors have temporarily disappeared and in these exceptional cases occurring in children the diagnosis becomes difficult. Comparing typical cases no great difficulty exists in the differential diagnosis. Insular Sclerosis occurring during adult life, mainly between 20 and 30, rarely after 30 and only occasionally during puberty and in childhood. There does not seem to be the same hereditary predisposition to nervous disease seen in Insular Sclerosis as in Friedrich's Ataxia. In the symptoms there are many points of resemblance such as rhythmical tremors increased on making any muscular exertion. Nystagmus, twitching of tongue and lips, while there is nearly always some defect in the speech, the voice being weak and monotonous and the words slurred. It differs in many cases by attacks of vertigo though this is occasionally seen in Friedrich's Ataxia, and by the presence of double vision, though this is as a rule temporary. As regards Nystagmus, this is said to be stopped by the patient fixing his eyes on the tip of his nose (18). There is also in many cases of Insular Sclerosis optic atrophy (19). On examining the reflexes the knee jerk in the majority of cases of Insular Sclerosis is increased. Both
are alike in the absence of any sensory atrophic disturbance until at all events near the end when bed sores are apt to form in Insular Sclerosis. The electrical contractibility is as a rule undiminished in the case of Insular Sclerosis. As the disease progresses it resembles Friedrich's Ataxia in the occurrence of contractions and the loss of muscular power going on almost to complete paralysis. The patient suffering from Insular Sclerosis offers intellectually, a considerable contrast to a case of Friedrich's as he is rather weak mentally and has a poor memory. They resemble one another in being subject to fits of uncontrolable laughter. Another important point in differential diagnosis is the absence of any form of deformity in Insular Sclerosis.

V. Chorea. Another disease which shows points of resemblance to Friedrich's Ataxia is Chorea. The diseases are alike in both occurring at an early age, i.e. before puberty. Both show inco-ordination of muscles giving rise to difficulty in walking and to the performance of anything requiring delicacy of manipulation, also nystagmus and affection of speech. There is also in both a family history of neurotic disease.

Points of difference are more numerous. Friedrich's Ataxia occurs about equally in the two sexes;
Chorea shows a predominance of females. The movements in the case of Chorea are to begin with unilateral and even after they have universal often show a predominance on one side. The muscles of the face are seldom affected in Friedrich's Ataxia commonly so in Chorea. The movements of the eyes seen in Chorea is not the slow lateral movement seen in Friedrich's Ataxia, but is sudden and without purpose in various directions. The movement of the tongue is also more sudden and jerky in chorea than in Friedrich's Ataxia. The tendon reflexes are not abolished in Chorea, while in Friedrich's Ataxia they are absent or almost so. There is no special deformity seen in Chorea as in Friedrich's Ataxia. As regards the mental faculties, in Friedrich's Ataxia they are always bright and often more so than usual, while in Chorea the patient has a dull vacant expression, impaired brain power and loss of memory. A very important difference is the amenability of the vast majority of cases of Chorea to treatment.

V. Ataxia Paraplegia. Yet another case with which Friedrich's Ataxia may be confounded is Ataxia Paraplegia. Points of resemblance between the two lie in the symptoms of inco-ordination, and muscular weakness ultimately existing in both along with the
fact of there being no sensory affections such as lightning or girdle pains, or visceral crises. In both there is a nystagmus, not so marked however, in the case of Ataxia Paraplegia, there being often more in this disease when the eyes are at rest (20). There is seldom any pupil abnormality or impairment of movement in the ocular muscles. There is often a tremulous movement of facial muscles and an impairment of speech. As in Friedrich's Ataxia also a history of syphilis is rare.

The points of difference are numerous and marked. Ataxia Paraplegia in the majority of cases occurs after puberty, generally between 30 and 40 and never as early as in Friedrich's Ataxia (21). It is also more frequently observed in males than females. There is not, except in a few cases a neurotic history. Coming to the symptoms, the gait is more spastic than ataxic. As regards sensory symptoms, there is very often sacral pain present while the reflexes on examination exhibit a marked difference there being an increase in the knee jerk, while ankledonus is also often present. The Sphincters are more often involved than in the case with Friedrich's Ataxia. There is no special deformity either present. Regarding prognosis, both are alike in being of long duration,
and in either being little influenced by treatment.

Pathology. Regarding the Pathological appearances of this disease, owing to the fact that comparatively few P.M. examinations have been made, while of these made many necessarily have been cases of long standing, considerable differences exist as to the exact situation of the primary lesion. All seem agreed that Sclerosis found in the Cols. of Burdach and Goll and in the vesicular col. of Clark are primary that found in the direct cerebellar and crossed pyramidal is declared by Dijerine and Le Tulle (22) to be secondary, the character of the lesion in these tracts differing from that in the post. cols., while Dr. Bramwell believes the lesion in these cols. to be primary. Friedrich himself believes the lesions in the cord to be primary and not secondary (23). In addition probably as the result of extension Sclerosis is occasionally found in the Ant. Cols. (24). Lesauri's tract is sometimes, but not as a rule, affected; the post nerve roots are generally affected. Gowers (25) mentions that cells in the anterior cornua may show signs of affection, while he believes that future observers will be able to report changes in the post cornua. The cord in all cases seems to have been smaller than normal in its whole extent as if due to congenital
atrophy, while Friedrich found shrinking of the Pons and Medulla. Atrophy of cerebellum has also been observed. Senator (26) is of opinion that in truly genuine cases of Friedrich, the most important phases of the disease are direct congenital atrophy of the cerebellum with which probably is associated atrophy of the spinal cord.

As regards the character of the Sclerosis itself Dejerine and Le Tulle say that it is a purely neuroglial one. "The appearances of the posterior cols." they say "is quite different from that which one sees in ordinary tabes, the fibres crossing one another in different directions and forming true whorls. The processes of pia mater which pass into the cord are perfectly normal, and in no way more apparent than in health. Under a high power one can see that the whorls consist of very fine fibres, pressed one against the other and arranged in different plans. In the midst of these fibres may be found a few nerve tubes, which seem in many cases to follow the direction of the whorls in which they are situated (27).

Gowers mentions a case (28) in which the neuroglial overgrowth suggested a pure increase of the normal tissue, rather than as a result of inflammation, and the peripheral nerve fibres although not degenerated showed a considerable number of embryonal
nerve fibres in the fasciculi. He considers that Dejerine and Le Tulle's idea of the Nemoglial increase being the sole element in the morbid process incorrect.

**Prognosis.** As far as life is concerned the prognosis is distinctly good, as with few exceptions life is prolonged for many years, 20, 30 or even more. Regarding any hope of improvement it is distinctly bad; as so far no cases have been reported as cured. It is quite possible for many patients however to continue in their employment for many years so slow and gradual is the progress of the disease. Unless some complication arises which hastens death, the patient will in all probability end life as a hopeless cripple.

**Treatment.** As yet no treatment has been followed by any definite improvement in this disease. Many remedies have been tried such as Arsenic, Phosphorous and Silver, the constant current and suspension but only with very temporary success and in some cases, while in severe cases, the two latter have seemed to do harm. A Sayres plaster jacket may be tried to give support to the back. As regards prevention in future members of the family, the infant should not be nursed by the mother, great care should be taken to keep it from infectious diseases and from cold, while in
later years a quiet life without any physical strain should be led. Dr. Ewart reports a case in which improvement in speech, standing and walking is said to have occurred under tonics, rest and massage (29).
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24. Fagge System of Medicine, p. 540.
26. Sajous Annual of the Universal Med. Sciences 1894, B. 33 vol. II.

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