A STUDY

of

CONGENITAL AND HEREDITARY PTOSIS

with special attention to

accompanying

CONGENITAL MALDEVELOPMENT OF THE OS SPFENOIDALE

by

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INTRODUCTION.

While working in an Edinburgh hospital in 1936 my curiosity was aroused by a ward-maid who appeared to go about with her eyes more than half-closed. She appeared to do her work efficiently and even attended hospital dances, although her appearance was a little grotesque, as she was obliged to throw her head back in order to see through the narrow slit to which her palpebral aperture was reduced. Investigation brought to light the interesting fact that her father and several of her brothers and sisters were similarly afflicted. On examination these people were found to be healthy in other ways, and to have no other neurological signs or symptoms, and in particular no syphilitic taint. Moreover, the condition of the eyelids was seen not to be a true "ptosis" but rather a smallness of the palpebral aperture, the eyelids being tacked down at the edges. On Dr. Ritchie Russell's suggestion I had X-Rays taken of the skull of these patients, and found that they all showed varying degrees of a definite developmental abnormality in the shape and form of the sphenoid bone, of a type that does not appear to have been described before. Later I was able to examine another family, entirely unrelated to the previous one, several of whose members
had a similar smallness of the palpebral aperture, accompanied this time by epicanthus. X-Rays of their skulls showed anomaly of the sphenoid of precisely the same nature as that seen in the first family examined. I then proceeded to investigate as many of the cases dubbed "congenital ptosis" as opportunity offered, and found that they were not all of the same nature, but could be divided into various fairly well-defined clinical groups. It is on this investigation that the following chapters are based. As the literature on the subject is relatively scanty, and often difficult to find, I have tried to reproduce as fully as possible the descriptions of previously published cases. It is a difficult subject to approach from the point of view of etiology, as the patients, being otherwise healthy, do not generally die in hospital, so that few autopsies have been performed. Many workers have published isolated cases or families, but the only composite works on the subject are that of Willbrand and Saenger in their textbook on the Neurology of the Eye, and an article by Pergola. In the few post-mortems that have been done the conformation of the sphenoid bone has not been noted, and in none of the published cases do X-Rays of the skull appear to have been taken.

A chapter is included on Hereditary Non-Congenital
Ptosis, of which I have had the opportunity of seeing two cases.

For convenience a short summary is added at the end of each chapter, and each chapter is followed by its own bibliography.

There is also a final summary at the end.
CHAPTER I.

CONGENITAL AND HEREDITARY PTOSIS WITH MALDEVELOPMENT OF THE OS SPHENOIDALE.
The following is an account of the D----- family, in whom congenital ptosis with blepharophimosis has occurred throughout three generations; of the forty persons comprising these three generations, fifteen were affected, of whom eight out of twenty-four were males and seven out of sixteen were females. The first member to be affected is said to have had normal parents. The explanation believed in the family is that this person's mother, on the eve of his birth, was terrified by being told tales of how many soldiers were being blinded in the Crimean war. Apart from this case, unaffected members did not transmit the condition to their offspring. There is no history of consanguinity.

The following is the genealogical tree of the D----- family

- = affected members.
The numbers refer to the members who have been examined personally and correspond to the number placed opposite each name in the ensuing text.
Twelve of the fifteen affected members have been examined.

J.D. (1) male, aged 57. Occupation: woodworker.

He has bilateral ptosis, two-thirds of the pupil being covered in the right eye and three-quarters in the left. The length of the palpebral fissure is 13 mm. s in both eyes. The width at the widest part is 4 mm. s in the right, and 3 mm. s in the left eye. The external ocular movements are full. There are some slight nystagmoid jerks on lateral deviation, accompanied by blinking movements of the upper and lower lids. The pupils are regular and equal, react to light directly and consensually, and react to accommodation. The fundi show no abnormality. Both eyelids show the line of an old incision. Eyelashes are present on the upper lid, with the exception of the inner \( \frac{1}{2} \) cm., and are entirely absent from the lower lid. The width of both orbits is 3\( \frac{1}{2} \) cm. s. The head is not thrown back, and there is no abnormal wrinkling of the forehead. The hair and eyebrows are dark brown, turning grey.

Uncorrected vision in the right eye is 5/60 J.1, but if the lids are held open it is only 2/60. This suggests that the drooped eyelid exercises the effect of a stenopaeic slit.
Refraction - 3 D. Sph. V = 6/18, - 6 D. Sph. v = J.1. A cylindrical error is present, but putting in a cylinder appears to make no difference to visual acuity.

Uncorrected vision in the left eye is 5/60 J.4. Refraction: -2 D. Sph. V 2 6/36. The poor vision present in spite of this low degree of myopia is probably due to amblyopia consequent on the convergent strabismus.

The patient has never been ill. He is mentally normal, and is of average intelligence. He suffers no incapacity from the condition, and is able to perform his work satisfactorily. In childhood he was operated on by Dr. Argyll Robertson at Edinburgh Royal Infirmary. He thinks that this operation lifted his eyelids a little higher than they were before.

For photographs, see following pages.
J.D. (1).
(Close-up of eyes)
X-Ray photographs of skull of J.D----- (1)
(for remarks, see next page)
X-Ray photographs of skull of J.D. (1)

1. Abnormal configuration of the superior orbital fissure (wide base)
2. Increase in length of lesser wing of sphenoid.
3. Shallowness of middle fossa.
4. Increase in size of spheno-maxillary fissure seen in lateral view.


There is bilateral ptosis, three-quarters of the pupil being covered in the right eye and one-half of the pupil in the left eye. The length of the palpebral fissure is 18 mm.s in the right eye and 17 mm.s in the left eye. The width of the palpebral fissures at their widest part is 4 mm.s in the right eye and 6 mm.s in the left. The external ocular movements are full.

There is no nystagmus. The pupils are regular and equal, and react to light and accommodation. He has high myopia and gets 6/24 vision with a -14 D. lens in the right eye. The left eye has nebulae and vision is not improved by glasses. The fundi are healthy except for the usual choroidal degeneration round about the optic disc. X-Rays of the skull show the familial abnormalities. Eyelashes are present in normal amount on the upper lids and are absent from the lower lids.
The head is not markedly tilted back, but there is considerable wrinkling of the forehead. The hair and eyebrows are brown in colour. The width of the orbits is $3\frac{1}{2}$ cm.s.

This man is mentally normal. His deformity does not incapacitate him.

X-Ray photographs of the skull of J.D. (2)

Both upper lids are ptosed, half of the pupil being covered in both eyes. The length of the palpebral fissure is 18 mm.s in both eyes, and the width 4 mm.s. The external ocular movements are full. There are coarse nystagmoid jerks on both lateral deviation and central fixation. These jerks are accompanied by blinking movements of both lids. The pupils are regular and equal, and react to light and accommodation. The fundi are normal. Vision uncorrected in the right eye is 6/60 and J.14 and in the left eye finger-counting at 2 metres and J.16. He is highly myopic and wearing 11.00 D.Sph. sees only 6/36 in either eye. This is accounted for by the nystagmus, and by the presence of early refractive change in the lenses. The eyelids are very thin, there being no evidence of the presence of the levator palpebrae superioris muscle. There is marked hollowing above the eyelids and absence of orbital tissue. The eyelashes on the upper lids are normal, but are absent from the lower lids. The patient always carries his head tilted backwards, and there is marked wrinkling of the forehead. The width of the orbits is 3½ cm.s. The hair and eyebrows are mouse coloured.
This patient is not greatly incapacitated in his work by the presence of ptosis. He remarks that if he tries to throw an object upwards it always goes over the back of his head. He is mentally stable and very intelligent.

X-Ray photographs of skull of R.D---- (3)

This case has the familial configuration of the sphenoid, but shows the least departure from normal of any of the family.

(for photographs, see following page)
X-Ray photographs of skull of R.D.(3)

This patient has a moderate degree of bilateral ptosis, about one quarter of both pupils being covered. The length of the palpebral fissure is 18 mm.s in the right eye and 17 mm.s in the left eye. The width of the palpebral fissures at their widest part is 7 mm.s in both eyes. The external ocular movements are full. There is horizontal nystagmus on lateral deviation. The pupils are regular and equal, and react to light and accommodation. Eyelashes are entirely absent from the lower lids, but are normal on the upper lids. Both eyelids show evidence of previous operative interference. The width of the orbits is 3½ cm.s The head is not tilted back, and there is no undue wrinkling of the forehead. The hair and eyebrows are mouse-coloured.

This patient is exceedingly intelligent. She states that when she was a child two operations were performed on her eyelids, one in Edinburgh Royal Infirmary, when she was an infant, and one in the Berkeley Street Eye Infirmary, Glasgow, when she was thirteen. She now shows the least degree of ptosis of any member of this family observed. She is married, but has no children. She says that she was
once told by a doctor that she was physically incapable of having a family. This matter has not been investigated.

**M.D. (5)** Female, aged 45. Occupation: housewife.

She has bilateral ptosis, half the pupil being covered in both eyes. The palpebral fissure measures 17 mm. in the right eye and 15 mm. in the left eye. The width of each palpebral fissure is 4 mm. The external ocular movements are full. There is slight nystagmus on lateral deviation. The pupils are regular and equal, and react to light and accommodation. The fundi are normal. Eyelashes are present as in a normal person on the upper lids, but are absent from the lower lids. The width of the orbits is 3½ cm. She always has her head slightly tilted backwards, but there is no marked wrinkling of the forehead. The hair and eyebrows are brown.

This patient is intellectually very feeble.

**I.S. (6)** Female, aged 41. Occupation: housewife.

There is bilateral ptosis of a very marked degree, more than three-quarters of the pupil being covered in both eyes. The right palpebral fissure measures 15 mm., the left 16 mm. The width of the
palpebral fissure in each eye is 3 mm. The external ocular movements are full. There are coarse nystagmoid jerks on lateral deviation and central fixation, accompanied by blinking movements of the eyelids. There is entropion of both upper eyelids. The eyelashes irritate the surface of the bulb, and the patient has chronic conjunctivitis. She was admitted to the Royal Infirmary to Dr. Traquair's wards, where the cautery was applied to the upper lids with a view to relieving the entropion. The narrowness of the palpebral apertures, cicatricial contraction of the lids, corneal opacities, and the presence of a thick exudate, combine to make examination of the pupillary reactions and of the fundi impossible. Eyelashes are entirely absent from the lower lids. This patient sees very little, vision being reduced to finger-counting at about a foot distance. Vision is not improved by spheres for near or for far vision. The width of the orbits is 3 cm. The hair and eyebrows are brown.

Z.S. is of average intelligence and mentally stable. Her vision is so bad that she very seldom goes outside.

(For photograph, see the following page)
X-Ray photographs of skull of I.S. (6)

1. The roofs of the orbits are low.
2. The sella turcica is abnormally developed, and is of the type typical to the family.
3. There is some hyperostosis in the frontal region and vertex on the right side.

(for Photographs, see the following page)
X-Ray photographs of skull of I.S. (6).

There is bilateral ptosis, three-quarters of each pupil being covered. Each palpebral fissure measures 17 mm. in length and 4 mm. in width. The external ocular movements are full. There is lateral nystagmus, which is present on both central fixation and lateral deviation, and is accompanied by blinking movements of the upper and lower lids. The pupils are regular and equal, and react to light and accommodation. Examination of the fundi shows myopic choroidal changes at the outer side of both discs. Eyelashes are present on the upper lids, but absent from the lower lids. The patient always has his head thrown backwards, and the forehead is very wrinkled. The width of both orbits is 3½ cm. The hair and eyebrows are mouse-coloured.

Uncorrected vision in the right eye is 6/24—,
J.1. When the eyelids are held open V. = 3/60. With
-12 D. Sph. -4 D. Cyl. axis 180° V = 6/18 J.1.

Vision uncorrected in the left eye is 6/24—,
J.1. When the eyelids are held open V = 3/60. With
-10 D. Sph. -4 D. Cyl. axis 180° V = 6/18 J.1.

This patient is mentally normal, and very intelligent indeed. He has never been ill. He is able
to do his work satisfactorily, plays football, and suffers no incapacity from his condition.
X-Ray photographs of skull of V.D. (7)

The anterior and middle fossae appear normal.

The lowermost portion of the sphenoid is wide and this is the only region that shows the familial abnormality.

She has a severe degree of bilateral ptosis, three-quarters of the pupil being covered in both eyes. She is able to raise her eyelids until about half of the pupil is exposed. If the observer's fingers are pressed firmly over the eyebrows so that the frontalis muscle is not allowed to come into action, only a very slight upward movement of the eyelids can be carried out. The length of the palpebral fissure of each eye is 18 mm., and the width at the widest part is 5 mm. in the left and 4 mm. in the right eye. The external ocular movements are full. On lateral fixation there are coarse jerking movements of the eyeballs in a direction upwards and away from the point of fixation. On central fixation the eyeballs jerk in a vertical direction. These movements of the eyeballs are accompanied by twitching of the upper and lower lids. The patient states that she has never seen double. The pupils are equal and regular and react equally to light, both direct and consensual, and to accommodation. The light reaction is well sustained, and there is no hippus. The fundus oculi shows no abnormality. The eyelids are smooth, and there is noticeable hollowing and absence of tissue above the orbits.
Eyelashes are entirely absent from the lower lids, but are present on the upper lids, being sparse in the inner 1 cm. on both sides. The width of the orbits is 3½ cm. The patient always carries her head tilted backwards, and there is pronounced wrinkling of the forehead. The hair and eyebrows are mouse-coloured.


Considering the degree of myopia the distant vision without glasses is surprisingly good. This is probably due to the drooped eyelid narrowing the palpebral aperture, and so producing the effect of looking through a stenopaeic slit.

Routine examination of the nervous system, and of the other systems of the body, revealed no abnormality. The electrical reactions of the muscles of the face have been tested and found to be normal. The blood Wassermann reaction is negative.

This patient states that for as long as she can remember she has been unable to open her eyelids. The condition has never caused her any inconvenience. She can read comfortably, and in childhood was able to
play games freely with her companions. Although the drooping of the eyelids gives her a somewhat sleepy expression, she is a normally intelligent and mentally alert girl, who carries out satisfactorily her duties as a housemaid in a hospital.

Instillation of one drop of 4% cocaine hydrochloride into the eyes is followed by considerable dilatation of the pupils, but by no alteration in the position of the eyelids.

**X-Ray photographs of skull of M.D---- (8)**

1. Increase in width of base of superior orbital fissures.
2. The lesser wings of the sphenoid are longer than normal in their antero-posterior diameter.
3. The middle fossa of the skull is much flatter from above downwards than normal.
4. The roofs of the orbits are nearer the level of the cribiform plate of the ethmoid than normal.
5. The pituitary fossa is very small, and the dorsum and posterior clinoids are much larger than normal.

(For actual photographs, see following page)
X-ray photographs of skull of M.D--- (8)

There is a moderate degree of bilateral ptosis, about half of each pupil being covered. The palpebral fissure of each eye measures 17 mm.s in length and 5 mm.s in width. The external ocular movements are full. There are coarse nystagmoid jerks similar to those described in Case 10. The pupils are regular and equal, and react to light and accommodation.

Eyelashes are entirely absent from the lower lids, and are absent from the inner $\frac{1}{2}$ cm. of the upper lids. This patient carries her head perfectly straight, and there is no marked wrinkling of the forehead. The orbits measure $3\frac{1}{2}$ cm.s across. The hair and eyebrows are brown. E.D. has a distinctly Mongoloid facies. She is taciturn, and of a somewhat sullen disposition, but appears to be mentally normal. She refused to be photographed.


This child lost her right eye as the result of an accident three years ago. Both upper lids have been ptosed since birth. The left pupil is half covered by the lid. The length of the right palpebral fissure is 18 mm.s, that of the left 21 mm.s. In each
eye the width of the palpebral fissure at its widest part is 6 mm. The external ocular movements of the left eye are full, and there is no nystagmus. The left pupil is regular and equal, and reacts to light and accommodation, and the fundus is normal. Eyelashes are absent from the lower lids and are normal in the upper lids. The child does not keep her head tilted back, and the forehead is not abnormally wrinkled. The width of the orbits is $3\frac{1}{2}$ cm. The hair and eyebrows are fair.

E.D. is a bright, intelligent child, finding herself incapacitated in neither work nor play by her congenital deformity.


There is bilateral ptosis of a considerable degree. Only one-third of the right pupil and half of the left pupil are uncovered. The palpebral fissure of each eye measures only 16 mm. The width of the right palpebral fissure at its widest part is 3 mm. and of the left 4 mm. The external ocular movements are full. There are frequent coarse rotatory nystagmoid jerks accompanied by blinking movements of the eyelids. The pupils are regular and equal, and
W.D. (11)

react equally to light and accommodation.

The eyelashes of the upper lids are normal, but there are no eyelashes on the lower lids. He does not carry his head tilted backwards, and there is no undue wrinkling of the forehead. The width of the orbits is $3\frac{1}{2}$ cm. The hair and eyebrows are mouse-coloured.

This patient is very intelligent. He states that when he was four years old an operation was performed on his eyelids at the Glasgow Eye Infirmary,
but that this led to little improvement in the condition. He reads very little because he feels that "it strains his eyesight"

X-Ray photographs of skull of W.D. (11)

The views show the typical familial deformity of the sphenoid bone, with shallow middle fossa, and rather low roofs of the orbits. The anterior clinoid processes are large, and the sella turcica relatively shallow.

There is calcification of the pineal gland.

There is an extreme degree of ptosis of both upper lids, more than three-quarters of the pupils being covered. The length of the palpebral fissure is 15 mm. in each eye, and the width 2 mm. Downward movement of both eyes is very limited. Otherwise the external ocular movements are full. There is no nystagmus. The pupils are regular and equal and react to light and accommodation. The cornea is unhealthy, showing a superficial infective keratitis with superficial vascularisation. The boy has chronic conjunctivitis and mucopotas is present in the conjunctival sac. The lens is healthy. The fundi and discs are normal as far as can be ascertained through the unhealthy cornea.

Uncorrected vision with the lids lifted is 6/60 in the right eye and 5/60 in the left. There is a high grade of mixed astigmatism with the rule in both eyes. The eyelids show evidence of previous operation. Eyelashes are normal on the upper lid, but are absent from the lower lid. He does not hold his head noticeably tilted back, and there is no marked wrinkling of the forehead. The width of the orbits is 3½ cm. The hair and eyebrows are fair.

This boy has perhaps the most marked degree of
W.S. (12)

ptosis of all the D------ family. He can find his way about remarkably well, but is greatly incapacitated. A Canthoplasty was performed some years ago, with no improvement in the condition. In fact, the parents maintain that it has made the condition worse.

The boy is of average intelligence.
X-Ray photographs of skull of W.S. (12)

1. Increase in width of bases of sphenoidal fissures
2. Increase in length of lesser wings in their antero-posterior dimension.
3. Shallowness of middle fossa due to unusual shape of lesser wings of sphenoid.
4. The sella turcica is relatively small in comparison with the size of the head.
All of the members of the D——— family examined had ptosis and blepharophimosis i.e. the dimensions of the palpebral apertures were diminished in both the longitudinal and the vertical diameters. In them, as in many of the cases described in the literature as having "congenital ptosis", the word "ptosis" is in reality a misnomer. "Ptosis" means a drooping or literally falling (πτωσις) of the eyelid and in many of these cases it is the narrowness of the palpebral apertures that renders lifting of the lids impossible. This point was stressed by Cockayne and also by Dimitry who suggested that the term "congenital blepharophimosis" be used instead of "congenital ptosis". Verwey thought the condition in the cases he described differed from congenital ptosis and that it corresponded in many respects to cryptophthalmos. He thought, however, that the relatively slight diminution of eye movements in his cases showed that they were not cryptophthalmos.

I have, however, retained the term ptosis, using it in the wider and more useful, although etymologically incorrect, sense of narrowness of the palpebral aperture in its vertical diameter, reserving the term blepharophimosis for cases in which there is
diminution in the horizontal length of the palpebral aperture. It must be remembered that this latter condition may occur without narrowness of the palpebral aperture in the vertical dimension, as has been shown by the work of John, Van der Hoeve and Halbertsma.

John has reported a family in several of whose members throughout three generations there existed shortness of the palpebral fissures although the width of the fissures was within normal limits.

*John's Pedigree.*

In those marked × besides uncomplicated blepharophimosis there was also heterochromia simplex.

In case 1 the length of the palpebral fissures was 22 mm.s. The distance between the inner corners of the eyelids was 44 mm.s i.e. greater than normal, while the distance between the outer corners of the eyelids was 68 mm.s.

In case 2 the length of the palpebral fissures was 22·5 mm.s, the distance between the inner
corners of the lids 39 mm. s, i.e. greater than normal, and that between the outer corners of the lids 84 mm. s.

The width of the fissures was normal, but was greater in proportion to the length. There was no epicanthus, and the outer corners of the lids were in the same horizontal plane as the inner. There was no consanguinity in the family.

John mentions that this condition had previously been described by Van der Hoeve in two deaf and dumb twin sisters. In Van der Hoeve's cases there was abnormal broadening of the root of the nose, and marked shortening of the distance between the outer corners of the lids with increase of the distance between the inner corners. The upper punctum lacrimale was much nearer the nose than the lower one, which was normal in position. The caruncle lay on the inner surface of the lower lid, and was much bigger than normal.

John also mentions that Halbertsma described a family afflicted with this condition.

The D----- family clearly belong to the "ptosis-with-blepharophimosis" group, i.e. they are unable to raise their upper eyelids because the lids are tacked down at the edges, not because the lids are paralysed.
Moreover, except in the case of W.S. (12) in whom there was some limitation of downward movement, the external ocular movements are full. This group seldom show paresis of the external ocular muscles. If they do, it is nearly always the upward movement of the eyeball that is affected and this is almost certainly, as Wilbrand and Saenger suggest, due to lack of use, because, as the patient can see nothing on looking upwards, he gradually loses the habit, and eventually the ability, to turn his eyes upwards.

Loeb has described a case of congenital bilateral ptosis associated with almost complete paralysis of the superior rectus muscle. The patient was a girl aged 11 who consulted him because of drooping of the eyelids, which had been present since birth. She carried her head thrown back, and the normal wrinkling of the cutaneous surface of the eyelids was absent. There was no upward movement of either lid, even on extreme effort accompanied by contraction of the frontalis muscle. There was no upward movement of the eyes, but when the lids were lifted up it was seen that the eyes could be raised voluntarily slightly above the horizontal plane. All other ocular movements were normal.

Congenital ptosis similar to that found in
the D—— family, was observed by Briggs occurring in a large family of six generations. This family lived in a lonely part of Western N. Carolina and Eastern Tennessee, and it was possible for Briggs to make a very thorough study of the condition.

The genealogy consisted of 128 persons composing 23 families. 64 persons were affected with ptosis and 64 were normal. Of the affected 33 were males, 30 females, and one of unknown sex. Of the 20\(^{+}\) affected parents 16 were male and four female. The 16 fathers had 100 children, 47 of whom had ptosis, and 53 were normal. The four mothers had 21 children, of whom 14 had ptosis and 7 were normal.

Inheritance in every case was direct, no generation in the lineage of the affected being skipped, with the exception of one case whose mother, a member of the genealogy, and father were both said to have been normal. In no case did affected parents fail to transmit ptosis to one or more of their offspring.

The following are descriptions of some of Briggs' cases:

W.H. had bilateral ptosis. His forehead was greatly wrinkled, his eyebrows high, and in order to fix objects above the horizontal he had to tilt his head.

+ For certain reasons Briggs leaves three families out of this calculation.
back. When the thumbs were pressed firmly against the eyebrows, so as to prevent the associated action of the frontalis muscles, the patient was totally unable to raise the lids. The length of the palpebral fissure was 24 mm. The upper lid was unwrinkled. The pupillary reflexes and external ocular movements were normal.

This patient's son had a very similar condition, but was more able to raise his lids by means of the frontalis muscles, without which he was unable to lift the lids at all.

C.H. had, in addition to ptosis, some impairment of the action of the superior recti. His palpebral fissures measured 20 mm.

Mrs. McK. had the least amount of ptosis observed in any member of Briggs' pedigree. She was able to raise the upper lid slightly even when the frontalis muscle was put out of action by pressure over the supraorbital region. Her palpebral fissures measured 23 mm.

A male infant aged 6 weeks, affected with ptosis, had very narrow palpebral fissures to the extent that blepharophimosis existed.

A girl aged 15 had in addition to ptosis congenital contraction of subcutaneous tissue over the malar bone,
which caused the lids to be drawn towards the outer canthus.

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* One sex unknown.

True congenital ptosis, due to congenital paralysis of the levator palpebrae superioris also occurs, and cases have been reported, among others by Dimitry, Stuckey, and Caudron.

Dimitry records from New Orleans 21 cases of hereditary ptosis which he observed in five known generations. Of 38 living children, 21 showed the condition.
The condition was never transmitted by a normal individual. Male and female transmitted in equal numbers, and were equally affected. There was no paralysis of any of the external ocular muscles. The bony orbits were properly developed and normal in size. The eyelids covered from one-third to one-half of the cornea. Slight elevation of the lids occurred when the frontalis muscles were contracted.

H. P. Stuckey in an article entitled "The Slit-eyed People" has described a family which he encountered in the mountains of north-eastern Georgia, in which congenital ptosis occurred in four generations.

The first affected individual in this family
who could be traced was the great-grandfather. He
transmitted the condition to his son, who married and
had nine children, of whom six were affected and three
normal. One of these six children, a man, married a
normal woman, and they had four children, three boys
affected and one girl normal.

The eyesight of these people was perfect. The
affected members of the family earned their living in
competition with normal individuals. They had difficulty
in getting clear vision unless they tilted their heads
back or turned them to one side.

Stuckey's Pedigree.

A case of congenital ptosis of the paretic
type has been described by Caudron, who carried out the
interesting experiment of instilling cocaine into the
conjunctival sac.

The patient, a boy of 14, had bilateral
congenital ptosis, complete on the left side, not quite complete on the right. There was no trace of activity of the levator palpebrae superioris muscle. The child could only open his palpebral aperture by relaxing the orbicularis oculi and contracting the frontalis to the utmost of his ability. Even then the opening of the palpebral fissure was only 4 mms. on the left side and 6 mm.s on the right. The child could only see by throwing back his head and depressing his eyeballs. He had convergent strabismus in the right eye, and upward movement of both eyes was very defective.

Instillation of cocaine into the conjunctival sac produced widening of the palpebral fissure, and moreover enabled the boy to open his eyelids feebly but with little effort. The effect was transitory.

Caudron attributes this result to the action of cocaine on the smooth muscle fibres of the upper eyelid.

Congenital ptosis of the paretic type, however, is most frequently seen as part and parcel of congenital external ophthalmoplegia which will be more fully discussed later. (Chapter V, page 154).

In all the patients described here the condition was present at birth. Hereditary, non-congenital ptosis may also occur, but this too will be discussed in a later i.e. the D------ family
chapter. Some showed coarse nystagmus. None of them had epicanthus. They had all the peculiarity of having no eyelashes on the lower eyelids. Abnormal distribution of the eyelashes was also noted in Pergola's cases, in which the lashes of the upper lids were irregularly planted, some of them outside the normal line of insertion.

Examination of the photographs of the patients shows the wrinkling of the forehead and the high arched eyebrows noted by Wilbrand and Saenger in several of their cases. This is of course caused by excessive contraction of the frontalis muscle in an attempt to open the eyes further. The D-—— family always walk about with their head well thrown back, in order to bring the pupil opposite the slit. This hyperextension of the head was also observed by Wilbrand and Saenger and was referred to by Hirschberg as the "astronomer appearance". The D-——'s all show varying degrees of diminution of visual acuity - possibly due to congenital amblyopia, as in the cases of Bach and Lanhofer.

Mental retardation does not appear to be by any means a constant accompaniment of congenital ptosis. Several members of the D-—— family are extremely

--- Cited by Wilbrand and Saenger.
intelligent. One, on the other hand (25) is intellectually very feeble, like the mentally retarded woman mentioned by Wilbrand and Saenger.

None of my cases suffered from convulsions, which have rarely been seen in conjunction with this condition, although one of Lamhofer's cases was also an epileptic.

The family belonging to Kiew, described by Ginzburg in 1913, is exceptional in that one of the affected members had unilateral congenital ptosis, while in the others the condition was bilateral.

His first patient was a female aged 16, who had been unable since birth to raise her right upper eyelid. The skin of this ptosed lid was smooth and had no folds. When the patient tried to open that eye the skin of the forehead on the right side went into broad folds, but the eyelid scarcely moved. The fundi were normal, and the left eye normal. There was no movement of the ptosed lid associated with chewing.

This patient's brother, who had had rickets, had congenital bilateral ptosis. His right palpebral aperture was wider than the left. When asked to open

# A special chapter is devoted to unilateral congenital ptosis (p.102.)
his eyes the skin of the right half of the forehead became very wrinkled.

The younger sister had very slight congenital bilateral ptosis, equal in both eyes. She could raise her eyelids a little by contraction of the frontalis muscle.

The external ocular movements were normal in all three cases. The parents of these children were normal, as also were two other sisters, the eldest and youngest members of the family respectively. There was no history of this condition having been present in any of the immediate ancestors. There had been no consanguinity in the family.

X-Ray examination of the skull was carried out in eight members of the D family. Varying degrees of abnormality of the sphenoid bone were observed in all of the eight. These bony abnormalities of the skull in association with congenital ptosis have not, to my knowledge, been reported previously. They may be summarised as follows:—
The anomaly lies chiefly in the wings of the sphenoid, the lesser wing being longer antero-posteriorly than normal and the greater wing having lost part of the curvature of its upper surface, so that the middle fossa
of the skull in lateral view appears to be more shallow than in the normal skull, the base of the superior orbital fissure is wide, owing to the unusual shape of the lesser wing of the sphenoid. In some of the cases this is so marked as to make the roofs of the orbits unusually low and unusually near the level of the cribriform plate of the ethmoid, as in the cases of M.D. (8) and I.S. (6). In I.S. (6), M.D. (8), W.D. (11) and W.S. (12) there is anomaly of the sella turcica, which consists chiefly in shallowness of the sella itself, and undue thickness of the anterior and posterior clinoid processes.

That congenital bilateral ptosis (of the blepharophimotic type) may occur sporadically and unassociated with any bony anomaly in the skull is shown by the following case:--


This patient has bilateral ptosis of a moderate degree, about half of each pupil being covered. Each palpebral fissure measures 23 mm.s in length and 7 mm.s in width at the widest part. There is no epicanthus. When asked to open his eyes as widely as he can he wrinkles his forehead and violently contracts the frontalis muscle, but this produced no further elevation of the lids.
The external ocular movements are full, and there is no nystagmus. The pupils are regular and equal, and react to light and accommodation. The fundi are normal. The colour of the irides is grey-blue. The eyelashes are normal. The width of the orbits is 4 cm. The forehead is greatly wrinkled, the eyebrows arched, and the patient goes about with his head tilted slightly backwards. The hair is black.

The condition has been present since birth, and the patient thinks that it is now less marked than it was when he was a boy. He enjoys excellent health, and apart from an attack of tonsillitis, has never been ill. General examination with particular attention to the nervous system reveals no abnormality.
He knows of no relation who had this condition. His parents are dead, but their eyes, he says, were normal. He is the youngest but one of a family consisting of four boys and five girls, all of whom have normal eyes. The patient is not married and says that he has no children.

The Blood Wassermann Reaction is negative.

No abnormality was detected by X-Ray examination of the skull.

Little, unfortunately, can be said as regards the aetiology of this condition. Several post-mortems have been carried out in cases of congenital external ophthalmoplegia, some in cases of congenital ptosis of the paralytic type, but few in the blepharophimotic type. Various workers have found a thin or poorly developed levator palpebrae superioris, but this cannot be regarded as necessarily primary. In the phimotic type of case, where the patient is unable to raise his eyelid, the levator palpebrae muscle cannot develop properly. Syphilis is only very rarely the cause and then only, of course, in the paretic type of case.
A case of syphilis in which both upper eyelids became paralysed was reported from Baltimore by Mackenzie in 1864. His patient was a female congenital syphilitic in whom complete paralysis of the upper lids had come on at the age of three, accompanied by occipital headache and convulsive movements occurring during sleep. These symptoms and the bilateral ptosis disappeared completely after a month. It was then noticed that the child had developed an internal strabismus of the right eye, which persisted. What treatment, if any, had been carried out during the month Mackenzie had not been able to find out. He considered that there had possibly been a gummatous deposit in the 3rd...nerve.

In Mackenzie's case the ptosis was not congenital, and it does not, of course, really come into the category at present under discussion.

In the case of the D---- family one can only come to the unsatisfying conclusion that there had been a developmental error leading to the abnormal configuration of the sphenoid and of the tissues forming the external palpebral aperture.

One of the most naive of the writings on this subject is that of Münden of Hamburg. He has described a case of congenital bilateral ptosis which was believed
to have been inherited from a woman who acquired the condition.

The patient was a male with congenital ptosis of both upper lids, so severe that only a slit was left at the outer part of each palpebral fissure. In order to see the patient had to keep his pupil opposite this slit, and move his head about, although actually, when the lids were passively lifted, the external ocular movements were seen to be full. The pupils were large, but reacted to light.

The story was that this man's grandmother, while pregnant with his mother, was endeavouring to cross the Elbe in a boat, when the boat capsized. In her fright she shut her eyes, and when brought to land was found to have bilateral ptosis, which she retained for the rest of her life. When her child was born it had bilateral ptosis. This daughter had two normal children, and a boy with bilateral ptosis, who was Minden's patient.
SUMMARY

1. A family in which congenital ptosis with blepharophimosis occurred throughout three generations is described.

2. In eight members of this family, X-Ray examinations of the skull have revealed a developmental abnormality of the os sphenoidale not previously reported.

3. It is concluded that the lesion in these cases is not true congenital "ptosis" of a paretic type, but rather a developmental anomaly involving the os sphenoidale and the tissues round the eye, causing the palpebral apertures to be diminished in size both vertically and horizontally.

4. It is pointed out that congenital "paretic" ptosis also occurs, and cases are quoted from the literature.

5. A personal case of congenital phimotic ptosis, unaccompanied by abnormality of the os sphenoidale is described.
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"Ein Fall von erworbener und vererbter Ptosis palpebrarum."
"Ptosi Congenita Associata A Malformazioni Oculari"


Wilbrand (H) & Saenger (A)
CHAPTER II.

CONGENITAL AND HEREDITARY PTOSIS

WITH EPICANTHIUS AND MALDEVELOPMENT

OF THE OS SPHENOIDALE.
The following is an account of the Sl----- family in which congenital ptosis and epicanthus has occurred in three generations. Four members of the family have been examined, including radiological examination of the skull in three of them. In all of these three gross developmental abnormality of the sphenoid bone, of the same nature as that observed in the D----- family, was found to be present.

Genealogical tree of the Sl----- family

- = affected members.

Numbers correspond to those opposite names of patients in the text.


Bilateral ptosis of a fairly severe degree has been present since birth, about two-thirds of each
pupil being covered. Each palpebral fissure measures only 21 mm.s. in length, and 3 mm.s. in width at the widest part. There is true epicanthus, a fold of true skin passing over the inner canthus, uniting the lower to the upper lid. Upward movement of both eyes is very defective. The external ocular movements in other directions are normal and there is no nystagmus. The pupils are equal and regular and react to light and accommodation. The colour of the irides is gray. The eyelashes are normal. There is some opacity over the upper part of the conjunctivae.

Vision uncorrected is 6/18 (J.4) (R.E.) and 6/36 J.8. (L.E.). Corrected vision for distance is R.E. 1.00 D.Sph. -1.50 D.Cyl. axis 15°, 6/18 and L.E. 3.00 D.Sph. -3.50 D.Cyl. axis 155°, 6/36, and for near vision, adding 1.25 D.Sph. is J.1 (R.E.) and J.2. (L.E.)
The eyebrows are very high and arched, and the forehead shows marked wrinkling. The patient carries his head erect. By contracting his frontalis muscle he can raise his eyelids by about 2 mm. This is prevented by firm pressure against the upper part of the orbits. This patient is not much incapacitated by his infirmity. He is a keen boxer, and is regarded as a particularly formidable opponent, because his antagonist is unable to foretell the direction in which his blows are intended. His general health is excellent and he has suffered only from infectious diseases of childhood.

X-ray photographs of skull of J. Sl---(1)

1. Broadness of base of superior orbital fissures.
2. Abnormal configuration of lesser wings of the sphenoid, which are longer than normal anteroposteriorly, and convex upwards instead of downwards.
3. The roofs of the orbits are at almost the same level as the cribriform plate of the ethmoid.
4. The ant. clinoid processes are very poorly developed.

(for X-Ray photographs, see next page.)
X-ray photographs of the skull of J. S1---- (1)
H. Sl---- (Senior) (2). Male. Aged 43.

Occupation - Cinema Operator.

H. Sl.---- (2)

Bilateral ptosis has been present since birth.
About one-third of each pupil is covered. The length of
each palpebral fissure is only 21 mm.s. Width of the left palpebral fissure at its widest part is 5 mm.s, that of the right 4 mm.s. There is true bilateral epicantus, a fold of skin passing from the lower to the upper lid over the inner canthus. The pupils are regular and equal and react to light and accommodation. He has 6/12 vision in each eye with normal fundi. There is a slight vascularisation of the conjunctiva at the upper margin of each cornea. The refractive error present is $2\text{D}$ of hypermetropic astigmatism in the right eye and $2\frac{1}{2}\text{D}$ in the left.

**X-ray photographs of the skull of H. Sl---**
*(Senior. 2)*

1. Wideness of bases of the superior orbital fissures
2. Lesser wings of the sphenoid longer than normal antero-posteriorly.
3. Poor development of the anterior clinoid processes.
4. There is an abnormal crescentic ridge at the outer end of the sphenoidal ridge.

(for X-Ray. photographs, see next page)
X-ray photographs of skull of H. Sl---Senior. (2)
J.S. (3). Female. Aged 19 months.

This baby's eyes are said to have remained unopened for four days after birth. There is now a marked degree of ptosis, only about half of each pupil being exposed. The left palpebral aperture is slightly wider than the right, and the length of each is 15 mm. The external ocular movements are full, and the pupils normal. A wide fold of skin passes outwards from the root of the nose over each inner canthus. The eyelashes are normal. The eyebrows are very arched.

The patient is otherwise normal, and apart from bronchitis has never been ill. Instruments were used at birth.


He has bilateral ptosis, about half of each pupil being covered. There is bilateral epicanthus, a vertical fold of skin passing up from the lower to the upper lid at the inner canthus. The length of the palpebral fissure is 21 mm.s in each eye, and the width is 5 mm.s. By making an effort he can raise the lids by
about 2 mm.s., but this cannot be done if the observer's thumbs are pressed firmly against the upper part of the orbits, thus preventing the frontalis muscle from acting. The external ocular movements are full, and the pupils and pupillary reactions normal. There is no nystagmus, and the fundi are normal. He carries his head tilted backwards and there is marked wrinkling of the forehead. The hair and eyebrows are mouse-coloured. The eyelashes are normal.

This patient is very intelligent. He
has had no illnesses except an inguinal hernia. He complains that when visiting a cinema, he develops a crick in his neck from having to bend his head backwards in order to see the screen below his drooping eyelids.

X-ray photographs of the skull of H. Sl--- (Junior) (4)

1. The base of the superior orbital fissure is wider than normal.
2. The antero-posterior length of the lesser wings of the sphenoid is longer than normal.
3. The middle fossa of the skull is very shallow, indicating abnormal configuration of the greater wing of the sphenoid.
4. The spheno-maxillary fissure is very wide, and the pterygoid processes very short.
5. The sella turcica is grossly deformed and its outline cannot be defined.

(for X-Ray photographs, see next page)
X-ray photographs of H. Sl---- (Junior)

(4)
Epicanthus was first described by von Ammon in 1831 when he gave the following general definition of it:
A semilunar fold passing from the tissue between the edge of the lid and the eyebrow over the inner corner of the eye, which it covers like a sickle, and ending in the region of the caruncle, generally below it.

Von Ammon has distinguished the following three types of epicanthus:

1. Supraciliary epicanthus which begins at the eyebrows and extends downwards towards the lacus lacrimalis or to the side of the nose, merging imperceptibly with the skin.

2. Epicanthus palpebralis, the best known type, begins in the skin of the upper lid above the tarsal fold, and stretches in a sickle-like fold down to the lower edge of the orbit where it ends in the skin.

3. Epicanthus tarsalis which begins from the tarsal fold of the upper lid, and either passes along the lower edge of the orbit, or ends under the inner corner of the eye in the skin covering the lower eyelid or loses itself in a small ridge of skin close to the inner corner of the eye.

Epicanthus may occur alone in otherwise normal people, or may be accompanied by other abnormalities.

Schmidgall has differentiated six types of
epicanthus, according to the abnormalities with which he had found it to be associated.

1. Epicanthus with Entropion.
2. Epicanthus with Disease of the Tear Ducts.
3. Epicanthus with Ptosis.
4. Epicanthus with Strabismus.
5. Epicanthus with Muscular Paresis.

Brückner adds a type of epicanthus associated with epiblepharon.

Braun mentions that Komoto described the type of epicanthus associated with congenital ptosis and shortening of the palpebral fissure as epicanthus inversus.

Kassowitz and Neumann (quoted by Brückner) stressed the fact that in about half of all cases of Mongolian Idiocy epicanthus was present. A patient with epicanthus and partial external ophthalmoplegia reported by Brückner was a Mongolian idiot.

Epicanthus may be uni-lateral or bilateral, and Braun has reported one case of unilateral right-sided congenital ptosis and epicanthus without hereditary history.

Steinheim, in reporting the family tree of a Bavarian family, many of whose members had this condition, mentions a local belief that Mongolian blood
was present in these families "as a memento of Attila's invasion".

A case of congenital bilateral incomplete ptosis with epicantius and blepharophimosis occurring in an idiot girl was reported from Wurzberg by Bach in 1893. The width of the right palpebral fissure was 3.5 mm.s, that of the left 2.5 mm.s. The right could be opened to 6 mm.s and the left to 5 mm.s. The length of each palpebral fissure was 15 mm.s and the distance between the internal commissures was 33 mm.s. The bridge of the nose was low, and the puncta lacrimalia were poorly developed. There was defective action of the lateral recti, and of the right rectus superior. The pupils reacted normally. Staphylomata were observed on ophthalmoscopic examination.

There was no history of a similar condition having been present in this girl's family.

Cockayne has described a case of epicantius associated with bilateral ptosis, occurring in a member of a family in which the same condition was transmitted as a simple Mendelian dominant through five generations.

Cockayne's patient was a boy of eight. He had an increased broadening of the skin-fold extending from above the upper eyelid to the skin of the lower eyelid,
over the inner canthus on both sides. The palpebral fissures were short, and the boy had bilateral ptosis. Slight hypermetropia was present, otherwise the eyes were normal. The boy was mentally defective.

\textbf{Cockayne's Pedigree.}

\begin{center}
\includegraphics[width=0.8\textwidth]{pedigree.png}
\end{center}

* Died young.

Hirschberg described in 1895 at a meeting of the Berlin Society of Psychiatry and Neurology a Russian who had been born with bilateral ptosis and total external ophthalmoplegia. This man's mother and his child had the same condition. His grandmother was said to have been similarly afflicted as a result of trauma, but this could not be proved.

He had high eyebrows, and his forehead was
corrugated by contraction of the frontalis muscle. The only surviving external ocular movement in either eye, was depression with some rotation towards the temporal side, due to the surviving superior oblique muscle. There was slight divergent strabismus, but no diplopia. The pupils reacted normally to accommodation. Astigmatism was present. No abnormality was detected elsewhere in the nervous system.

This patient's son had epicanthus and ptosis on both sides. The length of the palpebral fissures was 20 mm. and the width 8 mm. The distance between the inner corners of the eyes was 25 mm. The eyes could not be elevated, but adduction, abduction, and depression were satisfactory.

A family in which ptosis and epicanthus could be traced through three generations was described by Hüttemann at Strasbourg in 1911.

See next page for Pedigree.
Cases 2 and 3 were the illegitimate sons of Case 1, who had ptosis and epicanthus. Their mother had normal eyes, and there was no history of any abnormality having been present in any other member of the father's family. It was said that when this man's mother was pregnant with him she was frightened by seeing a dead person with half-opened eyes. This picture had haunted her mind, and to this she attributed the abnormality in her child's eyes.

Cases 2 and 3 both married wives whose eyes were normal. Case 2 had seven children, of whom three died at an early age. Two of these had had ptosis and epicanthus. Of the survivors three were affected, and one normal.
Case 3 had four children, of whom the youngest, an affected female, died in infancy. Of the three others, two were affected and one normal.

Case 2 showed wrinkling of the forehead, and held his head tilted backwards. The length of his palpebral fissures was 23 mm.s and the width 4-5 mm.s His fundi were normal. There was a suggestion of a Darwinian cartilage (Darwinscher knorpel) on the right.

In case 4, who was aged 14, the forehead was wrinkled, and the eyelids showed absence of wrinkling. The length of the palpebral fissures was 23 mm.s and the width 3-4 mm.s. The eyeballs could not be moved upwards spontaneously, but they could be elevated if the lids were passively lifted. Ophthalmoscopically a small conus was seen on both sides inferiorly.

In case 5, aged 6, the forehead was not markedly wrinkled, but there was no wrinkling of the upper lids. The length of the palpebral fissures was 20 mm.s and the width 3 mm.s It was doubtful whether upward movement of the eyeballs could take place. The fundi were normal.

Case 6, aged 2, carried his head tilted backwards, and his forehead was wrinkled. So severe was the degree of ptosis in the left eye that that eye appeared
almost closed. The length of the palpebral fissures was 20 mm.s. The width of the right palpebral fissure was $2\frac{1}{2}$ mm.s, that of the left 1$\frac{3}{4}$ mm.s. The external ocular movements were full, and the fundi normal.

Case 3 showed absence of the normal wrinkling of the upper lids, wrinkling of the forehead, and held his head tilted back. The left upper lid showed the scar of an old ptosis operation. The length of the palpebral fissures was 25 mm.s and the width 5 mm.s. Upward movement of the eyeballs was doubtful.

In case 7, aged 9, the forehead was wrinkled, and the upper lids unwrinkled. The length of the palpebral fissures was 20 mm.s and the width 5 mm.s. There was apparent deficiency of upward movement of the eyeballs, but this could not be proved. On ophthalmoscopic examination, a posterior staphyloma was seen in both fundi. The auricles were misformed.

In case 8, aged 6, the forehead was wrinkled and the eyelids unwrinkled. The left eye showed upward deviation and slight convergence. The right palpebral fissure measured 22 mm.s in length, the left only 20 mm.s. The width of the right palpebral fissure was 5 mm.s, that of the left 7 mm.s.

All of these patients were intelligent. They
all had attached lobes of ears, high gums and a Darwinian cartilage on one or both sides. They all showed the strange phenomenon that on internal rotation of either eye the eyelid of that side became almost closed. When the right eye looked to the left, and when the left eye looked to the right, the corresponding eyelid fell.

Pergola has recently described the following three cases of congenital ptosis, in two of which the condition was hereditary, and in two of which epicanthus was present.

**Margherita S. Aged 39.**

Ptosis of both upper lids had been present since birth. The width of the palpebral aperture in both eyes measured 2 - 3 mm. Upward movement of the eyeballs was limited, and even when she lifted the eyelids with the finger the patient was unable to rotate the eyeballs upwards. In order to see she tended to throw her head backwards. There was no real epicanthus, but the internal commissure was displaced laterally in such a way that the distance between the internal angles was much greater than normal. The eyelashes of the upper lids were implanted along the margin in an irregular way. About the middle of the margin there were some hairs of
the same length as the usual eyelashes, but more
delicate. These proceeded upwards and outwards in
an oblique direction to reach the eyebrows. No other
members of this patient's family showed the abnormality.

Antonietta M. Aged 16.

This patient had a Mongoloid appearance. She
had epicanthus, in consequence of which the distance
between the internal commissures in relation to the roots
of the nose appeared much greater than normal. The
length of the palpebral apertures in both eyes was only
20 mm.'s. The ocular movements, including upward move-
ment, were normal. The free margins of the upper lids
were almost rectilinear. The position of the eyelashes
was irregular, a few of them being implanted outside the
normal line of insertion, and, as in the last case, they
were distributed in an oblique line outwards and upwards
towards the eyebrows. The line of the eyebrows was
higher and more curved than in a normal person, due to a
contraction of the frontalis muscle. In order to see
she tilted her head backwards.


This boy had had since birth the same ocular
abnormalities as his sister, i.e. ptosis of both upper
lids with epicanthus, increase of the distance between
the internal commissures, shortening of the palpebral openings, which measured only 19 mm. in length, almost rectilinear margins of the upper lids, with irregular arrangement of the eyelashes, and eyebrows much more arched than normal. He had a Mongoloid appearance.

Sattler in 1897 reported from Cincinnati a family in which he observed congenital ptosis with epicanthus in two generations, a father and his four children.

The father had complete loss of power of both the levator palpebrae muscles, there was little compensatory action of the frontalis muscle, and in order to see he had to incline his head backward. The superior recti and the other extrinsic ocular muscles acted poorly.

This man had been afraid to marry in case he should transmit the condition to his children. He finally decided that marriage would be justifiable on the grounds that he himself had not inherited the condition, none of his ancestors, immediate or remote, having had it. He married a woman who was perfectly healthy and who had a normal family history, but none the less their four sons all had congenital ptosis and epicanthus.
In all four boys the ptosis was pronounced. There was no independent action of the levator palpebrae muscles. There was good compensatory action of the frontalis muscles, by means of which the palpebral fissure could be opened to some extent. The inner portion of the lids became inverted, so that the eye-lashes caused much irritation and epiphora. In all of these boys Sattler noticed that the cranial conformation was brachy-cephalic, the facial diameters broad, and the auricles inserted low down.

The father had a high degree of compound myopic astigmatism, but in the sons the refraction was hypermetropic with varying degrees of astigmatism.

McIlroy has recorded a family in which congenital ptosis associated with epicanthus occurred in four generations, twelve members being affected and seven unaffected.

The following is a description of the cases personally examined by McIlroy -

K.F. Aged 13. Female. She had well marked bilateral ptosis with slight epicanthus. The pupils were more than half covered by the eyelids and the head was thrown back. There was absence or poor development of the levator palpebrae superioris muscle, the eyes being kept open by
the action of the frontalis muscle. On examination
of the profile of this patient, McIlroy noted a distinct
concavity over the supraorbital region due to maldevelop-
ment of the bone. The normal furrows of the forehead
were absent. The child had nystagmus and divergent
strabismus. Myopia of a considerable degree was present,
the refraction being as follows:-

Right eye: -5 D. Sph.  -1.5 Cyl. ax. 30°
Left eye : -6 D. Sph.  -1.5 Cyl. ax. 150°
The best vision obtained was 6/36.

The girl's mother is said to have been
mentally defective. The girl's progress in school had
been unsatisfactory, but she had not at any time been
regarded as suitable for a class for mental defectives.
She was emotional, rather nervous, but was said never
to have fits.

McIlroy also saw this child's aunt who had
ptosis, which was of a slighter degree, this being
attributed to an operation which had been performed when
she was nine. The aunt was of normal intellect, and
stated that she had recently had fits "as a result of
severe shock".
**McIlroy's Pedigree.**

(Further history unavailable)

* - Personally examined by McIlroy.

The first generation could not be fully traced: descent was through an affected male. Other members of this or previous generations are believed to have been affected.

The noteworthy feature of McIlroy's cases is that the condition was transmitted only through males. Several females were affected, but did not transmit further. McIlroy, however, does not inform us whether these affected females were married, and if so, whether they had any offspring at all. Males and females appear to have been equally attacked in McIlroy's
family. This transmission of ptosis with epicanthus by the male only was also a feature of Usher's cases. McIlroy points out that the colour of the irides in both cases examined by him was brown. The girl's hair was reddish brown, and the aunt's hair was said to have been dark. This, as McIlroy mentions, bears out Usher's findings that brown pigmentation is the rule in these cases.

It is of interest too, that in McIlroy's cases there was an element of mental deficiency.

There was no history of consanguinity.

Pockley has described a case of congenital bilateral ptosis associated with epicanthus. The ptosis was so pronounced that the child had to raise his eyelids with his fingers in order to be able to see. The cornea of each eye was partly covered by the epicanthal fold.

Pockley treated the condition by a two stage operation, first remedying the epicanthus, and some weeks later dealing with the ptosis by Hess's operation, i.e. subcutaneous fixation of the lid to the frontalis muscle.

An instance in which congenital ptosis with epicanthus occurred in three members of a family of six
children has recently been described by Rodin of San Francisco. Three of the children were males and three females. Two males and one female showed this defect.

**Rodin's Family of Congenital Ptosis with Epicanthus.**

![Family Chart]

There was no history of any similar condition in parents or relatives.

The eldest of the family, a male, was examined by Rodin. He had marked ptosis with epicanthus. In order to see he had to tilt his head backwards. The eyelids could be raised slightly by contraction of the frontalis muscle. The eyelids were irregular in shape, with abnormal distribution of the lashes. The external ocular movements were normal.

Ross has described a family in which the condition of congenital ptosis with epicanthus was transmitted through four generations.

The boy seen by Ross had marked congenital ptosis with epicanthus. In order to look at the ceiling the child had to flex his neck on his back to the utmost. There was no defect in visual acuity. There was no history of intermarriage.
Wilbrand and Saenger describe a child normally developed in other respects and without hereditary taint, who had congenital ptosis and epicanthus. It was said to have had closed eyes at birth, the lids only gradually opening after three weeks. There was a fold of skin arising on the two sides of the bridge of the nose and passing over the inner angle of the eye, partly covering it.

Steinheim saw a father and two children who had epicanthus and ptosis in varying degrees. The father's great-grandfather had had the condition, and had 5 children, of whom three, one female and two males, were affected. All three were married, and of these one female and one male went to America and of their descendants
nothing is known. The remaining affected male became the father of two girls, of whom one was normal, the other affected. The latter became the mother of five children, of whom two boys and one girl were affected, the other two being normal. The three affected children were all married. Of the girl's offspring two were normal and one affected. One of the boys had four children, of whom again two were free and two affected. Of these one was a girl who had one affected child. The other affected boy had six children, of whom three were normal and three affected. Two of these latter were seen by Steinheim.

For purposes of convenience the following tree has been constructed from Steinheim's description of his cases.

(See the following page)
Verwey of Rotterdam has described two cases of congenital ptosis associated with epicanthus.

The first patient was a girl of six. She had a vertical skin fold covering the caruncle and puncta lacrimalia. The palpebral apertures were both 17 mm.s long and 4 mm.s in breadth (Fuchs gives 24 mms. and 10 mms. as the normal dimensions for a child of eight). The eyebrows were high and arched. There was no
depression of the bridge of the nose. The upper lid could only be elevated by contraction of the frontalis muscle, and if the skin was fixed to the orbital margin no movement of the upper lid could take place. Upward movement of the eyes was somewhat defective, and a slight convergent strabismus was present. The fundus was normal.

This patient's sister was a deaf mute. Her mother had had three abortions, followed by healthy children, all older than the patient.

Verwey's second patient was a boy of four. His appearance was similar to that of the girl. An interesting point in this patient was that he kept his mouth perpetually open, by this means keeping the palpebral fissures a little wider. His palpebral apertures were 18 mm. in length, the right 4 mm. high and the left 3 mm. Upward movement of the eyes was limited, and movement of the lids was dependent on the action of the frontalis muscle.

Braun has described several cases of congenital ptosis associated with epicanthus, which he observed at Prague.

One patient he saw was a boy of seven, whose mother, maternal grandfather, and two maternal aunts, all had this condition.
In this boy the length of the palpebral fissures was 23 mm.s as compared with 26 mm.s which Braun gives as the normal measurement for a child of that age. The distance from the bridge of the nose to the inner corner of the eyelid was also greater than normal, namely, 24 mm.s as compared with 21 mm.s. The medial end of the upper lid was bordered by a bow-like fold of skin which was directly continuous with the lower lid. The upper lid could be raised only by contraction of the frontalis muscle. Upward movement of the eyeballs was very limited. This Braun attributed to that movement not having been used; he found that it improved with exercise. Numerous vascularised blemishes were present in this patient's cornea.

Braun also reports a family in which three
sisters and their father had congenital bilateral ptosis and epicanthus, the mother and four other siblings being normal. He has observed also two girls, each of whom got it from her father, the mothers and siblings being in each case normal, and there being no history of the condition having been present in any other relatives.

Braun further observed four patients who had congenital ptosis and epicanthus in whom the condition was not hereditary. One of these was a twelve year old girl. The length of her palpebral fissures was 23 mm.s and the width 4 mm.s which contraction of the frontalis muscle could increase to $7\frac{1}{2}$ mm.s There was no action of the levator palpebrae muscle. The inner corner of each eyelid was 22 mm.s from the bridge of the nose, i.e. greater than normal. In addition to these four Braun reports three other cases in which the condition was not hereditary. In all of them and in the six familial cases mentioned above, complete congenital ptosis was present, the bridge of the nose was broad, and the distance from the inner corner of the eyelid to the middle of the nose was greater than normal. There was slight vertical expansion of the upper lid, upward movement of the eye-balls was defective, and the palpebral fissures slanted from within downwards.
Braun considers that this condition has no special racial incidence. His patients included Germans and Czechs in equal numbers, and one Jewess.

Bruckner has described two interesting cases of epicanthus with slight ptosis which he observed at Wurzburg. One of the cases was a Mongolian Idiot.

The first case was a fifteen year old boy who had Epicanthus Tarsalis, with paresis of both lateral and of both superior recti, and of the right inferior rectus. He had myopia, and a persistent pupillary membrane in both eyes. He had diplopia on looking straight forward or to the left. There was epicanthus of a slight degree on both sides. There was only slight ptosis and no wrinkling of the forehead, but in both upper lids the tarsal fold was little in evidence. The gums were high. The boy was intelligent and a good scholar. The following is the pedigree of his family

```
  ♂     ♀    ♀    ♀    ♂     ♀    ♀     ♂
   |            |            |
  ♂     ♂     ♂     ♂     ♂     ♂     ♂
```

The patient was the eldest but one of nine siblings, of whom three, in addition to himself, were known to have the same condition, and two were known not to have it. The patient's mother had normal eyes, but
two of her brothers were known to have the condition, two were known to be normal, while the condition of the fifth was not known.

Bruckner's second patient was a girl of \( \frac{5}{2} \)\text{ft}, who was a Mongolian Idiot. She had bilateral epicanthus with limitation of lateral movement of both eyes, and limitation of vertical movements. The bridge of the nose was flat, and its skin extended out into a reduplicated fold surrounding the inner corner of the lid from above. There was a slight degree of ptosis, but voluntary raising of the eyelid was not greatly impaired, and the tarsal fold was present in the entire breadth of both eyelids. She had a convergent strabismus and nystagmus. Her face was broad and flat. The terminal phalanges of the little fingers were blunted and the fingers and toes could be bent back over 90°.

The possibility of congenital ptosis and epicanthus being related to a bony abnormality was envisaged by several of the first writers on the subject. Von Ammon himself, in 1831, found that true epicanthus was sometimes associated with malformation of the skull, and considered it to be due to arrested development of all the parts of the face within the region of the orbit i.e. of the nasal and frontal bones.
Manz, in 1876, said "It is not improbable that for all these anomalies, epicanthus and muscular paresis, there is a common cause in the construction and development of the parts of the facial skeleton concerned, but this has not yet been proved anatomically. Lowness of the root of the nose may perhaps be the basis of a temporary or permanent excess of skin at this point. Shortness of the distance between the orbits would be an additional factor tending in this direction, but this has not yet been determined by measurement."

"All writers," says Brückner (in 1906) "will agree that in, at any rate, a large number of cases of epicanthus, the cause is to be found in disturbance of bony and sometimes of cartilaginous development. To what extent the disturbance in the innervation or formation of the paretic muscles can be attributed to the same cause remains undecided."

Sattler noticed that the cranial conformation in his cases was brachy-cephalic, the facial diameters broad, and the auricles inserted low down. In one of Mallroy's patients there was a distinct concavity over the supraorbital region due to maldevelopment of the bone.

Braun gives the following as having been Von Graefe's views on the subject. He found that epicanthus was generally associated with ptosis and defective upward
movement of the eyeball. For this reason he believed that the chief feature of this malformation was not the fold of skin, but an insufficiency of certain branches of the oculo-motor nerve. This explanation can be dismissed because epicanthus without ptosis and ptosis without epicanthus exist. One cannot imagine how epicanthus could arise as the result of purely motor nervous involvement.

In the cases reported by Bach, Hirschberg and Brückner some external ophthalmoplegia was present in addition to epicanthus and blepharophimosis, but it is a noteworthy fact that paresis of the external ocular muscles rarely occurs in association with epicanthus. This is additional evidence that these are not cases of true ptosis due to paresis of the levator palpebrae superioris muscles, or to a lesion of the 3rd. or 6th. nerves, but rather cases in which there has been faulty development of the tissues immediately surrounding the eye.

It will be seen, moreover, that the abnormalities of the cranium as shown by X-rays of the skulls of the Si--- family are precisely similar to those found in the D---- family, viz. broadness of the base of the superior orbital fissures, shallowness of the

As regards the defective upward movement in Verwey's cases, see p. 41 or p. 198.
middle fossa of the skull, and increase in the antero-posterior length of the lesser wings of the sphenoid. This suggests that epicanthus in itself is not particularly related to the unusual conformation of the skull, but rather that epicanthus and blepharophimosis are merely varying forms of expression of the same congenital anomaly. Whether this anomaly is itself secondary to the disturbance of the skull conformation or whether both are dependent on an unknown error of development cannot yet be said. The second hypothesis appears to be the more probable.
SUMMARY

1. Four members of a family in which congenital ptosis associated with epicanthus occurred in three generations are described.

2. X-Rays of the skull of three of the four members reveal anomalies of conformation of the os sphenoidale similar to those found in the D---- family described in the previous chapter.

3. The literature of congenital ptosis associated with epicanthus is reviewed; the abnormalities of the skull mentioned above have not previously been observed.

   External ophthalmoplegia is rarely present.

4. It is concluded that these cases are due to a developmental error involving the os sphenoidale and the tissues forming the palpebral aperture, and that the "ptosis" is not a paretic one.
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Wilbrand (H) & Saenger (A)

CHAPTER III

UNILATERAL

CONGENITAL PTOSIS.
Congenital ptosis may be unilateral, although this is much more rare than the bilateral form.

I have had the opportunity of observing the Sc--- family in whom congenital ptosis of the left upper eyelid has occurred throughout three generations. It has been unaccompanied by blepharophimosis, external ophthalmoplegia or epicanthus, and X-ray examination of the skull has shown no anomaly.

**Genealogical tree of the Sc--- family**

![Genealogical tree diagram]

- = Affected members.
The numbers refer to the patients mentioned in the text.
John Se---(1) aged 32. Occupation: Mercantile Marine Seaman.

He has very slight drooping of the left upper lid, the width of the left palpebral aperture being about 7 mm.s, that of the right about 9 mm.s. Each palpebral aperture measures 27 mm.s in length. Both upper eyelids show the normal degree of wrinkling. The forehead is not abnormally wrinkled, but the left eyebrow is higher than the right. The head is not tilted back. There is no epicanthus. The distance from the middle of the bridge of the nose to the inner canthus is 24 mm.s. That the left levator palpebrae superioris muscle is able to function a little independently is shown by the fact that the left upper lid can be elevated even when the thumb is pressed into the orbit to prevent the action of the frontalis muscle. The external ocular movements are normal and there is no nystagmus. The pupils are regular and equal and react to light and accommodation. Examination of the nervous system reveals no abnormality. The patient is left-handed. Apart from mumps in childhood he has never been ill. Ptosis was present at birth. It began to improve when he reached the age of 20, and the improvement has continued since then. The condition has never caused him any
inconvenience. X-Rays of skull are normal.

This patient, who is illegitimate, has no brothers or sisters. His father had normal eyes. His mother had ptosis of the left upper lid. Shortly after he was born she was removed to a mental home where she died. Her three half-brothers, sons of her father, are all stated by J.S. to have had congenital ptosis of the left upper lid.

M.S. (2) female. Aged 6.

This child has ptosis of the left eyelid which was noticed by the mother at birth. About one-third of the left pupil is covered. The length of both palpebral fissures is 22 mm. The width of the right palpebral fissure at its widest part is 9 mm, that of the left only 6 mm. By strongly contracting the frontalis muscle the child can raise the left eyelid by about 1 mm. If the observer's thumb is pressed firmly against the upper part of the left orbit this eyelid cannot be raised at all. There is no epicanthus. The external ocular movements are full. There is slight lateral nystagmus. The pupils are regular and equal and react to light and accommodation. The fundi are normal. The eyelashes are normal. The left eyebrow is more highly arched than the right.
She apparently does not find it necessary to tilt her head back in order to see, and the forehead is not unduly wrinkled. General examination with particular attention to the nervous system reveals nothing abnormal, apart from very large tonsils. Skull normal.

The mother thinks that the condition is now more evident than it was at birth. She has never noticed any other abnormality in the child, whose only illnesses have been measles and broncho-pneumonia at the age of 18 months.
The child's father and his mother have the same condition. She has no sisters, and her only brother aged 4, has normal eyes.

The dimensions of the palpebral aperture is in these cases normal, and there is no epicanthus. X-Ray examination has shown no abnormality of the skull.
The condition is not that seen in the D—— or S1—— families. The ptosis is a paralytic one, probably due to aplasia of the levator palpebrae superioris muscle. It is rare for congenital paralysis of the levator to occur independently of congenital paralysis of the external ocular muscles, but some such cases have been observed.

Unilateral congenital ptosis occurring in four generations of a family was reported by Alessi (quoted by Hüttemann) and Rodin has recently reported from San Francisco the pedigree of four generations of hereditary congenital unilateral ptosis. The remarkable feature of this family was that the eye affected with ptosis alternated with each generation.

In the first generation one female had ptosis affecting the right eye. In the second generation one male and one female had ptosis of the left eyelid in each case. In the third generation there was one male with ptosis of the right eyelid, and in the fourth generation one female with ptosis of the left eyelid.
Rodin's Pedigree of Unilateral Congenital Ptosis.

In case 6 the margin of the right upper lid reached to about the centre of the cornea. The eyelid could be elevated slightly by contraction of the frontalis muscle. The left eye was normal.

In this man's daughter, case 7, the ptosis was more pronounced than in the father, and affected the left upper lid. Contraction of the frontalis muscle had no effect on the ptosis.

In neither of these cases was there any paralysis of the superior rectus muscle.
The next case is that of a patient who suffered from right-sided congenital ptosis unaccompanied by epicanthus or blepharophimosis.

J. Sp----- Female. Aged 18.

Occupation - Typist.

She has a marked degree of ptosis of the right upper eyelid. By contraction of the frontalis muscle she can raise this lid by about 4 mm. If the observer's thumb be pressed against the upper part of the orbit, the eyelid cannot be raised at all. The external ocular movements are full in all directions. She had also a right concomitant convergent strabismus. This was treated by tenotomy of the internal rectus four months
prior to examination. The pupils are regular and equal and react to light and accommodation.

Vision in the right eye with -5.00 D. Sph. -1.50 D. Cyl. axis 160° = 2/60; and in the left eye with -2.00 D. Sph. -2.00 D. Cyl. axis 20° = 6/6 J.1.

The patient states that the ptosis of the right upper lid has been present since birth. Delivery was instrumental. Her periods are regular and give her no trouble. She has had no illnesses. There is no history of ptosis in any of her relations or ancestors.

It was found that cocaine dilated both pupils equally, and caused equal elevation of both upper lids. The Blood Wassermann Reaction is negative. Birth injury appears to be the most probable aetiology in this case.

The following two cases are children who have had since birth a mild degree of ptosis on one side only. The condition is unaccompanied by epicanthus, blepharophimosis, ophthalmoplegia or abnormality of the skull on X-Ray examination. In both of them delivery was difficult and instrumental, and birth injury would thus appear to be the most probable aetiology, the forceps having possibly injured part of the third nerve supply to the levator
palpebrae superioris muscle. Other cases of unilateral ptosis due to birth injury have been reported by Michel and by Berger. In Berger's case there was associated paralysis of the rectus superior muscle.

† Quoted by Pergola.
§ Quoted by Wilbrand and Saenger.

G.H. Male.
Aged 9 months
Slight drooping of the left eye has been present since birth.
The mother states that the eye was shut for the first three days of the baby's life and then gradually...
opened to reach the state of partial ptosis now present, about one-third of the left pupil being covered. External ocular movements are normal. There is no nystagmus. The pupils are equal and regular and react to light and accommodation. There is no epicanthus, and no blepharophimosis. The right eye is normal. No relative or ancestor has had any similar condition. There has been no consanguinity in the family. The patient was born after a difficult labour, in which instruments were used. The mother says that when he was born there was a mark, believed to be due to an obstetrical instrument, in the left frontal region of the skull. No such mark is now visible. The child is otherwise perfectly healthy.

J. Sh---. Male. Aged 11.

This boy has ptosis of the right upper eyelid which was present at birth, and is not of a marked degree. About one-third of the right pupil is covered. The length of each palpebral fissure is 26 mm.s. The left palpebral fissure at its widest part measures 10 mm.s while the right measures only 6 mm.s. The right eyelid shows complete absence of wrinkling. When the boy is asked to open his
right eye as widely as possible he strongly wrinkles his forehead, and succeeds in lifting the lid by about 1 mm. He cannot do this if firm pressure is exerted against the upper part of the right orbit. The external ocular movements are full, and there is no nystagmus. The pupils are regular and equal, react to light both directly and consensually, and to accommodation.

Examination of the fundus oculi shows no abnormality. The discs are very highly coloured. The eyelashes are normal. There is no epicanthus. The right side of the face looks smaller and less well-developed than the left, but no weakness of this side of the face can be detected. Apart from this observation, routine general and neurological examination reveals nothing abnormal. The boy has always been healthy. His tonsils were removed some
years ago. The father thinks that the condition is now no better and no worse than it was when the child was born. Instruments were used at the delivery, and a red mark above the right eye was noticed at birth.

There is no history of ptosis in any other members of the family. The parents are in good health and have normal eyes, and the patient is an only child.

This boy's Blood Wassermann Reaction was found to be negative, as also was that of his father.

The next case is that of a baby who has had ptosis of the right upper eyelid since birth, unaccompanied by any other abnormality. The aetiology in this case must remain a matter of conjecture. Unfortunately it was not possible to obtain X-ray photographs of the skull. The most likely explanation is a congenital developmental anomaly of the levator palpebrae superioris muscle which may be under-developed or abnormally inserted.
This baby has had ptosis of the right upper lid since birth. The length of the palpebral aperture is 21 mm. in both eyes. There is no epicanthus. The external ocular movements are full. The pupils are regular and equal, and react to light and accommodation. He is a very healthy child and has never been ill. General routine examination revealed no abnormality.

The mother says that the condition was the same at birth as it is now, no better and no worse. The degree of ptosis never varies. There is no history of ptosis in any other members of the family. He has one elder sister, who
is normal. The mother has a very slight external strabismus in the left eye. Delivery was not instrumental.

A similar case of unilateral congenital ptosis was described by Putnam-Jacobi in 1895. The patient was a year-old baby, who, for a few weeks after birth could not open the left eye at all. It then began to raise the lid a little and when seen by Putnam-Jacobi the eye was about half open. The eyelid was frequently twitching, and the eyeball often showed nystagmic oscillations. The external ocular movements were full; and the pupils were equal and reacted to light. The vision appeared to be normal. The orbicularis oculi muscle seemed ill-developed, a fact which suggested to Putnam-Jacobi that the paresis of the levator was due to imperfect development of the muscle rather than to paralysis of its nerve. The baby was perfectly healthy and well developed both mentally and physically. Putnam-Jacobi does not mention the sex of the child.

Putnam-Jacobi refers to the case of Armaignac.* This was a child of two in whom the lid of the left eye was thin, contracted weakly, and

* Rev. Clinique d'Oculistique VI, 1886.
could not be raised properly. It was associated with complete paralysis of the rectus superior, atrophy of the orbicularis, and facial asymmetry, the left external orbital region being flattened.

Caudron describes a case of unilateral congenital ptosis in a boy of 12. The upper eyelid of the affected side was smooth and unwrinkled, and even by contracting his frontalis muscle, the child was unable to widen the palpebral aperture by more than 4 or 5 mm.

Other somatic anomalies may accompany the unilateral, as the bilateral, form of congenital ptosis. Thus, in a case of left-sided congenital ptosis, without hereditary history, seen by Wilbrand and Saenger, there was under-development of the terminal phalanges of all five fingers of the left hand, on which the nails were absent, as they also were on the little toes of both feet. There was in addition slight hypospadias.

Unilateral congenital ptosis occurring as part of the picture of unilateral congenital external ophthalmoplegia is considered under that heading.

To the peculiar type of unilateral congenital ptosis associated with Jaw-Blinking (known as the Phenomenon of Marcus Gunn) the following chapter is devoted.
1. The following are described:—

(a) A family in which unilateral congenital ptosis of the paralytic type occurred in three generations.

(b) Three cases of unilateral congenital ptosis probably due to birth injury.

(c) A case of unilateral congenital ptosis probably due to congenital dysplasia of the levator palpebrae superioris muscle.

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CHAPTER IV.

UNILATERAL CONGENITAL PTOSIS

WITH JAW-BLINKING OR THE

PHENOMENON OF MARCUS GUNN.
An unusual and especially interesting form of unilateral congenital ptosis is that in which involuntary movements of the affected eyelid occur accompanying some other voluntary movement. The best known form of this type of disturbance is that known as Jaw-blinking, or the Phenomenon of Marcus Gunn, in which the ptosed eyelid which the patient is unable to move voluntarily, or which he can move voluntarily to a slight degree only, moves up and down automatically in unison with movements of the lower jaw.

The first case of this type was described by Marcus Gunn in 1883. The patient was a girl of 15. She had congenital ptosis of the left eyelid, the upper quarter of the pupil being covered. When she looked upwards the left eyelid did not rise quite so high as the right, and when the eyes were gently shut the left upper lid did not close quite so perfectly as the right. She could not raise her left eyelid while looking straight forward without contracting the frontalis muscle. All external ocular movements were normal in both eyes. The left pupil was smaller than the right, but contracted well to light and accommodation.

When the patient moved her jaw to the right, i.e. contracted her left lateral pterygoid muscle, the left upper lid rose at once, and stayed there as long as the jaw was kept deviated to the right. If the eyes were kept closed when the jaw was moved to the right, the levator palpebrae superioris on the left side still contracted. Elevation
of the left eyelid also took place when the lower jaw was projected forwards. If the mouth was opened gently the lid did not move. The patient's chief complaint was upward jerking of the lid when she was eating and speaking, especially in pronouncing words containing "s" or "x". It had been noticed when she was sucking the breast at the age of five weeks that the left upper lid moved upwards.

The cilio-spinal reflex was present on both sides, and no unilateral sweating or blushing had ever been observed.

She had slight asymmetry of the two sides of her face, which was more evident when she smiled, the dimples which then appeared being different in situation and depth.

Otherwise this patient was normal, although slightly anaemic.

Her paternal grandfather and all his family were reputed to have had asymmetrical eyelids. Marcus Gunn saw her father and his sister. They had very slight fulness of the right upper lid, but no true ptosis, and no abnormal associated movements of the eyelids.

The pupillary inequality present in Gunn's case, to which I shall refer more fully later, is not a usual feature of the syndrome.

The following is an account of a personally observed case :-
When this child was 12 weeks old it was noticed that the left eye was not so wide open as the right, and that the left eyelid moved up and down when she took her feeds. When she was sucking the breast, the eye opened wider with each mouthful. When later she was given a dummy teat she opened the eye so wide when sucking it that the mother thought it "was going to pop out of her head".

On examination she has ptosis of the left eyelid
Photographs showing elevation of left upper eyelid accompanying opening of the mouth.
as seen in the photographs. The lid moves upwards when the child opens her mouth, the degree of lid elevation being proportional to the width to which the mouth is opened. Movement of the lower jaw to the right also causes concomitant raising of the left upper lid. Movement of the lower jaw to the left has no effect on the lid. The right eyelid is normal. The external ocular movements are full and the pupils and pupillary reflexes normal. No abnormality is detected on examination of the nervous or other systems. The child is in all other respects perfectly healthy and her eyesight is normal. Wassermann Reaction is negative. No similar condition has occurred in the family. The parents are both in good health, and have had no serious illnesses. Patient is an only child. The mother had a miscarriage one year before her birth. She was born at full time and delivery was instrumental.

This patient was examined again four years later, when she was aged 6. No change was found in the condition, the upward flick of the eyelid accompanying movement of the lower jaw being if anything more noticeable. She was in good health, attending school, and making excellent progress with schoolwork.

In the great majority of the published cases, as in the case reported here, the drooping eyelid is elevated both on depression of the lower jaw and when it is moved
towards the side opposite to that of the ptosis.

Villard's patient, a man of 26 with left-sided congenital ptosis, the left palpebral aperture being 2 mm. narrower than the right, showed elevation of the left upper lid on lowering of the jaw, chewing, or movement of the jaw to the right. The height to which the ptosed lid rose was proportional to the extent to which the jaw was lowered, extreme lowering causing it to rise higher than the normal lid. Forward movement of the jaw caused the left lid to become slightly elevated. No movement of the eyelid occurred on swallowing, laughing, whistling or compression of the lips, and it was not marked on speaking. Elevation was maximal when the jaw was moved downwards and to the right. Passive depression of the jaw produced no lid elevation until the mouth was opened to its maximum dimension. The lid remained up only as long as the mouth remained open. The left pupil was slightly larger than the right, but reacted normally. Electrical examination of the muscles of mastication revealed no abnormality.

In the case observed by Rodrigo and Perez Llorca (quoted by Marquez) there was right sided ptosis, and when the patient opened his mouth the right upper lid rose almost to the normal position. When the patient at the same time moved his jaw to the opposite side, the elevation of the upper lid was greater. If, while the mouth was open, the jaw was moved towards the side of the ptosis, it
made very little difference, although the elevation of the right eyelid was then somewhat greater than when the mouth was shut, as then the eyelid did not change position at all.

Fischer, in 1899, reported from Moorfields Hospital a case showing this phenomenon. In his patient the ptosis affected the right upper lid, and was apparently more pronounced than in Marcus Gunn's case, as the right eye was almost closed when the patient looked at a distant object. Unlike Gunn's case, too, the eye closed equally on the affected as on the unaffected side. The lid was raised if the mouth was opened or the jaw moved over to the left. Fischer says that "reversing this movement caused the lid to droop". There was no facial asymmetry.

Fischer says that the condition had existed since the child was a few months old, but does not tell us whether it was actually congenital. The grandfather of the patient was said to have been similarly affected, also on the right side.

Thomson and Souter in 1912 reported a case of left congenital ptosis in which the lid became elevated when the mouth was opened, and when the jaw was moved to the right or protruded forwards.

The following are some of the main points from Thomson and Souter's very complete description of this case.

The patient was a female who had a slight degree of ptosis of the left upper lid, the left palpebral aperture
measuring 7 mm. at its widest part when the eyes were
directed forwards, the right measuring 9 mm. There was
no epicanthus. The external ocular movements were full,
and there were some coarse lateral nystagmoid jerks on
lateral movement. The pupils were equal and reacted to
light and convergence. The fundi were normal. The cilio-
spinal reflex was present on both sides, the left pupil
dilating less vigorously than the right. There was no
unilateral sweating or flushing.

When the mouth was opened rapidly there was
marked elevation of the left upper lid. When the mouth
was opened slowly the upward jerk of the lid was faint and
inconstant. When the jaw was moved to the right the
upward movement of the left eyelid was so marked that
2-3 mm. of sclera became visible; the width of the
palpebral fissure becoming 12-13 mm. This was even more
evident if, while the jaw was deviated to the right the
patient looked down or converged. The lid did not rise
in proportion to the degree of opening of the mouth, but
did rise in proportion to the degree of lateral deviation
of the jaw to the right. When the jaw was pushed over to
the left there was no movement of either upper lid. When
the jaw was protruded there was an upward movement of the
lid which was at first very pronounced, but soon became
exhausted. When the patient yawned there was a slight,
slow, upward movement of the left eyelid. When she chewed an apple the lid jerked up and down slightly when she looked forwards, and markedly when she moved her jaw sideways, especially if she looked down. There was no movement of the upper lid during laughing, singing, speaking, clenching the teeth or sucking. The lids shut to an equal extent on both sides both when they were gently and when they were firmly closed.

No other abnormality of significance was discovered on examination of the nervous and other systems.

Instillation of 5% cocaine hydrochloride caused dilatation of both pupils, to 8 mm. in the left eye, and to 7 mm. in the right.

Delivery had been normal and without instruments. The ptosis was first noticed a week or two after birth, and the winking of the lid when the child was 2½. It may, however, have been present before then.

The child suffered from time to time from enlarged cervical glands on both sides. No history of a similar condition in relatives was elicited. The child's mother had congenital dislocation of both hip joints.

Two brothers suffering from left-sided congenital ptosis and jaw-blinking were reported from Utrecht by Blok in 1891. In both elevation of the affected eyelid took place when the jaw was moved downwards or to the right. The movement of the lid was greatest when the eyes were directed
downwards, and absent on looking upwards.

L. G. Parsons has described the case of a male child afflicted with left-sided congenital ptosis covering the upper one-third of the pupil. When he opened his mouth or deviated his jaw to the right, the left eyelid was raised to a higher level than the right. The eyelid was continually raised and lowered when the child was feeding at the breast. Every time he attempted to look upwards he opened his mouth thus allowing the lid to be raised. The left pupil was slightly larger than the right, but the pupillary reflexes were normal.

Harman's patient was a female who had partial left-sided congenital ptosis in whom the drooping lid was raised when she chewed or diverted her jaw to the right.

Sym has reported the following case:—
A. J., aged 30, female, had partial ptosis on the left side. The external ocular movements were full. When she chewed or sang the left upper eyelid sprang upwards, disclosing perhaps as much as 3 mm. s of white sclera above the corneoscleral junction. There was distinct but partial elevation of the eyelid on use of the left lateral pterygoid muscle, i.e. moving the jaw to the right. When the patient suddenly opened her mouth by actively depressing the lower jaw, the lid sprang up. There was no corrugation of the forehead, nor any unusual movement of the eye itself, nor was there
any corresponding movement of the right upper lid. There was no evidence pointing to affection of the sympathetic on that side. The condition had been present all the patient's life.

In some cases the involuntary elevation of the ptosed eyelid occurs not only in association with movement of the jaw, but also with certain movements of the eyeballs themselves.

Coppez has reported a case of this type. His patient was a female with right-sided congenital ptosis. She could raise her right upper lid voluntarily only by contracting her frontalis muscle, and even so, the lid was very easily fatigued. It became raised involuntarily, however, when she depressed her jaw, when she moved it to the left or forwards, and also when she directed her eyes downwards or to the left. There was also an association between the ptosed and the normal lid. When the patient blinked the drooping lid sometimes became lowered and at other times executed the same movements as the normal one although lagging behind. Pick has observed a similar case—that of a female with very marked right-sided congenital ptosis in whom elevation of the abnormal eyelid occurred when the mouth was opened and shut, when the jaw was moved to the left and when the eyes were turned to the right. If the mouth was opened while the eyes were turned to the right,
the right upper lid rose still further. When the patient looked to the left the right eyelid became almost entirely closed.

Cases have been described in which there was an association between movement of the eyeballs and of the upper lids without a permanent ptosis being present in the resting state, and unconnected with movement of the lower jaw. Such are the two cases reported by Phillips in 1887. The patients were two brothers in both of whom the condition had been present since birth. On direction of the gaze laterally, laterally and upwards, or laterally and downwards, to either side, the upper eyelid of the side opposite to that to which the patient was looking simultaneously drooped, producing an almost complete ptosis. The forehead became transversely wrinkled on the side towards which the patient directed his eyes.

The parents and two other siblings were normal and there was no evidence of hereditary syphilis.

Phillips' explanation of the phenomenon was that in addition to the usual relaxation of the medial rectus on looking laterally, there also occurred relaxation of the levator palpebrae superioris, and that this, probably because of the unusually close commissural connection between the two 3rd. nuclei, occurred bilaterally. In order to overcome the bilateral ptosis which would thus be produced, the patient would contract the frontalis muscle on the side.  

*Hüttemann's cases of ptosis with epicanthus showed downward movement of the eyelid on internal rotation of the ipsilateral eye (p. 79).*
towards which he was looking, thus overcoming the ptosis on that side, the other eye remaining covered by its ptosed upper lid.

Browning in 1890 described a case showing exactly the opposite phenomenon to that present in Phillips' cases. His patient was a cheesemonger of 46. When he moved his eyes to either side the upper lid of the side to which he was looking dropped, whilst that of the other side became elevated, the elevation being more marked in the case of the left upper lid. In convergence both the upper lids were raised above the horizontal, the left more than the right. When he looked down, the upper lids followed the eyeballs as far as the horizontal plane, but as the globe rotated further downwards the lids remained stationary, while on extreme downward deviation the lids, especially the left, became somewhat raised. Coarse nystagmus was present on all extreme movements.

The right pupil reacted sluggishly to light, but reacted to accommodation. The left pupil did not react to light, but reacted to accommodation. The fundi were normal. 2% cocaine was put into the right eye, but made no difference to the condition.

It would appear that contraction of either internal rectus was associated with contraction of the levator palpebrae superioris of the same side, and relaxation of the levator of the opposite side.
Browning makes no attempt to explain the absence of the pupillary light reflex. He does not describe his patient's general health, nor does he mention whether the condition was congenital or hereditary.

Sometimes cases of the phenomenon of Marcus Gunn are associated with congenital paresis of one or more of the external ocular muscles. This was the case in patients seen by Uhthoff (rectus internus) Cantonnet (obliquus inferior), Hubbell (rectus superior and rectus internus), Vossius (all the external ocular muscles on both sides), Sinclair, Helfreich, Proskauer, Goldzieher and Marin Amat (rectus superior).

Uhthoff's case was a female of 19. When she was six years old her mother noticed that her left eyelid drooped, and that it moved jerkily up and down when she opened her mouth wide and also when she chewed. When she kept her mouth open the lid remained up. Lateral movements of the lower jaw to the right produced elevation of the left eyelid, but movement of the jaw to the left had no effect. She had insufficiency of the left medial rectus. When she attempted to look at a near object the left eye could not converge but instead turned outwards. The pupils were equal and the pupillary reactions normal. The sight was good.

There was some contracture in the region of the left facial nerve, the left naso-labial fold being deepened,
and the mouth drawn to the left. There seemed to be some association between the left eye and the facial muscles. For example, if the lips were pouted to whistle, the left palpebral aperture became smaller. The patient had noticed that her left ear was often redder and warmer than the right. Electrical stimulation of the sympathetic had no effect on the ptosis.

There was a glandular swelling on the left side of the neck, in front of and behind the left sternomastoid muscle. Since childhood she had often had left-sided headaches. There was some degree of scoliosis.

Unthoff had no doubt that in his case the condition was congenital.

Proskauer's patient, a male, had very marked left-sided congenital ptosis with paralysis of the left superior rectus muscle. The palpebral aperture at rest was only 2 - 3 mm. wide, and the eyeball could be moved upwards only a little. By using the frontalis muscle the patient was able to widen the palpebral aperture to 6 mm. As soon, however, as he opened his mouth the left eyelid was raised without any other muscle being thrown into action. The palpebral aperture then reached 10 mm. Occasionally the eyelid remained in this position as long as the mouth remained open, but it generally dropped before the mouth shut. Only on shutting the mouth, however, did it come into its position of rest. The opening and shutting of
the lid was very marked with each movement of mastication. It was less marked on speaking. When the lower jaw was pushed forward or to the right the left eyelid was raised, but no movement of the lid occurred when the jaw was moved to the left. The movement, therefore, followed the action of the digastric or of the int. pterygoid muscle of the left side. As it happened this patient had developed a complete right facial paralysis of rheumatic origin a month prior to consulting Proskauer. It was not, therefore, possible to ascertain whether shutting right the eyelid caused elevation of the left. The pupils were equal. There was hypermetropic astigmatism. There was a small antimacular crescent.

The case described by Vossius is particularly interesting and unusual. His patient had almost complete congenital bilateral external ophthalmoplegia with bilateral ptosis. The left upper eyelid was lower than the right; it could not be raised voluntarily, either by the levator or by the frontalis, but became markedly elevated when the mouth was opened. There was no movement of the right upper lid.

Marin Amat's case was a female with almost complete right congenital ptosis and paralysis of the right rectus superior with downward deviation of the right eye. When she opened her mouth, or moved her jaw to the left, there was excessive elevation of the right upper
eyelid. She had also a congenital capillary angioma on the right cheek.

In another group the eyelid rises concomitantly only with downward movement of the jaw, but not with movement of the jaw to the side. Into this category fall the cases reported by Hubbell, von Laqueur, Bernhardt and Sinclair.

Hubbell's case (in 1893) was reported from Buffalo. The patient was a boy of 7. He had complete right congenital ptosis with a divergent strabismus and downward deviation. When told to open his eyes as widely as he could, the right upper lid was raised a little, by contraction of the frontalis muscle. When he opened his mouth the right upper lid was raised immediately. The more the lower jaw was depressed, the higher was the upper lid lifted. It was possible, however, for the boy to overcome this involuntary opening of the right eye if, while opening his mouth, he at the same time contracted his orbicularis oculi. Upward movement of the eye was impossible, inward movement very slight, but downward and outward movement was full. The pupils were regular and equal, and reacted equally to light and accommodation.

Vision in the right eye was amblyopic, being reduced to finger counting at 4 ft. Vision in the left eye was normal. The refraction in both eyes was emmetropic. The fundi were normal.
In Bernardt's case (a boy of 7) the ptosis was on the left side, and the lid was raised when the patient chewed. The affected lid could also be raised voluntarily to some extent. The pupils were equal and reacted to light and accommodation, but there was slight enophthalmos of the left eye. There were no vaso-motor phenomena or other symptoms to suggest sympathetic involvement.

Von Laqueur of Strasbourg, in 1890, described a case of this type. His patient had partial ptosis of the right upper lid, which was altered in vertical movement of the eyeball, but not in lateral movement. When the patient looked up the ptosis became greater, and when he looked down it became less. When the mouth was opened, especially in chewing, the lid was elevated so far that a strip of sclera 2 mm. wide became visible above the cornea.

Walfreih's case was a female of 17 with right-sided ptosis, who could voluntarily raise the affected lid only by contraction of the frontalis muscle. The lid also rose involuntarily when she opened her mouth or chewed. There was also in this case insufficiency of the right rectus superior.

Occasionally the ptosed lid is raised by lateral movement of the jaw, but not by downward movement.

# quoted by Bernardt:
Marcus Gunn's own case may be included in this category and Sinclair mentions also the case of Schrapinger.

Curious is the case reported by Krauss in which there was an association between movement of the drooping eyelid and (a) movement of the other eyelid (b) movement of the tongue and (c) movement of the lower jaw. The patient was G.N., aged 6 years, a male, who had complete ptosis of the upper lid of the left eye. This was instantly removed by closing the right eye, and returned when the right eye was open. Protrusion of the tongue caused extreme elevation of the left lid, lasting about a minute, when the ptosis gradually returned. When the mouth was open the left lid became partially elevated. Movements of the left eye were somewhat restricted upwards and inwards. Movements of the right eye were normal in all directions. When the patient attempted to look upwards, especially with the right eye open, the lower edge of the left cornea was seen to be about 3 mm. below the lower edge of the right cornea, i.e., the left superior rectus muscle failed to elevate the eye synchronously with the slight elevation of the deficient lid. When the left lid was forcibly elevated the left eye turned downwards and outwards, and upon increased convergence of the right eye the left eye diverged.

The pupils reacted normally to light and
accommodation. There was slight facial asymmetry, the left side of the face being somewhat smaller than the right.

The parents and their three other children were normal. The birth of the child had been uncomplicated. Ptosis of the left eyelid was present at birth, and the left side of the face was thinner than the right.

Krauss treated the condition by administration of strychnine sulphate 1/120 gr. t.i.d., and making the child shut his right eye for a portion of each day.

Three months later the patient was able to keep the left lid elevated voluntarily even with the right eye open. Upon close fixation however, after one or two minutes, the left lid began to droop, the eyeball following it downwards so that the left cornea became lower than the right cornea. After momentarily closing, the left lid elevated again, the eye following it incompletely. Close fixation still resulted in convergence of the right eye and divergence of the left.

Krauss points out that many normal people when protruding the tongue or opening the mouth very wide, increase the palpebral fissure by extreme elevation of the lids. He suggests that this is due to the presence of connecting fibres between the 5th and 3rd nerves, and that in the condition he describes there may be an excessive
development of these fibres, and defective development in the 3rd. nerve fibres to the levator palpebrae superioris. He regards the improvement which followed on exercising the left lid by shutting the right, as proof of the presence of these latter fibres in his case.

Jaw-blinking may occur in the absence of ptosis.

Just in 1838 reported from Zittau a case in which movement of the left upper lid was associated with movement of the jaw, but in which there was no ptosis. The patient was a female of 13, in whom, when she chewed, the left upper eyelid rose and fell concomitantly with the action of the lower jaw. A similar movement of the eyelids occurred when she spoke very loudly. Lateral movement of the jaw caused no movement of the upper lid, which was also influenced by lowering of the line of sight. The eyes were healthy and emmetropic.

Sinclair mentions cases in which contraction of the levator palpebrae superioris was associated with contraction of the internal rectus, and cases of external rectus paresis in which contraction of the orbicularis oculi and retraction of the globe of the eye occurred in association with inward movement of the affected eye. A peculiar case in which the levatores palpebrarum were involved in an associated movement is that of Adamäck (quoted by Blok). Adamäck's case was a nun in whom powerful movements of the jaws in mastication caused not
only elevation of both upper lids, but at the same time bilateral exophthalmos. On cessation of jaw movement the position of the eyes returned slowly to normal. This was believed by Gurwitsch to be due to the fact that some of the veins from the orbits passed through the muscles of mastication and were obstructed when these muscles contracted. Blok points out that this explanation would not account for the upward movement of the lids, although he admits that such an effect might be produced by the exophthalmos widening the palpebral fissures.

Blok considered that the phenomena in Adamück's case might be due to double innervation of both upper lids combined with the abnormality of the veins suggested by Gurwitsch.

Very occasionally there exists an external epicanthus. MacKenzie in his "Traité pratique des maladies des yeux" - Paris, 1856 - quotes two cases, one of Sichel and one of Chevillon.

Jaw-blinking is rarely hereditary or familial. Blok's cases were brothers, and the grandfather of Fischer's case was reported to have had the condition, the same side being affected in both.

The aetiology of the Phenomenon of Marcus Gunn remains obscure, although numerous conjectures have been made.

Considerable interest was aroused by Marcus Gunn's
first case, and a committee consisting of Sir William Gowers, Stephen Mackenzie, William Lang and John Abercrombie was appointed to investigate the problem. They came to the conclusion that in these cases the levator palpebrae superioris muscle must receive a nerve supply both from the nucleus of the 3rd nerve and from the part of the nucleus of the 5th nerve responsible for the Lateral Pterygoid muscle. The ptosis would be due to the Levator Palpebrae receiving a deficient number of nerve-fibres from the oculo-motor nucleus, the remaining fibres arising instead from the motor nucleus of trigeminal. In other words the Levator receives less than its normal innervation, and there is therefore a certain degree of ptosis, but no paralysis of the muscle. It receives some fibres which were "intended for" the lateral pterygoid and when that muscle is put in action the levator is stimulated. The myosis seen in Gunn's case was explained as due to increase in the tonic contraction of the sphincter pupillae, due in turn to the sphincter receiving the Levator's share of impulses from the 3rd nerve nucleus in addition to its own. The committee considered that a sympathetic defect, which might theoretically be held responsible for myosis and ptosis, was unlikely to be present, as the cilio-spinal reflex was active and equal on both sides.

The above theory is perhaps the most plausible one that has been put forward, although of course it has
not been proved, as no post-mortem has been done on such cases.

Hubbell considers that the connection between the nuclei of the 5th and 3rd nerves may vary in different cases, but appears always to be from some part of the 3rd to some part of the descending root of the 5th. He thinks that in his case the origin of the fibres supplying the levator muscle has been transferred from the nucleus of the 3rd nerve to that part of the nucleus of the 5th which sends fibres to the anterior belly of the digastric muscle. The ptosis and deficiency of external ocular movements he would explain as due to imperfect development of the nucleus of the 3rd nerve, in that portion from which arise the fibres supplying the levator palpebrae superioris, the superior rectus and the internal rectus.

Harman would explain the phenomenon as a "revival of an old-time and long-acclimatized associated movement". He points out that in man the pterygoid and orbicularis oculi muscles are homologous with the deep and superficial muscles of the branchial arch of the fish's spiracle, and when the one is contracted the other tends to relax, and the weak levator, taking advantage of the quiescence of its too powerful opponent, lifts the eyelid.

According to Sanchis Banus the phenomenon of Marcus Gunn is due to variation in the tone of antagonistic muscles, while according to Rodrigo and Perez Llorca it...
is a conditioned "tic".

Marquez thinks that it is simply an exaggeration of the normal synergy existing between opening the mouth and closing the eyes. Some patients cannot overcome blepharospasm even by trying to open their eyes, but instead they open their mouth. This Marquez considers to be similar to the Phenomenon of Marcus Gunn.

Marin Amat points out that one frequently meets with patients, who when told to open their eyes, also open their mouth. He concludes that Marcus Gunn's Phenomenon is present physiologically in these people, and that cases showing the phenomenon with no ptosis are due merely to exaggeration of this physiological action. He thinks that the action is brought about by the presence in the superior division of the 3rd nerve of normal people of motor fibres, which pass to the 3rd nerve from the ophthalmic division of the 5th nerve. He considers that these fibres from the 5th nerve reach the 3rd nerve peripherally, and decries the view that the abnormal connections are situated centrally. This latter theory, he considers, does not account for those cases in which Marcus Gunn's Phenomenon exists without ptosis, those in which the phenomenon came on during life, and those in which it tends towards recovery. He believes that there is insufficient anatomical evidence to support the views
of Lütz and Demaria who thought that the condition was due to connections passing between different cortical centres.

Marin Amat suggests that this mechanism of double innervation may be a wise provision of nature to ensure proper functioning of the eyes. The most common cases of Marcus Gunn's Phenomenon are, he points out, those in which there is ptosis, and in them the substitution appears at the same time as the ptosis which it is going to remedy, whether the latter be congenital or acquired. He offers no suggestion as to what may be the cause of the ptosis in such cases.

He thinks that in those cases in which the condition improves, the will has been able to overcome the association between the eye movements and the movements of the jaw.

Marin Amat's theory that Marcus Gunn's Phenomenon is due to peripheral connections between the 5th and 3rd nerves is based largely on observations he made on a curious case which exhibited what he calls the "Inverted Phenomenon of Marcus Gunn."

The patient was a man of 56, who, after a severe wound to his left cheek, developed optic atrophy on the left side, and on the other side deafness, paralysis of the facial muscles with Reaction of Degeneration, lagophthalmos, and suppression of tears. These symptoms Marin
Amat attributed to fracture at the level of the Left Optic Foramen, and of the Right Fallopian Canal. Four months later, to Marin Amat's surprise, the orbicularis oculi and the other facial muscles of the right side, had recovered their function. Marin Amat saw the man about a year after this and then observed that when he chewed he suffered from hypersecretion of tears, and moreover, with each movement of the lower jaw, there took place visible involuntary contractions of the right orbicularis oculi. The eye became closed with every downward movement of the lower jaw, and remained closed until the mouth shut again. The same thing resulted from lateral movements of the jaw during mastication, but not in weeping, laughing, coughing or yawning.

Marin Amat found that the facial muscles contracted when the individual nerves were electrically stimulated, but not when the facial nerve itself was stimulated at its exit from the Stylo-mastoid Foramen. Moreover, electrical stimulation of the Mandibular Nerve produced not only contraction of the muscles supplied by the motor division of the 5th nerve, but also of all the muscles supplied by the facial nerve; the orbicularis opening and shutting the eye synchronously with the lowering and elevation of the lower jaw, tears and saliva being secreted in abundance the while.
Marin Amat thought that this phenomenon was due to peripheral connections between the 5th and 7th nerves by way of the Auriculo-temporal division of the former. That such an anastomosis exists was, he says, proved by Testut. Marin Amat thinks that the Auriculo-temporal in this case took over both the motor and tear-secreting functions of the facial, and that it must contain some motor fibres.

That the facial muscles contracted on stimulation of the branches of the 7th nerve and not on stimulation of the main trunk, was evidence against there being in this case any central connections between the nuclei of the 5th and 7th nerves.

It is clear that unilateral congenital ptosis with Jaw-blinking is an entirely separate condition from the cases of congenital ptosis previously discussed. The ptosis is a true "paralytic" one, is practically always unilateral, and is very rarely associated with epicanthus. It is a developmental anomaly, but one in which the abnormality is of a purely neuro-pathological nature. The most probable explanation is that suggested by Pergola, namely, that such cases are due to a persistence in extra-uterine life of embryological connections between certain nerve centres in the brain stem, e.g. between the 3rd and 5th nerve nuclei, in the cases of the Phenomenon of Marcus Gunn, between the two oculo-motor
nuclei, the 5th nerve nucleus, and the 12th nerve nucleus in the case of Krauss, etc. There is little evidence to support Darin Amat's suggestion that the abnormal nerve connections are peripheral, but until some post-mortem material has been examined this cannot be excluded.

Delivery has been instrumental in some of the cases but too seldom to be regarded as other than coincidental.
SUMMARY

1. A case of Unilateral Congenital Ptosis with Jaw-Blinking (Phenomenon of Marcus Gunn) is described.

2. Cases of this syndrome may be divided into various clinical groups. These groups are described with reference to the literature.

3. The aetiology of the condition is discussed, and it is concluded that it is probably due to abnormal arrangement of nerve fibres in the brain stem.
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CHAPTER V

CONGENITAL. EXTERNAL OPHTHALMOPLEGIA

(INCLUDING CONGENITAL AND HEREDITARY PTOSIS)
There is no doubt that certain cases of congenital and hereditary ptosis are due to a true "falling" of the eyelid. This is the case in the group now to be considered, namely, congenital ptosis associated with congenital external ophthalmoplegia.

Congenital ptosis unaccompanied by epicanthus or blepharophimosis may occur as part of the picture of congenital external ophthalmoplegia, which may be hereditary or sporadic, unilateral or bilateral.

The following is a personal case of unilateral congenital ptosis with external ophthalmoplegia.


This patient was born with the right eyelid drooped, and the mother noticed at birth that the right eye could be moved outwards, but not inwards, and that it was always in the outer corner of the palpebral aperture.

She has had whooping cough, measles, and chickenpox. Her periods are regular, but sometimes give her much pain. Otherwise she has always been perfectly healthy, and has very seldom suffered from headaches. Delivery was not instrumental.

She has two sisters and one brother, all of whom are well, although one of the sisters is said to be neurotic. No siblings have died. The parents are
Joan S.

alive and have always been healthy. The mother has never had a miscarriage or a stillbirth.

On examination she is seen to have ptosis of the right upper eyelid, about half of the right pupil being covered. By contracting the frontalis muscle she is
Joan S.

able to raise the right upper lid to almost the same height as the left. The external ocular movements of the left eye are full. In the right eye there is full lateral rotation, very slight vertical movement, and very slight inward movement to just beyond the midline.
The pupils are equal and regular, react to light, both directly and consensually, and to accommodation. Each palpebral fissure measures 31 mm. in length. The width of the left palpebral fissure is 12 mm., that of the right 7 mm.

On examination of the nervous system the abdominal reflexes are found to be absent. Otherwise there is no abnormality. X-Rays of the skull are normal.

It will be noted that in this patient the palpebral aperture is of normal dimensions, and that the ptosis is a true "falling" of the eyelid.

The case is therefore in an entirely different category from those of the D---- and S1--- families, and the lesion must be either nervous or muscular. On neurological grounds it is most likely in this case to be one involving the 3rd nerve and we must suppose either (a) a congenital aplasia of the 3rd nerve nucleus itself, not including the Edinger-Westphal nucleus, or (b) involvement of the nerve fibres, sparing those for irido-constriction.
Many cases of congenital external ophthalmoplegia have been described, and in a large number of them the condition is hereditary.

Four cases, a mother and three of her children, affected with congenital external ophthalmoplegia and bilateral ptosis, were reported from Heidelberg by Heuck in 1879. The following account of these cases is taken from Lawford's translation of Heuck's paper.

**Case 1**, the mother. She had complete bilateral ptosis, absence of vertical and of outward movement of both eyes, depression of the optic axes, and a moderate amount of convergence. Attempted movement to right or left resulted in both eyes being moved slightly inwards. The fundi were normal.

**Case 2**, had complete bilateral ptosis with loss of vertical movement of both eyeballs, and defective movement of the internal and external recti.

**Case 3** had bilateral ptosis, and slight convergence and depression of the optic axes. Vertical movement of the eyeballs was absent, and when it was attempted slight rotation occurred. Lateral movements could be effected, but were deficient in extent.
Case 4 had complete bilateral ptosis, depression of the optic axes, absence of vertical movement and defective lateral movement.

Case 2 died of an infectious illness, and Heuck was able to examine the contents of the orbits. The levator palpebrae muscle was entirely absent from the left side, and on the right consisted only of an ill-developed strip 2 mms. broad. The superior, inferior, and external recti were inserted too far back, in some cases by as much as 2½ mms. The superior recti were very poorly developed. The superior obliques were inserted anteromedially to the insertion of the superior recti, instead of postero-laterally as is normally the case. The inferior and internal recti were shorter than normal. The nerves of the orbits appeared normal.

Heuck regarded the defective upward movement as due partly to lack of use of the superior recti, and partly to their small size and backward insertion. The defective outward movement would, he thought, result from the backward insertion of the lateral recti and the wrong insertion of the superior obliques.

The shortness of the inferior recti would explain the downward deviation of the optic axes, and poorness of downward deviation.
Rodin and Barkan, in 1935, reported from San Francisco three cases of hereditary congenital ptosis associated with partial external ophthalmoplegia. The patients were a mother and her two daughters.

The mother had previously been seen by Wilmer when she was seven years of age. The following are some of the observations Wilmer then made: There was marked drooping of both lids, and the patient held her head thrown back at an angle of $45^\circ$. Movements of both eyes were very defective. The motion of the right eye was entirely along a horizontal plane; that of the left eye, down and in. When she tried to close the lids, the right eye diverged and the left eye converged. When she tried to raise her eyes, both eyes converged.

Skin and cartilage were removed from the upper lids, and the superior recti muscles were advanced. This was followed by some improvement.

When the woman was seen by Rodin and Barkan 22 years later she was observed to hold her head tilted backwards at an angle of $15^\circ$. The margin of the upper lids reached the upper margin of the pupils. A fine horizontal nystagmus was present. There was no movement of the eyes upwards. The right eye abducted fully, but adduction was very defective. The left eye abducted $20^\circ$
and adducted $15^\circ$. When closing the eye there was no movement of the upper lids. The lower lids came up and there was a horizontal movement of the lids nasally. The right eyeball was fully covered by the lids, while in the left a narrow strip of sclera was left exposed. There was no history of any ptosis in this patient's family.

The first daughter showed moderate drooping of the upper lids, whose borders reached the upper margin of the pupils. There was a fine rotatory nystagmus. There was no upward movement of the eyes. The right eye abducted well, but adduction was poor. In the left eye adduction was normal, but abduction showed some limitation. The left eye showed an external strabismus.

The younger daughter had complete ptosis of the right upper lid, moderate ptosis of the left. There was a fine rotatory nystagmus. The eyes could not be moved upward at all. The right eye showed an external strabismus, but could not be adducted beyond the midline; in the left eye adduction was normal, but abduction could not be carried out beyond the midline.

In both cases the mother had noticed the condition soon after birth. The fundi were normal.

Heard in 1901 reported a family who were
afflicted with hereditary ptosis and external ophthalmoplegia.

Heard's Cases.

The cases marked 4 and 5 were personally examined by Heard.

Case 4 had marked ptosis in both eyes, with inability to move either eye in any direction. He had slight rotatory nystagmus. Capsulo-lenticular cataracts were present in each eye. Heard gives the vision in both eyes as 15/200.

Case 5 had no movement of the eyeballs. Vision in the left eye equalled light perception, and in the right eye 15/100, with divergent squint. Heard mentions that there was ptosis in this eye, but does not say whether or not it was present in the other. There was no movement of the eyeballs, except slight rotatory nystagmus.
The father of these patients, two sisters, and two sons of case 4, had the same condition — ptosis and no movement of the eyeballs.

No history of other abnormalities was obtained.

Guende describes two brothers afflicted with congenital external ophthalmoplegia.


This patient had a moderate degree of ptosis which could be overcome by energetic contraction of the frontalis muscles. The external ocular movements were very limited in both eyes, and there was a slight convergent strabismus in the left eye. He had previously had exophthalmos, which had disappeared as he grew older. The pupillary reactions were normal. He had no binocular vision. His general condition was normal.

His brother, aged 17, a hairdresser to trade, had paralysis of all the extrinsic muscles of both eyes, the intrinsic musculature being normal. He had convergent strabismus which was the same on both sides, and more marked than his brother's. Binocular vision appeared to be present in a region about 1 metre in front of the eyes.

There was a third brother, aged 20, whose condition was similar.
The father was an alcoholic, the mother normal. They had five children, of whom the eldest, a girl of 25, and the youngest, a boy of 9, were normal.

Tirelli has reported a family in which congenital bilateral ptosis was observed in four generations. It was transmitted in direct line, and always appeared in the children of affected individuals. A man and three of his four children had it. In the third generation four out of five had it, and in the fourth generation five out of six. In all, thirteen members of sixteen were affected. All affected members who had issue transmitted the anomaly to their children, male or female, a high percentage being affected. In all the members of this family personally examined by Tirelli, there was marked disturbance of ocular motility, not only of the superior rectus muscle, but also of the other extrinsic ocular muscles. Two of the patients had complete paralysis of the extrinsic muscles. The five members seen by Tirelli all presented astigmatism of various types, always showing a higher refraction in the vertical meridian of the cornea, than in the analogous horizontal meridian. This, as Tirelli mentions, corresponds to the findings of Kretz, who pointed out that astigmatism is very frequent in the eyes of patients with congenital
ptosis, occurring in 53.8% of the cases observed by him.

In Tirelli's cases the abnormality was transmitted as a Mendelian dominant.

Tirelli's Pedigree.

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\( \circ = \text{affected.} \)

The following is a description of Tirelli's cases:

**Maddalene A.** Aged 37. Housewife.

She had congenital bilateral ptosis. The palpebral aperture in both eyes measured 6 mm. Vigorous contraction of the frontalis muscle increased the height of the aperture by a little more than 1 mm. When the patient was told to look into the distance the optic axes were seen to be not parallel but slightly converging and displaced downwards. There was complete paralysis of the extrinsic muscles. The patient could not look upwards, downwards or sideways. The only
thing noticed during the effort made to perform the ordered movements was a slight accentuation of the convergence mentioned above. There was no diplopia. The cornea was normal, the pupils round and regular, the pupillary reflexes normal. The fundus oculi was normal. V.A. -2/10 sc. (R), -5/10 sc. (L.). There was simple hypermetropic astigmatism according to the rule, with axis not deviated. The visual fields were normal. W.R., Meinicke, Kahn were all negative.

The patient had four children, the two first and the youngest showing eye changes very similar to those of the mother.

**Antonio B. Aged 10**:

He had bilateral congenital ptosis. There was considerable contraction of the frontalis muscle. The palpebral aperture measured only 6 mm. the upper two-thirds of the pupil being covered. The optical axis was slightly deviated downwards. The external ocular muscles were all paralysed. There was no diplopia, the cornea was normal, the pupils regular, and the pupillary reflexes normal. The fundus was normal. V.A. was 7/10 sc (R) and 5/10 sc. (L.). There was simple hypermetropic astigmatism according to the rule, axis not inclined. The visual fields were normal. W.R., Meinicke, Kahn were all negative.
Caterina B. Aged. 8.

She had bilateral congenital ptosis. The palpebral apertures measured 5 mms. the lower quarter of the pupil remaining uncovered. There was slight convergent strabismus of the right eye and paralysis of all extrinsic muscles. There was no diplopia. The cornea was normal in size and shape, the pupils round and regular, the pupillary reflexes normal and the fundus normal.

V.A. was 4/10 sc.(R) and 6/10 sc.(L). There was mixed astigmatism, axis not inclined. W.R., Meinicke, Kahn were all negative.

Margharita B. aged 4.

She had bilateral congenital ptosis, the lower one-third of the pupils remaining uncovered. There was paralysis of all extrinsic muscles. She had no nystagmus, the corneæ were normal and the pupils round and regular. The pupillary reflexes were normal and the fundi normal. There was simple hypermetropic astigmatism according to the rule, axis not inclined. W.R., Meinicke, Kahn were all negative.

Isabella B. aged 6.

The eyelids were normal in size and position. The palpebral apertures were of normal dimensions. Astigmatism according to the rule was present. V.A. was
10/10 sc in both eyes. W.R., Meinicke, Kahn were all negative.

From accurate enquiries and clinical data, confirmed by assertions from the family and relatives, together with photographs, Tirelli was able to establish the existence of ocular changes, analogous to those above mentioned, in various other members of this family belonging to various generations.

Maddalena A. was the first born of five children. Giorgio was also afflicted with congenital bilateral ptosis, and was a bachelor. Antonio died at the age of 16, and was normal. Maria, examined by Tirelli, was afflicted with ocular changes in every way similar to those in her sister Maddelena, with physical and psychological abnormalities in addition.

Maddelena was 32. She had marked congenital bilateral ptosis. The palpebral aperture measured only 4 mm. the pupils being almost completely covered. The right eye was deviated about 10° downwards, the left eye about 15° outwards. There was complete paralysis of the extrinsic muscles, no diplopia, the corneae were normal, the pupils round and regular, the pupillary reflexes normal and the fundi normal. V.A. was 1/10 sc (R) and 4/10 sc (L). There was simple hypermetropic astigmatism according to the rule. There was concentric constriction
of the visual field of about 10° for white and for colours. W.R., Meinicke, Kahn were all negative.

The last sister, Caterina, also had congenital bilateral ptosis, as also had her two only children, Caterina and Maria.

In the preceding generation to that examined there were four siblings, and with the exception of the eldest one, Maria, the three others Margherita, Giuseppe and Antonio were afflicted with congenital bilateral ptosis. The first two had no children, the last two had five children who have been described. Going back Tirelli was able to ascertain the existence of bilateral ptosis in Giorgio A. who was an only son. He was able to exclude the existence of this anomaly in members belonging to the families related to the issue of Giorgio A. He could also assert that there was no consanguinity.

Weisenburg and Sweet showed a case of this type at a meeting of the Neurological Society of New York in 1908. Their case was a girl of 14. She had a considerable degree of ptosis on both sides, and was unable to move her eyes in any direction. The pupils were normal and the child was otherwise normal and healthy.
The child's mother had the same condition. This woman had had two children, the patient described above, and one other child who had died in infancy and who was similarly afflicted.

Rampoldi has described two cases of congenital hereditary ptosis associated with external ophthalmoplegia. His patients were a brother and sister whom he observed in the Pavia Hospital in 1887.

**Luigi P.** aged 13. Occupation: tailor.

Both upper lids were ptosed and unwrinkled. He was unable to raise them at all, and could see only by bending back his head. Slight inward movement of the right eye was the only external ocular movement left. The pupils were regular, and reacted to light and accommodation. The discs were oval, with the greater diameter in the horizontal meridian. The eyeballs themselves appeared slightly flattened from above downwards. There was slight hypermetropic astigmatism against the rule. No marked abnormality in the visual acuity or visual fields was noted.

**Angela P.** aged 15. Occupation: servant; sister of Luigi.

There was no movement of the Levator Palpebrae Superioris, and the extrinsic movements of the eyes were completely absent. Both eyeballs were rotated downwards
and inwards. In order to see she had to throw her chest and head backwards, and adopt various attitudes. She had hypermetropic astigmatism according to the rule, of a greater degree than in her brother's case. With a considerable effort she could read at a distance of 12 cm.s. In reading she used her eyes time about, inclining her head this way and that. The optic discs were depressed and the physiological pits were well-marked.

The father of these children had the same condition. The story ran in their family that this man's mother, during her pregnancy, had been so horrified at the spectacle of this abnormality in the daughter of someone with whom she had been staying that her child had been born in like state.

Aurand described, at a meeting of the Ophthalmological Society of Lyons, a girl of four who had ptosis and external ophthalmoplegia in both eyes.

The eyes were rather prominent, the right more so than the left, which had a convergent strabismus. The right palpebral aperture was rather bigger than the left. The right eyelid could be raised 2 mms. higher than the left, but neither could be raised above the cornea. No movements of the eyes could be carried out in any direction, but when the child made violent efforts
to raise its eyelids the effort gave rise to slight horizontal nystagmoid movements. The mother said that the eyes had been immobile since birth. The eyelids could be shut in the normal way, the pupils were regular and equal and reacted equally to light and accommodation, and the fundus oculi was normal. There was a high degree of myopia with myopic astigmatism. The child had no other congenital abnormality.

This patient had two sisters. The elder had slight ptosis, complete absence of vertical movements of the eyes, but slight lateral movement. The younger sister had normal eyes. The maternal grandmother and one of her cousins had had slight ptosis. There was no external ophthalmoplegia in the ancestors.

It is noticed that in this family the condition occurred only in females, although Aurand mentions that several cousins of the patient's father had slight ptosis.

Bradburne in 1912 described a family in which congenital ptosis, of varying degree, and accompanied by almost complete external ophthalmoplegia, occurred throughout five generations. Of thirty-seven members of this family, sixteen were affected, eleven females and five males. Seven of them were personally examined.
by Bradburne.

**Bradburne's Pedigree.**

The following is a description of some of the more important features of the patients examined by Bradburne.

**E.B. female.** (III. 2) She had bilateral ptosis, almost complete on the right side, and partial on the left side. Both eyes were practically immovable. The pupils reacted to light and the fundi were normal. The refraction was hypermetropic. The head was somewhat thrown back.

**P.B. male.** (IV. 3) There was bilateral ptosis, of an equal degree, on both sides. The right eye was completely paralysed, and the left possessed only a
limited side-to-side movement. The axis of the right eyeball was directed downwards and outwards, that of the left downwards and inwards. The pupils and fundi were normal, the the refraction hypermetropic.

**E.B. female. (IV.6.)** Complete ptosis on right, partial on left. Both eyes were absolutely immobile.

**L.B. Female. (V.1)** This child had complete ptosis in the right eye, but no ptosis in the left. The left eye was completely immobile, and lateral rotation only was preserved in the right.

**E.B. Female. (V.2)** This patient had no ptosis at all. Movement in both eyes was very limited, and seemed to be produced by the Superior Oblique muscles only.

**R.B. Male. (V.3)** Died in infancy; said to have had drooping of both eyelids.

**E.B. Female. (V.4)** An operation had been performed for bilateral ptosis, but even so two-thirds of both corneae were still covered. External ocular movements were almost completely absent on both sides. The pupillary reactions were present.

**E.B. Female (V.5)** There was bilateral ptosis, half of each cornea being covered. Lateral movement only was preserved in both sides. The left eye had a
convergent strabismus.

In this family transmission of the condition was in every case direct. In no case was a generation skipped, nor did any affected member fail to transmit it. It was transmitted four times through the female line out of a possible six, and twice through the male line out of a possible three. It is thus seen, as Bradburne points out, that both sexes were equally liable to transmit it.

The condition was congenital and non-progressive, and unaccompanied by any other congenital defect, though all members of the family had high arched palates. There had been no consanguinity.

Bradburne's cases are remarkable in that the degree of ptosis and ophthalmoplegia varied in the two eyes, and did not run parallel to each other in any one eye. Of the fourteen eyes in the cases examined by Bradburne, ptosis was complete in only four, in each affecting the right eye. Partial ptosis was present in eleven eyes, and in three eyes ptosis was absent. Immobility was practically complete in eight eyes, limited to abduction in two, these being a right and a left eye in different patients.

Bradburne considers that in all his cases the superior oblique muscles were normally present but ill-
developed, and that the other muscles were more or less absent. In some of the cases there was evidence of the active presence of Müller's muscle, and no sign of its presence in others.

Chaillous and Pagniez showed at a meeting of the Paris Neurological Society in 1905, three patients, mother, daughter and grand-daughter, all of whom had congenital bilateral external ophthalmoplegia. Two other children in this family, a boy and a girl, had the same condition.

The mother had complete ptosis of the right eyelid and almost complete ptosis of the left. In the daughter the ptosis was somewhat less pronounced, and in the grand-daughter, aged 20 months, the paralysis was limited to elevation of the upper lid and vertical movements of the eyes. All three had marked nystagmus. The intrinsic muscles of the eye were normal.

Four members of this same family were shown by Caillaud at a meeting of the Paris Ophthalmological Society in 1912.

Flieringa has reported a family of Frieslanders who had congenital ptosis associated with defective ocular movements. This family consisted of seven people,
two parents, two sons and three daughters. The father was normal, and there was no history of ptosis in his ancestors. The mother had been the first member of her family to have ptosis, and she attributed the condition to the fact that her mother, when pregnant with her, had heard of the death of one of her sons, and had wept with her head on the table. All had paralysis of elevation of the eyelids, and of elevation and depression of the eyeballs. Some had defective functioning of the superior obliques. The intrinsic ocular muscles were normal, and in all there was nystagmus. Flieringa states that it was not until a few days after birth that these children were able to open their eyes.

In the case of the mother ptosis was more marked on the right than on the left. The upper lids could not be raised at all, the forehead was very wrinkled, and she kept her head tilted backwards and to the left in order to see. Horizontal and rotatory nystagmus was present, which increased when she tried to look upwards. The eyeballs could be moved neither upwards nor downwards. When she looked laterally the heteronymous eye lagged, and failed to cross the midline. Convergence was impossible. A slight pterygium was present in the left eye. The vision was normal.
The daughter showed a condition similar to that of the mother, the chief difference being that in her case the internal recti functioned normally and that the degree of prosis was less pronounced. The lids could be elevated slightly by contraction of the frontalis muscle. There was slight horizontal and rotatory nystagmus and the left eye showed a divergent strabismus. The vision was normal.

In the case of the eldest son the left upper lid could be raised a little by strong contraction of the frontalis muscle, but the right could not be raised at all. Horizontal and rotatory nystagmus was present, and there was a slight internal strabismus of the right eye. When he endeavoured to look upwards the eyes exhibited several to-and-fro horizontal movements, and then came to rest in the left corner of both palpebral apertures, the left eye only showing any elevation. When he tried to look downwards the left eyeball came to rest in the nasal corner of the palpebral aperture, while the right eyeball remained in the median position. The left eye finally occupied a more inferior position than the right. When he looked to the right there was some lagging of the right eyeball. This patient also suffered from congenital cataract.
The second son, aged 14, showed the least disturbance of ocular movements of all the family. His internal and external recti functioned normally, but there was no action of the levators, only slight action of the superior and inferior recti, and poor functioning of the obliques. Flieringa states that this boy had also proptosis. There was horizontal and rotatory nystagmus.

The youngest member of the family had relatively marked ptosis. When she looked upwards the right eye moved to the temporal corner of the right palpebral aperture, being slightly elevated. It was not, however, followed by the left eye. When she tried to look down the eyes moved backwards and forwards, but showed no depression. Convergence was weak, and when she looked laterally the eyeballs could not be brought to the nasal corners of the eyes. Like the rest of the family she had nystagmus.

One daughter, who was in a Sanatorium for diseases of the chest, was not seen by Flieringa, but he was able to report from a photograph that she had ptosis and divergent strabismus.

Lawford in 1887 reported four cases of congenital hereditary partial external ophthalmoplegia.

The patients were a father and three of his seven
children. They all showed almost complete bilateral ptosis, with loss of upward and downward movement of the eyeballs, and very defective lateral movement. Their vision and accommodation were good, and the fundi were normal. They were otherwise perfectly healthy. The three affected children were numbers 2, 4, and 7. The remaining four children were normal. The following is an account of Lawford's patients.

Case 1. The father. He had complete bilateral ptosis, carried his head slightly thrown back, and his brow constantly wrinkled in a vain attempt to raise the eyelids. There was no upward and very little downward movement of the eyeballs, while lateral movements were of moderate extent. The internal recti acted better in convergence than in conjugate lateral deviation. There was no action of the oblique muscles. There was nystagmus on lateral deviation. Lawford states that the right pupil measured 3-3½ mm's, the left 2½-3½ mm's.

Case 2. The second son. He had complete bilateral ptosis, very little aid being given by the constant contraction of the frontalis muscle. There was no upward or downward movement of either eyeball. The right eye was in a position of slight divergence, the left in one of slight convergence. There was moderate outward
movement and the right internal rectus acted better in conjunction with the left internal rectus than with the left external rectus. When he was told to look to his left, he moved both eyes outwards. There was nystagmus, the nystagmic movements of the left eye being neither synchronous with nor of the same kind as those of, the right eye. The pupils were equal.

Case 3. The fourth member of the family. There was complete bilateral ptosis, but by the aid of the frontalis muscle the palpebral fissures could be opened to a width of about 7 mm. There was complete loss of vertical movement and of outward movement. The internal recti could be used only in convergence. There was constant irregular nystagmus. The left pupil was larger than the right, and they reacted to light and accommodation.

The eldest member of the family (not seen by Lawford), was said to have slight drooping of the left upper lid, but no defect of ocular movement.

A noteworthy point in Lawford's cases is the better action of the internal recti in convergence than in conjugate lateral deviation.

Gourfein in a communication to the Academy of Medicine of Paris in 1896, described a family in which nine people were afflicted with complete congenital
external ophthalmoplegia associated with a greater or less degree of amblyopia.

The patient F. consulted Gourfein at the Hôpital Ophtalmique de Genève, complaining of conjunctivitis. He had inherited the condition of external ophthalmoplegia from his mother, who had been similarly afflicted since birth. The mother had had, by a second marriage, a daughter who had a convergent strabismus. The mother's father had also married again, and had had by his second marriage six healthy children. F. had seven children, two girls and five boys. Of the latter, one had died at the age of 8 months, and had had, like his father, congenital external ophthalmoplegia. The other four boys were similarly afflicted, but the two girls were perfectly healthy.

The following is a detailed account of Gourfein's patients:

F. aged 42. Occupation, pedlar. He had almost complete double ptosis. He carried his head tilted back, and when asked to open his eyes, he was able, by using his frontalis muscles, to raise the upper lid 3 or 4 mm., but in order to see objects he had to throw back his head. There was complete absence of voluntary movement of all the external ocular muscles, but the
light and accommodation reflexes were normal. Rotatory nystagmus was present. The uncorrected vision was 1/10 in both eyes; with -3 D. Sph., it was 1/3.

The fundus oculi was normal, apart from a small posterior staphyloma. The physiological pit was slightly more marked than in the normal. The visual fields were normal. The man had had no other illnesses, and, in particular, he had never had syphilis. His nervous system and intelligence were normal, his musculature badly developed and flaccid.

N.F. aged 12. Eldest son of F.; a very intelligent child. His ptosis was less pronounced than his father's but in order to see he had to tilt his head back. By making an effort with his frontalis muscles he was able to enlarge the palpebral fissure by two or three mm.s. He had complete immobility of all the external ocular muscles, but the light and accommodation reflexes were normal. He had rotatory nystagmus. Vision in the right eye, uncorrected, was 1/6, with -3 D. Sph. it was 1/3. Vision in the left eye was 1/3 and lenses produced no improvement of vision in that eye. The fundus oculi was normal, apart from the fact that the disc was somewhat pale and deeply excavated. No other abnormality was noted, but he was a
miserable looking child with a flabby, poorly developed musculature.

J.F. Aged 9. This child was better developed physically than his brother. He had marked bilateral ptosis, and always carried his head tilted back, using his frontalis muscles to raise his upper lids a little. He had complete immobility of all the extrinsic muscles of the right eye, and rotatory nystagmus. In the left eye movements of the internal and external recti were preserved, so that he could look outwards and inwards without moving his head. All other movements of the left eye were lost, and there was rotatory nystagmus. The light and accommodation reflexes were normal in both eyes. The fundus oculi showed no abnormality. He had had no previous illness of note, and all systems, including the nervous system, were normal.

It is noteworthy that in Gourfein's family the condition appeared especially in individuals of the opposite sex to the person first affected. It appeared first in a woman and then affected chiefly males. The unaffected daughters, aged 6 and 3 respectively, were not the youngest of F.'s family.

Congenital external ophthalmoplegia may be accompanied by abnormalities elsewhere in the body, as
in the cases described by Yealland, by Gazépy, and by Bach.

Yealland presented at the section of Neurology of the Royal Society of Medicine in 1931, a case of congenital partial ophthalmoplegia associated with absence of facial movements and with malformation of the tongue.

The patient was a female aged 16. She had complete absence of all ocular movements, except those in a vertical direction. Her pupils did not react to accommodation but reacted to light. There were no nystagmus, diplopia or squint.

There was almost complete bilateral absence of facial movements, and no response to electrical stimulation in muscles supplied by the facial nerves. The tongue was abnormally small, deformed, and wrinkled. No other abnormality was noticed, except some imperfection of the speech. Delivery had not been instrumental. The Blood Wassermann Reaction was negative. One sister was epileptic.

Gazépy of Athens reported in 1894 two cases of congenital external ophthalmoplegia occurring in a brother and sister, natives of Andros. The following
is a description of Gazépy's cases:

**J.I. Gardener. Aged 25.** He had marked ptosis of both upper eyelids and paralysis of the superior and lateral recti of the right eye, and of the superior, inferior, and lateral recti of the left eye. His pupils were normal, and reacted to light and accommodation. Both eyes were myopic.

This patient had also a congenital maldevelopment of the index and little fingers of both hands, which were smaller in proportion than the other fingers. He had syndactyly of the middle toes of both feet, and paresis of the sphincter of the bladder. He was also found to have hemianaesthesia of the right half of his head, he was deaf in his right ear, his memory was poor, and his expression was vacant.

**A.I. Servant. Aged 18.** She had ptosis of both upper eyelids, paralysis of both superior recti, and of the internal rectus of the right eye and of the lateral rectus of the left eye. She had myopic astigmatism in the right eye and hypermetropic astigmatism in the left eye. She had the same congenital abnormality of the fingers and toes as her brother. She was completely deaf in the left ear following an abscess, at the age of 12, and her memory was poor.
The parents of these children had normal eyes. Their father had a brother and sister who were also normal, but whose offspring showed these same ocular abnormalities. Gazépy concludes that the condition was probably inherited from the grandfather, i.e. was an atavistic phenomenon. He was not, however, able to obtain any exact information about the health of this grandfather.

A case of congenital bilateral ptosis with partial external ophthalmoplegia and other somatic abnormalities was reported from Würzburg by Bach in 1893. The patient was a man of 27 who belonged to Lichtenau. Ptosis was incomplete, the width of the right palpebral fissure being 6 mm. that of the left 5 mm. There was greater contraction of the frontalis muscle on the left side than on the right, and the left eyebrow was higher than the right. The length of the palpebral fissures was 23 mm. Movements of the eyeballs were very limited in all directions, and there was some degree of divergent strabismus in the right eye. There was horizontal nystagmus when the patient was rotated. The pupils reacted normally.

The condition had been noticed soon after birth. It was not present in any other members of the patient's
family. There had been no consanguineous marriage in the family. This man was unintelligent, being slow to learn. The lower part of his face was expressionless and weak, although no real paresis could be proved and the electrical reactions were normal. He was easily fatigued when chewing, and had difficulty in swallowing. His tongue was abnormally small and short. All the muscles of his body were very poorly developed, and there was atrophy of both upper arms, especially affecting the deltoids. Muscular power was poor, but the reflexes were normal. The facial part of the skull was poorly developed, and the forehead receded. There was some bulging of the back of the skull on the right side. The infraorbital margins were badly developed. The gums were abnormally small and arched, and the "pharyngeal bone" was very narrow. The lobes of the ears were poorly developed. The length of the skull in Rieger's Plane (through the ext.occip.protub. and the arcus superciliaris) was abnormally long, being 17.2 cm.s. The greatest width of the skull was 14.2 cm.s and the greatest height above Rieger's Plane was 11.4 cm.s.

Operation on this case revealed that the tendon of the levator palpebrae was normally inserted, and was of normal length and breadth. The rectus internus was
broad, normally inserted and looked normal.

We have seen that it is very rare for the intrinsic musculature of the eye to be affected in congenital external ophthalmoplegia. I have found only three such cases in the literature, one reported by Guende, and two by Li.

Guende's patient was a female of 20, who had had since birth very marked, but not complete, ptosis of both upper lids, with divergent strabismus on both sides. The ptosis was so severe that if the patient kept her head erect, a great effort succeeded in uncovering only half of the pupil. She used her left eye only, and was obliged when looking at anything to incline her head both to the right and backwards. The external ocular movements were very restricted in all directions, lateral rotation being the most full. Each effort to move the eyes was followed by a series of nystagmoid-like jerks. These occurred on both near and distant fixation. The pupils were very small and did not react to light. Repeated instillation of atropine produced only moderate dilatation.

The right eye showed a moderate degree of myopia with astigmatism, and vision in it was reduced to light-perception. The disc was very small, and showed a
small posterior staphyloma. Vessels on the temporal side were very tortuous. The eye itself lay in the outermost angle of the palpebral aperture.

The left eye had a higher degree of visual acuity than the right, and its disc was normal in size.

Accommodation was intact.

The girl was otherwise healthy, and of normal intelligence. Her parents and eight siblings had normal eyes. The five youngest siblings were dead, three having died of malnutrition a few days after birth, and two at the beginning of their second year of life. The father was a confirmed alcoholic, but showed no evidence of syphilis.

At operation on this case Guende found that the left medial rectus was reduced to a fine whitish strand. The left lateral rectus was contracted but was of normal calibre.

Three cases of congenital total bilateral ophthalmoplegia were reported from Peking by Li in 1923. Two of the patients, who were Chinese, were personally examined by Li.

Case 1 was a female aged 4. Until about ten days after birth she had been unable to raise her eyelids at all, and then they could be raised only slightly. The
eyeballs had also been immobile since birth.

There was bilateral ptosis, the skin of the upper lids being smooth, the inner borders of the eyebrows elevated, and the skin of the forehead wrinkled. The patient preferred to fix with her left eye, tilting her head to the right. The left palpebral fissure then became wider and the right narrower, while the left side of the forehead became more puckered than the right. If the patient's head was tilted towards the left, she then used her right eye to fix, and the same phenomena appeared on the opposite side. Elevation of the lids was brought about entirely by the frontalis muscle. Apart from slight movement of both internal recti the external ocular movements were completely lost, both eyeballs being practically fixed in a divergent position. Lateral nystagmus was present. Both pupils were contracted, the right which was pear-shaped being slightly larger than the left which was diagonally oval. The pupils did not react to accommodation, nor did they react normally to light, although strong light directed on to the macular region of each eye caused slight alternate contraction and dilatation. Atropine produced semi-dilatation of the pupils. There was slight pannus involvement of the upper corneo-scleral margin. The refractive error was as follows:–
(R.E.) +4.50 D.Sph. C +0.50 D.Cyl. ax. 105° = 6/20+
(L.E.) +5.00 D.Sph. C +0.50 Cyl. ax. 90° = 6/15.

The fundi were normal. No other abnormality was observed on examination of the nervous system.

At operation the internal recti were found to be light in colour, flabby and hypotonic, and the tendons thin and narrow. They were inserted about 5 mm.s from the limbus. The external recti were in a similar condition, and were inserted about 11 mm.s from the limbus.

Case 2 was the younger sister of Case 1. She used her left eye almost exclusively, because the left upper lid could be elevated more than the right. As in the sister, slight action of the internal recti was the only surviving external ocular movement. There was lateral nystagmus. The pupils were contracted and did not react to light or accommodation, although slight alternate contraction and dilatation followed the direction of a pencil of light on the macular regions. Atropine produced semi-dilatation of both pupils. Vision in the right eye was 4/30 and in the left 3/30, and the vision was not improved by glasses. The fundi were normal. No other abnormality was found on examination of the nervous system.

The internal and external recti were found to
be in a similar state to those of the sister. The internal recti were inserted about 5 mm. from the limbus and the right external rectus about 9 mm. from the limbus.

The parents of these children were normal, and their blood Wassermann reaction negative. The mother had never had an abortion or miscarriage. There had been no consanguinity in the family, and as far as they remembered none of their ancestors had had this condition, although Li is rather doubtful of the truth of this statement. Of their five children the middle three were affected, the eldest, a boy, and the youngest, a girl, having normal eyes. The eldest affected member was a boy who died at the age of two, and the other two were the patients described above.

Congenital ptosis with external ophthalmoplegia may, as in the case of Joan S. (above), be unilateral. Thus Wilbrand and Saenger have reported two cases of left-sided congenital ptosis, the one associated with absence of upward movement of the left eye, the other with absence of movement upwards, upwards and inwards, and upwards and outwards. Loeb observed a case of unilateral congenital ptosis associated with paralysis of the superior and inferior recti and of the inferior oblique.

Natale has reported a case of right-sided
congenital ptosis in which both eyes were deviated towards the right, and in which abnormal insertions of the recti were found at operation.

The patient was an Argentine aged 26. He had ptosis of the right upper lid, which he partially corrected by wrinkling his forehead by contraction of the frontalis muscle. There was slight inclination of the head towards the left. Both eyes were strongly deviated towards the right. The right eye could not be moved upwards or downwards. The right pupil was smaller than the left and reacted only slightly to light and accommodation. Vision in the right eye was 3/10, and in the left eye it was normal. The fundi were normal, and there were no diplopia or nystagmus. There was no disturbance of gait, and the general condition was good.

There was no history of this or any similar condition having existed in the patient's family. He had eight brothers, all of whom had normal eyes.

The following conditions were revealed at operation: - the left medial rectus was inserted at barely 2 mm.s from the corneal margin. The left lateral rectus was replaced by tendinous bundles, inserted 8 - 9 mm.s from the limbus. The right lateral rectus had a double insertion 8 mm.s from the limbus.

The right medial rectus was inserted at 8 mm.s
from the corneal margin and its muscular belly was partly replaced by tendinous bundles.

The right superior and inferior recti were found to be absent.

After an operation in which the deviation of the eyes to the right was to some extent corrected, the rotation of the head disappeared.

Examination of the cases reported above shows that the ophthalmoplegia is not always complete and that generally it seems to affect the external ocular muscles indiscriminately and in a haphazard manner, independently of their nerve supply. This, coupled with the fact that the intrinsic ocular musculature is practically invariably intact, suggests that the fault is muscular rather than nervous. This tends to be borne out by the few post-mortems that have been done. In most of these abnormal insertion of the extrinsic ocular muscles has been the most common finding, although atrophy or absence have also been seen.

Thus, Olbers and Wrisberg\(^{\text{II}}\) found union of the superior rectus and external rectus on the right and of the superior rectus, superior oblique and internal rectus on the left. Ahlstrom\(^{\text{III}}\) could find no trace of the levator palpebrae. Silex\(^{\text{III}}\) performed a post-mortem examination on a girl with congenital ptosis who died

\(^{\text{II}}\) quoted by Wilbrand and Saenger.
at the age of 27 and found uniform atrophy of the levator palpebrae superioris muscle. The findings of Heuck (p. 154) and of Natale (p. 194) were similar.

In certain cases the lesion must no doubt be of the nature of a congenital aplasia of the oculo-motor nuclei, as in the case described by Bach (p. 186) in which operation revealed normal eye muscles. In the few cases in which internal ophthalmoplegia has been observed (Guende, Li) congenital aplasia of the motor nerves of the eye themselves must be assumed. The atrophy of the left medial rectus found by Guende at operation was probably a disuse atrophy.

In the case of Joan S... the lesion would appear to be in the 3rd nerve nucleus, or in the 3rd nerve itself, sparing the irido-constrictor fibres.

Congenital external ophthalmoplegia may, as we have seen, occur in cases of congenital ptosis with epicanthus and blepharophimosis, although it is rare for it to do so. (Cases of Bach, Hirschberg, Brückner, Hüttemann, Verwey, Braun, Fergola). In the great majority of these cases the ptosis is not a paralytic one, but is due, as has already been pointed out, to smallness of the palpebral aperture. It is a noteworthy
fact, borne out by perusal of published cases, that congenital ptosis of the "phimotic" type is much more rarely accompanied by external ophthalmoplegia than is congenital ptosis of the "paralytic" type. When congenital ptosis of the "phimotic" type is accompanied by ophthalmoplegia it is nearly always upward movement only that is limited, and this was so in the cases of Hittmann, Verwey, Braun and Pergola.

Of all the cases of congenital ptosis in the families D------ and Sl---- (all of the "phimotic" type) one only (J.S.----) showed external ophthalmoplegia and then it was limitation of upward movement of both eyes. The explanation of this is not far to seek. Owing to the narrowness of their palpebral apertures, these patients must tilt their heads backwards when they desire to look up. It is useless for them to turn their eyeballs upwards, as their view would then be obstructed by the overhanging lids. It is thus not surprising that they should lose, or indeed, never acquire, the upward movement.
1. Congenital ptosis may be part of the picture of congenital external ophthalmoplegia. It is then a true paretic ptosis, unaccompanied by blepharophimosis, epicanthus or cranial deformity.

2. A case of unilateral congenital ptosis and external ophthalmoplegia is described.

3. Cases from the literature are quoted.

4. Congenital external ophthalmoplegia is most frequently due to a congenital dysplasia or malinsertion of the external ocular muscles.
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CHAPTER VI.

CHRONIC PROGRESSIVE EXTERNAL

OPHTHALMOPLEGIA

(INCLUDING HEREDITARY NON-CONGENITAL PTOSIS)
The preceding chapters have been devoted to various forms of congenital and hereditary ptosis of the upper eyelids. Familial ptosis may also supervene in adolescence or in adult life. It may or may not be accompanied by paresis of external ocular muscles. Consideration of this syndrome forms the subject of the following chapter.
Paralysis of the external ocular muscles has been described by numerous authors (Graefe, Hutchinson, Parinaud, Wernicke, Panas etc.) and under a variety of names, Superior Chronic Polioencephalitis, Progressive Nuclear Ophthalmoplegia, Progressive External Ophthalmoplegia. It may follow a vascular lesion in the brain-stem, it may occur in the course of tabes dorsalis, cerebral syphilis, disseminated sclerosis, or pseudo-bulbar palsy. Many cases in which external ophthalmoplegia has been congenital have also been reported and these have already been discussed. It is not my intention in this chapter to deal with any of these conditions, but to confine my remarks entirely to a group of cases in which paralysis of some or all of the external ocular muscles, including the levator palpebrae superioris, comes on in extra-uterine life, is slowly progressive, is unaccompanied by other signs or symptoms of neurological disease, and occurs frequently as a hereditary and familial (although not congenital) phenomenon. I think that these cases may rightly be included in the larger group of "Chronic Progressive External Ophthalmoplegias". I shall not employ the less comprehensive terms "Superior Chronic Polioencephalitis" of "Progressive Nuclear Ophthalmoplegia" which postulate the nature and site of the lesion.
Nine cases in which paralysis of the external ocular muscles of this type was a hereditary and familial manifestation have been reported (Lawford; Ayres; Homen; Beaumont; Pasetti and Salani; Spencer; Roger, Aubaret and Siméon; Crouzon, Christophe and Braun-Vallon; Aguilera). Of the condition occurring sporadically, Wilbrand and Saenger reculled twenty-two cases in the literature and six others have since been published. (Altschul; McLellen and Hines [two cases]; Collins; Langdon and Cadwalader; and Schäffer).

Ptosis of the upper eyelids occurring as an isolated lesion has even more rarely been observed. It is also found either as a hereditary and familial condition, or occurring sporadically. Of the former, seven families have been described (Fuchs; Dutil [two families], Delord, Boulanger, Meumann and Forsberg) and of the latter may be cited the cases of Fuchs, of Goldsieber, of Batinev, and others. Two cases are here described, one of progressive ptosis accompanied by slight external ophthalmoplegia, and one of chronic progressive external ophthalmoplegia involving all the extrinsic eye muscles. An attempt is made to review rapidly the literature of the subject, and to emphasise the salient clinical features of the condition. I shall conclude by discussing the various hypotheses which have

Detailed descriptions of these cases are given in the Appendix to this chapter.
been put forward as to the pathogenesis.

**Case 1. Miss Annie X——, aged 41. Occupation, Millworker.**

The patient complains of drooping of both upper eyelids of 21 years' duration. When she was twenty years of age her friends told her that her eyelids were drooping slightly. The condition became very gradually worse, and it was not until she was about thirty that she became subjectively aware of it, and at that time it was chiefly towards evening that she noticed it. The condition is now present all the time, and the patient is convinced that it is gradually becoming worse. For some years she has noticed that when visiting a cinema her neck becomes painful with the effort of tilting her head backwards in order to see the screen. She has at no time seen double.

The patient has had no previous serious illness. She works all day in a mill, and enjoys her work. She still has her periods, which have always been regular, although they give her some pain. She occasionally suffers from attacks of "flushing."

Her mother's eyelids droop slightly, but to a less degree than the patient's. Apart from this she knows of no other members of her family who have had ptosis.
On examination the patient is seen to have a severe degree of ptosis of both upper lids, slightly more pronounced on the left side than on the right. The width of the palpebral fissure is 4 mm. on the left side and 5 mm. on the right. The lids are thin and atrophic, and the forehead furrowed. By contracting the frontalis muscle she can widen the palpebral aperture by about 2 mm. If the observer's thumbs are pressed into the orbit above the upper lids to prevent the action of the frontalis, the lids can scarcely be raised at all. Vertical movement of the eyeballs is normal. Lateral movement in either direction is very limited in both eyes, but there is no complete paralysis of any external ocular muscle. The pupils are equal and regular, and react to light, directly and consensually, and to accommodation. Instillation of two drops of 4% cocaine hydrochloride causes dilatation of the pupils, but no change in the position of the upper lids. Ophthalmoscopic examination shows no abnormality. The vision uncorrected is (R.E.) 6/18 and (L.E.) 6/36, and corrected with -0.50D.Sph. -0.25 D.Cyl. axis 180° V = 6/9 J.1, and with -1.00 S. Sph. -0.00 D.Cyl. V = 6/12 J.1.

There are weakness and atrophy of the orbicularis oculi muscles, the patient being unable to keep the eyes shut against resistance.
The tendon reflexes are equal and active, and the plantar responses in flexion.

Examination of the other muscles of the body shows no weakness, atrophy, fibrillation, abnormal movements, hypotonia, or inco-ordination. The patient can keep both arms outstretched at a right angle for five minutes. She can look upwards to the ceiling for the same length of time. She can count up to 500 without any weakening of the voice. The dynamometer readings, taken in rapid succession with the patient in bed, were as follows:

**Right Hand.** 60 lbs. 70 lbs. 70 lbs.
**Left Hand.** 60 lbs. 52 lbs. 60 lbs.

Electrical examination of the muscles with the faradic current showed no myasthenic reaction, even with 20-25 closures. An attempt was made, using the galvanic current, to elicit an isolated reaction from the levator palpebrae superioris muscle. No contraction was obtained and no attempt was made to examine with a stronger current. The Wassermann Reaction is negative in the blood and in the Cerebro-spinal fluid, which shows no abnormality as regards chemistry or cytology.

1.5 mgm. Prostigmin was administered subcutaneously but this had no effect on the ptosis.

General examination of the other systems of the
body shows no abnormality. There is no enlargement of the thyroid gland.


This patient came to the Consultation Externe of the Salpêtrière in February 1926. She then stated that in October 1925 she had begun to see double. Several months later she had observed that she squinted with the left eye which was directed outwards. Examination showed an incomplete paralysis of the third nerve on the left side. She returned to the Salpêtrière in 1929, complaining of drooping of the right eye. She had also had some difficulty in going up and down stairs, because of inability to raise and lower her eyes. Examination at that time showed, on the left side, ptosis, divergent strabismus, vertical movements limited, inward movement impossible, and outward movement normal, and on the right side limitation of vertical movements, and normal lateral movements. In 1931 it was observed that inward movement of the right eye showed some limitation.

Examination at the present time shows complete external ophthalmoplegia and ptosis on the left side, and complete paralysis of vertical and inward movement on the right side, outward movement being to some extent preserved. The pupils are equal and regular, and
The photographs of this patient were unfortunately lost with the candidate's luggage during the Dunkirk evacuation. As the negatives are at the Salpêtrière, they are at present unobtainable.
react to light, direct and consensual, and to accommodation. There is no nystagmus, and no diplopia; the fundus oculi and the visual fields are normal. The visual acuity is 9/10 on the right side and 8/10 on the left side. There is slight exophthalmos of the right eye. The width of the palpebral aperture is 5 mm on the left side and 8 mm on the right. There is no oedema of the loose tissues of the upper or lower lids. The ocular tension is normal and equal on both sides. There is weakness of both orbiculares oculi, the patient being unable to keep the eyes shut against resistance. The abdominal reflexes are absent but as the patient is aged 49 and is parous, this is without significance. No other abnormality has been detected on examination of the nervous system. The tendon reflexes are present and symmetrical and the plantar responses are flexion. X-Ray examination of the skull shows no abnormality. Electrical examination of the facial musculature shows no abnormality.

A soft blowing systolic murmur is detected in the mitral area, but no murmur is heard at the base of the heart. The Blood Pressure is 150/90, and the Pulse Rate 70-80. There is no cutaneous pigmentation, the thyroid gland is not visibly enlarged, and the Basal Metabolic Rate is -6%.
The patient suffers from slight breathlessness on climbing stairs. She has not lost weight, and is not getting thinner. There is a very slight tremor of the outstretched hands. Her periods are beginning to be irregular.

The Blood Wassermann Reaction has repeatedly been found to be negative.

Lumbar Puncture was performed in May 1929, and gave the following results: Fluid clear and of normal tension; Protein (Sicard's method) 22 m.gms. per 100 cc.s; Pandy's Reaction, Negative; Weichbrodt's Reaction, negative; Wassermann Reaction, Negative; 0.6. lymphocytes per cub.mm; Colloidal Benjoin: precipitation in the 7th and 8th tubes.

Repetition of the Lumbar Puncture in 1938 gave exactly the same results, except that the cell count was 3, 6 lymphocytes per cub.mm.

The patient is an only child, and there is no history of ptosis or of other nervous disease in her family. She has had two children, both of whom are in good health, and she has never had an abortion or a miscarriage. She had varicella at the age of 6, intercostal herpes at the age of 38, and cystitis at the age of 42. At the time of the onset of her eye trouble, she had no infective or febrile illness, no
sore throat, and no somnolence.

That paresis of the external ocular muscles of gradual onset during adolescent or adult life, and of slow evolution, may occur as an isolated syndrome has long been recognised, and cases have been published from time to time since the end of the last century. Of the thirty-two cases quoted by Wilbrand and Saenger, twenty-four only deserve to be included in this category. The others must be eliminated for the following reasons: those of Eliasberg, Kunn and Dufour because of the intermittent course, those of Marina, Strumpell, Galezowski and the cases referred to by Lehmann because of the pupillary abnormalities, and that of Hanke because the condition appears probably to have been congenital. In nine of the published observations the condition has been familial and hereditary; two of these, (Lawford and Ayres) are mentioned in Wilbrand and Saenger's list, and the others were reported by Homen (twin brothers affected); Beaumont (eleven persons affected in four generations); Pasetti and Salani (eleven persons in four generations); Spencer (six persons in three generations); Roger, Aubaret and Siméon (two brothers affected, condition transmitted from a maternal grandfather); Crouzon, Christophe and Braun-Vallon (five persons affected in two generations); and Aguilar (six persons in three
generations).

That isolated paresis of the levator palpebrae superioris muscle may supervene in extra-uterine life is less well-known, because more rare. Attention was first drawn to the subject by Fuchs in 1890, when he published five cases of isolated bilateral ptosis occurring in adult life. In one of these the condition was familial. In 1892 two families in which isolated ptosis beginning in later life occurred in several members belonging to different generations were observed in Charcot’s clinic at the Salpêtrière, and reported by Dutil, who described the affection as "a type of paralytic ptosis, coming on in adult life, as an isolated symptom, unconnected with the development of any ascertained affection of the nervous or muscular systems, and liable to be transmitted hereditarily through two or several generations". In Dutil’s first family seven persons were affected with ptosis in four generations, and in the second four persons in three generations. Four other such families have since been described; from Nimes by Delord (eight persons affected in three generations), from Lille by Boulanger (six persons affected in two generations), from Munich by Meumann (twenty-two persons in five generations) and from South Dakota by Forsberg (thirteen persons in four generations). Various
non-familial cases have also been reported, for example, that of Goldzieher, who named the affection "Ptosis Amyotrophica". Crouzon employs the term "Ptosis familial tardif de Dutil". In Italian literature the condition is referred to by Tirelli as "Ptosis tardiva del Dutil"

All of these cases, whether the condition be or be not hereditary, and whether the paralysis affects the levator palpebrae superioris only, or involves all the external ocular muscles, should probably be considered as belonging to the same group. This view was held by Apert, who speaks of "the cases of ptosis which come on only during extra-uterine life, and which may be associated with progressive atrophic paralyses of the eye muscles"

Little can be said as regards the pathology, as few post-mortem examinations have been carried out. Langdon and Cadwalader were able to examine the brain and cranial nerves in their case, but not the external ocular muscles. They found diminution in the size and number of nerve cells in the nuclei of the 3rd, 4th and 6th cranial nerves. In the 3rd nerve nucleus it was difficult to determine whether the variation observed in the size of the cells was much greater than similar variations in normal specimens. Some of the cells in
Some of the cells in this nucleus had undergone chromatolysis, and many of them were deeply pigmented. No changes were observed in the nucleus of Perlis, in the nuclei of Edinger–Westphal, or in the nuclei of Darkschewitsch. In the 6th nerve nucleus at least one-third of the cells were distinctly smaller in diameter than the others. The fibre bundles of all these nerves appeared smaller in diameter than normal.

Fuchs excised a piece of the levator palpebrae muscle of one of his patients, and examined it microscopically. He found that certain groups of fibres showed atrophy, in some cases only the empty sarcolemma being left. There were practically no hypertrophied fibres. There was increased pigmentation in the diseased muscle fibres, the colouring appearing first in the middle of the fibre, and later spreading through its entire thickness. There was great increase of nuclei in the diseased muscle, and also increase of the nuclei of the interstitial tissue and of the sarcolemma. There was increase of the connective tissue between the atrophic muscle fibres. Collins excised a piece of muscle from his case of chronic external ophthalmoplegia, and found it atrophic, but did not examine it microscopically.

I shall now attempt to describe briefly the
chief clinical features of this form of chronic progressive external ophthalmoplegia.

The age of onset is generally in adolescent or early adult life, occasionally in childhood or middle life. Considering first the cases of isolated ptosis we find that in all of the non-familial cases described, onset has been in middle life, with the exception of one of Goldzieher's, who was a young man. In the familial cases onset was in childhood and adolescence in Forsberg's family, in adolescence in Fuch's families, and in middle and late middle life in the families of Dutil, Delord, Boulanger and Meumann. Turning to the cases in which there was not only ptosis but also paralysis of the other external ocular muscles, in seventeen of the non-familial cases onset was before the age of twenty, in ten between twenty and forty, and in three over forty. In the familial cases the age of onset is very variable. Thus in Ayres' family it was in childhood, in Homen's 15-17, in Aguilar's about 20, in Spencer's 15-25, in Beaumont's 20-30, in Roger, Aubaret and Siméon's 30-40, in Pasetti and Salani's 35-45, in Crouzon, Christophe and Braun-Vallon's 40-50, and in Lawford's 50.

There is no special sex-incidence, the proportion of males to females in the reported cases being roughly equal.
The onset is gradual, so much so that it is frequently difficult to ascertain when exactly the symptoms began, or to know which muscles were first affected. The earliest indication is generally that the patient's friends notice that one of his eyelids is drooping slightly. The condition is not symmetrical; in the great majority of cases it begins unilaterally, but always becomes bilateral. In one observation only, that of Pasetti and Salani, has the order in which the muscles were involved been noted. Pasetti and Salani divided the external ocular muscles accordingly into three groups:

1. the levator palpebrae superioris, the rectus superior, the rectus internus and the obliquus inferior, i.e. the muscles responsible for upward and inward eye movement were first involved.

2. the rectus externus was next affected, and lastly,

3. the rectus inferior and the obliquus superior, i.e. those which depress and abduct the eye. In my second case inward movement and vertical movements were the first to be affected, and outward movement was affected last. It has not been possible to determine with any degree of certainty whether upward or downward movement was involved first. The left eye was involved before, and to a greater extent than the right. In Case 1, lateral movement is equally limited in both directions, vertical movement not at all.
Diplopia is very rarely complained of. It was present at the beginning in Langdon and Cadwalader's case, in one of McMullen and Hines' cases, in one of Beaumont's cases, and in one of my cases. The absence of diplopia is almost certainly due, as Pasetti and Salani have suggested, to the slow march of the affection.

Progression is always exceedingly slow, lasting over a period of years (Wilbrand and Saenger mention as a feature of isolated ptosis that at the beginning the ptosis is often more marked in the evenings, and is worse after exhaustion or mental depression. This was present in one of McMullen and Hines' cases.

Otherwise the condition progresses steadily and uninterruptedly. It may become arrested at any stage or may proceed to complete external ophthalmoplegia.

The patients have a characteristic appearance, carrying their head tilted slightly backwards, and having the forehead wrinkled, due to contraction of the frontalis muscle, which attempts to compensate for the ptosis. The expression is typically "sleepy" (Hutchinsonian facies" of some authors). The pupils and pupillary reactions are invariably normal. This point was emphasised by Paton.

Weakness of the upper facial muscles with paresis of the orbicularis oculi has been remarked by Forsberg, by Homan and by Beaumont, and is present in
both of my cases.

Bilateral exophthalmos was observed in Altland's case and in one of Forsberg's cases, and unilateral exophthalmos in one of my cases.

No other concomitant sign of organic disease of the nervous system has been observed.

**Differential diagnosis.** The most important diagnosis is that from myasthenia gravis, especially in the cases in which diplopia is present at the beginning, or in which the ptosis is worse towards evening or where the patient is tired. The diagnosis is not difficult if a family history of ophthalmoplegia is obtained. Otherwise it is made by the absence of other symptoms, by the uninterruptedly progressive course of the ophthalmoplegia, and by the absence of response to prostigmin.

It has to be remembered that certain rare cases of labio-glosso-laryngeal paralysis or even of progressive muscular atrophy may begin by progressive external ophthalmoplegia. This is referred to by Déjerine as follows "Les lésions des noyaux oculo-moteurs peuvent en se propageant aux noyaux des nerfs bulbaires déterminer une paralysie labio-glosso-laryngée, ou encore descendre du côté de la moelle et produire une atrophie musculaire à marche progressive". Charcot, Guinon and Parmentier
(quoted by Guillain and Alajouanine) have also stressed the relationship between progressive external ophthalmoplegia, progressive muscular atrophy and labio-glossolaryngeal paralysis. In presence of a sporadic case of chronic progressive external ophthalmoplegia coming on in adult life it is impossible to know whether one is dealing with the isolated syndrome which forms the subject of this chapter, or with an early case of pseudobulbar paralysis. Only the evolution of the condition will give the diagnosis, and the prognosis in such cases must therefore at the onset be guarded.

A syphilitic condition may be ruled out by the absence of other signs or symptoms, and by the negative serological reactions.

These cases must be distinguished from cases of congenital external ophthalmoplegia and of congenital ptosis, which may also occur in either a sporadic or a hereditary and familial form, but which are encountered very much more frequently, may be accompanied by other congenital abnormalities and are non-progressive. The opinion that congenital ophthalmoplegia may be progressive is held by McMullen and Hines. Numerous cases of this condition have been published in detail, for example, those of Rodin and Barkan, Aurand, Bradburne, Rampoldi, Guende, Gazépy, de Schweinitz, Gourfein, etc. and a careful
perusal of this literature reveals no case in which progression had been observed. Three cases of congenital bilateral external ophthalmoplegia were reported by Chaillous and Pagniez in 1905, and the same patients were observed by Crouzon and Béhague fifteen years later, when no advance in the ocular condition was found. In a series of some twenty cases of congenital ptosis seen by myself, no progression was observed.

**Prognosis.** McLellen and Mines, defending the recognition of chronic progressive external ophthalmoplegia as a disease entity, point out how important it is from the point of view of prognosis, to realise that in these cases there is no threat to life, and no development of further serious organic disease. I agree with this view, but must point out that such an unreservedly good prognosis cannot always be given at the outset in sporadic cases beginning in adult life because of the difficulty of the diagnosis from early pseudo-bulbar paralysis, as discussed above.

What is the nature of the lesion in these cases? Are we dealing with a central lesion in the nuclei of the 3rd, 4th, and 6th cranial nerves, or with a primitive dystrophy of the eye muscles similar to that of the muscles of the body in the primitive myopathies?
The first view is held by Langdon and Cadwalader, who base their opinion on the histological findings in their case. They point out the striking resemblance of the changes they found to those seen by Greenfield and Stern and others in cases of the Werdniq-Hoffmann type of progressive spinal muscular atrophy of childhood and believe that their case must be regarded as a similar form of chronic neurone degeneration, entirely different from the case of isolated ptosis described by Fuchs, which they consider to be cases of primary myopathy. They point out that while in Fuchs' cases the muscles of the eyelids were very wasted, in their patients there was no atrophy of these muscles. On the other hand Langdon and Cadwalader did not carry out a post-mortem examination of the muscles in their case.

McMillen and Hines think that progressive ophthalmoplegia developing in later life may be related to the hereditary infantile form of progressive bulbar paralysis described by Oppenheim. They suggest that the condition may be due to an "abiotrophy" or "lack of inherent vitality of the cells of the cranial nerve nuclei involved," the time of onset depending on the degree of vitality with which these cells were originally endowed. A similar view was held by Delord.
Pasetti and Salani considered the condition to be nuclear in origin, and regarded the age of onset in their cases (35-45) as a factor against its being due to a primary muscular involvement.

Beaumont thought that the lesion was probably nuclear, and placed it just posterior to the centres for accommodation and pupil-contraction.

The view that the condition is due to a central lesion is also held by Neumann.

In certain of the sporadic cases the lesion must almost certainly be central, as in the case reported by Wilbrand and Saenger, who carried out a post-mortem examination on a patient who had suffered from generalised arteriosclerosis, and who had had bilateral ptosis unaccompanied by other neurological symptoms. They found haemorrhages of different size and age scattered throughout the region of the 3rd nerve, the ganglion cells being normal in form and number. I have had under observation a hypertensive patient who suffered from a slowly progressive ptosis, in whom the vascular origin of the ptosis appeared highly probable, although it was not verified. For cerebral vascular disease to be thus isolated and strictly localised is not, however, common, and of course the pathology in the hereditary cases cannot be of this

Salpêtrière, 1938.
nature.

Certain authors (Rimbaud, de Lapersonne and Cantonnet) put these cases into the category of "Superior chronic polioencephalitis". It is indeed conceivable that certain of the sporadic cases may possibly be due to a virus having a special affinity for the cells of the paired, large-celled lateral nuclei of the 3rd nerve, and for those of the 4th and 6th nerves. Léri and Weissmann-Netter saw a case of progressive ophthalmoplegia which began during a febrile illness accompanied by meningeal symptoms, and considered the possibility of this being due to a specific virus. There is, however, no resemblance between the cases which we are considering and the case of Léri and Weissmann-Netter. The gradual afebrile onset and slow march of chronic progressive external ophthalmoplegia appear to rend improbable an infective aetiology.

Finally there is the possibility of the condition being regarded as due to an inherent weakness of certain cells of the oculo-motor nuclei. No concrete evidence for or against this view has yet been produced.

The second theory, that chronic progressive external ophthalmoplegia is due to a primitive dystrophy of the eye muscles, was held by Fuchs, who regarded the condition in his cases as being a primary muscular
dystrophy sui generis, i.e. a muscular dystrophy confined to the levator palpebrae superioris muscle. Silex (quoted by Wilbrand and Saenger), who performed a biopsy in one of his cases, regarded the findings as identical with those of muscular dystrophy. Collins considered that both the cases of ptosis only and of ophthalmoplegia were due to a primary degeneration of the muscle fibres, and this view was also held by Batirew.

It is rare for the ocular muscles to be involved in the muscular dystrophies. Such cases have, however, been observed. Winkler and Van der Weyde have described a case which showed the features of both facio-scapulo-humeral dystrophy of the Déjerine-Landouzy type, and juvenile dystrophy of Erb, and which presented in addition bilateral ptosis, limitation of inward, outward and upward movement in the left eye, and of inward movement in the right eye, the pupillary reflexes being normal. Ptosis had appeared when the patient was 24, five years after the onset of the other symptoms. Another case in which ocular muscles were involved in myopathy was reported by Gowers, and Oppenheim mentions Lombroso, Oppenheim, Marie, Baeg and Jendrassik as having described others. The histological findings of Langdon and Cadwalader need not necessarily be taken as evidence against the myopathic nature of this
affection. Their patient, a woman of 81, had suffered from ophthalmoplegia for 30 years and it is not surprising that some dropping out of nerve-cells should have been observed in the nuclei subserving the unused muscles. That such a secondary retrograde atrophy of nerve cells occurs in the spinal cord in the muscular dystrophies has been shown by Gordon Holmes. In a case of muscular dystrophy he found the nerve cells of the anterior horns reduced in number and in size, and atrophy and diminution of fibres in the anterior roots. He considered this to be due to injury or destruction of the axis-cylinder nerve endings, left bare by disappearance of the muscle-fibres, and perhaps compressed by the proliferating connective tissue. He refers to similar observations made by other workers.

That the intrinsic musculature of the eye is invariably spared is an argument in favour of the myopathic nature of the affection. It is difficult to imagine a central lesion which leaves intact the median, unpaired nucleus of Perlia and the lateral, paired nucleus of Edinger-Westphal, which are generally regarded as the centres controlling the intrinsic ocular muscles.

Indeed, in the absence of more certain evidence in favour of a central lesion, I think that the myopathic
theory should not perhaps be entirely discarded. The myopathies cannot be separated into "types" according to their distribution. Apart from the pseudo-hypertrophic form, these so-called types overlap and merge into one another. Thus in the case reported by Winkler and Van der Weyde, the muscles of the shoulders, trunk, lower limbs, face and the external ocular muscles were involved.

May not chronic progressive external ophthalmoplegia be a form of muscular dystrophy of slow progression tending to limit itself to the external ocular muscles, but sometimes extending to involve other muscle groups, for example, the upper facial musculature, as we have seen is sometimes the case? It is interesting to refer here to the family described by Bartok, in which three members in three successive generations were affected by ptosis of the right lower eyelid coming on in adolescence. These may be analogous cases, in which the dystrophic process was confined to the right orbiculans oculi muscle.

Of interest in this connection is the work of Yuma Sanaga (quoted by Langdon and Cadwalader) who, following removal of the ciliary ganglion in dogs found changes in the external ocular muscles greatly resembling the changes of muscular dystrophy. He considered
these changes to be due to the fact that the sympathetic supply to these muscles had been interfered with. The possible relationship between disease of the sympathetic nervous system and the muscular dystrophies was discussed by Edwin Bramwell in 1925. He put forward the suggestion that the muscular dystrophies may perhaps be determined by disease of the sympathetic supply to the skeletal muscles. Similar views have been expressed by Leschke, Babes, Kalindero, and Poix and Nicolesco.

It is possible that the cases here considered may be related to the important group of "Exophthalmic Ophthalmoplegias" recently described by Russell Brain. We have seen that exophthalmos was present in several of the cases of chronic external ophthalmoplegia described in the literature, and in one of our cases. This latter differs from Brain's cases in that the exophthalmos is unilateral, while the ophthalmoplegia is bilateral, and more severe on the side opposite to that of the exophthalmos, and also in that the exophthalmos is slight. Brain suggests that the orbital changes in exophthalmic ophthalmoplegia may be produced by the thyrotropic hormone of the pituitary, and that the ophthalmoplegia may be due to the mechanical effect of the intra-orbital tension upon the muscles. Examination of the ocular muscles in certain of his cases showed aedema and enlargement of the muscle
fibres.

The syndrome of chronic ophthalmoplegia here discussed, and the exophthalmic ophthalmoplegia of Brain, may both be dependent on some form of endocrine upset, possibly acting on the orbital muscles by way of the sympathetic. Bramwell has suggested that a relationship may exist between disorder of the pituitary and the muscular dystrophies. He mentions personal cases in which muscular dystrophy has been associated with adiposity and defective sexual development, and cites others. He thought it more probable that the muscular dystrophy and the endocrine disturbance were due to a common cause, than that the former was secondary to the latter.
SUMMARY

1. Two cases of chronic progressive external ophthalmoplegia are reported.

2. The course and characteristics of this syndrome are described, with reference to the literature.

3. The aetiology of the syndrome is discussed, and it is suggested that some of the cases may be due to a form of muscular dystrophy dependent on some endocrine derangement, which may act on the external ocular muscles by way of the sympathetic nervous system.
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DUTIL. - Progrès Médical. 1892, Nov. 12, Vol. 16, p.401.

Note sur une forme de Ptosis non congénital et héréditaire.

Dutil, in 1892, reported a family in seven of whose members, throughout four successive generations, bilateral ptosis appeared in their 50th year. The paralysis of the levator palpebrae muscles was neither preceded nor followed by any other disturbances of the musculature of the eye, by no true visual disturbance, and in fact by no symptom that would lead one to suspect the existence of any disease of the nervous system. One member of the family, indeed, developed signs of tabes dorsalis three years after the onset of his ptosis, but this was almost certainly, as Dutil points out, coincidental.

The following is an account of Dutil's cases :-

X, a lace designer, then aged 66, consulted Dutil at the Salpêtrière. He had at the age of 30 contracted a venereal infection, almost certainly syphilitic, which had been imperfectly treated. He had enjoyed perfect health from that time until the age of 50, when he had developed bilateral ptosis. The left eye was involved first, and the condition had gradually progressed until at the age of 53 ptosis was so complete that he could scarcely, even by throwing back his head, see the people he was speaking /
speaking to. Between his 53rd and 66th year, three surgical operations (of which Dutil gives no details), were performed for the relief of the ptosis. After the first two of these the condition relapsed, but the third produced a permanent cure. During this time he suffered from time to time from diplopia, and from hyperaesthesia of the left hand.

When first seen by Dutil he was complaining of lightning pains in his legs and his left knee jerk was diminished. Three years later there was no doubt that he was suffering from tabes dorsalis. He then suffered from constricting sensations on the trunk, lightning pains in the legs, tingling in the fingers of the left hand and loss of pain sensibility on the back of this hand. He had marked Ronbergism, his knee jerks were absent and his gait inco-ordinate. His pupils were unequal and showed the Argyll-Robertson phenomenon. There was slight impairment of associated eye movements upwards and laterally, and his diplopia had returned.

This man's ptosis would almost certainly, as Dutil remarks, have been regarded as part and parcel of his tabetic condition, had not the following family history been elicited.

Throughout four generations all members of his family, on the maternal side only, had developed bilateral ptosis towards the age of 50. In the case of his greatgrandmother ptosis had never become complete. His grandfather had had to raise his eyelids with his fingers in order to see an object placed in front of /
of or above him. His granduncle had developed the same condition towards the age of 50.

In the case of the patient's mother, and of one of two maternal uncles affected with the condition, it was, as in the patient, the left eye that had been the first to develop ptosis. A third maternal uncle had died at the age of 45 and had then had no ptosis. A brother of the patient, who had died at the age of 50, had no ptosis.

All these persons had excellent general health, no indication of any nervous disease, and lived to an advanced age.

There was nothing of note in the family history on the paternal side.

Pedigree of Dutil's first Case.

Dutil's second case was a man of 57, in whom bilateral ptosis had begun at the age of 45. It had developed over a period of fourteen months, the right eye having been affected first. When seen /
seen by Dutil the eyeballs were almost completely covered by the upper lid, and the forehead furrowed. The patient was in the habit of lifting one eyelid with a little pair of pincers, his vision, apart from the ptosis, was normal, his general health perfect, and he showed no signs or symptoms suggestive of the presence of syphilis of the nervous system.

This patient's father had the same condition of bilateral ptosis, but in him it had never become as pronounced as in his son. It had begun in his case at the age of 42, and, as in the son, the right eye was affected first. His general health and his vision, apart from the inconvenience caused by the ptosis, were excellent.

The patient's grandfather also had ptosis. His brother, aged 53, had no ptosis, and his mother, although neurotic, had normal eyes.

**Pedigree of Dutil's second Case.**

![Pedigree Diagram]

Dutil concludes that "there exists a type of paralytic ptosis, coming on in adult life, as an isolated symptom, unconnected with the development of any ascertained affection of the nervous /
nervous or muscular system, and liable to be transmitted hereditarily through two or several generations.

(Note sur une forme de Ptosis non congénital et heréditaire.)
The fourth case of this type of ptosis was described by Delord in 1903.

The patient was a man of 66, who had noticed that for the last five years he had gradually had difficulty in raising his eyelids, especially in the evenings. Soon the disability persisted during the day, and became progressively worse. Both the upper lids were affected at the same time. The patient had continued with his work until a few months prior to consulting Delord, but had eventually been obliged to give it up altogether. He had to raise his eyelids with his fingers in order to see clearly, or else throw back his head. His frontalis muscle was not able to supplement the action of the levator sufficiently to give adequate vision. No abnormality in the nervous system or elsewhere was detected on examination. His family history was as follows: -

His maternal grandmother was affected like himself with bilateral ptosis. She had three children all of whom had the same condition, the age of onset varying from 40 to 50 years. These three children were the aunt, uncle and mother respectively of Delord's patient.
patient. The aunt had two sons, one of whom died accidentally aged 20, and had normal eyes. The other died at the age of 70 and had incomplete bilateral ptosis. The uncle had two children. One died at the age of 30, and had no ptosis; the other had bilateral ptosis. The patient himself had three sisters of whom only one had ptosis.

Delord's Pedigree.

* Delord's Patient.

Delord suggests that the condition may be due to a congenital and hereditary weakness of the centre concerned with elevation of the upper lid, such their advancing age would not be long in destroying its function altogether.

Delord. La Presse Médicale, 19th Aug. 1903, p.592.
(Sur une forme de Ptosis Non Congénital et Héréditaire.)
recently Forsberg reported thirteen cases of hereditary ptosis coming on in adult life in five generations of a family of sixty-seven individuals, which he observed at Sioux Falls, South Dakota. Of these thirteen cases of ptosis, two appeared at the ages of six to eight years; four at the ages of ten to twelve years; three at the ages of fourteen to seventeen years, and four at the ages of twenty to twenty-two years.

One ancestry was formed by the marriage of a woman from Michigan to a man whom she met there. Her husband later remarried. The other ancestry was begun in Wisconsin by the union of a man of Irish /
Irish descent and a woman of English descent. None of the five grandparents had ptosis.

Case 1 developed ptosis when he was fourteen. He lived to the age of seventy-three.

Case 2 had severe ptosis which developed at the age of seventeen. Her twin brother did not develop ptosis.

Case 3 developed ptosis at the age of twenty-two. It gradually came on over a period of two years and was getting worse each year.

Case 4 developed ptosis gradually when he was twenty-two. He could scarcely talk above a whisper.

Case 5 developed ptosis when she was eleven, and the condition had been getting worse for the previous twenty years. When she was thirty-one her voice began to trouble her, and this had gradually become worse. She also had keratitis marginalis.

Case 6 - Ptosis began when she was about twenty.

Case 7 - This patient showed, in addition to a severe degree of ptosis in both eyes, paralysis of the orbicularis, no movement of the frontalis, and a certain degree of exophthalmos. Movement of the eyeballs in all directions was limited. The pupils reacted to light and accommodation. The nasal portions of the visual fields were distinctly contracted. The vocal cords were seen to approximate normally.

Case 8 - In this patient ptosis began at the age of twelve.
Case 9 - This patient had three children, a boy of twenty-two, a girl of seventeen, and a girl of ten. The last two were developing ptosis.

The cousins of the second generation may be divided into two groups. A - Those who could have received the condition from one source only. B - Those who might have received it from both sources. It is interesting to observe that four out of seven in the first group and four out of six in the second group were affected.

Of the total males, six were affected and eighteen were not. Similarly, of the total females, six were affected and eighteen were not. The sex of the remaining offspring was not known.

Forsberg's family is remarkable in that the condition was transmitted through unaffected persons. In one case an unaffected female transmitted the condition to a male child. In another case an unaffected male transmitted it to two male and two female children.

The condition was transmitted through both males and females.

Forsberg's cases differ from most of the previously described cases of late hereditary ptosis in that the condition began in childhood or early adult life. Three of them experienced difficulty in speaking, and one of them had paralysis of the orbicularis oculi. Cases 5 and 7 strongly suggest a diagnosis of myasthenia gravis.
Meumann, in 1928, described a family whom he observed at Munich. With one exception, all members of this family, who had reached a certain age, and who could be adequately investigated, suffered from ptosis which developed in middle life and was of a gradually progressive nature. In the pronounced cases the ptosis had developed to such an extent that it was impossible for the patient to raise the eyelid without external aid.

Meumann's Pedigree.
The first case observed (Case 28 in the Pedigree), was a man who had marked bilateral ptosis which had come on between 40 and 50. He considered that the condition was transmitted through his great-grandmother (A), his great-grandfather (I) not having been affected with the condition. Of the family of A the living members were too young to have developed ptosis, but he knew a number of them who had died and all of whom had suffered from ptosis. E had had normal lids, but his grandfather C had had to lift up his lids with his fingers in order to be able to see.

In this family the age of onset of the ptosis was after 40, and in cases 33 and 30 it was slightly apparent at the ages of 30 and 24 respectively. As Meumann points out, it is probable that the patients did not notice the ptosis until it was of a sufficiently marked degree to interfere with their vision. It would thus appear that the age of onset in Meumann's family was considerably earlier than in the family described by Boulanger, in which ptosis did not appear until after 50. On the other hand, there is no doubt that in Meumann's cases the condition was not congenital. In many of the small children in the descendants of Nos. 19 and 26 no trace of ptosis could be detected.

No.s 19, 21, 25, 26, 27, 28, 30, 33 and 35 were personally examined by Meumann.

No.s 2-14, 20, 22-24, and 29 had ptosis in later life.
No.s 31, 32 and 35-42 were too young to show the condition.
Case 27 was 52 years of age, and had no ptosis although his father had marked ptosis.

No.s 19, 30 and 35, the two grandchildren of No.19, and the sister of No.35 had strabismus.
Beaumont in 1900, described a family who showed the most remarkable condition of hereditary non-congenital external ophthalmoplegia, beginning between the ages of 20 and 30. As far as can be ascertained, this is the only case of this type that has been described.

Beaumont's Pedigree.

* = Eleven children - sex unknown.
† = Sixteen children - sex unknown.

Case 1 was the first member of the family known to have had ptosis. It was not known whether she had paralysis of any other ocular muscles.

Case 2 /
Case 2 had ptosis, and reached the age of 90.
Case 3 had ptosis, and reached the age of 85.
Case 6 has also been described by Lawford (Trans. Ophth. Soc. U. K. Vol. 7, 1887, p.260). Ten years prior to consulting Lawford the patient had noticed that his upper eyelids were beginning to droop. This had become gradually worse, and when he was seen by Beaumont thirteen years later, i.e. twenty-three years after the onset of the condition, he had complete paralysis of the levator palpebrae superioris on both sides, and there was very little action of any of the recti. He could not say when the loss of movement of the eyes began, but thought it came on simultaneously with the ptosis. The light and accommodation reflexes were normal, perfectly and he was otherwise healthy. He had had no headache, vomiting, fits or paralysis elsewhere. None of his five children had ophthalmoplegia up to the time of examination by Beaumont. Lawford gave him potassium iodide in gr. 810 doses for five months without the slightest amelioration of the condition.

Case 7 began to have bilateral ptosis when he was 30 years of age, since when it had been slowly progressive. His lids almost completely covered his pupils, and only the very slightest movement of the eyes in any direction was possible. The light and accommodation reflexes were normal, and he had never had syphilis. Beaumont remarks that this patient's orbicularis oculi muscles were very feeble.

Case /
Case 8 had sixteen children, none of whom were affected.

Case 9 had ptosis and external ophthalmoplegia, which had begun at the age of 39.

Case 10 consulted Beaumont at the age of 38, and stated that her ptosis had been coming on for ten years. Richardson Cross had seen this patient eight years previously, and the ophthalmoplegia had then been well marked. She could move her eyes moderately well inwards, outwards and downwards, but poorly upwards. Her chief complaint was that she had to turn her head to look at anything not in the direct visual axis. She was otherwise perfectly healthy and normal.

Case 11, aged 30, showed definite symptoms of commencing external ophthalmoplegia, although she was unaware of having anything wrong with her eyes. The eye movements were limited in all directions.

In this family one sister had epilepsy.

Case 12 was one of the sixteen children of Case 8. Her photograph showed ptosis, and some paralysis of ocular muscles was said to have come on recently.

Lawford thought that the lesion in the case described by him was central, and suggested that it had involved the nuclei of origin of the nerves to the external muscles, leaving the nuclei concerned with the light and accommodation reflexes intact.
Cases of Congenital Ptosis may be divided into the following Categories:

1. Those in which the palpebral aperture is abnormally small, preventing the eyelids from being fully opened. The condition may be accompanied by epicanthus, and there may also be a congenital malformation of the os sphenoidale. There is rarely external ophthalmoplegia, although paresis of the rectus superior may be present owing to lack of use of that muscle.

   In this Category it is rare for the condition to be unilateral.

2. Those in which the palpebral aperture is of normal dimensions, and the ptosis is due to paresis of the levator palpebrae superioris. In the majority of such cases there is also external ophthalmoplegia and the commonest cause is maldevelopment of the levator and of the other external ocular muscles involved.

   Such cases may be unilateral although the unilateral is much less common than the bilateral form.
3. Cases due to birth-injury. These are generally unilateral.

4. Cases of Jaw-Blinking, or the Phenomenon of Marcus Gunn. There are always unilateral.

Ptosis of the upper eyelids may supervene in later life, and this is sometimes hereditary. It may be an isolated symptom, or it may be accompanied by chronic external ophthalmoplegia. The aetiology of this condition is unknown, but it is suggested that it may be due to a dystrophy of the levator and external ocular muscles.