

A STUDY OF PROGRESSIVE MUSCULAR ATROPHY

WITH

SPECIAL REFERENCE TO THE IDIOPATHIC FORM

AND

CLINICAL NOTES OF TWO CASES OF THE LATTER VARIETY.

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BEING THE THESIS SUBMITTED FOR GRADUATION

BY

DAVID ROBERT DOW, M.A., M.B., C.M., EDINR.

1895.



_____ A SHORT SKETCH OF THE HISTORY OF _____
PROGRESSIVE MUSCULAR ATROPHY
OF
MYELOPATHIC ORIGIN.

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A Short Sketch of the History of Progressive Muscular Atrophy of Myelopathic origin.

The subject of Progressive Muscular Atrophy has been a fruitful source of discussion amongst Neurologists since the middle of the present century when Cruveilhier, Aran and Duchenne first described it. At intervals during that time many clinical and anatomical observations have been recorded and these, aided to a certain extent by the results of such physiological experiments as those of Yeo and Ferrier, have been the means of considerably subdividing what was originally an apparently homogeneous group.

Under the head of Progressive Muscular Atrophy was included a number of forms having the common characteristic of Atrophy but due to various causes.

Cases of the disease were published by some of the older writers, as Cooke (1), Abercrombie (2), Bell (3), Parry (4), and Dubois (5), but it was not till 1849 that it became recognised as a distinct type of disease. Duchenne (6)* gave the first systematic account in 1849, Aran's (7) Memoir followed in 1850, and three years later Cruveilhier (8)† read a paper upon it before the Académie de Médecine. The latter however had described it in his lectures for several years.

There at once arose considerable difference of opinion as to the Morbid Anatomy and Pathology of the disease. Cruveilhier (9)†† found no disease of the cord in his first autopsy, as he expected, but in 1853 he found the anterior roots of the spinal cord consider-

*Duchenne- Atrophie Musculaire avec transformation graisseuse
Memoires de l'Academie du sciences 1849.

†Cruveilhier- "Sur la Paralyse Musculaire Progressive

††Cruveilhier, atrophique." Arch. Gen. de Med. 1853.

†† Cruveilhier-Loc cit.

A of the cervical enlargement of the cord increase

ably atrophied and similar changes have been described by Reade (10) Duménil (11), Chareot and Joffroy (12). The anterior nerve roots however, have been found normal by Clark (13) Fromman (14) and others so that we must look upon this as secondary, and not the essential morbid alteration as Cruveilhier supposed.

Duchenne then concluded from this inconstancy that the disease was one primarily of the affected muscles and this opinion was shared amongst others by Aran and Virchow and even as late as 1873 Friedrich (15)† in an able monograph strenuously defended the myopathic origin of the disease. He considers the disease to be a primary chronic myositis, the *intra* muscular nerves being secondarily implicated and that there is an ascending neuritis, but a fatal objection to this theory is that we may have the most advanced Atrophy in muscles with the peripheral nerves and anterior nerve roots normal; moreover the ascending neuritis has not been shown to exist. However, when we come to speak of the Myopathies it will be seen that there is some justification for the view maintained by Friedrich.

The theory that the disease was due to a lesion of the Sympathetic was advanced in 1854 by Schneevogt (17) who found it diseased in a case examined by him. Jaccoud and Duménil also found it affected but Hayem, Vulpian and Chareot have found the Sympathetic perfectly healthy, and, therefore, the Pathogeny of Progressive Muscular Atrophy cannot be explained by a lesion of this part of the nervous system.

We now come to the last theory given forth as the cause of the disease.

Valentiner in 1855, Schneevogt and Fromman had observed changes in the upper part of the Spinal cord but Luys (17)† was the first to demonstrate the morbid changes in the grey substance. His patient with advanced atrophy of the muscles of the left hand and forearm having died, there were found in the grey substance of the capillary vessels and thickening of their walls—a large number of the ganglion cells of the anterior cornua had disappeared in the

*Friedrich—"Ueber Progr. Muskelatrophie." Berlin 1873.

†Luys "Gaz. Med de Paris" No. 32. 1860.

part of the cord affected and were replaced by granular masses. The degeneration affected chiefly the left anterior cornu, corresponding with the seat of the muscular atrophy. The left anterior nerve roots were also atrophied.

Lockhard Clarke, in 1861 and 62 (18) described the existence of atrophy of the cells in the anterior cornua in three cases. Haymen reported the result of the Examination of the cord in another case. These observations were confirmed by similar ones from examinations made by Chareot and Joffroy (19), Duchenne and Joffroy (20) Prévost and David (21), Pierret and Troisier (22). Chareot in particular was a strong supporter of the spinal origin of the disease; and chronic degeneration of the anterior horns of the grey matter with destruction of the ganglion cells was laid down as the essential cause.

In support of this view we have the constancy of the changes in the ganglion cells and the fact that acute lesions of the anterior horns of the grey matter will cause atrophy of the muscles.

Associated with this degeneration of the cells we have a degeneration of the white matter, indeed Schultze (24) expresses the opinion that only two autopsies (those of Chareot and Com-bault, and Pierret and Troisier) are as yet recorded where this was not the case. Gowers (25)* says that he has not met with a single case of Progressive Muscular Atrophy in which the pyramidal tracts were unaffected and that such cases are extremely rare; but that one has been published by Strumpell. Dr Leach published two cases in which were will marked changes in the motor ganglia and sclerosis of the lateral columns of the cord. Thus we have established a Myelopathic type of Progressive Muscular Atrophy of which the thenar is the most common form, that is, the atrophy commencing in the small muscles of the hand, the thenar and hypothenar particularly. We may however have irregular forms in which the atrophy begins in other muscles such as those of the shoulder, serratus magnus, the arm and neck muscles.

The commencement of the disease in the small hand muscles has been elucidated by the experiments of Professors Ferrier and Yeo† on monkeys which show that the small hand muscles are connected

*Gowers "Nervous System 1892.

†Ferrier and Yeo. Proc Roy Soc 1881.

"Localisation Atrophic Paralysis." Brain 1881.

with centres in the dorsal cord at about the level of the second dorsal roots. The other features of the disease are:-

- (a) the progression of the wasting to the rest of the limb, to the muscles of the trunk and lower extremities.
- (b) the onset of the disease after twenty one
- (c) presence of fibrillary twitchings
- (d) certain contractures giving rise to the well known claw shaped hand
- (e) diminution of faradic irritability in proportion to the wasting, ^{and} the late development of the reaction of degeneration.
- (f) the slight tendency to heredity.

PROGRESSIVE MUSCULAR ATROPHY

of

MYOPATHIC ORIGIN.

Progressive Muscular Atrophy

of

Myopathic Origin

All the forms of Progressive Muscular Atrophy came to be included under the Myelopathies; but the investigations of the last ten years have been the means of separating from that group a large class of cases clinically resembling but pathologically distinct from them.

This is the class of atrophies in which the seat of the disease lies solely in the muscles themselves, which is independent of all lesion in the nervous system, central or peripheral, and to which the names of Myopathic Atrophies, Muscular Dystrophies and, in this country, Idiopathic Muscular Atrophies have been applied.

One of the earliest recognised forms of progressive muscular atrophy of Myopathic origin is the well known Pseudo-hypertrophic muscular paralysis. Cases of what we know to be examples of this disease were described by ^{the} older writers. It was first described by Greisinger but Duchenne (26) was the first to thoroughly investigate the nature of the disease and to consider it as a pure myopathy.

A series of cases was published by Meryon (27) and Oppenheimer and they considered it identical with "Progressive muscular atrophy".

Friedrich as already stated, judging from the results of his autopsies, looked upon all forms of progressive muscular atrophy as of Myopathic origin, and regarded the Pseudo-hypertrophic paralysis as identical with it, and his view was to a certain extent justified when in 1883, Erb, (28) described at the congress at Fri-

bourg, his variety under the name of the "Juvenile form" of Progressive Muscular Atrophy, based upon the clinical observations of 20 cases, and since proved by the autopsies of Schultze and others to be apparently unconnected with any lesion of the nervous system.

Another variety recognised by Landouzy and Déjérine (29)* in 1885 as a pure myopathy is that described by Duchenne (30) in 1855 as the "Infantile Form of Progressive Muscular Atrophy" so called because it always begins in early childhood with affection of the muscles of the face. Landouzy in 1874 recorded the cases of two brothers suffering from the disease and was able to watch the cases for a period of ten years. This observer along with Déjérine showed that in this disease there exists a simple Atrophy of Muscles and not an Atrophy of Myopathic origin as Duchenne believed. The first observation of Progressive Muscular Atrophy with participation of the muscles of the face is really due to Cruveilhier (31) who in 1853 described the case of Legrand, and the autopsy on his body showing the nervous system, both central and peripheral, to be intact.

Remak (32) Mossdorf (33) Trosier and Guinon (34) Marie and Guinon† and others have recorded cases of this disease.

Leyden (35) proposes another clinical variety under the head of "Hereditary Progressive Muscular Atrophy" to which he assigns the following characteristics:-

- (a) Invariable heredity
- (b) Commencement in early childhood, in the muscles of the lower extremities and the back
- (c) Male sex chiefly affected
- (d) Hypertrophy of the muscles of the calf and sometimes of other Muscular groups.

Möbius reunites this hereditary form to the ~~pseudo-hypertro-~~

*Landouzy and Dejerine- Rev. de Med. 1885

† Marie and Guinon Rev. de Med. 1885.

~~phic form~~ to the pseudo-hypertrophic form of which it is only a variety and thus became established the Leyden-Möbius type.

As however, heredity is an important feature in pseudo-hypertrophic paralysis, Erb's form and the peroneal type of Atrophy to be mentioned later;— and as we may have undoubted examples of these varieties commencing in the lower extremities, another variety can hardly be established on these grounds.

"ACCOUNT OF TWO CASES

of

MYOPATHIC ATROPHY

OCCURRING

IN THE AUTHOR'S PRACTICE

WITH

FAMILY HISTORY."

Account of Two Cases Of Myopathic Atrophy, Occurring In The Author's Practice, With Family History.

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Before proceeding to give a more detailed description of the varieties of Myopathy, I propose to narrate the family history of two cases which in my opinion belong to the group of Myopathies.

The family history goes back to the great Grandfather, whom the father of my patients distinctly remembers, stating that "he had a withered hand and arm", and though not very robust, was healthy and able to move until he died at the age of 72.

The great grandmother was a hale, hearty, active energetic woman, who lived to be 80 years of age.

The children of the great grand parents were four in number:-

J.C. The paternal grandfather of my patients;

T.C. The maternal " " " "

G.C. A son, who was not very strong and died young; and

A.C. A daughter, who was a little wiry, very active woman, dying at the age of 96.

J. C. The paternal Grandfather, was a healthy, ordinarily muscular man. At the age of 45 "he took a fit of some sort, which the Doctor called a rush of blood to the head." After this attack he grew weaker, and complained much of giddiness, though still able to work a little. About two years later he died from a third attack.

The Maternal Great Grand Mother was not strong, and died comparatively young leaving only one child.

The Paternal Grandmother died of what was then called a "decline", some months after the birth of her sixth child-whether it was really consumption, or merely a gradual decrease of strength, my patient does not know.

The Paternal Grand Parents had six children, three sons and three daughters. Two of the sons died in childhood. Of the surviving children:-

* The Maternal Grandfather, a brother of the Paternal, was a stout, able-bodied man. For three years prior to his death he suffered from a growth at the back of the neck - ' at the base of the skull ' (sic) - which was removed; but the part never healed.

- (1)- M. C. when young was fairly strong and healthy but about 12 or 13 years of age she received a wound in the foot, which suppurated and never healed. Sores also broke out upon the leg, hand and cheek. Hence she was more or less constantly confined to bed, and died aged 50.
- (2)- S. C. a strong, active woman, always healthy, and still alive, aged 85.
- (3) E. C. fairly strong when young, but became less robust some years after marriage, and suffered from disease of the heart, of which she died, aged between 50 and 60.
- (4) J. C. the father of the patients, was born in 1811. He had always been healthy, strong and hardy. In 1834 he had fever in Batavia, and in 1835 was very ill with scurvy. In 1838 he had yellow fever at Demarara. In 1841 he married, and a few months later had small pox. From that time he has enjoyed excellent health, and does so still at the age of 83.

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The Maternal Grandmother was a little, stout woman very muscular. All through her life she was subject to sick headaches, and complained of her back for many years before her death at the age of 95

The Maternal Grand parents had three children, two sons and a daughter:-

- (1)- One son died when 3 or 4 years old
- (2)- a son, is still living, aged 80. He has been very healthy, but now suffers from a weak heart.
- (3)- the mother of the patients- N. B. It will be remembered that the parents of the patients were cousins-When still a young woman she hurt her back, while doing some heavy work, and ever afterwards complained of it. She had small pox at the same time as her husband, some months after marriage and while pregnant with first child, who will be referred to immediately. She was very subject to headaches, and suffered from pains in the side. She was, however, fairly strong and muscular, and died of aortic disease at the age of 72.

She had nine children:-

- (1) F. C. could walk when very young, but with a "rolling, rocking

thighs had lost "their roundness and become very thin in front" though the lower parts of the legs were still full and round. What his arms were like at this period he cannot remember. He did not, however, notice much of any deficiency in them, though he recalls that his shoulders were thin. By the time he left school and entered an office at the age of 14, all these difficulties were increased. He could not then run at all, but could walk as well as ever, and used to take a great deal of walking exercise, suffering from cramps when he over-did it. A year or more after joining the office, he has, so he says, a second attack of scarlet fever, and he thinks that, following on this, he noticed that the upper parts of his arms were thinner than they should have been. For many years he continued to suffer from cramps in the calves and behind the knees after prolonged walking, and, indeed, he is still affected in this way. Before his apprenticeship of four years was over he found that the difficulty of going up steps was increasing, likewise that of rising from a seat or walking uphill against the wind. His leg power, in a word, was growing less. From this time onward during the twenty odd years he was in City Offices he remarked a continual loss of muscular power, but not in any marked degree at any particular time. He took regular walking and dumb-bell exercise, but nevertheless failed to restore or maintain his strength. He grew very bilious when in town, and his appetite was never very good; yet on the whole he enjoyed fair health. He used to "firm up" during his holidays, but invariably lost ground throughout the Winter. He worked as long as he was able, but found his occupation growing too heavy for him, and ere he gave it up he felt as if he would collapse altogether. After his return home he improved a good deal, and though still losing strength notes that he is not doing so^{as} rapidly as when he was in business. He is now 48 years of age.

- (9) G. C.—the other patient, is the second youngest in the family. At the age of 13 or 14 he first found a difficulty in running down a steep hill, but had otherwise no trouble in running as fast as his companions, and could jump as far and as high as any of them. When about 16 the running power was greatly

modified, but could still be exercised "in a sort of a way" He was sensible of no other deficiency. His shoulders and upper arms had become thinner, but he felt no want of power therein. Some three years later he began to experience difficulty in rising from the ground (indeed, one frosty day he found that he could not rise at all without help) and subsequently in getting upstairs and rising from a low seat. His arms now began to feel weaker. A few years passed, and he noticed a want of power in going downstairs, and while he could still walk and climb up a steep hill, he could scarcely descend without fear of his knees giving way. The arms gradually grew weaker, the back less strong, and he required help in rising from an ordinary seat. At the same time he was conscious of an ever increasing weakness throughout his entire muscular system. To-day the lower part of the arms, the calf and the legs are still strong and firm and he can still go up steps pretty well, though not so easily as before.

I have given the family history somewhat in detail, for it is interesting as showing that though an element of weakness runs through the family, many of its members have been very healthy and survived to a ripe old age, which would lead one to conclude that the morbid weakness in one ancestor has been neutralised to a certain extent by the excessive vigour and robustness of the other, as seen in the great grand-parents. Although there is no history of direct transmission of the disease, yet there is good evidence of a neurotic tendency, witness the great grandfather, who was affected with a withered hand and arm—the grand-father who died from cerebral mischief—the maternal grandmother and mother who suffered from sick headaches ^{one} of the sisters who was subject to neuralgia. The consideration of these facts would lead one to look upon the Atrophy as of spinal origin.

To sum up:—

- (1)—In the case of the great grandfather it is of course impossible to say whether the wasting was of Myelopathic origin or a local palsy.
- (2)—It is also possible that the paternal grand-mother may have been atrophic, as her wasted appearance would be likely enough to give rise to the idea of a "Decline". It is mentioned that she died of

- this decline some time after the birth of her sixth child. This is also an important point, as Chareot and Joffroy (36) have observed Progressive Muscular Atrophy occurring immediately after child-bed.
- (3) Another curious point in the history is the prevalence of affections of the heart, and in the case of the paternal aunt (E.D.) may not the symptoms of heart disease have arisen from the implication of the respiratory muscles? For it is stated that she gradually grew less robust.
 - (4) From the history of the paternal aunt (M.D.) having been confined to bed more or less during her whole life, one would be inclined to imagine that the injury to the foot had set up an Atrophy which gradually spread and left her almost helpless.
 - (5) It will also be noticed that the father suffered severely from fevers, and had there been any predisposition to Atrophy in his case it would have likely been developed by these external influences, for, as will be seen further on, Atrophy has frequently been noticed to follow acute infectious diseases. In the case of the elder of the patients we find the morbid process precipitated after an acute febrile attack, and after another attack manifesting itself in a different part of the body.
 - (6) It has been mentioned that the mother suffered from small pox during her first pregnancy, which resulted in the birth of an atrophic son; and perhaps this disturbance in the course of her gestation may have determined the defective evolution of the affected parts.
 - (7) A further point to be observed is that the father and mother of the patients were first cousins, and in this case at least we may look upon consanguinity as a factor in the development of the disease. Hence perhaps, the explanation of the appearance of the disease in the earlier members of the family, and of the fact that more than one member was affected. The reappearance of the disease in the younger members of the family may have been caused by the weakening tendency of frequent parturitions.
 - (8) The first child born of this marriage is evidently an example of Pseudo-hypertrophic paralysis, since at a very early age his gait is described as having been of a "rolling, rocking character," and for several years he could move about with the aid of crutches, until the disease had seized the muscles of the shoulder, which, as we shall see, are Atrophied sooner or later in the course of the

- malady. Lastly, his death at the age of 20, which is apparently the most fatal period in the life of a hypertrophic patient.
- (9) From T.C.'s description of his case when he speaks of his early difficulty in running and being easily knocked over, and of his being round and ruddy and as well developed as his playfellows, one would be apt to infer that he, too, was suffering from pseudo-hypertrophic paralysis; but such is not the case, as his calves even at the present time are well developed and he can raise himself on the balls of his toes.
- (10) The beginning of the atrophy in the elder brother was in the muscles of the thigh and hip, while in the younger, although he noticed the weakness in the legs first, the shoulder muscles were really first attacked as will be evident when the state of the muscular system in the two cases is described.
- (11) It is rather curious that the younger brother complained first of difficulty in going down a hill while he could go up steps easily enough, the conditions being reversed in the elder.
- (12) As is so commonly the case we find the disease affecting only the male members of the present generation.

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Present Condition

The elder Patient, T. C. is of average height and a healthy looking man, with a chest measurement of 38 inches, that of the abdomen being 34. There is nothing abnormal about the thoracic or abdominal viscera. Patient suffers from frontal headaches, but not so frequently now as at an earlier period of his life. His sight both for near and distant objects is perfectly good, his hearing, taste and smell are normal. So also his sensibility to touch, pain, heat and cold. He is a very intelligent man, and his memory is good. The sleep function is somewhat disturbed. Organic reflexes quite normal. Skin reflexes, plantar, epigastric and abdominal are present. The knee jerk is abolished on both sides, the ankle jerk can be elicited.

The condition of the muscles and their movements are as follows:-
 The muscles of the face are apparently unaffected as all the various movements are perfectly performed. The muscles of the neck are well developed and very powerful, probably from the fact that they are so frequently brought into action in assisting the patient to turn on his side while in the recumbent posture. The head can readily be flexed and extended and the sterno-mastoids act equally well on both sides.

The Atrophy of the muscles acting on the shoulder is masked by a considerable deal of obesity, especially in front of the chest, where the Atrophied pectorals are concealed under a thick layer of fat, so that the existence of the atrophy can only be detected by the feebleness of the movements when the muscles are brought into play.

Of the muscles moving the shoulder joint we can only prove the existence of the atrophy by the eye in the case of the Deltoids, which are small on both sides.

There is no marked hollowing over the Trapezius, nor in the inter-scapular region, and as the shoulder can be elevated with but slight effort, there cannot be much wasting of the Trapezii (upper parts) *Levatores Anguli Scapulae* or Rhomboids.

When the arm is slightly abducted and the shoulder moved forward, the Scapula at its lower angle stands out in a marked way from the chest wall, showing some weakness of the Serratus Magnus, which is further proved by the difficulty the patient has in crossing the arms in front of the chest.

There is a depression over the supra-spinous region, showing slight Atrophy of the supra-spinatus. This is not seen in the infra spinous region, and as the Infra-spinatus, and Teres minor ^{rotate} rotate the humerus freely out-wards, we may consider them ^{as} being only slightly affected. Hence the explanation that the patient can write as quickly and freely as ever, both these muscles being concerned in this operation.

The Latissimus Dorsi, Pectoralis major and Teres major are all atrophied to a considerable extent, as the patient finds it impossible to adduct the arm against very slight resistance, and while doing so, the inferior angle of the scapula projects.

The Subscapularis is also atrophied, for rotation inwards of the humerus is very feeble, and the patient cannot pass the extended arm behind the back. The Teres major and Latissimus are also rotators inwards. Elevation and depression of the shoulders are weakened to a lesser extent than are the other movements. Movement of the Scapula backwards is more forcible than that forwards.

Adduction of the upper arm is much weaker than abduction, which is strongest in the left, but the slightest resistance checks either movement, and during the former the Scapula projects. Though the arm can be moved both forwards and backwards, the latter movement is very weak. Rotation outwards of the arm is very well, and rotation inwards very imperfectly performed.

The Patient can raise the arm vertically by means of the Trapezius and Serratus magnus with some effort.

The Upper Arms are very small, the Biceps, Brachialis anticus and Triceps on both sides being markedly Atrophied and very flabby.

Measurements at the middle third are:-

Right, 8 $\frac{1}{2}$ inch: Left 8 $\frac{1}{2}$.

Movements:- Flexion is more powerful than extension.

The Muscles of the Fore Arm are atrophied and soft, the extensors being less affected than the flexors, over which there is distinct hollowing on both sides.

The Supinator longus and Extensor Carpi radialis longior and brevior are less wasted.

Measurements (over Supinator):-

Right, 8 $\frac{1}{2}$ inch: Left 8 $\frac{1}{2}$.

Movements:- Supination and Pronation are fair, but flexion is extremely weak.

Dynamometer

Right 5 Left 0

Muscles of the Hand: There is slight atrophy of the left thenar eminence; both hypothenars are wasted, and even during contraction the thenars and hypothenars on both sides are very soft. When writing the Patient suffers from cramp in the muscles of the hand. (writers cramp)

The Muscles of the Back and Abdomen are in good condition.

The Gluteal Muscles ^(maximus) are atrophied ^{and} very soft.

The Muscles of the Thigh are extensively wasted. The Quadriceps extensor on either side is much Atrophied, and there is a falling in of the inner and upper part of the thigh, showing marked atrophy of the Adductors.

The Flexors (Psoas and Iliacus etc.) must also be atrophied as Flexion is almost in abeyance.

At the back of the thigh, the Flexors of the Knee are firm and do not appear to be affected to the same degree as the other thigh muscles.

Measurements of the thigh a little above the knee:-

Right $12\frac{1}{2}$ inches: Left $12\frac{1}{2}$ inches.

Adq. upper part:-

Right $14\frac{1}{2}$ inches:- Left $14\frac{1}{2}$ inches

Movements at the Hip-joint:- The extension of the hip is very weak, owing to the atrophy of the Gluteus maximus, and hence the difficulty experienced by the Patient in going upstairs. The power to stoop is almost lost, for if the Patient ventures the body beyond a certain angle, he invariably falls forwards. There is no Lumbar Lordosis, which one might expect to find if this were due in all cases to weakness of the Extensors of the hip, so that there can be little doubt, that, in some individuals at least, weakness of the Spinal muscles may be assigned as the cause, as suggested by Duchenne.

Adduction of the thigh is lost to such an extent that the Patient can only approximate the legs by rotating the thighs outwards and inwards aided by flexion of the toes, the foot being ^{moved along} fixed to the ground.

Flexion of the thigh is very feeble indeed, and when lying on his back in bed with the legs fully extended, Patient has no power of flexion. When sitting with the knees flexed, he cannot lift the feet from the ground.

Owing to the absence of these movements the power of crossing one leg over the other is completely lost.

There is considerable power of abducting the limb.

Rotation outwards is weakened in a lesser degree than rotation inwards.

Movements of the Knee:- Extension is distinctly weak, but flexion is good, and hence the great assistance the flexors give as extensors of the hip-joint.

The Muscles of the Legs:- Are perfectly normal and the calves are hard and well formed.

Measurements:-

Right, $13\frac{1}{2}$ inches :- Left $13\frac{1}{2}$

There is thus a difference in the volume of the muscles of the legs, which may be accounted for by the strain being so frequently thrown on the left leg in rising from the sitting posture.

All the movements at the ankle are performed without difficulty.

Fibrillary twitchings are present, particularly after exertion and sudden exposure to cold, and patient states in illustration of this that they had been lately very marked after going to bed, even the muscles of the

calf being affected.

The Patient G.C. is of Medium height, dark complexion and, his Atrophy notwithstanding, very healthy. The internal organs appear to be perfectly normal.

Circumference of chest 34 inches, of abdomen, 31 inches.

Intelligence good; Memory not so strong as it was two years ago.

Sleep function somewhat disturbed

Sensibility to touch, pain, etc., normal

Sight not so good as it used to be, the print becoming somewhat blurred after he has read for some time. At the same time there is nothing abnormal to be observed in the eyes themselves.

Hearing, taste, and smell unimpaired.

Organic reflexes, plantar, epigastric and abdominal are present.

Deep reflexes, knee jerks equal and of average strength. There is a lively ankle-jerk.

The muscles of the face and neck unaffected

The muscles of both upper limbs are small

The wasting is more marked in the muscles of the shoulder and upper arms than in the forearms and hands.

Pectoralis major small with the sternal part more atrophied than the clavicular on each side.

The Deltoids are also small, but both they and the greater Pectorals are firmer than the muscles of the arm.

In marked contrast to that of the brother, the atrophy around the Scapula is very evident.

The Trapezii while little affected in the upper part, are considerably diseased below.

There is a marked hollowing over each Supraspinous region, particularly on the right side

The Infra-Spinati are only slightly affected.

There is a depression observable over the, Teretes which are wasted.

The Rhomboids are much atrophied

The posterior borders of the Scapulae project, leaving a distinct hollow between.

The Latissimus Dorsi and Serratus Magnus are fairly strong on each side.

Movements of the Upper Arm at the Shoulder are in no way diminished in range. Abduction is very weak, and so also is adduction, while if the arm is raised to the level of the horizontal and ~~is~~ brought down ~~is~~

against some resisting force, the lower angle of the Scapula is turned strongly outwards and approximated to the humerus, owing to the absence of the usual fixation of the Rhomboids and Trapezius. Rotation outwards of the arm is much weaker than rotation inwards. Forward movement of the Scapulae is diminished to a less extent than that backwards. In the brother the reverse of this obtained.

The Upper Arms are very small.

The Biceps is small in each arm, and the Brachialis Anticus is more atrophied

The Triceps is small, soft and weak, much weaker, indeed, than the Biceps
Measurement-at middle third-

Right 8½ :- Left 9

Movements:- Flexion and extension of the elbow are very weak, especially the latter.

The Muscles of the Forearm do not exhibit marked Atrophy, but are very soft. The flexors are more affected than the extensors.

The Supinator longus and Extensor Carpi

Radialis longior and Brevior are less affected than the other muscles.

Measurements:-

Right 9 :- Left 9½

The left hand is stronger than the right as shewn by the Dynamometer:-

Right 40 Left 50

The Thenar and Hypothenar Eminences are of normal size but very soft, even during contraction of their muscles.

The muscles of the Back and Abdomen are unaffected.

Fibrillary twitchings are present in the muscles of the upper limbs, especially those of the shoulders, and this occurs more readily on exposure to cold.

Lower Limbs:- The muscles here are not wasted to nearly the same extent as those in the upper limbs.

The Gluteals are fairly well developed and firm.

Both Thighs are somewhat atrophied, the Quadriceps extensor being affected on both sides, as also are the adductors and flexors. At the back of the thigh the Flexors of the Knee show but little appearance of atrophy and are very firm. This excess of development of the Flexors of the Knee

ELECTRICAL EXAMINATION

CASE OF T.C.

FARADISM:-

In the upper part of the body the muscles acting on the shoulder react to strong currents. Next in order, the Deltoid, Biceps, Brachialis Anticus and Pectoralis all react to currents of about the same intensity. The Triceps requires a stronger current (4 cells more). In the fore-arm the Supinator Longus reacts more readily than the extensors and flexors (3&4 cells more, respectively). The Thenars react to weaker currents than the Hypo-thenars (a difference of 3 cells).

In the lower limbs the Gluteus Maximus and Quadriceps Extensor react to very strong currents. Next come the adductors and flexors, the latter reacting to a weaker current than the former (3 cells less).

GALVANISM:-

The Supra-Spinatus, Serratus and Latissimus show the highest resistance to the constant current, the Deltoid and Biceps to a less extent. In the lower limb the order is Gluteus Maximus, adductors, and, finally, Quadriceps.

CASE OF G.C.

The muscles on the left side react more readily than those on the right to both currents. The Supra-Spinatus and Triceps require much stronger currents (both faradic and galvanic) than any of the other muscles in the upper part of the body.

In the lower limbs the Quadriceps and adductors react more feebly than do the Gluteus Maximus and flexors of the knee.

NOTE:- In both patients electrical contractility, though diminished, persists in all the muscles; but in none could the Reaction of Degeneration be made out.

over the Extensors explains the inability of the Patient to descend a stairs, or steep road without great difficulty.

Measurements:- (at upper part)

Right 17 inches:-Left 18 inches.

Movements of the Hip-joint:- These are principally diminished in the direction of flexion and adduction. Less so in that of extension.

The Patient cannot cross his legs without the assistance of his hands. To do so he lifts his leg by laying hold of the lower part of the trousers. Extension of the knee is considerably weakened while flexion is remarkably good.

the Muscles of the Legs are well developed and firm.

Measurements:-

Right 15 inches:- Left 15 inches.

All the movements at the ankle-joint are well performed.

Fibrillary twitchings are seen in the muscles of both thighs

It will be observed in this patient that the volume of the muscles, both in the upper and lower limbs, is larger on the left side.

Diagnosis:-

The diagnosis of these cases is based upon the following grounds:-

- (1) The fact that more than one member of the family is affected with the disease.
- (2) The age at which the disease began (in the one case in childhood, and in the other in early youth) it being very rare to find spinal atrophy beginning under the age of 20, and still more so before the age of 10. Indeed, when both these characteristics are found to be present, it is reasonably certain that the atrophy is idiopathic.
- (3) The point of attack being at the proximal ends of the limbs.
- (4) The simple diminution of electric irritability to both currents, and the absence in both cases of Reaction of Degeneration.
- (5) The slow progress of the disease (in the case of the elder patient a period of forty four years) without the appearance of any distinctive spinal symptoms—such as excess of knee jerk which results from lateral sclerosis. Indeed, the only point, which the cases have in common with atrophy of spinal origin is the existence of Fibrillary Twitchings, which are

^ to the right side and is firmly pressed against-

^ so that the sole is firmly planted on the floor. The body is then gradually pushed re-wards

rarely if ever, absent in Myelopathies.

To make a distinction, these partial contractions of muscle where present in Myopathies may be considered to be of local origin, while in Myelopathies they indicate a disturbance of the centre.

Peculiarities in the Movements of the two Patients:-

There is nothing very remarkable in the attitude of the Patients except that the shoulders are thrown well backwards. There is no marked projection of the abdomen, nor any lumbar lordosis.

The manner in which the elder patient rises from the recumbent posture is very striking. When lying in bed on his back he first of all turns upon his side, which he accomplishes by pressing the balls of the toes of the left foot against the front of the right foot (i.e. extending the left foot against the right which is flexed) by which means the left side of the pelvis becomes raised. Next the left shoulder is brought forward (sometimes assisted by the left hand) till the Patient is fairly on his side. He then rolls on to his face with the right arm flexed against the front of the chest and at the same time extends his head as far as possible, the weight being thrown upon the chest and abdomen. His next endeavour is to get the arm from underneath the body and to raise the right shoulder high enough to allow him to extend the right arm. By pressing his face upon the pillow he raises the shoulder high enough to allow him to place the chin in the palm of the ^{right} hand, and by a series of jerks, to the right, on the point of the elbow, gradually brings the weight of the body to bear upon the left elbow. The right arm is then extended in front and a little ^{to} the head of the bed (underneath the pillow) by which means the shoulders are raised to a certain height. The left arm is then extended and thus the body is pushed over on to the right hip. While the weight of the trunk is on the left arm, the right is drawn nearer to the body, so that the trunk is raised still higher, then with a push from the right arm, aided by the extensors of the hip on the left side, and by the rotators outwards of the thighs the trunk is extended, and the Patient attains a sitting posture. The legs are then lifted and hung over the side of the bed, and with the arms extended behind him, the Patient pushes the hips to the edge of the bed and so gets the feet to the floor. He next allows the weight to fall on the right hip, extending the right arm and the left leg, with the foot turned inwards, and at the same time rotated upon the left hip joint, so that the right leg can be drawn back underneath the Patient, and then rotated outwards, so as to bring the front of the right thigh against the side of the bed facing the latter. The left leg is now brought into line with the right and slightly abducted, and is also pressed against the side of the bed.

Finally, by pressure on the bed with the right arm, aided by the extensors of the hip, the patient attains the erect posture.

In sitting down the Patient seizes the arms of a chair and thus lowers himself, but even with this aid the trunk falls somewhat suddenly.

Rising from the sitting posture:- Patient uses a large arm chair. With the right hand he lays hold of the front part of the right arm, and with the left the back part of the left arm with the body slightly rotated in this direction, and the elbow flexed against the back of the chair. Then by pushing the latter with the point of the elbow, aided by the right hand, the hips are brought to the edge of the chair. The legs are then extended to the front, and slightly to the left, when, the arms being extended, the body is so far raised, and the legs are drawn towards the chair and kept extended. The body is then pushed over by the left arm until the weight is thrown upon the right arm, and the left arm slipped forward to the front of the arm of the chair. The left leg is slightly abducted, which brings the weight on to the left arm, and the trunk is then elevated by a kind of rotatory movement, the body being drawn first to one side and then to the other, the extensors of the body on the thigh being aided by the abductors of the latter.

In ascending a Stair the Patient lays hold of the rails with the left hand and flexes the arm so that the forearm rests against them. The weight of the body is then allowed to fall to the left side, the right leg abducted far enough to enable the right foot to be placed on the step, and then with a push from the left arm the left side of the body is raised and the foot placed upon the step.

The gait of this patient (P.C.) has an oscillating character the body being drawn alternately from side to side. His difficulties are caused by the weakness of the flexors of the thigh and the extensors of the knee. The patient, therefore, keeps the toes of the passive leg clear of the ground by dorsiflexion of the foot, and when the leg is to be moved forward the body is dragged over to the side away from the passive leg (i.e. the leg off the ground). The cause of this movement has given rise to considerable difference of opinion. According to Duchenne it is due to weakness of the Gluteus Medius muscle. "This muscle normally" says Cowers "counteracts the tendency of the pelvis at each step to incline towards the leg which is off the ground; and if the muscle is weak, the weight of the body

has to be thrown further over the supporting leg than in health, and hence the oscillating gait". Ross however, believes that the gait instead of being due to paralysis of the Gluteus Medius is necessarily effected by contraction of the muscles. The present case bears out Ross's view for the patient can abduct his thigh with great freedom when reclining on his side (action of the Gluteus Medius) and the muscle can be felt contracting while doing this. Therefore, we may consider that this muscle drags the body over the active leg by its contraction. The thigh of the passive leg is next flexed on the body aided by contraction of the abductors (on the same side) which also at the same time assist the extensors of the knee in throwing the leg forwards.

The younger patient has also certain peculiarities in movement, for example in rising from the recumbent posture, when possible he ~~was~~^{uses} surrounding objects as a means of drawing the body upwards by the hands. When he has nothing to take hold of he goes through the following series of movements to attain the erect posture.

If lying on his back, the patient can get into the sitting posture without any great difficulty. Next, putting the weight of the body on the left arm, which is extended, he draws round the right leg and flexes it under him; then transferring the weight to this knee he flexes the left leg at a right angle in front of him. The body is next thrown forward on to the arms extended in front and slightly to the right side. The left leg is now extended as far as possible, then by drawing the hands along the floor towards himself the right leg is extended, and finally, with a push from the hands, he aids the extensors of the hips in bringing the body into erect posture with the feet far apart. His method resembles that adopted by pseudo-hypertrophies in getting up, except that he has not reached the stage when it is necessary to climb up his thighs. The elder brother, however, required to adopt this method at a very early stage of the disease.

When about to sit down on a low seat the patient G.C. steadies himself by the hands placed on the seat of the chair or he would go down suddenly. In rising off a high seat he bends the body slightly forwards^{and} with a jerk attains the erect posture, but in getting off a low seat he extends and abducts the left leg; Then by pressure with both hands in the chair pushes himself up. Patient can ascend a stair with very slight assistance from the hands^{and} only

when getting on to a high step does he require to pull himself up. Descending a stair is more difficult and while doing so he keeps the legs extended, each foot reaching successive steps with a soft^{of} thud

There is nothing very striking in his gait. The flexors being so weak the knee does not bend much in walking, and no doubt his Gluteus medius (anterior part) helps to throw the leg forward.

When asked to pass the arm behind the back both patients first rotate the upper arm inwards as far as their limited power will allow, then flex the forearm to a right angle and finally push it across the back.

We will now proceed to point out, *seriatim* (1) the clinical features of the three principal varieties of Myopathic or Idiopathic Muscular Atrophy. Then take up together (2) the pathological Anatomy, as the same essential morbid process^{es} are at the root of each (3) the Etiology, as most of the factors are common to all three, and finally, their prognosis and treatment.

CLINICAL FEATURES

OF

I. PSEUDO-HYPERTROPHIC MUSCULAR PARALYSIS

(LIPOMATONS MUSCULAR ATROPHY)

Clinical Features of

I. Pseudo-Hypertrophic Muscular Paralysis.

(Lipomatous Muscular Atrophy)

This disease is essentially one of childhood generally manifesting itself between the ages of two and seven by an impairment of power in the lower extremities. The child is clumsy in his walk, is apt to fall and has considerable difficulty in rising, while in going upstairs he hauls himself up by the aid of the banisters. This period of weakness may have continued for a few months or a year when there is observed an hypertrophy in certain muscular groups and this change is most frequent in the muscles of the calf. The Gluteal muscles become affected soon after and then the disease spreads to the lumbar muscles and to some of the muscles of the thigh. The flexors of the hip are unusually feeble. In the upper extremity the deltoids are usually the first to suffer, indeed in some cases the Muscular enlargement begins in those muscles. The Spinati, however, (especially the Infraspinatus) next to those of the calf are most frequently found hypertrophied.

The Biceps and Triceps sometimes suffer, as also the muscles of the forearm in a few cases. The temporals, masseters and muscles of the tongue have also been found hypertrophied.

In the majority of cases the enlargement is limited to a few muscles, chiefly those of the calves and buttocks and is always accompanied by atrophy in others, especially those of the trunk and upper extremities.

The Pectorales Majores are never enlarged indeed ^{they} are generally the first to show signs of wasting. Gradenigo (1883) ~~in 59 cases~~ found the pectorales majores atrophied, *twenty one times in twenty one observations.*

The sterno-costal portions with the Latissimi dorsi are very frequently wasted, and according to Gowers this is a point of considerable diagnostic value. The Trapezii, Serrati Magni, extensors of the back and the muscles of the thigh become atrophied in this order.

We thus see that a certain degree of atrophy of some of the muscles is a constant feature of the disease, being almost a more important one than the enlargement; for if one is to judge from the measurements of the calves which have been given in many cases, one would imagine that they were merely big from the strong contrast with the slenderness of the upper part of the body. Damaschio, in his lectures on pseudo-hypertrophic Paralysis and

its forms, gives as a distinctive character of the third form ~~of~~ atrophy of a greater or less number of muscles, and as a character of a fourth form more or less general atrophy taking the place of hypertrophy; this last form is well seen in the first group of cases published by Meryon in 1852, where enlargement of muscles was inconspicuous.

In five cases seen by Poore, there was no exceptional enlargement of the muscles although the calf muscles felt unduly solid.

As above stated this atrophy is not consecutive to the pseudo-hypertrophy as Duchenne believed, but accompanies it, if, indeed, in some cases it does not precede it.

Very few of the muscles will be found normal. The muscles of the neck are very seldom affected and as a rule the intrinsic muscles of the hand escape altogether.

The hyper-trophied muscles after a time begin to diminish in size and become markedly atrophied.

The muscular weakness of the lower extremities causes certain peculiarities in the Attitude and Movements.

The patient stands with the feet widely separated, the abdomen projecting forward and the shoulders thrown well backwards. The nates also project, and there is marked lumbar lordosis, due to weakness of the extensors of the hip, which allows the pelvis to be tilted forward on the thigh bones. The peculiar attitude is due to the weakness of the extensors and Flexors of the hip and the extensors of the knee which maintains the equilibrium in the upright posture. The weakness of the extensors of the hip ^{gluteus max. et min. p. 24.} is also the cause of the waddling gait, the body being thrown over to the side of the active limb so as to bring the centre of gravity over each foot. Gowers (37) has pointed out how on rising from the floor the patient places the hands on the knees in order to assist the extension of the hip-joints.

We have also distortions from contractures of certain muscles, the most common being talipes equinus, the result of the retraction and shortening of the calf muscles. This also occurs at the knee and ankle and we may even have lateral curvature of the spine from weakness of the Spinal muscles.

On palpation of the hypertrophied and atrophied muscles we find changes in their consistence. The former are generally elastic and firm, at times soft and flaccid as in the case of those which are atrophied. When brought into action they sometimes form irregular nodes.

The Electric Irritability of the Muscles is lowered, even in the early stages of the disease to the faradic and galvanic currents both in the muscles which are apparently hypertrophied and those which are atrophied. The reaction of degeneration is never present, but some doubt is cast on this by Schultze, who found it in some of the atrophied muscles.

Fibrillary contractions are also said to be entirely absent, but these were also present in Schultze's case so that we can only say that their absence is the almost invariable rule.

The tendon reflexes are diminished and the knee-jerk usually disappears entirely as the disease advances.

The other functions of the nervous system are commonly normal, and although now and again symptoms of mental weakness are seen, we must consider it as one of the complications.

There are considerable variations in the rate of progress and duration of the disease.

The feebleness gradually increases so that by the time the patient reaches adolescence he has lost the power of standing erect. After this the disease seems to make more rapid progress, so that the patient becomes almost helpless and is confined to the recumbent posture. Still he may live for several years until death ensues either from exhaustion, some intercurrent malady, or from some chest affection, which is the more readily developed from the implication of the respiratory muscles.

If the patient passes adolescence with only slight impairment of power the disease is much slower in its progress as in the case recorded by Percy Kidd (38)* where the patient was 24 years of age and the calves had been noticed large for a long time.

The patient may survive in these cases to the age of 30 and in a few to the age of 40.

These late cases are most frequent amongst females, in whom also the disease progresses more slowly.

*Percy Kidd- "Case of Pseudo-hypertrophic Paralysis in an Adult,
Brain Vol. 1886-87

Clinical Features

of

II. The Juvenile Type of Myopathic Atrophy.

(Juvenile Form of Erb)

Clinical Features of

II. The Juvenile Type of Myopathic Atrophy

(Juvenile Form of Erb.)

We now pass to the consideration of the characters of a variety described by Erb, (39) in which the Pseudo-Hypertrophy is not such a constant feature as in the last, according to him it consists in "An atrophy and weakness of certain muscular groups, above all the Scapular girdle and arms properly so called, pelvis, thighs and back, commencing in infancy or adolescence, progressing in a slow and regular fashion, or with some interruptions; often remain^{ing} stationary. An atrophy which frequently combines with a true or false hypertrophy; but which progresses without presenting fibrillary tremors or any trace of reaction of degeneration, or any other trouble anywhere in the organism, either in the nervous system, organs of sense, vegetative organs, or external integument.

The disease generally makes its appearance about the time of puberty sometimes in childhood as in the case of T.C. and very rarely after 20 years of age, though cases are reported by Edgren (40) where the disease appeared at the age of 22 by Singer (41), at age of 34 by Dreschfield (42)*, at age of 42 by Musso (Riv. Clin. June 1887) at the age of 44

As in most atrophies the onset of the disease is so insidious that the patient does not notice its appearance until marked symptoms set in and the weakness has sufficiently developed to cause a difficulty in performing certain movements. Associated with this weakness we have distinct changes in the volume of the muscles of the affected parts. There is a wasting of certain muscular groups, most frequently those of the shoulder and upper arm (but sometimes those of the legs and back) the atrophy thus resembling in its distribution and time of onset that of Pseudo-hypertrophic paralysis.

The following muscles in the upper limb are generally more or less atrophied :-Pectoralis major (except usually the clavicular portion) and Latis-

*Dreschfield- "On some of the rarer forms of Muscular Atrophy

Brain July. 1886.

simus Dorsi (lower part). This also has been noticed as a feature of the Pseudo-Hypertrophic variety. The Trapezius, Rhomboids, and Serratus Magnus (often affected). In the upper arm the Flexors (Biceps and Brachialis anticus) along with the Supinator longus.

In the lower limb, the Glutei, the flexors of the hips and the extensors of the Knee. In the leg generally the peroneal and tibial muscles.

In the back, the Sacro-Lumbalis and Longissimus Dorsi; and in the abdomen, the oblique and transversales muscles have been found atrophied, and in the later stages, the intercostals and, sometimes, the diaphragm.

Along with this atrophy a certain number of muscles pass through a hypertrophic stage and (in some cases true, in others false), the Deltoids Supra and infra-Spinati and the Triceps in the upper limb, and in the lower the Tensor fasciae, Sartorius and Gastrocnemius.

Lastly the following are often normal, at least for a very long time. Sterno-Mastoids, Coraco-Brachiales, Teretes Levatores anguli Scapulae; Muscles of the forearm (when the atrophy spreads to them it usually attacks the extensors first). The small muscles of the hand (unless in very exceptional cases). The Rectus abdominis, muscles of the calf and foot are usually not involved.

The tongue has been always unaffected, as well as the muscles of the larynx, pharynx and mastication.

The facial muscles are described by Erb in the case of a man aged 22 as being affected; but when we come to the next variety of Myopathy it will be seen that this^{is} its characteristic feature, while the distribution of the Atrophy in the other parts of the body corresponds to the juvenile form of Erb.

As in the last variety we have peculiarities in the form produced by the wasting of the muscles. We may have the Scapulae standing out like wings (Scapulae alatae) from their not being fixed down to the Trapezii Latissimi Dorsi, Rhomboids and Serrati. The forearm becomes almost cylindrical in the upper part from atrophy of the Supinator longus.

The attitude resembles what we have seen in Pseudo-hypertrophic paralysis marked lumbar lordosis being present.

The movements also are peculiar—we have the waddling gait, the difficulty in getting upstairs and in rising from the ground, due to weakness of the extensors of the hip and Knee. Erb (43)* also points out the anomalous con-

*Erb—in Progressive Muscular Dystrophy." New Sydenham Society 1894.

dition which may exist in the thorax which is occasionally flattened and retracted in the middle from the action of the powerful Rectus Abdominis and prominent at the sides, the edges looking as if turned up from the weakness of the oblique and transversalis muscles. On attempting to raise the head in the recumbent position these latter muscles appear as two soft tumours in the lateral aspect of the abdomen.

Distortions from contractures are only occasionally observed in this variety.

The consistence of the muscles varies. They may be tough and with a feeling of induration while others again are quite soft and flaccid. The mechanical irritability is lessened often to an extreme degree. The electric Irritability is diminished, usually in proportion to the wasting. In one of Marina's (44) cases partial reaction of degeneration was present.

In Schultze's (45) case there was reaction of degeneration in the left Deltoid, Thenar and some Interossei. In one of Zimmerlin's (46) cases reaction of degeneration was present to a moderate degree in several of the atrophied muscles but as a rule there is no trace of reaction of degeneration. Fibrillary twitchings are usually absent but this is not invariable as they were present in both the cases recorded.

The Knee-jerks are present though diminished and many disappear altogether when the atrophy is extreme. In some cases they may vanish early.

The other functions of the nervous system:-

Sensibility; trophic functions of Skin, Sphincters etc are all normal.

Progress and Duration:-

The progress is essentially chronic; the disease going on for a considerable number of years, with periods of cessation, and then beginning again, always in a very insidious way. For example the right arm ^{may} become affected at 19 the left at 25 and the legs at 30 (Gowers) (47)*. Occasionally the progress is rapid, the atrophy becoming universal in the course of eight or ten years. The duration varies from ten to fifty years, death resulting from some intercurrent malady, and rarely in consequence of the disease.

*-Gowers- Disease of the Nervous System. 1892.

Clinical Features

of

III The Facio-Scapulo-Humeral Type of

Myopathic Atrophy:

(Landouzy-Déjérine type)

Clinical Features of

III The Facio-Scapulo-Humeral Type of Myopathic Atrophy.

(Landouzy-Déjérine Type)

We now reach the consideration of the last of the varieties of Myopathic Atrophy, variously described as the "Infantile form of Duchenne"—the Facio-Scapulo-Humeral type, or the Landouzy-Déjérine type, from the authors who definitely settled the nature of the affection.

If one is to judge from the number of recorded cases the disease is not uncommon in France, where it has been specially studied; while on the contrary, in Germany, America and Britain the disease has been little noticed. Wilks (48)* describes a case of this type under Progressive Muscular Atrophy.

Clinically this form may be said to be similar to Erb's juvenile form with the addition of Atrophy of the muscles of the face. Indeed this Atrophy may be more common than is supposed, for Landouzy and Déjérine (49)† state that, though no abnormality could be detected in life, yet after death, in a case examined by them, the muscles were degenerate or beginning to degenerate, a point they consider of importance as showing that sooner or later the face becomes involved in this progressive atrophy.

If we examine the cases recorded by Landouzy and Déjérine (50) (5 cases of atrophy in the family L. 2 cases of atrophy in the brothers M. 1885†† and 6 further observations in 1886†††) we find that in 7 the atrophy began in the muscles of the face, and, when it does so begin, it is nearly always at ages ranging between three and seven.

Duchenne (51) says "this commencement in the face of the Progressive Muscular Atrophy of childhood is a premonitory sign of its extension sooner or later to the muscles of the limbs and trunk"

*Wilks Lectures on Diseases of the Nervous System 1883.

†Landouzy and Dejerine "Nouvelles recherches cliniques et anatomie Pathologiques sur la myopathie atrophique Progressive." Rev. de Med. 1886

†† Landouzy et Dejerine. "Myopathic atrophique Progressive sans Neuropathic" Rev. de Med. 1885

††† Rev. de Med., 1886.

^ the atrophy of the face is consecutive to

It may be soon, as in an instance at the age of 10, or the atrophy may remain limited to this part for a long time as in one of their cases where the atrophy began in the face at the age of 3 and in the upper limbs at 16. Indeed when the face is affected early, the atrophy does not begin to spread as a rule till early adolescence; the upper limbs being in most cases attacked first, though in obs. 223 of Duchenne, atrophy of the muscles of the lower limbs and trunk followed the atrophy of the face at the age of 5, and that in the upper limbs between the ages of 6 and 7

This was also seen in case 3 of Landouzy and Déjérine (1886).

When however the appearance of the atrophy is postponed till later in life it begins in adolescence and generally affects the shoulder and arm muscles first, the atrophy of the limbs as in the cases recorded by Hemak (52) and Ladame (53) among others.

The atrophy may not show itself till adult life is reached, as in two of Landouzy and Déjérine's Atrophies, where the disease began in one in the shoulder at age of 28 and the face at 32; in another it did not develop till 40 in the shoulder, and at 45 in the face.

The atrophy commonly affects both sides of the face, it being very rare to find cases of asymmetry, though in Mossdorf's case (54) only the left side of the face contracted. One side may suffer a considerable time before the other.

The wasting of the muscles of the face produces a peculiar and characteristic expression to which the French observers apply the term "facies Myopathica." The countenance is smooth and immobile like a mask. Owing to the constant affection of the Orbicularis Oris the lips remain separated when at rest. The lower one projecting, and when thickened as ^{often} they are, ~~often~~ giving rise to the "Tapir mouth". The patient cannot "pout" or bring the lips firmly together and pronunciation of labials is indistinct; The naso-labial furrows disappear owing to atrophy of the Zygomatici; and when the patient laughs his mouth extends laterally (rire en travers) the upper half of the face not sharing in the change of expression. The wasting of the Orbicularis Palpebrarum causes the closure of the eye-lids to be incomplete even during sleep. The forehead becomes smooth and the patient cannot wrinkle the eye-

brows. In many cases the Euccinators have been affected. The muscles of the tongue, pharynx, larynx mastication and those of the eyeball are normal, though Westphal records a case with affection of the ocular muscles.

The atrophy of the muscles in the Scapulo-humeral region corresponds in every way to what has been described under the juvenile form so that it is not necessary to go into detail here. Atrophy of the muscles of the lower limbs is also frequently present, the atrophy, as in the upper, beginning in the proximal end of the limb. The muscles of the back and of the abdomen may participate in the atrophy and in the case of the latter giving rise to the deformations seen under the juvenile variety. We have here also the same peculiarities in the attitude and movements.

The electric irritability persists so long as Muscular fibres remain, and is usually diminished to both currents in direct proportion to the size of the muscle, although there may be extreme atrophy and the faradic contractibility only slightly altered.

Landouzy and Déjérine met with reaction of Degeneration ^{in the} ~~and~~ Orbicularis Oris in one of their cases and in the deltoid in another but it is very rare.

Fibrillary contractions are almost never present.

The tendon reflexes are present and vary in direct ratio to the volume of the muscular mass; although Landouzy and Déjérine (55)* observe that the patellar reflex may be abolished before there is any trace of atrophy in the thigh muscles; as in observations 3 and 4.

Contractures may be present, especially in the biceps and in the posterior regions of the thigh.

The other functions are normal.

Progress and Duration.

The disease is essentially chronic, but very variable in its progress; for example, the atrophy may remain confined to the face for a period of 13 years before spreading to the upper limbs (obs. 2 Landouzy and Dejerine 1885). In another the extension may begin a year or two after the face, and the atrophy become rapidly generalised (obs. 3 Landouzy and Déjérine 1886) but under favourable hygienic conditions the patient may live a long time, twenty, forty or even more years.

* Landouzy et Dejerine "Loc cit." 1885.

MORBID ANATOMY
OF THE
MYOPATHIC ATROPHIES-

Morbid Anatomy of the Myopathic Atrophies.

In the pseudo-hypertrophic condition the naked eye and microscopic appearances will vary according to the stage at which the disease has arrived. When the muscles are enlarged they are pale yellow.

In the earlier stages of the disease there is simple atrophy of the muscular fibres with an increase of the connective tissue between the bundles of muscular fibres and between the fibres themselves. These thick septa which now separate the muscular bundles from each other consist of fibres arranged parallel to the long axis of the muscular fibres, intermingled with a considerable number of embryonic nuclei or spindle cells.

In this stage then, if the atrophy is in excess, the muscles present an appearance of atrophy, when the connective tissue is in excess an appearance of hypertrophy. The hypertrophy, however, is really due to the changes which take place in the next stage of the process, viz:- the development of fat cells in the connective tissue and in the newly formed fibrous tissue. This fatty substitution may be slight at first, but gradually increases and as it does so, the connective tissue of recent formation, as well as the muscular bundles, tends to disappear, so that we have tracts of nucleated fibrous tissue amidst adipose tissue. These tracts contain muscular fibres, presenting great diversity in their diameter, some being normal, others much narrower, and others again broad and suddenly becoming narrow. In these narrow fibres the transverse striation may be found to have disappeared, or to have been replaced by granules distributed through them, or a simple fading of the striae. The fibres occasionally undergo fatty degeneration - a longitudinal striation - vitreous degeneration or vacuolation. In others, generally those of the smallest diameter, we find clusters of nuclei distending the sarcolemma sheath, or in others the latter may be quite empty.

Very rarely, in some part of a muscle, there has been only wasting of the fibres, without the interstitial change, present elsewhere (Singer, in the triceps, quoted by Gowers. loc. cit.)

We find normal fibres more frequently where the fat is abundant than in those cases where fat cells are sparse and the interstitial tissue is almost entirely fibrous.

The fat may be subsequently absorbed and connective tissue/

tissue with traces of muscular fibres be all that is left.

In the juvenile form of myopathic atrophy we meet with as varied alterations in the condition of the muscles as are seen in the pseudo-hypertrophic form.

The most important changes are in the muscular fibres themselves. Erb (56) examined pieces of muscle excised from the Deltoid, which was hypertrophied, and from the Biceps which was atrophied. In the case of the Deltoid nearly all the muscular fibres were increased in size, the nuclei were increased in number round the fibres, which showed no signs of fatty or granular degeneration, but the striae were faint. The connective tissue was also increased. In the Biceps the fibres also appeared to be hypertrophied and there was some increase of nuclei.

The apparent increase in size, however, may have been due to a contraction caused by the process of excision, but that some fibres are hypertrophied is evidenced in three cases examined by Hitzig (57) and in one examined by Schultze (58). In the latter there was an increase in the sarcolemma and muscle nuclei but little increase of the interstitial connective tissue.

There was some fatty infiltration in the interstices between the fibres. In muscles highly atrophied there was large infiltration of fatty tissue with a number of broad connective tissue fibres. The muscular fibres in some muscles were undergoing a process of vacuolation.

In a case reported by Dreschfield (59) simple atrophy of fibres with some increase of muscle nuclei was found. In other cases (Singer, 60) we may find splitting up of the fibres in a longitudinal direction with a tendency to split into plates horizontally.

The results of Roth's autopsies go to show that fibres may atrophy in their length even more than in their breadth. This he says is borne out by the clinical appearance of the atrophied muscle, the belly becoming shorter and shorter. In the Zygomatic he found that half consisted of fibrous material, the other half being natural. Much the same condition he also found in the Biceps.

From the above it will be seen that changes are found in the muscular/

muscular fibres very much resembling what we have noticed in Pseudo-hypertrophies.

Landouzy and Déjérine (62) in their infantile form found hypertrophy of certain muscular fibres, though simple atrophy was more common, the fibres not presenting any granular or fatty degeneration. "The striated fasciculi diminished progressively in volume, always preserving the striation intact, the atrophy evolving without any irritation of the sarcolemma or muscle nuclei - the alteration of the contractile substance, properly so called, being similar to pseudo-hypertrophic paralysis." Empty sarcolemma sheaths may be found and between these fibres on the road to atrophy either along their whole length or only in parts. They observed a trifling thickening of the perimysium, externum and internum. Fatty infiltration existed especially in the Orbicularis Oris, Frontal, Deltoid, Biceps and Supinator longus, but was completely absent in others as Trapezius, Radials, &c.

In their examination in case of obs. 6 (63) they found simple atrophy with great multiplication of nuclei - in some hypertrophy and in most a certain amount of interstitial fat.

Having now studied the condition of the muscles in the varieties of myopathic atrophy it is essential to inquire into the condition of the nervous system (spinal cord and nerves).

Notwithstanding the lesions that have been observed from time to time in the spinal cord we must consider it as a rule perfectly normal. The site and nature of the changes have been so variable that we cannot look upon them as the cause of the symptoms which have been described, more especially when we take into account the fact that so many cases have been reported where the nervous system, both central and peripheral, has been found intact. In the pseudo-hypertrophic variety, atrophy of cells in one case, sclerosis of the lateral columns in another, in others still alteration in the shape of the grey matter have been noticed. In the atrophic varieties, atrophy of the ganglion cells has been observed by Heubner (64) Frohmaier (65) and Singer, but the examinations of Schultze (66) Landouzy (67) Dreschfield (68) Westphal and many others in recent years have given absolutely negative results.

The/

THE PATHOLOGY

of

MYOPATHIC ATROPHIES.

The Pathology of Myopathic Atrophies.

It follows from what has been said as to the morbid anatomy that the varieties described are essentially muscular diseases, consisting of a wasting of the muscular fibres, of an overgrowth of the connective tissue, more marked in one and less in another, and in which fat may or may not be deposited.

Those who have found changes in the cord, looked upon the lesion here as the primary and the muscular as the secondary affection. Even Erb (69) is inclined to think that after all the affection may be dependent on the nervous system, for which he gives the following reasons:- (1) the muscles depend for nutrition to a great extent on trophic nerve centres, but the balance of evidence is against the existence of any special trophic nerves, and, by extension of reasoning, trophic nerve cells; (2) the atrophy often follows the exact course of the nerves in a plexus, but the fact that in the sphere of distribution of same nerve we so often find individual muscles absolutely destroyed in the midst of muscles intact, would appear to negative this view; (3) mental aberrations occur among the patients and other neuroses in their families, but if we look upon the disease as one of a morbid developmental tendency, there is no reason why there should not be a similar tendency in the diseases of the nervous system and hence congenital malformations in the arrangement of the grey matter and in the shape of the cord; (4) the fact that similar morbid changes (hypertrophy, proliferation of nuclei and division of the fibres) both in the muscles and the connective tissue occur in undoubtedly spinal cases, as acute anterior poliomyelitis; but these lesions do not belong peculiarly to Myopathic atrophy and cannot therefore be sufficient to differentiate it. He promulgates a theory that we may have a 'kind of tropho-neurosis having its origin in the trophic centres of the cord, a disturbance of the functions of these centres, which finds its expression in the very complicated muscle changes of the disease. While on this supposition there are as a rule no coarse nerve changes, it occasionally happens that, after the affection has lasted a long time or been very intense, such changes do become visible.'

That there may be a connection existing between
Progressive/

Progressive Muscular Atrophy of myelopathic and myopathic origin, receives some measure of support from the existence in the same family of both these forms as will be seen in the following cases reported:- (1) by Philip (70) where in a family of eight children four were pseudo-hypertrophics, while the father suffered from a form of spastic paralysis with wasting and contraction of muscles, which seems to have been of spinal origin; (2) by Cenas and Douillet (71) where we find a brother affected with the Landouzy Déjérine type and a sister with atrophy commencing in the thenar muscles of the right hand subsequent to severe neuralgic pains in the arm.

If we consider the atrophy in the case of the great-grandfather (recorded above) as a Myelopathy, we have both the forms occurring in one family.

The theory which I think best explains the pathology is advanced by Gowers (72) "that all the diseases of this class seem to depend upon a defective tendency of development of the germinal tissue which forms muscles. They are essentially congenital diseases, although they are in most cases merely potential maladies at the time of birth, and sometimes for years afterwards, even occasionally during a considerable part of life. Sometimes the morbid tendency attains actual development in the earlier years."

The primary shrinking of the muscles is due in all cases to this defective tendency. In the pseudo-hypertrophic we find the connective tissue chiefly involved and as the process advances damaging the fibres; in the atrophic forms the muscular fibres are principally at fault.

This explanation of the nature of the disease will account for there being almost invariably several members affected in a family, while the fact that the Latissimus Dorsi and lower part of the Pectoralis major (two muscles which stand lowest in functional importance) are sometimes congenitally absent, is also quite consistent with this.

When the knee-jerk is lost this can be accounted for apart from any lesion of the cord by the wasting of the muscles, when the atrophy is considerable. Further when it disappears before there is any trace of atrophy in the thigh muscles, the sensory muscle nerves, which receive the different impulses, may be destroyed by the overgrowth of the connective tissue.

Etiology/

ETIOLOGY

of

MYOPATHIC ATROPHY.

Etiology of Myopathic Atrophy.

Myopathic Atrophy (Idiopathic muscular atrophy) is essentially a congenital, and, in many cases a hereditary disease, as shown by its occurrence in family groups and in more than one generation.

In the hypertrophic variety two children in a family are often attacked, and Meryon (73) relates an instance where eight brothers died of the disease, while all the daughters escaped. Lutz (74) met with two sisters, a maternal uncle and aunt and a maternal half sister, issue of a first marriage, all affected with the disease.

In the atrophic variety Barsickow (75) records 24 cases in two unconnected families, distributed through five generations. Landouzy and Déjérine (76) traced the disease through five generations in the family L-, and of the nine cases in this family four occurred in one generation.

In Schultze's (77) cases heredity is very marked. The maternal grandfather and grandmother were cousins and of their children one son had muscular atrophy of some sort. A daughter who was unaffected and was married to a healthy man had five sons, three of whom had muscular atrophy; one died and in the other two the disease began at about eight years. The elder was evidently an example of the pseudo-hypertrophic disease, while the younger presented the appearance described by Erb.

In one of Westphal's (78) cases there was marked atrophic heredity, - younger sister, father and uncle were affected.

A peculiarity in the inheritance of the disease in the hypertrophic form is, that it is transmitted by the mother, she herself being unaffected. In this respect it resembles Haemophilia, a distinctly hereditary disease. The explanation of this is that females are rarely affected while the males die soon after puberty.

In a case quoted by Gowers in which four brothers suffered, the mother's brother and sister were likewise affected. Again a brother and sister were diseased, one daughter of a second sister and three daughters of a third sister.

Thus the congenital tendency is exclusively due to the maternal element in the embryo, - proved by the fact that children of the same woman by different husbands suffer in the same way (Gowers) 79

Duchenne states that maternal heredity was somewhat more frequent than paternal heredity in cases of the infantile form noted by him.

Sometimes isolated cases are met with in which no heredity can be traced, as in two cases recorded by Marina (80) two by Edgren (87) and in Obs. 224 to 227 of Duchenne (82).

Sex is also an important etiological factor, at least in the hypertrophic variety, where males mostly are affected.

Landouzy and Déjérine state that the atrophic form has been observed more frequently in boys than girls, but statistics are not sufficiently numerous to determine whether this is so.

The Age at which the disease first manifests itself has also an important bearing on the etiology. It is difficult in many cases to determine the exact date of onset, as the disease often begins so insidiously. The pseudo-hypertrophic variety shows itself during the period of development, most frequently during mid-childhood, sometimes in infancy, very rarely after puberty. Cases in adults have been recorded from time to time by Percy Kidd (83) Hughes Bennett (84) and others.

The atrophic generally appears during the later period of growth and the early period of adult life, sometimes it begins in childhood as early as 2 or 3, especially when the wasting shows itself in the face.

Barsickow's cases illustrate how variable the date of onset may be. In 17 cases it was as follows:—one at 12, four between 15 and 20, seven between 20 and 30, three between 30 and 40, two after 40.

In some instances neuropathic tendencies can be traced among the ancestors, as in the family history recorded, but Erb found it only in 8 out of 18 families, so that this can have little influence.

Consanguinity sometimes seems to act as a factor. It is difficult to say what part exciting causes play in this disease, but if on enquiry one finds the existence of a predisposition to the disease, exciting causes may be assigned a share in its evolution; as for example, the frequency of the appearance or acceleration of the morbid process after acute infectious diseases observed/

observed in the case of T.C., and in a case recorded by Treisier and Guinien. In this the face was affected at three, at seventeen the patient had typhoid fever which seemed to precipitate the evolution of the atrophy, and after that the shoulder muscles were completely gone.

DIAGNOSIS

of

MYOPATHIC ATROPHY.

Diagnosis of Myopathic Atrophy.

I. From Myelopathic Atrophy.

The difficulties of diagnosis of Myopathic and Myelopathic atrophies are well illustrated in Schultze's case which showed widespread atrophy of the upper extremities (including those of the hands) with fibrillary tremors, reaction of degeneration and absence of heredity, but which was proved post mortem to belong to the Myopathies. The main points in forming a diagnosis are:- (1) the point of attack in the typical form of Myelopathy is the small muscles of the hands; conversely in Myopathy the atrophy usually begins in the muscles of the shoulder, back or face. A predominance of the atrophy in the muscles of the proximal end of the limb persisting almost indefinitely is very characteristic of Myopathies. Slight implication of the intrinsic muscles of the hands has been met with in very rare instances (Gowers and case of T.C.)

(2) As already stated the white matter as well as the anterior horns is affected in the vast majority of myelopathies giving rise to paresis out of proportion to the muscular wasting, and hypertonus added to the symptoms of atrophy. Frequently also in myelopathies bulbar symptoms make their appearance, while neither of these symptoms are observed in myopathies.

(3) Hypertrophy of certain muscles is very often present in myopathies but has never been described in myelopathies.

(4) In myopathies the muscles are affected in an individual way as we have seen and we find muscles destroyed in the midst of muscles intact. This, which is not seen in the spinal form, is important, especially when the case is advanced.

(5) Reaction of degeneration is not uncommon in myelopathies in the later stages. In myopathies it is exceptional to find any other changes than diminished response to both currents.

(6) Fibrillary tremors are common in myelopathies but rare in myopathies.

(7) Pain in the neighbourhood of joints in the affected limbs is frequently complained of in myelopathies but exceptionally in myopathies.

(8) Trophic changes are frequent in myelopathies but do not occur in myopathies.

(9) Myopathic atrophy evolves more slowly than the myelopathic.

(10)/

- (10) The age of onset in myelopathies is about 30 or later, rarely earlier. In myopathies any age below 20, rarely later.
- (11) Heredity is rare in myelopathies but remarkably common in myopathies, especially that heredity in which members of the same generation are affected, often without ascendent or descendent heredity.

II. From Chronic Multiple Neuritis.

In this disease we often have a history of alcoholism or family tuberculosis. (1) It is never hereditary. (2) The paralysis is out of proportion to the atrophy. (3) There are marked sensory disturbances. (4) There is early appearance of reaction of degeneration and (5) absence of hypertrophy.

III. From Syringomyelia.

The localization of this affection is of the Spinal type. We have atrophic paralysis with reaction of degeneration, absence of hypertrophy and distinct sensory disturbances.

IV. From the Peroneal Type of Atrophy. (Neuropathic Atrophy).

In this type hypertrophy is never present, fibrillary contractions are common, degenerative electrical changes appear early, hands and forearms are affected soon after the lower extremities and sensory troubles are not uncommon.

PROGNOSIS

of

MYOPATHIC ATROPHIES.

Prognosis of Myopathic Atrophy.

The prognosis in all the forms of myopathic muscular atrophy is grave, for the disease does not appear to be arrested in its progress. Hence it deserves the name progressive muscular atrophy as it successively attacks different muscles, which gradually progress to the highest degree of atrophy.

The prognosis is most unfavourable in the pseudo-hypertrophic variety. Even though the disease develops late and the patient reaches his twentieth year before the condition becomes marked, there is only very slight hope that it will not advance to its ultimate degree. Gowers says that in any case, and at any age, it is unlikely that the patient will live more than seven years after the power of standing is lost.

In the atrophic forms when the symptoms are late of appearing the progress of the disease is slower. Sometimes the degenerative process seems to stop in its course and we have long periods of arrest, and this is a favourable circumstance in a prognosis already sufficiently grave.

We thus see that the prognosis is influenced by the period of commencement and the observed rate of progress.

The general health remains good, as the muscles of mastication and deglutition are unaffected; and, if the muscles of respiration remain intact, under favourable hygienic conditions the patient may live to an advanced age.

Treatment/

TREATMENT

OF

MYOPATHIC ATROPHIES.

Treatment of Myopathic Atrophies.

From what has been said under pathology it will be evident that treatment is of little avail in cases of Idiopathic Muscular Atrophy.

It is difficult to determine in a disease so variable in its progress what share the various methods of treatment have had in cases in which improvement seems to have taken place.

We have no positive knowledge that any drug is capable of exerting a beneficial influence, although the administration of such nervine tonics as arsenic and phosphorus have been considered efficacious in some cases in retarding the progress of the weakness.

Electrical treatment has been advocated and both Duchenne and Erb report cases of arrest following its employment, but this is very questionable when we take into account the variable tendency of the disease. Erb thinks that electricity is still the most trustworthy means at our command. He advises the galvanic current along the back and over the cervical and lumbar swellings of the cord, whether we consider them the proper seat of the disease or not, and the faradic, galvanic or farado-galvanic currents to the nerves and muscles, avoiding too strong currents or too long sittings. One of my patients was of opinion that in his case electric treatment instead of being beneficial tended to increase the weakness.

But by far the most powerful agent in improving the condition of the muscles is the physiological stimulus of Voluntary Effort, and as we have already seen in pseudo-hypertrophies, the muscular disease progresses more rapidly when this ceases. In families with a predisposition to atrophy, if systematic muscular exercise can be taken, it may assist in preventing the occurrence of the disease and in retarding the failure of power in those already attacked. This has been well marked in both my cases where gymnastics adapted to bring into play the weakened muscles has had the effect of strengthening them.

Hence every means should be adopted to maintain the power of moving about as long as possible.

Massage, combined with passive movements to prevent muscular contraction/

contraction and consequent deformities may be of use; and, if necessary, tenotomy should be performed in cases of contraction of the calf muscles.

Excessive exertion should be carefully avoided.

I may here mention that the brothers of my patients believe that they have escaped the disease because of the active life which they have required to lead, the one having been in the army, the other employed in a large store. As bearing on this point it is rather curious to note that the majority of patients have led a sedentary life.

The general health must be carefully attended to in the case of members of atrophic families, both of those who are affected and those who are not. They should have plenty of fresh air, good food, baths, &c., and every means should be employed to give tone to the system.

Concluding/

CONCLUDING REMARKS

upon the

SUBJECT OF MYOPATHIC ATROPHY.

Concluding Remarks Upon The Subject Of Myopathic Atrophy.

From the description which has been given of the Idiopathic Muscular Atrophies, it will be seen that there exist several types, all characterised by a progressive wasting of a greater or less number of the voluntary muscles of the body, but which, at the same time, present differences in their clinical features.

Three principal types have been selected as being most pronounced in their characters, and in this way the points of agreement and difference are more clearly brought out.

These points of difference constitute, according to some, sufficient ground for regarding the various types as distinct primary muscular affections, and not as essentially one disease, as the numerous points of agreement would more readily lead one to believe them to be.

The differences are, it must be remembered, merely differences of degree of the morbid process and in the initial localisation, both of which seem to be influenced by the date of onset.

Thus, when the disease shows itself in the early period of development, it is more apt to be attended by hypertrophy, especially in the legs and face; but when the onset occurs at a late period, wasting, especially at the roots of the limbs, is the conspicuous feature.

But it has been mentioned under the pseudo-hypertrophic form that atrophy is almost as important a feature in the disease as hypertrophy, and that the latter was due to a fatty infiltration. This fatty infiltration is also marked in the face in the Orbicularis Oris, which is always first affected, and which gives to the lips the volume they possess. Thus it may be looked upon as a phase of the pseudo-hypertrophic form.

The infantile form, again, affects more frequently the male sex, and, as in the pseudo-hypertrophic form, we find transmission occurring more usually through the mother.

Many observers in recording cases of the pseudo-hypertrophic form, speak of the stupid expression worn by these patients, even in cases in which the intelligence was undoubtedly unimpaired, and this, no doubt, was due to the greater or less implication of the muscles of the face, and the imperfect performance of/

of their functions.

Zimmerlin records a case in which the patient was attacked by that variety of the disease which first involves the muscles of the face, while the pseudo-hypertrophic form appeared in the lower extremities (Mendal's Centralblatt, 1885.)

That a connection also exists between the pseudo-hypertrophic form, and that of Erb is proved by the fact that when the former spreads to the upper part of the body, the muscles do not increase in size, but become markedly atrophied, and the atrophy here occurring has exactly the same localisation as in the 'juvenile form,' and begins about the same time.

Another point which brings them into contact is that in the juvenile we often find the same muscles in the upper part of the body (Deltoid, etc.,) hypertrophied, as we do in the pseudo-hypertrophic form.

Another character which they have in common is a diminution in the size of neighbouring muscles, namely, the lower part of the Pectoralis and Latissimus Dorsi. We sometimes meet with the pseudo-hypertrophic and the juvenile forms combined in one individual, as in Singer's (85) case when there was pseudo-hypertrophy of the calves and characteristic gait, with marked atrophy of the shoulder and upper arm muscles.

These two types may also occur in the same family as will be seen from the history of the cases recorded above and those of Schultze, (86) where the elder brother was a pseudo-hypertrophic, and the younger presented the appearance described by Erb. In Russell's (87) cases, one brother was a pseudo-hypertrophic and the other two were atrophics. This latter fact is an important argument in favour of the unity of the diseases.

We now take up the relationship of the infantile form and the juvenile form of Erb.

When the face is attacked in infancy, sooner or later, but very often about the time of puberty, the disease spreads to the shoulders and upper arms, as seen in Landouzy and Déjérine's cases, and then the juvenile form of Erb is produced, or the scapulo-humeral type of the French observers. The latter, however, argue that in 'Erb's' form there is hypertrophy of certain muscles, which feature is not present in the form, they describe; but Erb states that hypertrophy is not always present. It only exists/

exists for a longer or shorter length of time and gives place in the course of the affection to a pronounced atrophy. It is, in fact, a fleeting symptom. When the affection of the face does not appear till later in life, it does not precede but follows the atrophy of the shoulders as we have seen in Remak's and Ladame's cases. Remak, indeed, published his case under the juvenile form with participation of the face. This point brings these two types into close contact, and heredity here also strengthens the evidence, for we frequently find the two types present in the same family. In addition to Landouzy and Déjérine's cases, Duchenne describes the case of a father in whom the disease began in the shoulder muscles at the age of 48, while his two children were early affected with atrophy beginning in the face. In Troisième and Guinon's (88) cases the father was affected at 14 with atrophy of the shoulders, while his daughter, who began to suffer at 11, had atrophy of the face, shoulder and arm muscles.

In a case described by Friedrich we see a patient in whom all three varieties appear in combination - the infantile form, from the implication of the face muscles; the juvenile form, from the atrophy having a special localisation; and the pseudo-hypertrophic form, from the increase in the volume of the muscles.

The morbid anatomy also aids in establishing the unity of the three types described. In all we have a primary wasting of the muscular fibres, an increase of connective tissue and a deposition of fat. Those changes may vary considerably in different muscles in the different types and in different individuals; but this is only a difference in the degree of the morbid process, while its nature is the same in all.

The other points of agreement between the various types are:— (a) the gradual onset, (b) the disease generally attacking the proximal ends of the limbs, (c) the peculiarities in the attitude and movements, (d) the simple diminution of electric irritability, with almost invariable absence of reaction of degeneration, (e) absence of fibrillary twitchings, (f) lessening or complete loss of mechanical irritability and (g) of tendon reflexes, (h) frequent heredity, and (i) all the other functions of the nervous system unaffected.

Progressive/

PROGRESSIVE MUSCULAR ATROPHY

of

NEUROPATHIC ORIGIN

---oOo---

(PERONEAL TYPE.)

Progressive Muscular Atrophy Of Neuropathic Origin. (Peroneal Type.)

We now come to the last of the forms of Progressive Muscular Atrophy, bearing certain resemblances to the atrophies just described viz:- the age at which it begins, the gradual onset, more than one member of the same family being affected, and its slow but progressive course.

Charcot and Marie, however, (89) in 1886 recognised it as a distinct type and Tooth (90) in the same year proposed for it the name 'peroneal type of progressive muscular atrophy' from the tendency of the disease to attack first the peronei and extensors.

In this form males suffer about twice as frequently as females. In the majority of the cases the onset of the disease occurs between the ages of two and fifteen, though in a case reported by Osler it began unusually late in life, at the age of 47.

According to Tooth, the disease is most often first apparent in the Peronei and at about the same time, or soon after, in the Extensor longus pollicis and Extensor communis digitorum. It is not unlikely, however, that the small muscles of the foot are the first to undergo degeneration. The calf muscles are later in being involved, and still later those of the thigh, especially the Vastus internus. From two to five years after the beginning of the atrophy in the legs and feet the intrinsic muscles of the hands are attacked, after them the extensors of the forearm, and still later the pronators and supinators. In Charcot and Marie's cases, four in number, the Supinator longus escaped, as well as the muscles of the shoulders, upper arms, neck, body and face; but Hoffman (91) reports cases in which all these muscles were diseased.

We may have a claw-like deformity of the fingers and an early development of club foot from the unequal affection of the muscles.

Hypertrophy has never been observed.

Fibrillary tremors are common, especially in the hand.

The irritability to the faradic current is diminished, and in nearly all the cases reaction of degeneration appears early. Shooting/

Shooting pains occur in a few cases, and cramps of the muscles of the thighs are described by Charcot and Marie. Cutaneous anaesthesia is sometimes present.

Muscle reflex action is lessened or lost.

Heredity, especially collateral heredity is a marked feature of the disease.

In Osler's (92) case it could be traced through three generations.

Herringham (93) reports a case where the disease could be traced through five generations and the disease was transmitted through females to males, in this respect resembling pseudo-hypertrophic atrophy.

In Schultze's case two sisters and a brother, and in Ormerod's (94) a brother, sister and father were affected.

A striking peculiarity of the disease is its tendency to follow an acute specific disease such as measles. Out of six cases recorded by Ormerod (op. cit.) and Donkin (95) measles preceded the disease in five.

There is considerable difference of opinion as to the pathogeny of the disease. Both Herringham (op. cit.) and Sachs (96) are of opinion that it is one of the central nervous system, and that this form represents the leg type of myelopathy.

Dr. Ferrier's experiments (97) showing the centres in the lower cord and their corresponding groups of muscles strengthen this view. That it may be so in some cases is, I think, further supported by the History of the 'Wetherbee Ail,' reported by Hammond in his book on the Nervous System, where we have evidently a true myelopathy in the earlier cases, beginning in the legs, while the later cases developed the thenar type.

Hoffman (98) (who styles it Progressive neurotic muscular atrophy) and Tooth arguing from the autopsies of Virchow, Oppenheimer and Friedrich, where extensive nerve degeneration was found, advocate the nerve origin of the disease, so that we may have a Progressive Muscular Atrophy of Neuropathic origin. The other points in favour of a neuritis are:-

(a) the early appearance of the reaction of degeneration.

(b) /

- (b) the absence of reflexes.
- (c) the tendency to contracture and
- (d) pain, especially along the course of the nerves.

If the disease is really neuritic (and this can only be settled by further autopsies) and seeing that often many members of a family are affected, we may have a congenital weakness in the peripheral nerves, just as occurs in the case of the muscles in idiopathic muscular atrophy.

The atrophy in some cases may be idiopathic, so that we may have a peroneal type due to spinal, neural or muscular changes.

The prognosis and treatment mentioned under the myopathies apply here also, except that electrical treatment may be more beneficial.

Thus we see that Progressive Muscular Atrophy may have its origin in the three parts of the neuro-muscular system - the spinal cord, the nerves and the muscles, and not alone in disease of the cord as was originally believed.

David R. Dow.

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