Section of Neurology.

President—Mr. PERCY SARGENT, C.M.G., D.S.O., F.R.C.S.

Observations on Myopathy.

By EDWIN BRAMWELL, M.D.

I PROPOSE to refer to some experiences and problems in relation to myopathy. While some of my observations may appear to be suggestive rather than conclusive, they will, I hope, for this reason, stimulate discussion.

SYMMETRICAL ATROPHIC PARESIS OF THE QUADRICEPS MUSCLES OF PROBABLE MYOPATHIC ORIGIN.

The distribution of the affected muscles in the myopathies shows considerable variation, and a number of well known types are described in accordance with the incidence of the muscular involvement. Two cases characterized by progressive bilateral weakness and symmetrical wasting of the quadriceps muscles, which are, I believe, examples of myopathy, are of interest on account of the unusual distribution and limitation of the atrophy.

Case I: Bilateral Progressive Weakness and Wasting of the Quadriceps Muscles.—A married woman, aged 53, complaining of weakness in the legs, was seen on October 4, 1921.

History: The patient, for a year previously, had noticed difficulty in rising from a chair or ascending stairs. This disability had apparently developed gradually, had been unaccompanied by pain or other subjective sensory symptom, and had been progressive. The thighs had become appreciably thinner; otherwise, the patient stated, she felt in perfect health. There was no history of any obvious ætiological factor or of any similar condition in the family.

State: Symmetrical wasting of the thighs, especially of the vasti interni, was noted on examination. The movement of extension at the knee-joints was very easily overcome, and when the patient attempted to bend the knees she would have fallen had she not been supported. No weakness of any movement, apart from extension at the knee, was detected. The electrical response in the quadriceps muscles showed a varying degree of quantitative reduction but no qualitative change. There were no fibrillary tremors. A feeble knee-jerk was obtained on reinforcement, but no contraction was observed in the vasti interni muscles. The ankle-jerks were brisk and equal, there was no tendency to clonus, and the plantar reflexes were definitely flexion. No subjective sensory symptoms had been complained of, and no objective sensory disturbance was found. There was no sphincter trouble and no vertebral deformity. Radiograms of the spine showed no defect in the bony arches. No abnormality of the nervous system or of the thoracic or abdominal viscera was detected. The urine contained neither albumin nor sugar, and the Wassermann reaction, both in the blood and cerebro-spinal fluid, was negative.

The leading features of another case presenting a very similar clinical picture are as follows:—

Case II: Bilateral Progressive Weakness of the Quadriceps Muscles; Congenital Absence of the Brachio-radialis and Biceps Femoris.—A ploughman, aged 59, who complained of weakness of the legs, was seen on November 3, 1921.

History: The patient stated that some six or seven years previously he had first noticed a tendency for the knees to give way if, when ploughing in wet weather towards the end of the day's work, his boots became clogged with clay. Up to this time he said he had been "very smart on his feet," and could run and jump as well as any man of his age. About the same time he found that he was unable to walk upstairs carrying a heavy load on his back. A year or two later he found that when he bent his knees beyond a certain point he would collapse in a sitting position. At a later date he was only able to go upstairs one step at a time, and recently when ascending stairs he had been obliged to hold on to the banister. No history of any similar condition in the family was obtained.

State: The patient looked rather older than his years. His general muscular development was good. There was pronounced symmetrical wasting of the thighs, particularly in the lower two-thirds; the tensor fasciæ femoris muscles, on the contrary, were unusually well developed. Extension at the knee was very easily overcome. In rising from the ground and assuming the erect position, the patient was obliged to support himself by placing his hands upon his knees. The biceps femoris muscle could not definitely be demonstrated, and the brachio-radialis (supinator longus) muscle was absent on both sides. Apart from the weakness of extension at the knee, all the other movements of the body were powerfully carried out. The electrical examination showed a quantitative reduction in the wasted portion of the quadriceps muscles. No abnormality of the nervous or muscular systems was otherwise detected; no fibrillary tremors were observed; the knee-jerks were absent, while the other reflexes presented no abnormality. Neither pain nor other subjective sensory disturbance had been complained of. There was no sensory loss, no sphincter trouble, and no evidence of vertebral defect or disease. Movements at the hip-joints were quite free. The Wassermann reaction was negative. The arteries were slightly thickened and tortuous, but the heart was not enlarged, and the blood-pressure not unduly high. There was no polyuria, and the urine was free from albumin and sugar. The conformation of the abdomen suggested a degree of visceral ptosis, and a bismuth meal showed some delay in the large bowel. Sir Harold Stiles, whom I asked to examine the patient in relation to the condition of the muscles, kindly sent me a detailed report, from which I abstract the following remarks: "There is visible wasting, more especially in the lower quadriceps region of both thighs; the whole of these muscles are either partly atrophied or imperfectly developed. If one assists the patient to adopt the squatting position, he is quite unable to raise himself into the erect posture without great assistance, owing no doubt to the imperfect action of the quadriceps. I was unable to discover any functional impairment in any of the other muscles of the lower extremities (with the exception of the biceps femoris). I formed the opinion that the tensor fasciæ femoris had become hypertrophied to compensate for the weakness of the quadriceps, and I was inclined to think that the anterior fibres of the gluteus medius and minimus muscles had also participated in this hypertrophy. All the muscles of the upper extremities appear to be normal, with the exception of the brachio-radialis (supinator longus) muscles, which are either congenitally absent or so imperfectly developed as to be quite undemonstrable."

The two cases I have just described resembled each other closely. In both there was a symmetrical, atrophic and apparently progressive palsy, limited to the quadriceps muscles, with a quantitative reduction in the electrical excitability, and great diminution or loss of the knee-jerks. Again, no fibrillary tremors were observed, nor was there any subjective or objective sensory disturbance. Further, it is interesting to note that both patients were in the sixth decade of life. These cases, I submit, are to be regarded as examples of myopathy in which the process is limited to the quadriceps muscles. Although it is affirmed that the anterior thigh muscles may be the first to be affected in a case of myopathy, I have not hitherto met with a case in which these muscles were alone implicated.

The second case differs from the first, since in the former the supinator

longus and biceps femoris muscles were absent, while the tensor fasciæ femoris and the anterior fibres of the gluteus medius and minimus were abnormally well developed. The hypertrophy of the tensor fasciæ femoris and anterior fibres of the gluteus medius and minimus was, from its distribution, obviously compensatory.[1] There can, I take it, be little doubt that the absence of the supinator longus and biceps femoris is to be ascribed to a congenital defect. Indeed the question arises whether the disability might not be explained by a congenital muscular defect, the apparent progressive weakness being accounted for by the supervention of the general deterioration of the muscular system, which occurs with advancing years. While this possibility cannot perhaps be excluded with certainty, my strong impression is that there has been progressive localized weakness in the quadriceps group, for the patient, who was a muscular man, stated emphatically not only that up to the time he first noticed his disability he could run and jump as well as any man of his age, but that since that time his weakness had been steadily, if slowly, increasing.

Congenital anomalies of muscle, I may remind you, have been noted by several observers in the myopathies. Thus cases of the kind, in which muscles were congenitally absent, have been reported by Erb, Abromeit, Ziehen, and others. The late Dr. Batten remarked that a child might be born with a myopathic facies and yet for many years there might be no further developments. Apart from congenital absence of muscles, other muscular anomalies in development have also been described in cases of myopathy. Oppenheim, for instance, refers to a case in which he found a muscle with supernumerary digitations. Again, in two cases of the scapulo-humeral type, I observed a bilateral anomaly of the biceps muscle, with which you are very possibly familiar, and which I take it must be of congenital origin. When these patients voluntarily flexed the arm at the elbow, the biceps stood out as a round mass opposite the insertion of the deltoid muscle, while little if any contractile tissue was demonstrable below this point. The facts to which I have referred would appear to indicate that there is a tendency to congenital muscle defects in the myopathies. Other congenital stigmata, on the contrary, do not appear to belong to the picture of these disorders, although in this respect myotonia atrophica, which is commonly classed with the myopathies, is quite exceptional. A congenital muscular anomaly in a case of doubtful myopathy is, I submit, comparable in its significance to that of a spina bifida occulta in a case of central gliosis.

Muscular Dystrophy and Trauma.

Does trauma ever play a part in determining or aggravating a myopathy? This is a question which may come to be a matter of practical importance, as in two cases to which I shall now refer.

Case III: Myopathy with Facio-scapular Distribution attributed to an Alleged Trauma.—A soldier, aged 24, complaining of weakness in the arms, was seen upon September 27, 1916.

History: The patient had been enlisted on August 31, 1914, as an A1 man. He stated that before enlisting he had been in the best of health and that up to that time he had been doing a full day's work as a ship's plater. No comment was made, he said, as to his appearance either in August, 1914, or in October, 1915, when

[1] Hypertrophy of the tensor fasciæ femoris muscles was a striking feature in a case of myopathy of the juvenile type which has since come under my notice. The anterior thigh muscles were otherwise much wasted.

he was examined, stripped, by the medical officer before entering the firing line. In February, 1916, he had been buried, he told us, in a trench and had lain on his face covered by earth and sandbags for about an hour before he was extricated. Although he did not go sick at the time, the patient stated that for some months after this accident he was troubled with a good deal of pain about the upper part of the back and shoulders. Further, he told us that in May, 1916, some three months after he had been buried, he had first noticed some slight weakness of the arms, and that in June, when he reported sick with trench feet, the medical officer to whom he mentioned these pains drew attention to the peculiar appearance of his shoulders. When, after his discharge from hospital in September, 1916, he was returned to his depot, he stated that because of the weakness in the arms he could no longer handle a rifle. He was consequently sent to hospital for examination.

State: The patient was an exceptionally muscular man with very sloping shoulders. It was noted that the sternal portion of both pectorals was absent, and the clavicular portion, especially on the right side, very poorly developed; the upper third of both trapezii was much wasted, and the lower two-thirds undemonstrable; the latissimus dorsi on both sides was very weak and atrophied. The scapulæ projected to some extent when the arms were held horizontally forwards, but the serrati were acting powerfully. The spinati, rhomboids, sternomastoids, teres major and erector spinæ were all very well developed. There was no weakness or wasting of upper arms, forearms, lower limbs or trunk. The tendon reflexes were present and equal; the plantar reflexes were not elicited: there was no sensory loss. A symmetrical facial paresis of the myopathic type was present (the patient had not previously noticed anything amiss with his face). The patient could not forcibly close his eyes, he could not pout his lips, and he stated that he had never been able to whistle. Otherwise the cranial nerves showed no abnormality. Three brothers and two sisters older than himself, and two younger sisters, constituted the family. So far as the patient was aware, none of his seven brothers and sisters nor any other relative had suffered from any similar condition. When this man was examined again upon May 8, 1922, I failed to satisfy myself that the condition had definitely progressed. The patient informed me that he had been engaged in light work since his discharge from the Army.

This was obviously a case of myopathy affecting the face and certain muscles of the shoulder-girdle. Had the condition been determined or aggravated by trauma? Although the patient stated that he had noticed no weakness before enlisting and he had been accepted as an A1 man in 1914, it does not follow that the myopathy had developed since his enlistment for it is to be remembered that a myopathy may exhibit obvious evidence of its presence long before the subject complains of any consequent disability. Indeed in this case the wasting of the muscles was so extreme that it was almost impossible to believe that this could have developed in the course of a year or two, particularly since the impairment of movement complained of was comparatively slight. Hence it appeared almost certain that the myopathy had been in existence for a long period of time. The question of aggravation still, however, remained to be considered. Since no mention of the alleged accident was to be found in the patient's case sheets the veracity of his statements as to the trauma, to which he attributed his condition, may be called in question. The patient's account of the incident and his whole bearing were however such that I felt satisfied that his story was to be accepted as correct. Admitting this to be the case, several possibilities arose for consideration. Had there actually been an aggravation of the condition as the result of the accident, with a result that for the first time the patient had become conscious of a manifest interference with movement? Might it be that the pains from which the patient stated that he had suffered for some time after the accident had resulted in some impairment of movement which the patient attributed in part to muscular weakness? or was it possible that when

the medical officer drew attention to the nature of his trouble, the patient either recognized his weakness for the first time or decided to utilize the information thus obtained in order to obtain discharge. True, the patient stated that he had complained of weakness to the medical officer before the latter examined him, but was this statement correct? The circumstance that when I examined this man six years later I was unable to satisfy myself that there had been any distinct tendency for the condition to progress would seem to militate against the view that the trauma, admitting this to have occurred, had aggravated the pathological process.

The record of another case in which a progressive muscular atrophy, which I believe to be a dystrophy, was attributed to trauma, is as follows :—

Case IV: (?) Muscular Dystrophy determined by Trauma.—Miner, aged 53, was seen on August 7, 1913.

History: The patient stated that upon May 6, 1911, he had "racked" (strained) himself when shovelling coal; that he had been troubled from that time onwards with pain between the shoulder-blades; that in consequence he had been unable to work after the date mentioned: and that in the autumn of 1911, six months after the accident, he had first noticed some weakness of the shoulders which had gradually progressed, and had been followed at a later date by some weakness of the legs. The patient appeared to be an honest, straightforward man who was in no way attempting to mislead, and I was satisfied that his statements were to be relied upon so far as his observation was concerned.

State: Upon examination I found complete paralysis of the right and pronounced weakness of the left serratus magnus with some weakness and wasting of both sides of the trapezius (I have no note as to whether the whole of the latter muscles was involved). There was, too, distinct weakness and wasting of the latissimus dorsi and pectorals especially on the right side, but no paresis or wasting of the deltoids, upper arms, forearms, hand muscles or face. The weakness of the shoulder muscles at the time the patient was examined was undoubtedly such as to unfit him for work. The thighs were somewhat small as compared with the calves (although I have no note as to weakness of any particular groups of muscles in the lower extremities, I would almost certainly have recorded this had it been observed). When the patient lay on his back and was asked to get up, he turned over on his face and "climbed up his thighs." There were no fibrillary tremors. The muscles were not examined electrically. The knee-jerks were present and not exaggerated, and the plantar reflexes were flexion. No pain or other subjective sensory symptom was complained of and there was no sensory loss. There was a slight degree of kyphosis which was no doubt occupational. I have not been able to trace this patient and know nothing of the subsequent history of his case.

The question arose as to whether the condition of the patient was attributable to the injury upon May 6, 1911. In support of the contention of a relationship of cause and effect were the facts that the man was doing a full day's work up to the date of the accident; that he had not worked since; that according to the patient's statement he first noticed weakness of the muscles of the shoulders six months after the accident; that some weakness and wasting of the shoulder muscles had been observed when he was medically examined in February, 1912, nine months after the accident; and that there was good reason for believing that the weakness had been slowly progressive since that time. Although I was of opinion that the muscular atrophy in this case was of the nature of a dystrophy, I knew at the time of no similar cases in the literature in which trauma had been suggested as a determining or aggravating factor, as the facts in this case would seem to suggest. This case however presents a somewhat similar symptomatology to the cases described by Claude and others, in which the condition was attributed to war injuries.

A number of cases of myopathy are reported in which a trauma is affirmed to have played an ætiological rôle. Although I have made no attempt to search the literature exhaustively, I may briefly refer to some of these. "A Case of Juvenile Muscular Dystrophy in a Man after Trauma" which was demonstrated by August Hoffmann at a meeting of the Rhenish-Westphalian Medical Society held at Düsseldorf in 1904,[1] is of interest in this connexion. The patient, a labourer, aged 41, had been severely scalded over the arms and back. From this time onwards he stated that he had noticed progressive weakness in the upper arms and back, with the exception of the deltoids, which were hypertrophic and the supra- and infraspinati which were preserved. The pectorals were almost absent and the nates and thighs were distinctly atrophied. Although the history suggested that the dystrophy had been determined by trauma in this case, Hoffmann in a short communication made a few weeks later stated that he had since discovered that the patient had been exempted from military service twenty-two years previously on account of paralysis of the scapular muscles. It would appear that the dystrophy had remained stationary for twenty years, and during that time the man had done hard work. After the accident the condition had become rapidly worse. Consequently the dystrophy had not been determined although probably aggravated by the trauma. Hoesslin[2] in the same year reported the case of a youth aged 21 in whom two years after a trauma to the right shoulder a dystrophy developed, affecting the right, and later, the musculature of the left scapular region. An exhaustive inquiry showed, however, that some slight muscular weakness had existed before the trauma, and that the muscular weakness had merely progressed more rapidly after the injury. In this case, as in that reported by August Hoffmann, there was no heredity. Again, Erb in his work on "Juvenile Dystrophy" describes the case of a man in whose family there was no history of myopathy, who, at the age of 34, was rendered unconscious as the result of an accident in a stone quarry, and who a year and a half later developed a typical muscular dystrophy. Another case reported by Hitzig, and quoted by Erb, was that of a boy aged 13, who, following a blow in the region of the right temple, developed progressive weakness of the right, and three years later of the left, arm, with a typical picture of dystrophy. In this case there was no hereditary history. Two further cases are referred to by Erb. In one of these, reported by Boye, the symptoms of dystrophy were stated to have commenced in the right arm three months after an injury in the region of the right supraclavicular fossa. The other case, recorded by Israel, was that of a healthy labourer, aged 37, who sustained a severe contusion of the left hip which necessitated confinement to bed for three weeks. The dystrophy which was present at the time of examination was said to have commenced at the time of the injury and to have been accentuated by a second trauma. No hereditary history was forthcoming. Spiller, in introducing the discussion on the myopathies at the London meeting of the International Medical Congress of 1913,[3] reports an interesting case:—

It was that of a man, aged 19, who fell from a scaffold, a distance of 60 ft., and possibly struck his shoulders. He was able to resume his work the same day but the following day his arms were stiff and painful and remained so for two days. From that time he could not lift anything and could do only light work. He noticed about four months later that the muscles of both shoulders were wasting and that he had difficulty in using them. The muscles wasted, and when seen a year after the accident

[1] *Münch. med. Wochenschr.*, 1904, li, p. 1027.

[2] *Ibid.*, 1904, li, p. 1156.

[3] *Trans. Seventeenth Internat. Congr. Med.*, sect. xi, Neuropathology, pt. i, p. 129.

he could raise the upper limbs only to the level of the shoulders. The deltoid, biceps, triceps and pectoral muscles were much atrophied. No fibrillary tremors were observed and no sensory disturbance for touch or pain. The patellar reflexes were present but not increased. There was no family history. A positive Wassermann reaction was obtained. "The atrophy conformed to Erb's juvenile type and apparently had been started by the trauma, which is an interesting fact" (Spiller).

The case reports to which I have referred do not appear to afford definite proof that a true myopathy is ever determined by trauma. Indeed, a perusal of the records which I have been able to find in a cursory study of the literature indicates the caution which must be exercised before a conclusion as to a relationship of cause and effect can be legitimately arrived at. In order to establish such a relationship there must be definite proof: first, that the case is one of muscular dystrophy; secondly, that there has been a trauma of some severity; thirdly, that there was no evidence of muscular weakness preceding the accident; and, fourthly, that the symptoms of myopathy had developed within a comparatively short time after the trauma. The possibility of coincidence must always be borne in mind. In relation to this problem I would again emphasize the fact that a myopathy may exist long before the muscular defect becomes so pronounced as to give rise to occupational inconvenience. Thus Hoffmann's patient, who had been exempted from military service on account of wasting of the scapular muscles, was able to continue the arduous work of a labourer for twenty years after this date. The case of a man, aged 61, the subject of a pronounced facio-scapulo-humeral myopathy, at present under my care in hospital, illustrates the same point. This patient, who is still able to follow his occupation, that of a plumber, informs me that although his unusual build and sloping shoulders have attracted attention since boyhood, it was only some seven years ago, since which time he has found increasing difficulty in working with his arms above his head, that he has been conscious of any muscular disability.

While the evidence as to an ætiological relationship of trauma to true myopathy is, I think, to be regarded as inconclusive, it is necessary in passing that I should remind you of the group of cases to which attention has been drawn by Claude, Vigouroux and Lhermitte.[1] These cases, in which the pathological condition followed a bullet or shell wound, were characterized notably by a unilateral or bilateral wasting of the trapezius and serratus magnus muscles and, it may be, other muscles of the shoulder girdle. On account of the distribution of the wasting, the purely quantitative changes in the electrical responses, the normal condition of the reflexes and sensation, and the fact that the position of the wound was not such as to interfere with the nerve supply to these muscles, the authors class these cases under the term muscular dystrophy of the myopathic type. They regard the condition as due to a development under the influence of trauma of an "impairment of nutrition of certain muscles giving rise to atrophy and secondarily to paresis." Huet and Français have described somewhat similar cases following war injuries. Possibly the last of my series of cases (Case IV), Spiller's case, and some of the other cases referred to are of a similar nature. These cases, however, would appear to depend upon a pathological process quite distinct from the true myopathies. Madame Athanassio-Benisty[2] in discussing Claude's cases remarks:—

"We have seen three cases of unilateral paralysis of the trapezius and serratus magnus, the first following an attack of rheumatism of the shoulder (according to the

[1] *Presse médicale*, 1915, xxiii, p. 393.

[2] "Treatment and Repair of Nerve Lesions" (Translation). Edited by E. Farquhar Buzzard, 1918, p. 156.

diagnosis on the case sheet), the second from carrying a heavy burden fixed by a strap to the shoulder, and the third which had occurred after a wound in the scapular region. The symptoms were identical with those described by Claude, Vigouroux and Lhermitte."

The association of isolated unilateral paralysis of the serratus magnus and the lower portion of the trapezius muscles is well known. In 1903, J. W. Struthers and I recorded such a case in which the symptoms were first noticed six days after an operation for appendicitis and in which we could obtain no certain evidence of trauma. At that time we were able to find references to seventeen cases of paralysis limited to these muscles, and I have seen several examples since.

May Recovery take place in Cases of Myopathy?

This problem was discussed by the late Dr. F. E. Batten in the *Quarterly Journal of Medicine* for April, 1910. After reviewing the literature of reported recoveries, Batten selected three cases which, in his opinion, might be regarded as instances of myopathy in which recovery had occurred. Thus of a case reported by Marina he says: "This case must, I think, be accepted as a case of myopathy in which recovery had taken place." Regarding Jendrassik's case he remarks: "In this case, again, the evidence for the diagnosis of myopathy might be criticized, yet it is difficult to see in what other group the case could be placed." In Erb's case he thinks "the diagnosis can hardly be called in question." (Spiller in his introductory paper on the relation of the myopathies at the London meeting of the International Congress refers to a case of myopathy in "an adult in which so great improvement occurred that it amounted to recovery.")

The case which I am about to report is of particular interest and importance since the patient was examined by Dr. Batten.

Case V: Recovery in a Case diagnosed as Myopathy (? Toxic Neuritis).—W. C., aged 5, an only son, one of a family of four, one sister being older and two sisters younger than the patient, was seen on September 26, 1919.

History: As a baby the patient had appeared to be quite healthy, although his mother stated that when placed on his back he had unusual difficulty in turning over. I have no note as to when the child first began to walk, but according to his mother he showed a tendency to waddle when walking about Christmas, 1917, i.e., two years before I saw him. The onset would seem to have been gradual. In May, 1918, the boy was seen by Dr. Batten. I have a letter written to the patient's mother upon May 20, 1918, in which Batten expressed himself in the following terms: "Although I must admit that there is much in favour of the diagnosis of pseudo-hypertrophic paralysis already given you, yet there are several unusual features which make me hesitate to give that diagnosis unreservedly, and I should be prepared to consider the possibility of a toxic neuritis, from which recovery is possible after a period of about eighteen months or two years. I very much want to find out about the family history. If it can be shown that the members of the family suffered from a form of paralysis which corresponds in course and type with that now occurring in W., then I fear it would not be possible to accept any other diagnosis than that of myopathy."

The patient's mother informed me that Dr. Batten, after examining the boy again, had written her that he was satisfied that the diagnosis of myopathy was correct, and that only three cases were known in which recovery had taken place. Unfortunately I have not the letter in which Batten expressed this opinion. The patient's medical attendant wrote me, on March 24, 1922, as follows: "I am sorry not to be able to send Batten's last letter, which would have helped you more than these notes; there was a later one which the boy's mother has mislaid, but which I saw, in which he stated that he was inclined to change his opinion, and regarded the condition as a

myopathy. I may say that there was no family history of importance. The first time that Sir Harold Stiles saw the boy he was a typical pseudo-hypertrophic in gait and manner of getting up from the floor, and laxity of the shoulder girdle when lifted with one's hands in the armpits. Professor Gulland also saw him on the same day and diagnosed myopathy."

The child's mother stated that on August 2, 1919, she took him to a herbalist, from whom he had been receiving treatment since. She further told me that the boy soon began to improve under this treatment, whereas the massage and electricity he had been having previously had produced no effect. On the other hand, the patient's doctor writes me, on March 24, 1922: "The old herbalist woman gets the credit for a cure, but he had begun to improve before that."

State: My notes at the time I examined the patient (September 26, 1919), were as follows: "The boy is very active and alert mentally. When he walks there is a slight lordosis and a distinct tendency to waddle; he cannot stand securely on his toes, nor can he raise the toes from the ground and walk on the heels; if he bends his knees to any extent they give way, and he falls to the ground. When he lies on his back and is asked to get up, he turns over with a little difficulty, and rises very much like a case of pseudo-hypertrophic paralysis. Although he does not require to place his hands on his knees in so doing, he gives the body a sudden jerk backwards in assuming the erect posture. He can bend down and touch the toes with the knees very slightly flexed, and can raise himself from this posture without using his hands. The muscularity is poor as a whole, but I cannot say that there is any definite localized wasting, nor is there any hypertrophy. The scapulæ do not project, and I can detect no weakness of the upper extremities. When the patient is lifted from the ground with the hands under his arms, there is no tendency for the shoulder-girdle to move upwards. The right knee-jerk is obtained with a little difficulty, the left is absent, as are both ankle-jerks; the plantar reflexes are indefinite. There is no weakness of the face, and no defect of the ocular movements.

On March 24, 1922, the patient's doctor wrote to me as follows: "W. C. is, as far as I know, quite strong and well; it will be about a year since I have seen him, as the family have removed (from this neighbourhood for a time). He has gone through the minor children's ailments without any mishap. He is now able to ride a tricycle and a scooter; the only remains of his trouble is his gait, which has a distinct waddle."

An assertion to the effect that a case of a well-known disease which is commonly regarded as incurable has recovered demands most careful consideration before the evidence can be accepted as conclusive. The possibility of an error in diagnosis at once suggests itself as the most probable explanation of the facts, and the data must be capable of withstanding the most severe criticism before the contention can be admitted. Since Dr. Batten hesitated to give the diagnosis of pseudo-hypertrophy unreservedly when he first examined this patient, the case may appear to be of little value in relation to the question at issue: on the other hand, we have the statements of the child's mother and medical attendant that after Batten had seen the child on a second occasion he committed himself to a diagnosis of myopathy. While it is possible that the case was one of myopathy in which recovery has taken place, it seems on the facts much more probable that it was not an example of this disease, but that it so closely resembled myopathy that Batten was driven to a diagnosis by a process of exclusion. Had there been a family history of myopathy, the probabilities in favour of the diagnosis would have amounted to a certainty. This case illustrates the importance of a positive family history in any case in which the diagnosis is doubtful. Is it not possible that in the three cases of reported cure cited by Batten the diagnosis may have been incorrect, particularly since in none does there appear to have been a family history of myopathy?

REMARKS ON MYOTONIA ATROPHICA—REPORT OF A CASE WITH AUTOPSY.

In conclusion I shall allude briefly to some features of myotonia atrophica, an affection which has been classed with the myopathies. Thus the tendency to a symmetrical distribution, the myopathic facies, the absence of fibrillary tremors and of qualitative changes in the electrical responses, the occasional presence of hypertrophy and the familial tendency, are features common to the two conditions. On the other hand, in certain respects the symptoms of myotonia atrophica differ very notably I think from the myopathies. Thus, apart from the myotonia, a symptom which has not, so far as I know, been observed in the true myopathies, the distribution of the muscular atrophy does not correspond to any known myopathy, while, further, transitional types have not been described. I would also emphasize in this relation the frequency of congenital stigmata of various kinds in myotonia atrophica, while, in addition, I have been forcibly impressed by the peculiar mental state in several cases of this disease with which I have met. Again, it is to be remembered that these patients do not die of the disease. Myotonia atrophica cannot be regarded as a disease of extreme rarity. Batten and Gibb[1] collected twenty-nine, and Steinert[2] twenty-six cases from the literature in 1909, while Addis and I[3] were able to find reports of sixty-one cases four years later. Twelve cases in all—ten males and two females occurring in six different families—have come under my personal observation. The symptoms may exist for long without causing any appreciable inconvenience. Thus, one of my patients, who consulted me regarding a facial neuralgia, was annoyed—an instance of the abnormal mental state to which I have referred—when I proceeded to inquire regarding his muscular development. He practically told me it was no business of mine, since he had been born like that, and had never been caused any real inconvenience thereby. Another patient who presented the characteristic symptoms of the disease in a pronounced form was sent to me with an old-standing hemiplegia which had developed in early childhood. This patient's two younger brothers whom I also examined both exhibited the myopathic facies, with some wasting of the sternomastoids and the myotonic phenomena in the upper limbs. They persisted in saying there was nothing the matter with them. All three, but notably the younger brothers, were obvious psychopaths, and their parents, who were most respectable, hard-working people, had been caused much distress by the boys' inability to realize the meaning of truth and honesty. Another man who attended my clinique was constantly changing his situations, under the impression that his employers were endeavouring to get the better of him in one way or another. At one time he flatly denied that he had any brothers, and only admitted his mistake when I confronted him with a photograph of himself and two brothers which I had taken some years previously. The testicular atrophy first noted in myotonia atrophica by Steinert in his paper published in 1909 is of interest. Addis and I found this observation recorded in eight cases. I have now seen three instances in which testicular atrophy was a striking feature. In one or two other cases, too, I was inclined to think that the consistency of the testicles was unduly soft although they were not obviously reduced in size. The testicles were about a third of their normal bulk in the case of a married man, aged 43, the father of a family of six, who has just left my ward. In this case the first manifestations of the disease had shown themselves seven years

[1] *Brain*, 1909, xxxii, pp. 187-205.
[2] *Deutsche Zeitschr. f. Nervenheilk.*, 1909, xxxvii, pp. 58-104.
[3] *Edin. Med. Journ.*, 1913, n.s., xi, pp. 21-44.

ago ; the patient's youngest child was a year old, and for fourteen months he had lost all sexual desire. The possibility of an ætiological relationship between this testicular atrophy and myotonia arises in view of these facts. There are two other points in the symptomatology of the disease, such as the cold hands so frequently complained of, and the early loss of hair observed in some cases, which raise the possibility of a glandular disturbance as an underlying ætiological factor. The fact that testicular atrophy had been observed in almost 20 per cent. of the male cases reported up to 1913 is certainly striking. The arguments that in the remaining 80 per cent. no atrophy was observed, and that no disturbance of the primary sexual functions may be observed until the disease has been in existence for years, do not, you will probably agree, justify us in dismissing forthwith the possibility of a disturbance of testicular secretion at a much earlier date. This relationship, indeed, calls for elucidation. I may say that I have seen no obvious effects produced in two or three cases of the disease in which I have employed testicular preparations.

A pathological examination of the nervous system has only been reported, so far as I have been able to ascertain, in two cases up to the present time. One of these cases was recorded by Steinert in 1909, the other by Hitzenberger in 1920. I shall conclude by giving you a résumé of a case which was under my care and which was examined pathologically by Dr. Dawson.

Case VI: Case of Myotonia Atrophica with Autopsy.—W. H., aged 52, a valet and masseur, was seen in July, 1913. On inquiry it was elicited that he was the elder brother of a family three of whom I had previously examined and found to be affected by myotonia atrophica. These cases were reported, in conjunction with W. R. Addis, in the *Edinburgh Medical Journal* for July, 1913, pp. 21-44.

History: The patient had been married for seven years and had one child, a girl, aged 6, who was said to be well and strong. As a young man he stated that he had been particularly strong and a first-rate athlete; he had, he told us, jumped 5 ft. 10 in.; he used to play football and he had been a good boxer. As a younger man he was able, he said, to lift two 56 lb. weights above his head. Again, he stated that he was able to stand extremely strong electric currents; thus, he said that many years previously, when at a fair in America, he and two of his companions tested themselves on an electrical instrument, when he found that he was able to stand the full strength of the current without discomfort. The showman told him that his nerves must all be dead, and offered him, he said, five dollars a day to travel round America with him for purposes of exhibition. He stated that he had never had venereal disease, and the Wassermann reactions were found to be negative. As a young man he had been a fairly heavy drinker but he stated that be had taken practically no alcohol for the past ten years.

For some sixteen years he stated that he had noticed that after firmly closing the hand he had had difficulty in extending the little and ring fingers of the right as also the little finger of the left hand. He had been unable for some years to do any heavy work because of the weakness of the grasp, but for the past six years he said that his hands had been distinctly stronger. Eight years before he came under my notice his right eye had been operated on for cataract while the left eye had been needled for the same condition six years later. Two and a half years before I saw him he had had pneumonia: he had had a cough since, and upon examination I found that he was suffering from a bronchiectasis. For three months the patient said that he had been going downhill rapidly, and that he had lost almost two stones in weight.

State: The facial appearance was striking; there was great hollowing in the temporal and masseter regions on either side; he often sat with his eyes half closed and the eyeballs turned upwards so that only the whites of the eyes were visible. There was no ptosis. The palate was very high and narrow. He was almost bald. The voice was distinctly hoarse; articulation was a little thick, with a distinct nasal

intonation. He gave a history of occasionally having brought fluids through his nose when swallowing. He said that he had often noticed in the morning that he had difficulty in opening his eyes. The masseters and temporals were weak and much wasted. He could not close his eyes firmly and he was unable to whistle; the facial movements were indeed very defective. The sternomastoids were both distinctly atrophied; the general musculature, too, was very poor. The forearms were especially wasted, particularly on the ulnar aspect; the abductor pollicis was practically absent on both sides; otherwise there was no definite wasting of the hand muscles. No myotonic phenomena were observed in the grasp or on percussion of the other muscles though carefully tested for. (There were no fibrillary tremors nor was there any muscular hypertrophy.) Though he stated that he became rather easily tired after walking, there was no obvious localized weakness or wasting of the lower extremities. The deep reflexes were unaltered. There was no sensory loss or sphincter disturbance. The left testicle was rather smaller than the right though I could not say that it was definitely atrophied. For the past three years there had been no sexual desire. About the root of the right lung the physical signs suggested the presence of a cavity and there were some moist sounds. The sputum was tested repeatedly for tubercle with a negative result. There were two pimples, one on either side of the neck immediately below the larynx; he stated that from childhood up to eight years ago a discharge occurred from these pimples from time to time. *The patient died on March* 4, 1915, the immediate cause of death being an attack of erysipelas affecting the face.

The post-mortem was carried out by Professor Drennan on March 4, 1915; the following are some of the more striking facts observed: There was marked emaciation. The skull, membranes, brain and spinal cord showed no gross abnormality. The pituitary body was of normal size, as also was the thyroid gland. On section the thyroid was seen to be pale brown in colour, and showed no colloid to the naked eye. The left lung was smaller than the right, and the smaller bronchi, which contained thick pus, were distinctly dilated; extending from the root there were several larger bronchi, showing marked bronchiectasis; there was some recent fibrinous exudate over the lower lobe. The heart was small, the muscle of a dark brown colour. The abdominal organs showed nothing worth noting. The spleen was somewhat small and soft; the pancreas was also small; the kidneys showed a slight uniform diminution of the cortex; the suprarenals were both very thin, the medulla was small in amount, and the cortex thin and of a brown colour. The prostate was not enlarged. The left testicle appeared slightly atrophied; both testicles were soft, and on section of a light brown colour. In the situation of the thymus there was a small lobule of fatty tissue, resembling thymus in shape; in the left portion of this there was a small pocket of pus. The orbicularis oculi was very thin and pale yellow. The temporal muscles were thin and somewhat pale. The sternomastoids, which were uniformly small, were of the usual colour. In general, the change in the muscles consisted in a diminution in size; there was no evidence of fatty change, and their colour was practically normal; if anything, some muscles appeared slightly darker red than usual. Subcutaneous fat was almost absent.

The histological examination of the nervous system was carried out by Dr. James Dawson, whose conclusions were as follows: "The points to be noted in the investigation of the nervous system may be summarized thus: (1) No tabiform degeneration; no change in extra-medullary posterior or anterior nerve roots; no definite change in posterior entry zone nor in collateral fibres. No infiltration in the meninges nor walls of the blood-vessels. No uniform gross changes in cells or cell-groups of grey matter nor posterior root ganglia (except pigmentation); no changes in the nerves of the brachial plexus nor cauda equina. (2) Weigert and Marchi sections show only the slightest evidence of degeneration; there is marked pigmentation of the anterior horn cells and cells of the posterior root ganglia; and amongst these cells are very numerous amyloid bodies; in the column of Burdach, at level of the third cervical segment, occur a few heterotopic ganglion cells."

(The various tissues which were kept for histological examination, with the exception of the spinal cord which was handed over at the time to Dr. Dawson, have unfortunately been mislaid.)