

THE PERONEAL FORM OR LEG-TYPE OF PROGRESSIVE MUSCULAR ATROPHY.¹

BY B. SACHS, M.D. (NEW YORK).

Professor of Mental and Nervous Diseases in the New York Polyclinic.

DURING the past few years the subject of progressive muscular atrophy has been carefully investigated. As a result of these investigations we have learnt to discriminate between the spinal amyotrophies, and the various types of primary muscular dystrophies.²

The cases, which are still *sub judice* are those which Tooth has described as representing the peroneal form, and which I suggested a year ago might be said to represent the leg type of progressive muscular atrophy.

The clinical symptoms and pathology (whether of spinal or nerve origin) and the relationship of this peroneal form to Duchenne's atrophy—the arm type—are the points on which further evidence is needed.

There is a consensus of opinion however on this one point, that the cases in question do *not* belong to the category of primary muscular dystrophies.

At the outset let us note this one curious fact, that the hereditary or family element in cases of the leg type has been well established; indeed, so prominent a factor is it that many examples of this class are to be found among those cases which were described years ago by Eichhorst, Eulenburg, Hammond, Leyden, and others as hereditary forms of progressive muscular atrophy; whereas the influence of heredity is not so clearly proven in the cases of the arm type (Duchenne's type).

¹ Read before the American Neurological Association, June, 1889.

² The literature of the subject to be found in Schultze's Monograph, 1886; Tooth, *BRAIN*, vol. x., and Sachs, *N. Y. Med. Jour.*, December, 1888.

The peroneal form was first recognised by Charcot and Marie,¹ and independently of them by Tooth² in England. The German neurologists were originally opposed to the creation of another type, but this type has now received the sanction of Erb's clinic in an article recently published by his assistant Hoffman³ and entitled "Ueber progressive neurotische Muskel-Atrophie"—a title which indicates the author's views as to the pathology of this disease. To this able contribution of Dr. Hoffman I shall have occasion to refer repeatedly in the course of this article.

Hoffman has attempted to establish a most rigid and accurately defined type of progressive muscular atrophy, adding a number of symptoms to those described by former authors. In describing a new form of disease it is desirable that all the possible symptoms should be clearly set forth, but it is a mistake I think to confine the clinical limits so closely that a slight variation would take certain cases out of the given clinical group.⁴ Charcot's description of multiple cerebro-spinal sclerosis differs in many respects from numerous cases which undoubtedly belong to this same category. And so with the disease under question. Variations must be allowed for, particularly in the case of true family ailments. The three brothers whose affliction Hoffman has so ably described represent the disease as developed in their family. His fourth case in another family differs in some important respects, and the type of this disease can only be established by fixing upon the symptoms which occur most frequently in all reported cases. As the number of such cases is still very limited, I believe that the cases which I report upon will help to define the clinical aspects of this disease. Certain features which Hoffman described with great care could not be made out in these cases, and yet there can be no doubt whatever that these cases must be regarded as genuine examples of the peroneal form of progressive muscular atrophy.

¹ *Rev. de Med.*, 1886.

² *Dissert.* 1886; also *Critical Digest*, BRAIN, vol. x., 1888.

³ *Arch. f. Psych.*, vol. xx., 1889.

⁴ It seems doubtful for instance whether the sensory symptoms described by Hoffman will be found in a majority of these cases.

ILLUSTRATIONS TO THE PAPER ON THE PERO-
NEAL FORM OR LEG TYPE OF PROGRESSIVE
MUSCULAR ATROPHY, BY DR. B. SACHS, NEW
YORK.

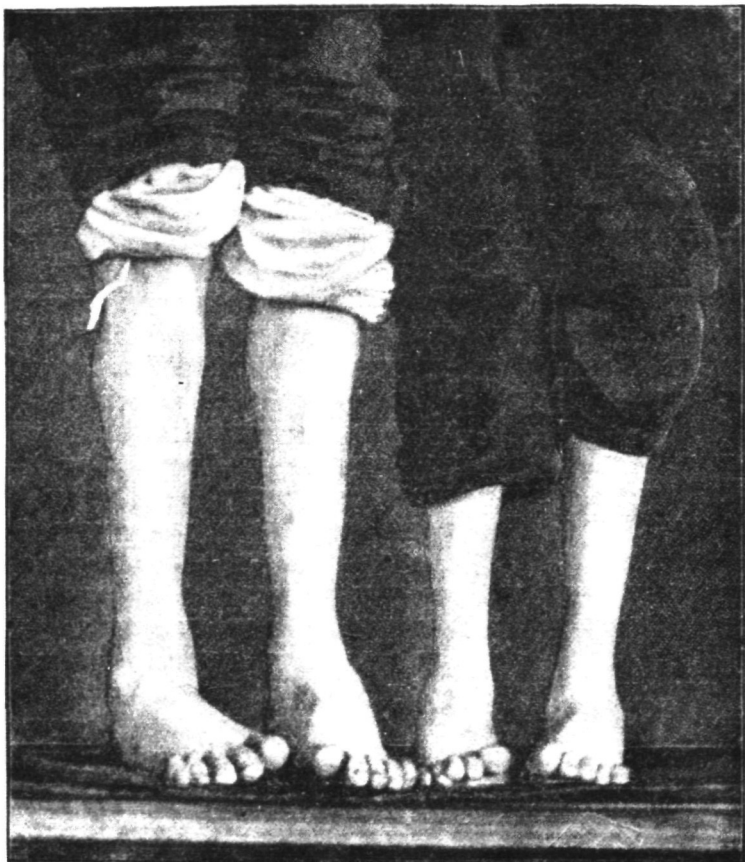


FIG. 1.—Showing condition of feet and legs in both boys, eight months and one year respectively after first operation.

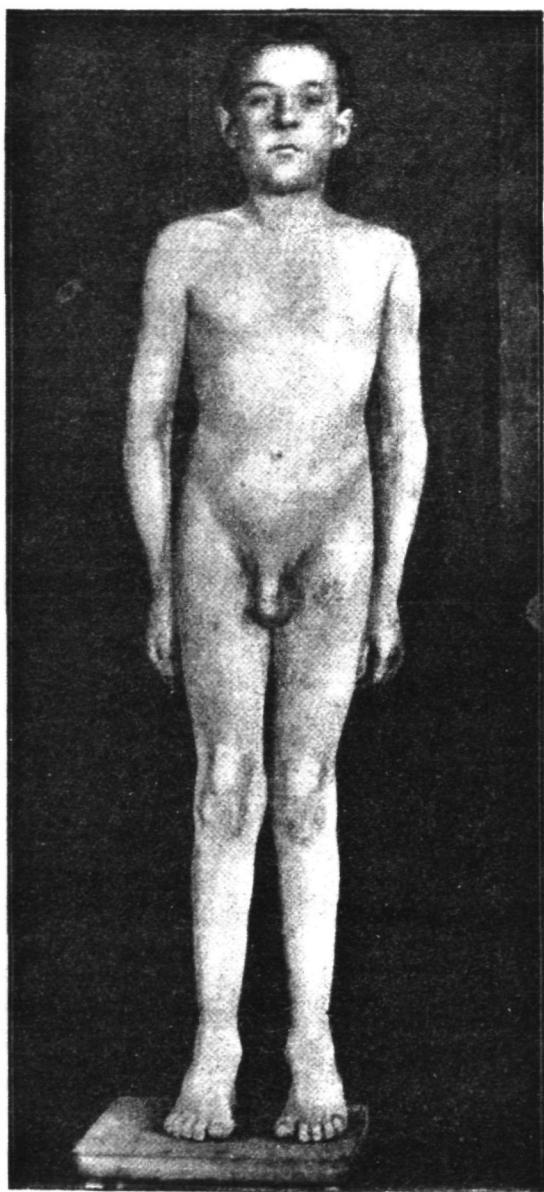


FIG. 2.—F. W. after second operation.

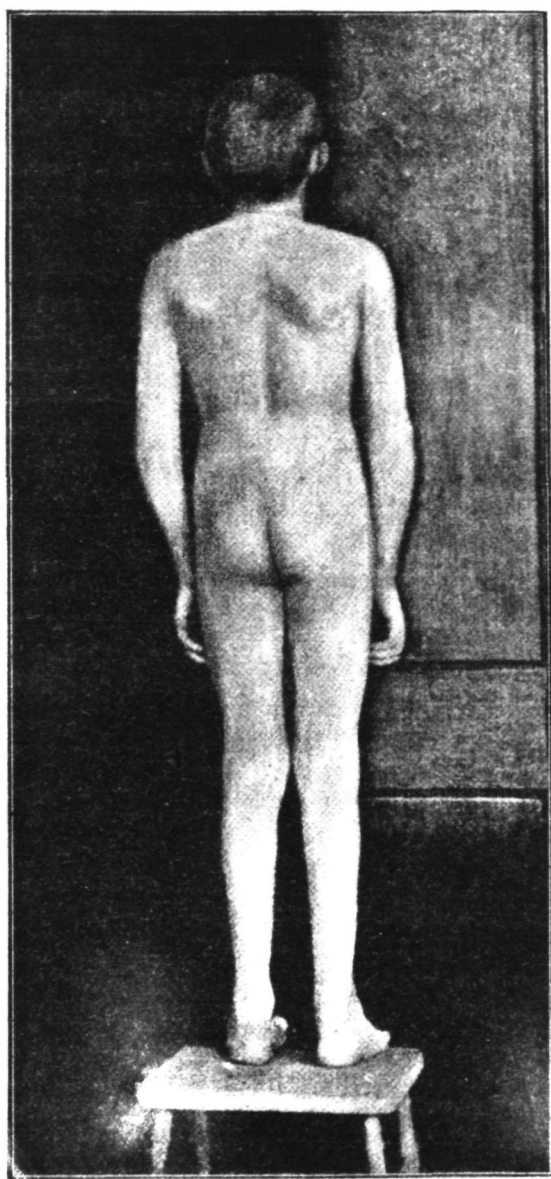


FIG. 3.—F. W., wt. 13.

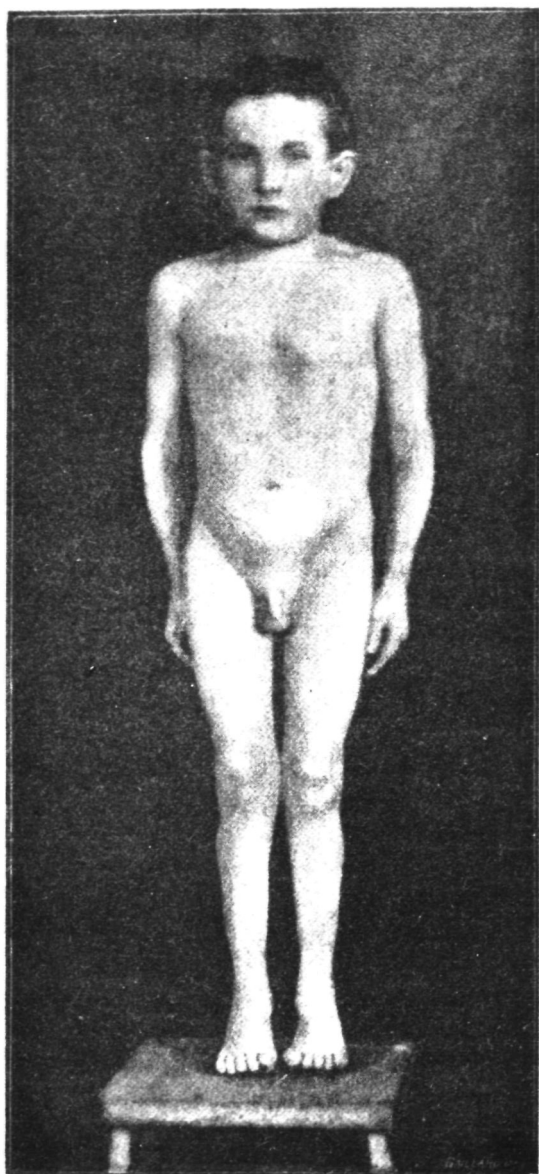


FIG. 4.—After second operation.

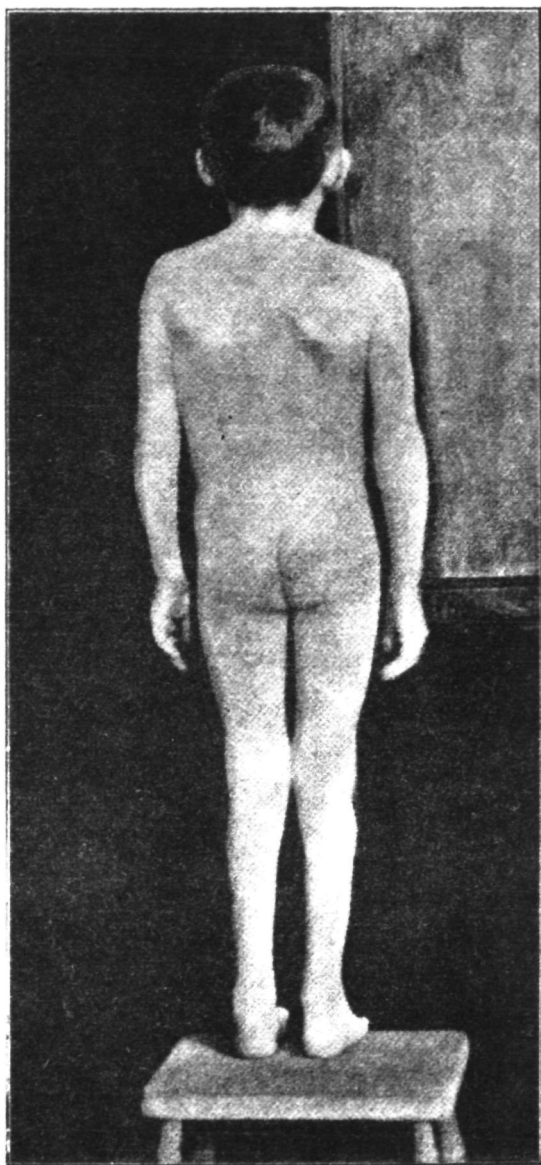


FIG. 5. W. W., *æt.* 10.

I am indebted to the great kindness of Dr. Gibney for the opportunity of studying the cases which I report, and to Dr. Milliken for the pains he has taken in looking up the records of the New York Hospital for the ruptured and crippled. The cases are as follows :—

The family history is very meagre. The father, a Bavarian, is dead; cause of death unknown. Mother living, healthy, thirty-two years of age. The two patients are the only children. Both children said to have been born healthy, to have shown disturbances in the use of the legs at an early day, and to have acquired double club feet at the age of five years. In both cases a thorough drenching of the skin by exposure to the wet is mentioned, but the exact relation of this factor to the development of the disease is not well established.

CASE I.—The older boy, Frank, is now thirteen years of age. He was admitted to the Hospital for the Ruptured and Crippled November 29th, 1887. The hospital record states that the boy stands on toes and balls of feet; that he walks with rolling outer part of feet, with knees in (marked genu valgum); pronounced kyphosis of lumbo-sacral region. Movements of spinal column restricted slightly. Thighs fairly developed, and movements of these muscles appear normal. No voluntary movements of peroneal group; when patient attempts to flex feet dorsally, hyperextension of great toes is the result. Little motion at the ankle joint. Both Achilles tendons shortened. Feet can be passably flexed dorsally by manual force to 150° . The head of the astragalus stands out prominently in both feet. Comparative measurements: right calf, $8\frac{1}{4}$ inches; left calf, $8\frac{3}{4}$ inches.

December 10th, double achillotomy was performed by Dr. Gibney; eighteen days later plaster of Paris splints removed. Feet can be flexed to angle of 90° (dorsal flexion). The patient was given proper shoes and discharged March 29th, three months after operation. At that time he walked well with sole flat on the ground; slight valgus position of feet. Only eight months later boy was re-admitted and it was noted that he again had the characteristic rolling gait of talipes equino-varus; stands with toes slightly inverted, the whole foot rolled outward. Shows rounding of outer surface of both feet, with special prominence of tarsal bones of right foot. Fig. 1 represents the appearance of the legs and feet at this time. Both feet can be forcibly flexed. Treatment

—double achillotomy, division of plantar fasciæ, application of Thomas's tarsiclast, and immobilized plaster splints. In November, 1888, immediately preceding the second operation, I had occasion to make a short examination of the patient. I noted very marked atrophy of thighs and legs, the atrophy being distributed almost equally over all the muscles. His walk was very much like that of a boy with a poliomyelitis affecting both legs. He could flex his toes but very little, on the left side somewhat better than on the right side, and plantar flexion was slightly better than dorsal flexion; but this difference was more marked on the right than on the left.

Comparative measurements: right thigh, 13 inches; left thigh, 13 inches; right calf, $9\frac{1}{2}$ inches; left calf, $10\frac{1}{2}$ inches. Patellar reflexes normal. Myotatic excitability of vasti greater on left than right side. At this time some attention was paid to the electrical examination, but the results can be best given by stating what I found on a subsequent but equally hurried examination, May 6th, 1889. In the course of this later examination the following notes were made. The boy is well developed with the exception of his lower extremities; the muscles of the face and arms appear normal. No hypertrophy anywhere. Shoulder muscles firm; a slight wasting however of infra-spinatus. Grasp of upper extremities good, and supinators stand out well. No deformities of hand. Measurements: Right arm at the largest circumference, $7\frac{1}{2}$ inches; left arm, $7\frac{1}{2}$ inches; right forearm over supinators, 8 inches; left forearm, 8 inches. Trunk and thorax muscles normal in every respect. Lower extremities: patient can raise heel off ground, but cannot get on tip toes; keeping heels on the ground, he cannot flex the foot upward, though he succeeds in moving the big toe. The gross muscular power of the legs and thighs decidedly diminished. Boys have great difficulty in walking up and down stairs. No mechanical excitability of leg and thigh muscles to be obtained. Patellar reflexes present and equal. Plantar reflexes absent. Upper extremity reflex weak but present. Measurements: right thigh, $12\frac{1}{2}$ inches; left thigh, $12\frac{1}{2}$ inches; right leg, $9\frac{1}{2}$ inches; left leg, 10 inches. Sensation: tactile sensation good all over the body; can distinguish figures written on skin. Temperature sense normal. Pain sense slightly diminished as determined by tolerance to faradic stimulation. Muscular sense perfect. Extremities slightly livid. No fibrillary contractions.

Electrical Examination.—Upper extremity: both currents give normal responses. Lower extremities: faradic examination, thigh

muscles respond to strong currents only; leg muscles, posterior group respond to strong currents in anterior tibial muscle and in extensors of toes and foot; no response on direct or indirect faradic excitation. Galvanic examination limited on account of want of time to muscles below knee. From peroneal nerve first KCC with 13 milliampères (ma.) in extensor hallucis longus; no ACC obtainable with current of 18 ma. Direct excitation of tibialis anticus KCC 14 ma.; ACC 16 ma. Extensor hallucis longus, KCC 13 ma.; ACC 16 ma. All contractions slow. No contraction could be obtained with the strongest currents at command in extensor communis digitorum. In the peronei muscles KCC at 13 ma.; ACC not at 20 ma.

The electrical examination, therefore, shows a diminution in the faradic responses of the *thigh* muscles; the galvanic examination of these muscles was not made at this time, but at a former examination I noted contractions with strong, almost unbearable currents, the anodic being equal to the cathodic effect. In the leg muscles we find that the posterior group alone respond to strong faradic currents, no response in all the anterior muscles, and in these muscles the galvanic examination shows that unusually strong currents are necessary to bring about contractions in the muscles, but that the formula is not altered; that the contractions are decidedly slow. Furthermore we may note this difference from typical poliomyelitis, that the muscles which were in a tolerable state of preservation could be excited by the indirect method fully as well as by the direct. We have, therefore, a partial re-action of degeneration in most of the muscles of the legs.

CASE II.—The younger brother was admitted to the hospital November 17th, 1887. He presented double pes equino-varus; when admitted to the hospital he walked on crutches, as he could not get along without their support. The hospital records read as follows:—There is a callosity on each foot over fifth metatarsal bone; foot extended to an angle of 135° , plantar flexion, and inverted to an angle of 142° ; considerable force needed to bring feet into normal position. Marked shortening of tendo Achillis and plantar fasciæ of both feet. Foot arched (pes cavus); when at rest, inner side does not touch floor. Measurements: right calf, 7 inches; left calf, 7 inches. Double achillotomy performed by Dr. Gibney November 25th, 1887; separation of ends $1\frac{1}{2}$ inches on the left, and almost the same on the right side. Feet were flexed dorsally to about 80° and plaster-of-Paris splints applied. Discharged four months after the operation with the note that the patient walks quite well, soles flat on the ground,

toes slightly inverted. He returned to the hospital November 12th, 1888, with paralytic limp. The right foot can be everted about 10° beyond the median line and flexed to about 85° . The left can be everted to median line and flexed to 90° . Stands with toes inverted; whole of outer surface rolled outward and downward (see Fig. 1). Double achillotomy was again performed. Division of plantar fasciæ was made. There followed application of Thomas' tarsiclast and plaster splints, with the result as shown in Figures 4 and 5. Two months after the operation it was noted that the patient walks on his feet in typical calcaneus when using his shoes, without apparatus squarely on soles of feet, with disposition of feet to roll inward. He can voluntarily flex ankle-joint a little beyond 90° , but in so doing toes are hyper-extended and the back of foot drops. Still walks with peroneal type of paralysis; marked disposition to pes varus. I had occasion to examine this boy before the second operation and again on May 6th of this year. I give the results of both examinations.

William is of stouter and shorter stature than his brother. Intelligence good. His external appearance peculiar in this that his broad chest and fat stomach are in curious contrast to his spindle-shaped extremities. The circumference of chest 26 inches; right arm, $6\frac{1}{2}$ inches; left arm, $6\frac{1}{4}$ inches; right forearm, $6\frac{1}{2}$ inches; left forearm, $6\frac{1}{4}$ inches. Grasp of both hands very weak, cannot move dynamometer more than two degrees. A general emaciation of all parts of upper extremities. Very distinct atrophy of infra-spinatus. In the legs, general atrophy is very well marked: right thigh (4 inches above patella), 11 inches; left thigh, $10\frac{1}{4}$ inches; right calf, 8 inches; left calf, $8\frac{3}{4}$ inches. Walks with a slightly waddling gait, and has most difficulty in walking up and down stairs. Keeping heels on floor, he can raise toes slightly on left side and less well on right side. He can raise left leg on tip toe; hardly succeeds in doing this with right leg, and in attempting to raise the whole body on tip toes falls forward.

Sensation.—Tactile sensation normal as determined by cotton, pin test, and the writing of numbers on skin. Temperature sense normal. Pain sense exaggerated as determined by faradic current. Muscular sense normal. Plantar reflexes present and knee jerks about normal. Slight lividity of legs; not as marked, however, as in the case of his brother.

Electrical Examination.—Faradic current: tests had to be made quickly on account of extreme painfulness. In the lower extremities no contractions could be obtained anywhere with the strong

currents used. In the upper extremities the faradic response of the median and ulnar nerves was decidedly diminished. In the median nerve first KCC with 13 ma; ACC not at 20 ma.

Galvanic current.—Right leg, no reactions could be obtained by excitation of the nerves with currents used. In the extensor hallucis longus the first KCC and ACC were obtained with a current of 14 ma. The tibialis anticus did not respond to currents of 20 ma. The anterior thigh muscles and posterior thigh muscles respond to strong currents of about 16 ma without reversal of formula. In the left leg, in the extensor hallucis first KCC with 16 ma; first ACC with 18 ma. No contractions could be obtained by direct excitation of the tibialis anticus with currents up to 20 ma. Extensor digitorum: no contraction except of the division going to the small toe. Further examinations could not be made for want of time.

The electrical examination in this case, therefore, shows that the reaction of degeneration is present in its typical form in most of the muscles below the patella, the galvanic excitability of the peroneal nerve being entirely lost. It shows also a change in the electrical behaviour of the nerves of the upper extremities, since the responses of the median and ulnar nerves were markedly diminished.

In addition to the two cases related above I can refer to two additional cases which I have also had the privilege of seeing in the Hospital for the Ruptured and Crippled. The one is that of a girl about fourteen years of age, the other a boy about twelve years of age, whose legs presented the characteristic appearance of this form of progressive muscular atrophy.¹ I related last year a case which, though not as typical as those described in full in this paper, yet surely belongs to the same category; taken together these observations would seem to prove that the leg-type of progressive muscular atrophy is not so very rare a disease. Illustrative cases will evidently be found most frequently in surgical wards and in children's hospitals. I should in future suspect this trouble in all cases of gradually acquired double club foot. Among the large number of paralytic club feet treated at the Hospital for the Ruptured and Crippled, the cases I refer to appeared exceptional as regards both their antecedent

¹ These patients left the hospital before a careful examination could be made; their histories are, therefore, omitted from this article.

history and their behaviour under treatment. My cases present further interest for the reason that we may note the peculiar appearance of these cases after treatment has been instituted. In both of these cases the condition for which the first operation was done reappeared after eight months; a second operation was thus made necessary and the progressive character of the trouble was most clearly proven. In all other cases which have been reported no mention has been made of any surgical or orthopædic treatment,¹ and for that reason the cases here described present some points of difference from those which have now passed into literature as the strict types of this disease.

We shall have no difficulty, however, in demonstrating the nature of the disease from which these two boys suffered.

We note first of all that the disease occurs in two brothers, and that it made its appearance in both at exactly the same age. In both boys difficulty developed gradually in the use of legs and feet, this difficulty increasing with the development of bilateral club foot; in the one case leading to an absolute impossibility to walk without the use of crutches. The deformity of the feet was evidently due to a paresis and atrophy of the peronei, the anterior tibial, to a greater or less extent of the extensors of the feet and toes, and of the small muscles of the feet. This atrophy appears to have developed symmetrically in both legs and in an upward direction, effecting a weakness of the thigh muscles as well as of the muscles of the legs. In the one case the atrophy did not extend beyond the hips, in the case of the younger brother we find a uniform wasting or at least a uniform weakness of most of the muscles of the upper extremities. In this younger boy the infra-spinatus also was affected. In both cases the knee jerks were present. In the one case plantar reflexes were present, in the other absent. The sensory disturbances were not very marked; in the one case there was a slight hyperæsthesia, in the other a slight anæsthesia to pain. Vaso-motor changes were

¹ The brilliant results of Dr. Gibney's operations should induce other orthopædic surgeons to undertake similar and, if necessary several, operations for the relief of this class of patients.

noticeable in both cases, but not more marked than we are apt to find in cases of poliomyelitis. The electrical reactions show an approach to a complete reaction of degeneration in one case, and to a partial reaction of degeneration in the other of most of the muscles of the lower extremities. The only other electrical phenomenon I was able to establish in these cases was a diminished faradic and galvanic excitability in the nerves of the upper extremity.

The symptoms in these cases are in perfect agreement with the symptomatology as given by Charcot and Tooth. The upper extremities were not, however, as distinctly involved as in some of the cases heretofore described, but this evidently depends upon the progress which the disease makes, and must be ascribed to the fact that in both instances the disease is still in its earlier stage. In several of the cases described by other authors, the arms were not affected until many years after the first symptoms appeared in the legs. I cannot, therefore, agree entirely with Hoffman in making clawed hand an early and characteristic feature of this disease. We must lay greatest stress upon the leg symptoms; and here an *atrophie individuelle* beginning in any one muscle of the lower extremities and affecting in succession others and possibly all the muscles of the lower extremities, would seem to me to be the cardinal factor of the case. The presence or absence of fibrillary contractions in the affected muscles will not help us much in the diagnosis of this type. While the disease is apt to appear in very young children, we must also bear in mind that it may appear much later, either at the age of puberty as in my own case last year; at the age of twenty or thereabouts as in one case of Charcot-Marie, or even as late as the age of forty-six, as in one case of Osler,¹ who described this form as it occurred in the Farr family of Vermont.

The disease is apt to be confounded with other chronic disorders of the cord and peripheral nerves. The points of difference between this disease and amyotrophic lateral sclerosis, syringomyelia, multiple sclerosis, transverse chronic

¹ Archives of Medicine New York, 1880. There is room for doubt whether these cases were strictly of this type.

myelitis, hereditary ataxia, and tabes dorsalis are too evident to require minute description. From chronic multiple neuritis the leg type will be distinguished by the fact that pain plays a very important rôle in most cases of neuritis, that neuritis rarely leads to double club-foot, that the atrophy and paralysis are apt to be developed more quickly, and above all that the neuritis is not apt to occur as a family affection. The entire absence of hypertrophy will distinguish it from the primary muscular dystrophies, and except in the very last stages the reaction of degeneration in these primary dystrophies will not be as pronounced even as in cases of the peroneal form. From congenital club foot this leg type can be differentiated by the absence of marked electrical changes, and by the antecedent history of the cases, also by the result of treatment which is apt to be much more satisfactory in the congenital cases than in these cases of acquired club foot. The early history, the mode of onset, the retrogressive character of the paralysis and the distinct electrical changes in the nerve trunks, and the lack of heredity will distinguish acute poliomyelitis anterior from this form. It will be more difficult however to draw the line between this affection and cases of chronic anterior poliomyelitis. The only points upon which we can rely are these, that here again heredity is not marked in cases of chronic poliomyelitis, and that this disease is more apt to begin in the hands, and that it is apt to become retrogressive rather than progressive after a certain lapse of time. The electrical changes will be more definitely marked, and above all the electrical responses of the nerve trunks supplying the paralysed and paretic muscles will not be preserved as they are in some of the cases of the peroneal form. Moreover, if such uniform wasting as we have seen in the two cases I have described were due to a poliomyelitis, the knee jerks would surely be absent. It is on account of this presence of the knee jerks, the progressive form of paralysis involving the entire leg, and the slightly altered electrical reactions, that I claim the case which I presented last year to belong rather to the peroneal form of progressive muscular atrophy than to poliomyelitis. The difficulties of diagnosis would

be experienced in just such cases, whereas it would be quite easy to distinguish from one another typical cases of either disease. This disease might also be confounded with two other conditions which I have seen. Take the case of pseudo-hypertrophy which I described last year, and in which just such changes were noticed in the upper extremities as are apt to occur in this peroneal form. In this case the hypertrophy is giving way to an atrophy of most of the muscles of the lower extremities. At a later period I can very well imagine that on superficial inspection such a case would appear to resemble the disease under consideration, but here the almost normal character of the electrical reactions, the additional involvement of the trunk muscles, and the antecedent history would supply the points of differential diagnosis. And now another question suggests itself—can we discriminate in every instance between this special peroneal form and those cases of Duchenne's atrophy in which the leg is affected almost simultaneously with the upper extremities? Two such cases I have in mind in which the atrophy developed gradually in the leg and appeared very soon thereafter in the upper extremities. I know of no other sufficient point of differential diagnosis than the deficient proof of heredity and the unilateral or a asymmetrical development of atrophy in these cases of Duchenne's type. I grant that the points of difference are few, and this strengthens me in the belief which I expressed in my former paper, that Duchenne's atrophy and this form may represent the arm and leg type of the same trouble, or that they are at least closely allied diseases.

As to the pathology of this disease we are still very much in the dark. No autopsy has as yet been performed upon any one of the cases that have been distinctly recognised as belonging to this special type. Hoffmann, in looking for similar cases in the literature of progressive muscular atrophy, has picked out two cases—one examined post-mortem by Virchow,¹ the other examined post mortem by Friedreich.² In both cases a degenerative atrophy of the

¹ Virchow's Archiv., 1855.

² 'Prog. Muskel Atro.' Monograph, 1873. Cases I. III.

nerves and a degeneration of the columns of Goll were found. No mention is made of any change in the ganglion cells of the anterior horns, but we must remember that these autopsies were made at a time long preceding the introduction of modern staining methods and Charcot's publications on disease of the anterior ganglion cells. While it is interesting to learn that both Virchow and Friedreich found changes in the peripheral nerves, it is impossible to deny that these changes may have been secondary to changes in the ganglion cells. Hoffmann, who styles this disease progressive neurotic muscular atrophy, on the strength of these findings of Virchow and Friedreich, argues from the physiological and embryological researches of Vignard, His, and Kölliker that the nerve origin of this form of muscular atrophy is made extremely plausible. The degree of development of nerve-fibres he quotes from the embryologists is directly proportional to their proximity to the nerve roots, and conversely he infers that degeneration of nerve fibres would be most apt to begin at the distal end of the peripheral nerves. It is from the periphery toward the centre that the atrophy progresses in most of these cases. He acknowledges, however, that upon the health or disease of the ganglion cells depends the health or disease of the nerve fibres, and so we see that we are led by his own reasoning to suspect the ganglion cells to be the primary source of all trouble. However enticing such physiological and embryological inferences may be, it will be safer for the present to defer judgment upon the true pathology of this leg type. A single autopsy will be worth far more than the most acute reasoning based upon the results of laboratory experiments. I am of the opinion that little is to be gained by labelling these cases neurotic muscular atrophy. It is extremely desirable to have accurate pathological designations, but it makes confusion worse confounded to connect an unproved pathological process with a disease which has at least the one merit of presenting definite clinical symptoms. I propose, therefore, to retain the name "peroneal form of progressive muscular atrophy," or to speak of these cases as belonging to the *leg* type of progressive muscular atrophy. In conclusion, let me

say that it is a question of some practical importance to be able to discern these cases from the ordinary cases of paralytic and congenital club foot. The prognosis in the latter cases will be far better than in those due to a progressive atrophy.

We shall be able to recognise this peroneal form if we keep in mind that it is characterised by a symmetrical atrophy of the muscles of the legs and feet, which will in most cases lead to double club foot ; that the process may or may not involve the upper extremities, and if it does extend to the latter may produce typical *main en griffe* ; that sensation may or may not be affected ; that vaso-motor disturbances are apt to occur ; that the reflexes are present up to a late day ; and above all that heredity plays a very important rôle in the development of this disease.