II. IDIOPATHIC MUSCULAR ATROPHY

(*Primary Muscular Dystrophy—Erb*).

Definition.—Muscular wasting, with or without an initial hypertrophy, beginning in various groups of muscles, usually progressive in character, and dependent on primary changes in the muscles themselves. A marked hereditary disposition is met with in the disease.

Before considering the primary muscular atrophies it may be well to summarize briefly the chief conditions under which muscular atrophy occurs. These are:

(1) Acute or chronic lesions of the nuclei of the motor path, which may be (*a*) cortical, as a direct result of a cerebral lesion; (*b*) medullary, as in chronic bulbar paralysis; (*c*) spinal, either acute, as in poliomyelitis of children, or chronic, as in the progressive muscular atrophy of the simple or of the spastic type.

(2) Neuritic muscular atrophy, following a local neuritis due to trauma, a multiple neuritis due to alcohol, lead, and the infectious diseases. In this same category probably may be placed the muscular atrophies associated with joint-disease, the progressive hemi-atrophy of the face, and the atrophy sometimes found in cases of hysteria.

(3) Conditions of the muscles themselves—primary muscular atrophy.

Etiology.—The most important factor is heredity. Many members of the same family may be attacked through several generations. Males, as a rule, are more frequently affected than females. The disease is usually transmitted through the mother, though she may not herself be the subject. As many as twenty or thirty cases have been described in five generations. Isolated cases, however, are not uncommon. The disease usually sets in before puberty, but may be as late as the twentieth or twenty-fifth year, or in some instances even later. No etiological factors of any moment are known other than heredity.

Clinical Forms.—Two chief types may be recognized: (1) With primary hypertrophy, the pseudo-hypertrophic muscular paralysis; and (2) with primary atrophy.

Pseudo-hypertrophic Muscular Paralysis.—The first symptom noticed is, as a rule, clumsiness in the movements of the child, and on examination certain muscles or groups of muscles seem to be enlarged, particularly those of the calves. The extensors of the leg, the glutei, the lumbar muscles, the deltoid, triceps, and infraspinatus, are the next most frequently involved, and may stand out with great prominence. The muscles of the neck, face, and forearm rarely suffer. Sometimes only a portion of a muscle is involved. With this hypertrophy of some muscles there is wasting of others, particularly the lower portion of the pectorals and the latissimus dorsi. The attitude when standing is very characteristic. The legs are far apart, the shoulders thrown back, the spine is greatly curved, and the abdomen protrudes. The gait is waddling and awkward. In getting up from the floor the position assumed, as so well known now through Gowers's figures, is pathognomonic. The patient first turns over in the all-fours position and raises the trunk with his arms; the hands are then moved along the ground until the knees are reached; then with one hand upon a knee he lifts himself up, grasps the other knee, and gradually pushes himself into the erect posture, as it has been expressed, by climbing up his legs. The striking contrast between the feebleness of the child and the powerful-looking pseudo-hypertrophic muscles is very characteristic.

The course of the disease is slow, but progressive. Wasting proceeds and finally all traces of the enlarged condition of the muscles disappears. At this late period distortions and contractions are common.

Primary Atrophic Form.—Here, too, there is the same marked tendency to involvement of different members of a family. Five or six different types have been described, but it seems more rational to group them together under the designation of idiopathic muscular atrophy. In all of the cases the atrophy begins, as a rule, before the twentieth year. According to the site of the primary atrophy different forms have been described. In the *juvenile type* of Erb the affection begins about the fifteenth or the twentieth year and involves the muscles of the upper arm and shoulder and the gluteal and thigh muscles. In the facio-scapulo-humeral type of Landouzy and Déjérine the muscles of the face are early involved with those of scapulo-humeral groups. This form occurs usually in families, and the onset may be delayed until the twentieth or thirtieth year. Leyden describes an *hereditary form*, beginning in the lower extremities and back, which may be associated with hypertrophy of the calves. Another type has been described by Charcot and Tooth—the *peroneal form;* but there is still some doubt whether this is not in reality a myelopathy and more closely related to chronic polio-myelitis anterior. In this form the atrophy begins in the muscles of the legs, usually in the extensors of the great toe, and afterward in the common extensors and the peroneal groups. The cases usually begin early, and the heredity through the mother has been traced in several remarkable series, particularly that of Herringham's. Fibrillary contractions and the reaction of degeneration are present. Nerve degeneration has been found in the peripheral parts, and ascending degeneration of the columns of Goll.

Morbid Anatomy.—The spinal cord and peripheral nerves have been found normal in cases of pseudo-hypertrophic muscular paralysis and in the forms of idiopathic muscular atrophy. The muscles in the pseudo-hypertrophic condition present great variations in the size of the muscle fibres, some of which may be hypertrophied and others wasted. In the early stage the hypertrophy of the fibres may be very pronounced and the nuclei of the sarcolemma are greatly increased. In some instances, too, the fibres have been fissured longitudinally. At a later stage the muscular

fibres are wasted and largely replaced by connective tissue and fat. In the primary atrophic form wasting of the fibres, increase in the interstitial tissue, and the development of fat are the most marked features. Except in the peroneal type, about which there is still doubt, no affection of the nerves or cord has been determined.

Diagnosis.—The primary myopathies can usually be readily distinguished from the cerebral, myelopathic, and neuritic forms.

(*a*) In the cerebral atrophy loss of power usually precedes the atrophy, which is either of a monoplegic or hemiplegic type.

(*b*) In the myelopathic or spinal muscular atrophy the distinctions are clearly marked. *Polio-myelitis anterior chronica* begins in the small muscles of the hand, a situation rarely if ever affected by the primary myopathies, which involve first those of the calves, the trunk, the face, or the shoulder-girdle. In the myelopathic atrophy the reaction of degeneration is present and fibrillary twitchings occur in both the atrophied and non-atrophied muscles. In many cases in addition to the wasting in the arms there is a spastic condition in the legs and increase in the reflexes. The myelopathic atrophies come on late in life; the myopathic forms develop, as a rule, early. In the primary muscular atrophies heredity plays an important *rôle*, which in the myelopathic is quite subsidiary.

(*c*) In the neuritic muscular atrophies, whether due to lead or to trauma, the general characters and the mode of onset are distinctive. In the cases of multiple neuritis seen for the first time at a period when the wasting is marked there is often difficulty, but the absence of family history and the distribution are important features. Moreover, the paralysis is out of proportion to the atrophy. Sensory symptoms may be present, and in the cases in which the legs are chiefly involved there is usually the *steppage* gait so characteristic of peripheral neuritis.

The outlook in the primary myopathies is bad. The wasting progresses uniformly, uninfluenced by treatment. Erb holds that by electricity and massage the progress is occasionally arrested. The general health should be carefully looked after, moderate exercise allowed, frictions with oil applied to the muscles, and when the patient becomes bedfast, as is inevitable sooner or later, care should be taken to prevent contractures in awkward positions.

III. THOMSEN'S DISEASE; MYOTONIA CONGENITA.

Definition.—An hereditary disease characterized by tonic cramp of the muscles on attempting voluntary movements. The disease received its name from the physician who first described it, in whose family it has existed for five generations.

Etiology.—All the typical cases have occurred in family groups; a few isolated instances have been described in which similar symptoms

have been present. The disease is rare in this country and in England; it seems more common in Germany and in Scandinavia.

Symptoms.—The disease comes on in childhood. It is noticed that on account of the stiffness the children are not able to take part in ordinary games. The peculiarity is noticed only during voluntary movements. The contraction which the patient wills is slowly accomplished; the relaxation which the patient wills is also slow. The contraction often persists for a little time after he has dropped an object which he has picked up. In walking, the start is difficult; one leg is put forward slowly, it halts from stiffness for a second or two, and then after a few steps the legs become limber and he walks without any difficulty. The muscles of the arms and legs are those usually implicated; rarely facial, ocular, or laryngeal muscles. Emotion and cold aggravate the condition. In some instances there is mental weakness. The sensation and the reflexes are normal. The condition of the muscles is interesting. The patients appear and are muscular, and there is sometimes a definite hypertrophy of the muscles. The force is scarcely proportionate to the size. Erb has described a characteristic reaction of the nerve and muscle to the electrical currents—the so-called myotonic reaction, the chief feature of which is that normally the contractions caused by either current attain their maximum slowly and relax slowly, and vermicular, wave-like contractions pass from the cathode to the anode.

The disease is incurable, but it may be arrested temporarily. The nature of the affection is unknown. There is an extraordinary increase in the size of the voluntary fibres. According to Hale White,* who has recently treated the subject in an exhaustive and critical manner, the fibres may be more than double the width of those of the normal muscles. The nuclei and the interstitial tissue may be increased and some of the fibres contain vacuoles. No post-mortem has been made. No treatment for the condition is known.

IV. PARAMYOCLONUS MULTIPLEX.

An affection, described by Friedrich, characterized by clonic contractions, chiefly of the muscles of the extremities, occurring either constantly or in paroxysms.

The cases have usually been in males and the disease has followed emotional disturbance, fright, or straining. The contractions are usually bilateral and may vary from fifty to one hundred and fifty in the minute. Occasionally tonic spasms occur. It is not accompanied by any sensory or motor disturbances. In the intervals between the attacks there may be tremors of the muscles. In the severe spasms the movements may be very

* Guy's Hospital Reports, 1889.