Progressive neuro-muscular Atrophy : (Peroneal Type of Charcot, Marie, and Tooth) : with report of three cases in a family without heredity / by Floyd F. Hatch, Boston.

Progressive Neuro-Muscular Atrophy, (Peroneal Type of Charcot, Marie, and Tooth): with Report of Three Cases in a Family without Heredity.

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PROGRESSIVE NEURO-MUSCULAR ATROPHY, (PERONEAL TYPE OF CHARCOT, MARIE AND TOOTH): WITH REPORT OF THREE CASES IN A FAMILY WITHOUT HEREDITY.*†

By Floyd F. Hatch, Boston.

[From the Medical Clinic of the Peter Bent Brigham Hospital, Boston, Mass.]

The impulse given to the study of this type of muscular atrophy came in 1886 when Charcot and Marie[1], and Tooth[2] coincidently in the same year published papers reviewing the myopathies. Separately they established from clinical and pathological observations that the peroneal form, beginning usually in early childhood and involving the extremities only, depended upon neural as well as muscular degeneration and isolated it between the myopathic (muscular dystrophies) and the myelo-pathic (anterior poliomyelitis) affections.

The characteristics of the affection as originally described by Charcot and Marie are:

1. Progressive muscular atrophy beginning first in the feet and legs, not appearing in the hands and arms until several years later, the progression of the atrophy being slow. 2. Relative integrity of the muscles near the trunk, or at least much longer preservation of these than of the muscles of the distal ends of the limbs. 3. Integrity of muscles of the trunk, shoulders and face.

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† Received for publication Dec. 22, 1914.

4. Fibrillary contractions in the atrophying muscles. 5. Vasomotor disturbances in the portions of the limbs which have atrophied. 6. Absence of pronounced contractions of the tendons. 7. Sensations usually intact but sometimes affected. 8. Reaction of degeneration in atrophying muscles. 9. Hereditary and familial tendencies.

The three cases reported here are members of a Jewish family which came to this country in 1899. The father, a well developed and nourished man, left his wife and children several years ago. Nothing is known of his family with the exception of one brother and his family, all of whom have always been well. The mother is living and well. There is no history of paralysis or other nervous disturbance in her family. The mother does not use alcohol. The father drank at times. There are three healthy sisters, aged 21, 13 and 8 years. All children were normal deliveries, healthy infants and walked before one year of age.

Case 1. Plates I and II. Sam F., 24 years old, has always lived at home, never having been able to work.

Complaint: "Weakness of legs and arms."

Past History: Internal strabismus of the left eye was present at birth. He had measles at two years, chicken pox at three years and whooping cough at four years of age. Entropion of both lower lids began at 17 years with falling out of the lashes and chronic irritation since that time.

Present Illness: Began at the age of seven years when weakness was noted in his feet, making walking difficult. Weakness and atrophy increased until at the age of 14 years the muscles above the knees also seemed to be involved and he fell down constantly when walking about. At 16 years the patient was

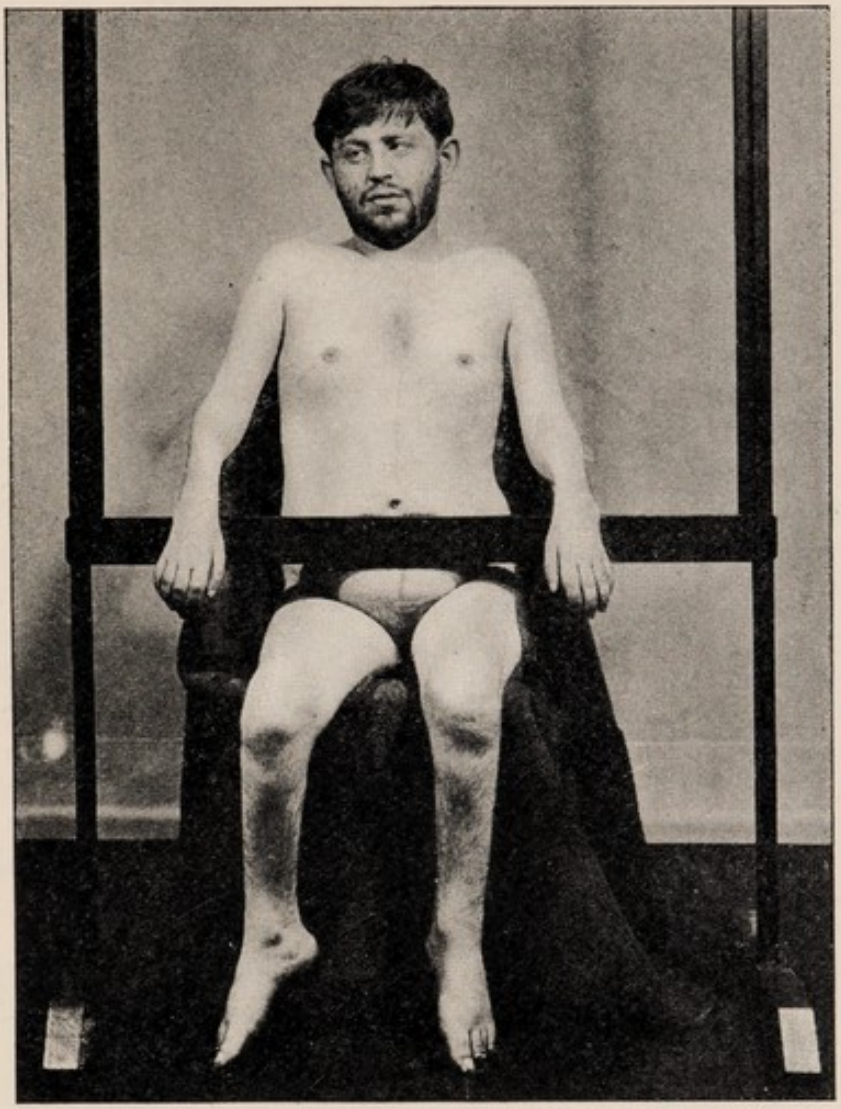

PLATE 1.

wholly unable to walk and moved about by crawling. At about this time the fingers of the left hand and almost immediately of the right became weak with gradual development of contracture so that they could not be straightened. Subsequently the whole lower arm on both sides became involved so that at the age of 20 years he was unable to use either hand. At present there remains ability to move the third and fourth fingers of the right hand to a slight extent. No sensation of pain, cramp or discomfort has ever been felt. He sleeps well, has a good ap-

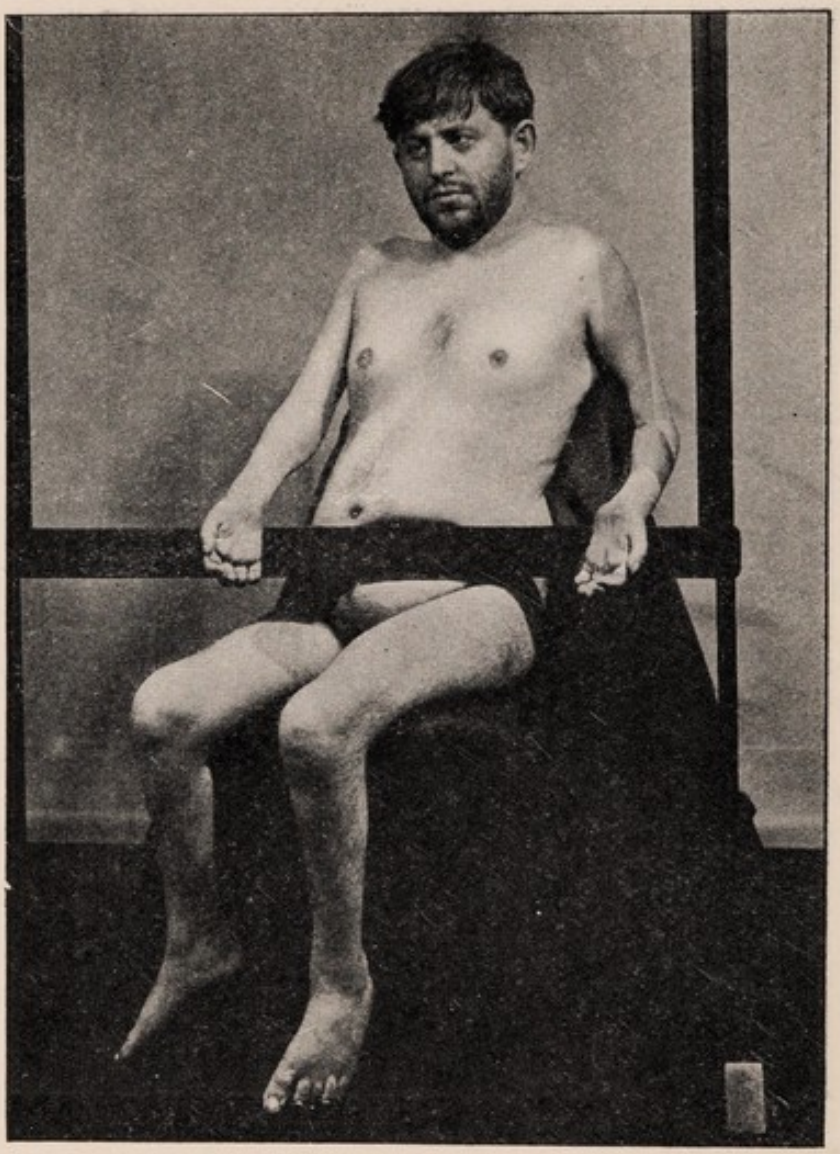

PLATE 2.

petite and never becomes nervous or irritable. At the present time he clumsily washes and dresses himself, using his hands in a flail-like manner but can perform no complex movements. His mentality appears normal.

Physical Examination: Shows head and body, including the shoulders and buttocks, well developed and nourished. Skin and mucous membrances are normal. Eyes show entropion of both lower lids with a low grade chronic conjunctivitis and a left

internal strabismus. Pupils react equally to light. Tongue is clear and protrudes without tremor. Chest is well formed, expansion good. Lungs and heart are normal. Systolic blood pressure is 120 mm. of mercury. Abdomen is fat and somewhat protuberant, no masses or tender areas are made out. Patient sits or lies with both knees flexed at right angles to the thighs from contracture. There is evident equino-varus of both feet, most marked on the right, also a marked degree of external rotation of the whole lower right leg from atrophy and weakness of the vastus medialis. The legs are short as compared with the body, giving the patient a dwarfish appearance. Atrophy of all muscles of the legs below the middle third of the thigh is evident but to some extent obscured by the thick layer of subcutaneous fat. The hands and arms show a marked degree of atrophy and paralysis below the middle third of the upper arm, comparable with that of the feet and legs. The thenar and hypothenar eminences of both hands are absent. The hands are flattened with the terminal phalanges slightly flexed. The biceps on the right is capable of flexing the arm. The left biceps cannot accomplish this.

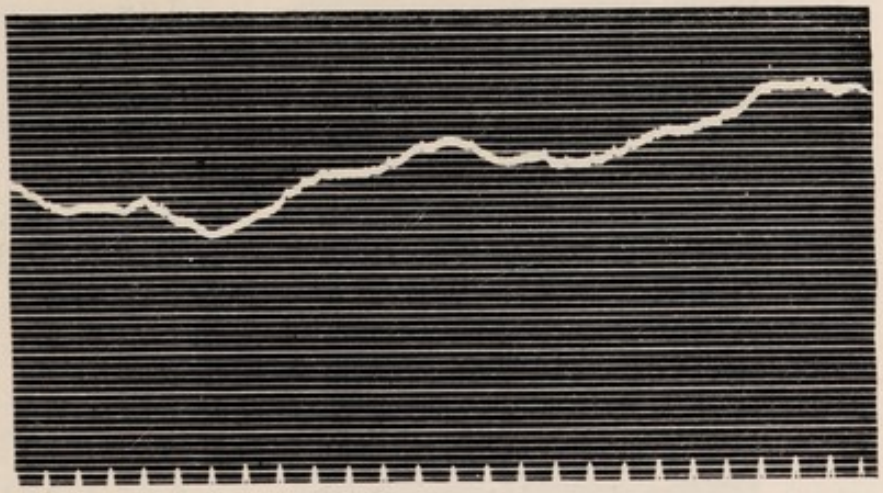

PLATE 3.

Tracings were obtained by placing the electrodes on the outer side of the left upper arm, one at the upper part and the other at the lower part of the triceps. There are fine movements of the string produced probably by muscular twitchings which result when patient tries to move his fingers.

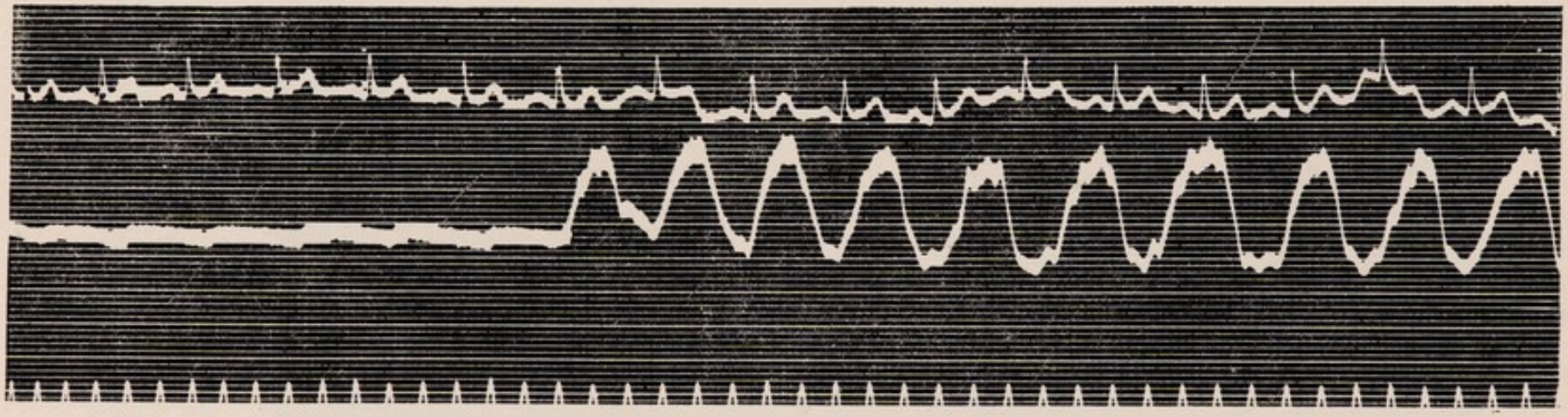

PLATE 4.

Upper curve is electrocardiogram showing normal cardiac complex. Middle curve is record obtained from a recording tambour connected with an air cuff around upper arm. First portion of the curve shows low waves produced by pulsation of brachial artery; second portion shows large and small waves produced by twitching of arm muscles. Lower curve is record of a time marker beating one-fifth seconds.

There is practically no muscular movement below the elbows. On the left there seems to be slight weakness of the shoulder muscles. The hands and feet are about two-thirds normal size and length. The knee jerks, Achilles jerks, plantar, biceps and triceps reflexes of both sides are absent. There is no ankle clonus or Babinski sign. The cremasteric and abdominal reflexes are present.

While the patient was in the hospital fibrillary tremor at a rate of about 8 per second was noticed in his pectoral, biceps and triceps muscles, especially on the left, brought on by any attempt to move the arm. A tracing was made, by connecting the arm by electrodes with the electrocardiograph galvanometer (Plate III), and by means of an air-containing cuff around the arm connected with a recording tambour (Plate IV). These plates show graphically the electric disturbance set up by the fibrillary twitching of the muscle and the rate and degree of muscle contraction. The muscle sounds during the period of tremor could readily be heard with a stethoscope. There was a similar tremor noticed in the muscles of the upper left cheek and those of the lower left lid which probably originated in the orbicularis palpebrarum. The location of these muscles was not suitable to obtain graphic tracings.

Case 2. Plates V and VI. Harry F., 12 years old.

Complaint: "Cannot walk."

Past History: No previous illness. Digestion good. Bowels regular.

Present Illness: Began at age of seven years when he noticed that the right foot became weak and easily tired when walking. The same difficulty was encountered in the left foot about four weeks later. The legs and feet have grown progressively weaker with marked atrophy of all muscles below the knees and six months ago he finally became unable to walk at all. About four years ago he noticed weakness followed by incoördination in his hands,

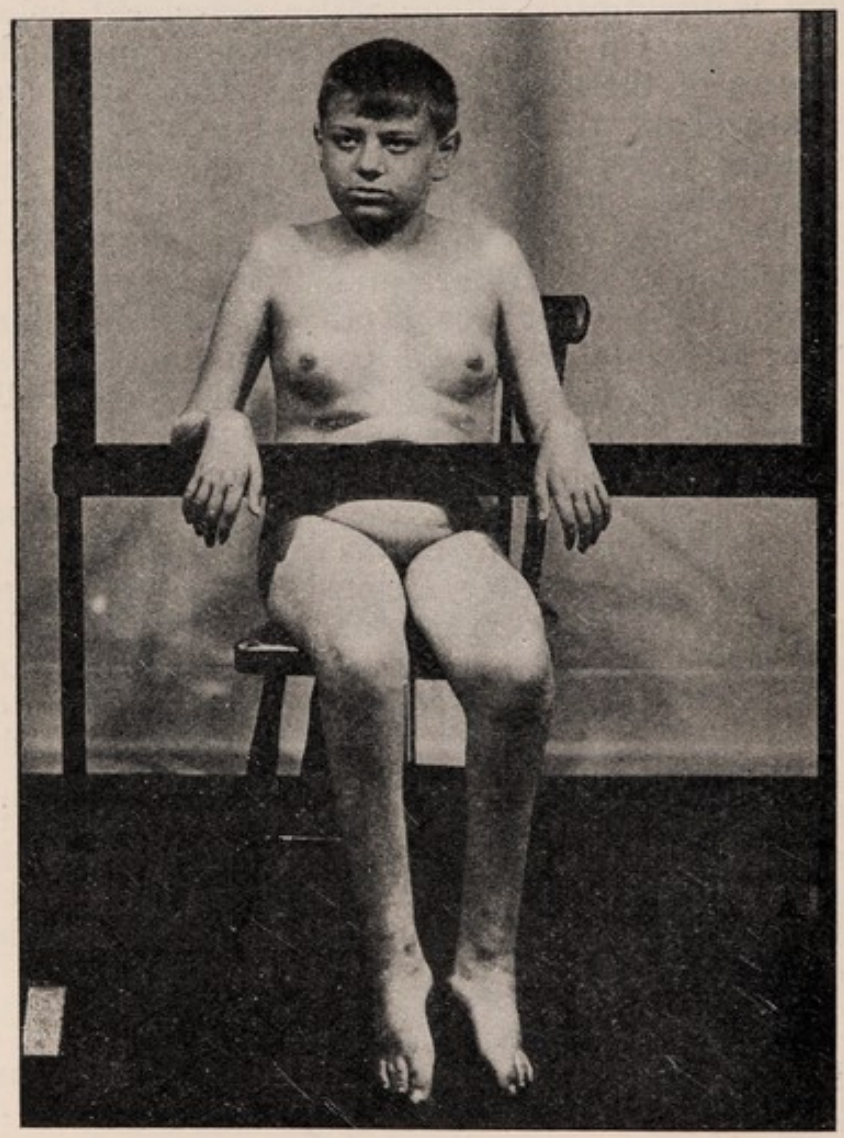

PLATE 5.

appearing in the first and second fingers of the right hand. Atrophy and paralysis have progressed until at present there is no muscular control below the insertion of the biceps. His mentality appears normal.

Physical Examination: Shows a normal appearing boy of 12 years, well developed and nourished except in the musculature of the arms and legs. Pupils are equal and react to light. Tongue is protruded in the mid line without tremor. Skin and

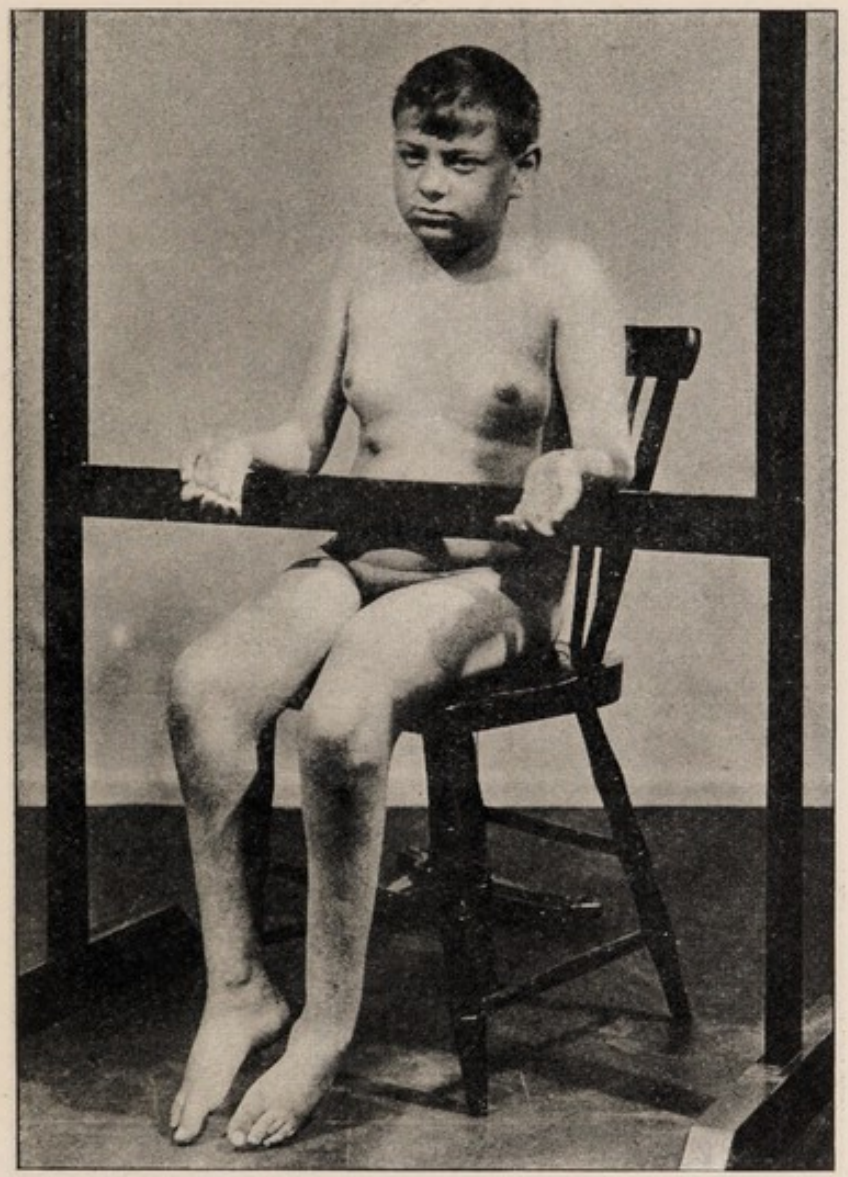

PLATE 6.

mucous membranes are normal. Chest is well formed, expansion good. Lungs and heart are normal. Systolic blood pressure is 120 mm. of mercury. Abdomen is fat and slightly protuberant, no masses or tender areas are made out. Upper extremities show a progressive tapering from the shoulders to the hands which are held in a position of pronation, flexed at the wrists. The lower arm muscles, interossei muscles, thenar and hypothenar

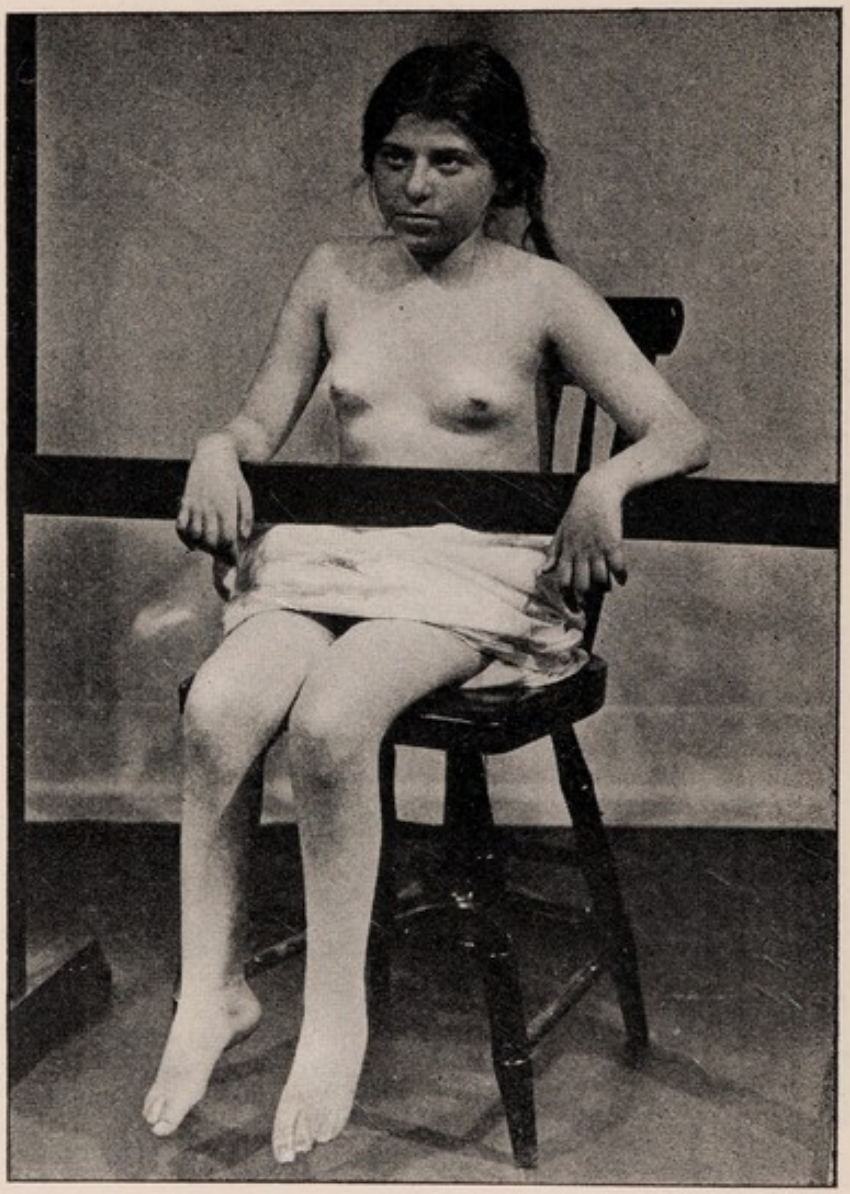

PLATE 7.

eminences show marked atrophy. The biceps and triceps reflexes are absent. Movements of the lower arms and hands are accomplished by flinging movements of the shoulder muscles. The lower extremities show the same type of progressive tapering. The right knee is held in position of genu valgus. Both feet show equino-varus which is especially marked on the left. The feet and hands are smaller

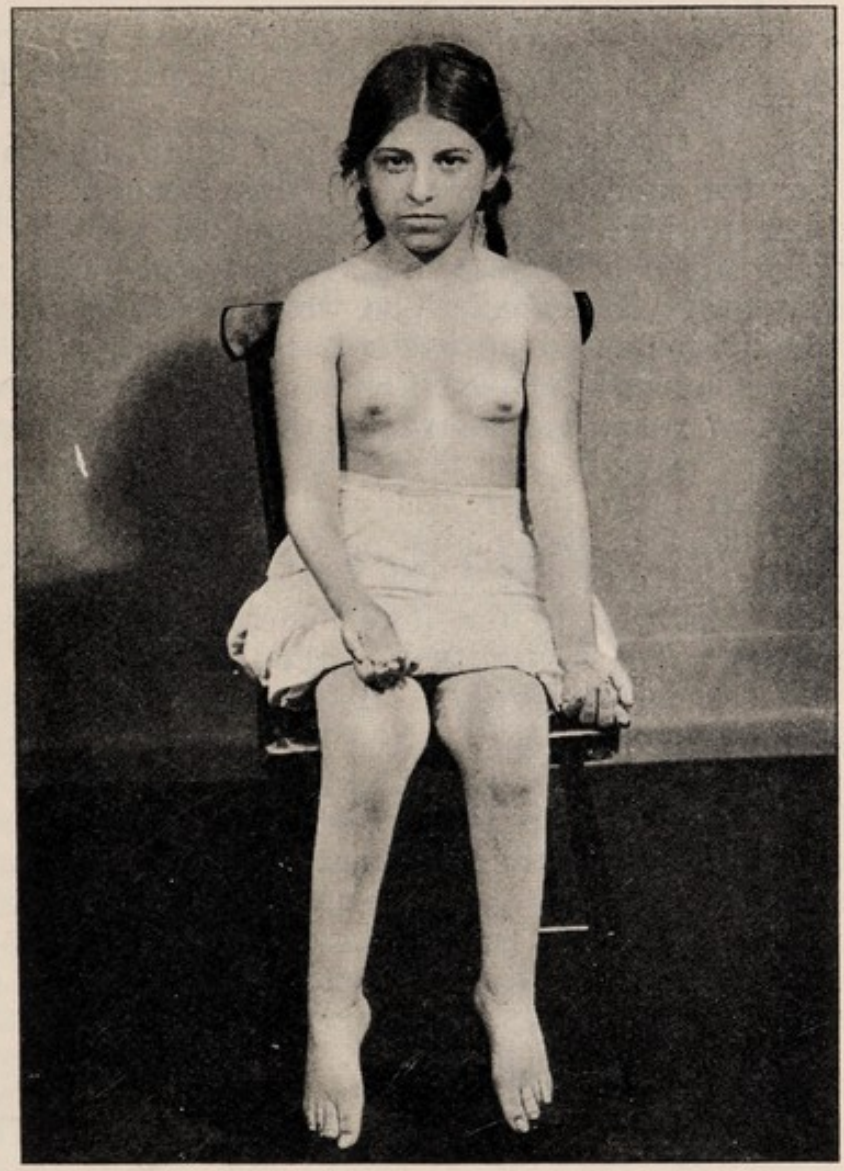

PLATE 8.

than those of the average boy of his age and size. The atrophy of the lower legs and feet is even more obscured by the thick layer of subcutaneous fat than in the arms and hands. All muscle power below the knees is gone. The knee jerks and plantar reflexes are absent. There is no ankle clonus or Babinski sign. No muscular tremor was observed.

CASE 3. Plates VII and VIII. Mary F., 10 years old.

Complaint: "Unable to use hands or feet."

Past History: Measles at four years followed immediately by whooping cough which persisted for six months.

Present Illness: Began at age of four years about one or two months after the whooping cough, when mother noticed that the child walked peculiarly, picking up feet higher than usual and bringing them down "flail-like." The weakness in her legs gradually increased until she is now barely able to walk. The weakness was first noticed in her hands about two years ago, at which time she could perform any complicated task, but paralysis and atrophy gradually progressed until now she is practically helpless, being able only to feed and dress herself with difficulty. The weakness and atrophy remain localized to the lower portions of the extremities. Her mentality is normal.

Physical Examination: Patient is well developed and nourished except in the musculature of the lower arms and legs. Pupils are equal and react to light. Teeth are in poor condition. Tongue protrudes in the mid line without tremor. Skin and mucous membranes are normal. Chest is well formed, expansion good. Lungs and heart are normal. Systolic blood pressure is 115 mm. of mercury. Abdomen is soft and rounded, no masses or tender areas are made out. The upper extremities appear normal as far down as the elbows. Flexion at the elbows seems weak but not limited. The muscles of the lower arms and hands show marked atrophy; especially noticeable is the absence of the thenar and hypothenar eminences. There is some contracture of the second, third and fourth fingers of both hands in the two distal phalangeal joints, giving the picture of an early "claw hand." Reflexes of the biceps and triceps are transient. No reflexes are present in the lower arms. The lower extremities show marked atrophy and practically complete paralysis of the muscles below the knees. The degree of atrophy is masked by thick subcutaneous fat. Both feet show

equino-varus, more marked on the right. Knee jerks, Achilles jerks, and plantar reflexes are absent. No ankle or Babinski sign.

The electrocardiograms in all three cases showed the heart mechanism to be perfectly regular and normal.

The special senses, smell, sight, hearing, taste and speech as well as the general sensations of pain, touch, position and temperature, were normal in all cases without exception. There is a well marked vaso-motor disturbance in the involved portions of the extremities in that their appearance is cyanotic and they feel cold as compared with the uninvolved portions.

Ophthalmoscopic examination showed normal fundi with the discs clearly outlined and vessels well seen throughout their course.

X-ray examination of the affected parts show evident bony atrophy, more or less failure of development of the sesamoid bones, shafts of long bones with small flaring extremities and some failure of the epiphyses to unite. In all three cases the sella turcica was small, antero-posterior diameter 10 mm., depth 6 mm.

The electrical reactions of the muscles in all three cases were very similar, varying directly with extent of the disease. The reaction of degeneration was found in muscles more recently involved (*i. e.* those about the knees and elbows). No reactions to electrical stimuli were obtained on muscles of the lower portions of the extremities in any case (*i. e.*, hands and feet). Partial reaction of degeneration was obtained in many muscle groups that appeared to be unimpaired. The muscles giving these partial reactions of degeneration were located above the atrophic muscles. This was most evident in the case of the elder boy where only very large amounts of faradic

current (60-70 mil. amperes) gave muscle response in the deltoids and pectorals of the left side, while relatively small amounts of galvanic current (15-20 mil. amperes) gave similar response in normal individuals.

The clinical pathology of the urine and blood in all three cases showed normal conditions. The Wassermann reactions on the blood serum and spinal fluid in each case were negative. The spinal fluids, were, however, very interesting as shown by the following table. The coincident increase of albumin and globulin with the stage of the disease in the three cases seems a logical finding, as well as the pathological gold chloride reaction. I have not found these tests previously reported. The gold chloride reaction, as indicated by the number of the tube with the strength of reaction following, is one that simulates the cerebro-spinal syphilitic reactions, falling nearest to tabes.

The cases of this type, with atrophy confined strictly to the distal parts of the limbs, are recognized as a distinct type of neuro-muscular atrophy, with a distinct pathology. Cases of this kind with necropsy are very rare and we are obliged to depend on the findings of Marinesco[3], Sainton[4], and Dejerine and Armand-Delille[5].

The lesions of the cases studied by Sainton were: sclerosis of the posterior columns, especially the columns of Burdach, a slight degeneration of both pyramidal tracts, alterations of the columns of Clarke, atrophy of the cells of the anterior horns, slight degeneration of the intramuscular nerves, slight sclerosis of the nerves of the forearms and legs very distinct in the peroneal nerves, atrophy of the muscle fibres even causing complete disappearance of some fibres

SPINAL FLUID.

	Gold Chloride Reaction.							Globulin.	Albumin.	Cells per Cu. Cm.
Tube	1	2	3	4	5	6	7			
Mary F.	±	+	+++	++++	+±	±	—	—	—	3
Harry F.	—	—	+±	+++	+±	±	—	±	+	2
Sam F.	—	—	++	++++	++	±	—	±	+±	2

with proliferation of connective tissue. In this case the atrophy began in the upper limbs. The lesions resembled those observed by Marinesco in his case, except that Marinesco found the antero-lateral columns intact.

The lesions in the case reported by Dejerine and Armand-Delille were degeneration of some of the nerve cells of the anterior horns of the cervical and lumbar regions without diminution in their number, chronic meningitis, degeneration of the muscles of the hands and feet (*i. e.*, many nerve fibres of small size, many empty nerve sheaths and a few nerve fibres in the process of degeneration). The nerve trunks, the cutaneous sensory nerves and the anterior and posterior nerve roots with slight exception were normal.

Siemerlings'[6] case is held in question as belonging to this type, as there was great atrophy of the muscles of all the extremities, the upper limbs as well as the lower, and of the trunk. There was flaccid complete paralysis of the lower limbs so that all voluntary movements were lost. He found degeneration of the posterior and lateral columns, most intense in the lower thoracic and upper lumbar regions, degeneration of the peripheral nerves and muscles, atrophy of the cells of the anterior horns, of the columns of Clarke, of the anterior roots and of the spinal ganglia.

In the original description the type was so sharply defined that confusion with other types was supposed not to occur, but literature contains many reported cases in which some atypical features are present. Sachs[7] reported the first two cases from this country which deviated from the original type in that the infraspinati muscles were atrophied. Oppenheim and Cassirier[8] re-

ported a case with involvement of the orbicularis palpebrarum. My first case is somewhat atypical in that the pectorals are weak and show the typical fibrillary contractions of early muscular atrophy and neural degeneration; otherwise the cases are absolutely typical clinical pictures of the original type descriptions and their classification cannot be questioned.

That the disease is a hereditary as well as a familial affection is well shown in the family reported by Dr. Herringham[9]. In his case, however, extending over three generations, males only were affected, though the disease was transmitted by females, following closely the hereditary features noticed in color blindness. In the family reported by Osler[10] thirteen individuals were affected in two generations, seven being males and six females. Both males and females transmitted the disease. This is by far the most common. Males are affected more frequently than the females in the proportion of two to one. (Some say five to one).

Concerning the prognosis it may be said to be good so far as life is concerned; the disease is of slow development and after reaching its height the patient may live for years without having additional symptoms. Thus in a typical case reported by Spiller[11], symptoms are said to have been present 45 years, and the patient's condition has not changed during the many years he has been under observation. There is no reason for expecting any tendency toward improvement.

REFERENCES.

[1] Charcot and Marie: Rev. de Med., Paris, 1886, vi, 97.

[2] Tooth: The Peroneal Type of Progressive Muscular Atrophy. Graduation Thesis, M.D. Cambridge, 1886.

[3] Marinesco: Archiv de Med. Exper., Paris, 1884, vi, 9.

[4] Sainton: Nouvelle Inconographie de la Salpetriere, 1899, xxii, 306.

[5] Dejerine and Armand-Delille: Review Neurol., Paris, 1903, xi, 1198.

[6] Siemerlings: Archiv für Psychiatrie, 1898, xxxi, 105.

[7] Sachs: Brain, 1890, xii, 447.

[8] Oppenheim and Cassirier: Deutsche Zeit. für Nervenheilkunde, xi, 143.

[9] Herringham: Brain, London, 1888, xi, 320.

[10] Osler: Arch. Med., N. Y., 1880, iv, 316.

[11] Spiller: Journal Nervous and Ment. Dis., 1907, xxxiv, 15.