INFANTILE AMYOTROPHIC LATERAL SCLEROSIS OF THE FAMILY TYPE.[1]

By CHARLES HENRY BROWN, M.D.,

New York City.

AT the April meeting of the New York Neurological Society, I presented a case for discussion as to its diagnosis and classification. It seemed to call out opinions more or less at variance. I have been persuaded to present the case more at length before this Association. I do so, in the hope that my study of the case and the added opinions of the members may aid in the better understanding and differentiation of a class of trophic disease in children. I believe that my case opens up a new type of cases for study and that it is a sign post not to be overlooked. Infantile amyotrophic lateral sclerosis of the family type heretofore has not been recognized, as far as I have been able to ascertain.

History—Boy, A. K., aged fifteen, of German parents, fifty-eight inches in height and weighs clothed seventy-one pounds. Family history reveals nothing of importance; the early history is meagre, on account of the stupidity and ignorance of the parents, and to the fact of the inability of the boy to hear well, and to articulate distinctly and also to the fact that, the child was raised in a charitable institution which refuses to send me information in relation to him.

About three years ago the very first symptoms were noted by the boy, which were the inability to whistle or to talk plainly. The onset was rather rapid and grew more pronounced every day so that in a week he could not freely move his tongue, whistle or swallow without an effort.

He soon was unable to laugh or entirely close his eyes. He grew hard of hearing and on the least exer-

[1] Read before the American Neurological Association, Washington, May 31, 1894.

tion he had difficulty in breathing. Soon after this, perhaps a week or two, he found it very difficult to button or unbutton his clothes, write or carry as heavy weights as formerly. He grew thin rapidly and felt twitchings of the muscles of the face, neck, and even all over the body from the beginning.

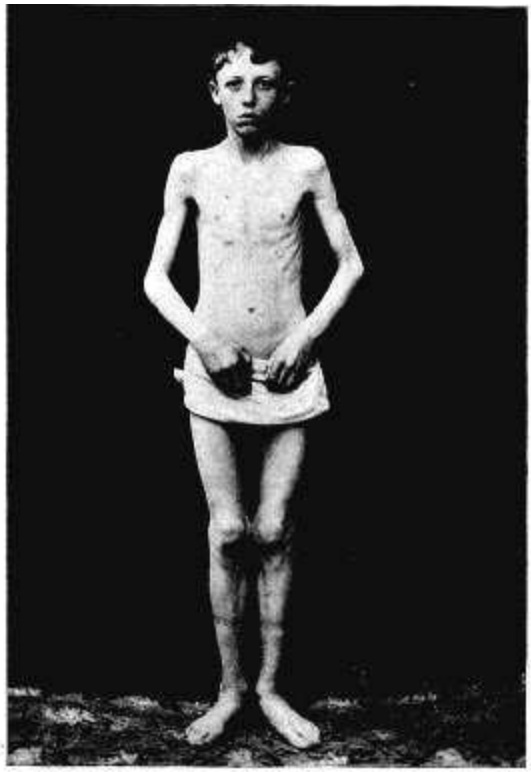

FIG. 1.

Status-presens.—As observed by the photograph (No 1) he presents an exceedingly emaciated boy, more markedly so, however, in the upper parts of the body.

Subjective symptoms are the same as stated in his history. He especially complains of the fact that he cannot laugh, swallow well or run.

Objective symptoms.—There is marked paralysis and atrophy of all the facial muscles excepting those of mastication. The lower facial atrophy is more marked than

tion he had difficulty in breathing. Soon after this, perhaps a week or two, he found it very difficult to button or unbutton his clothes, write or carry as heavy weights as formerly. He grew thin rapidly and felt twitchings of the muscles of the face, neck, and even all over the body from the beginning.

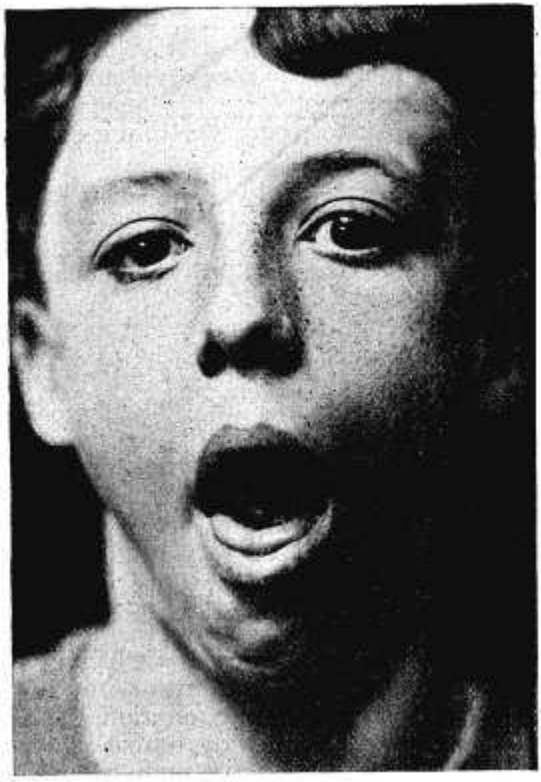

FIG. 1.

Status-presens.—As observed by the photograph (No 1) he presents an exceedingly emaciated boy, more markedly so, however, in the upper parts of the body.

Subjective symptoms are the same as stated in his history. He especially complains of the fact that he cannot laugh, swallow well or run.

Objective symptoms.—There is marked paralysis and atrophy of all the facial muscles excepting these of mastication. The lower facial atrophy is more marked than

Articulation is difficult, barely intelligible and strongly nasal; it is without variation whatever in tone.

Laryngoscopic examination.—The pharynx reveals two enlarged tonsils, a narrowed opening, the pharyngeal muscles thin but acutely active, sensation may be somewhat blunted, but the pharyngeal reflex is acute.

Closure of glottis is fair, examination of the larynx difficult, laryngeal bands relaxed, no action whatever muscularly in attempts at phonation. The sterno-cleido mastoid of the left side is atrophied to a greater extent than that of the right, both are less paralyzed and atrophied proportionately than the other front muscles of the neck.

The omo-hyoid is more prominent than any of the other muscles of the throat. The platysma myoides stand out in spastic thread like bundles when the neck is stretched by various movements of the head to the left or right.

The posterior muscles of the neck are strong and the humeral scapuli muscles firm, and when the boy is lifted by the arms the Erb symptom is not noted.

The intercostal muscles seem to be all weak. Expansion, except by a great effort, hardly exceeds one-half an inch and the diaphragm does not seem to possess very much power.

Breathing is extremely shallow, hardly a perceptible movement of the chest walls, occasionally there is a raising of the shoulders and with head thrown back an extra full inspiration is obtained, to be quickly followed by a collapsing explosive expiration.

There is no question of the serious envolvement of all the intrinsic muscles of respiration to more or less degree. The heart with the exception of occasional variation in rhythm accomplishes its work fairly well.

The digestive processes are somewhat weakened, pain and colic frequent. He is potbellied and physical signs suggest a dilated stomach.

The boy passes enormous quantities of urine and complains of constant thirst; urine analysis shows only a low specific gravity.

There is apparent paresis and atrophy of most of the arm muscles, but not so marked as to be seen in the hand muscles, especially the interossei and thenar group; adduction of thumb impaired.

Examination of the eyes reveals nothing of importance, except the inability to close the lids completely;

winking is fairly rapid; slight esophoria of two degrees present.

Dr. John L. Adams reported that the deafness noticed in the case was due to impairment in the function of the eighth nerve. On careful testing the ærial conduction of sound is more acute than the bone conduction, otherwise on careful examination the ears appear normal.

The electrical reactions show these facts that most all the muscles of the face, throat and upper extremities respond in part. In the intrinsic muscles of the throat it was difficult to decide, but many muscles must be beyond excitation in part, otherwise there is no marked degenerative reactions.

Fibrillary twitchings are observable all over the body and easily excited and intensified by cold and tapping.

Superficial and deep reflexes are very much brighter than normal throughout the body. The right ankle-clonus can be obtained quite constantly, the left occasionally.

There are many retracted tendons noticed; for instance, the flexor tendons, such as the biceps and tendo—Achilles are taut, and stand out too prominently, and a special effort is necessary to extend the limbs fully.

The patient in full extension of a limb feels the stretching of the flexors. The extension is uncomfortable.

He walks with a slight spring-halt sort of gait, and his shoes show they are not much worn at the heel. There is a double lateral curvature of the spine, and the boy stoops very much, and especially in walking.

Sensory symptoms.—The eighth nerve impairment is the only one noted. Temperature sense.—Normal Vasomotor symptoms.—Face always pale, hands cold and damp and are constantly of a mottled reddish hue. Feet always cold.

Mentally, the boy is fairly bright, reads, writes (with difficulty), and is good natured. Cries and is amused at trifles. He is microcephalic. He acts and looks about ten years old. Evident general arrested development.

This completes the picture of a rare case, and for the better ease in the consideration of its classification, I will present a concise resumé of the principal features.

A boy of twelve is taken ill, gradually with all the

symptoms of a labio-glosso laryngeal paralysis. This extends more or less rapidly to upper facial parts, one sensory nerve and proceeds down the body, implicating profoundly the muscles of respiration and markedly those of the upper extremities. There is marked atrophy, in facial, tongue, throat,respiratory muscles,and those of the hand and arm. There is a general arrest of development and general emaciation.

Fibrillary twitchings, exaggerations of the reflexes and retraction of tendons complete the necessary resumé.

Glosso-labio-laryngeal paralysis is a disease confined to adult life. Most writers affirm that it never occurs in childhood. Progressive infantile bulbar paralysis of the family type, however, is a fact.

Hoffman reported a case in 1891.

Remak reported a case in 1892.

Fazio reported a case in 1893.

Bernhardt reported three cases in 1889.

Londe reported two cases, in brothers, in 1893.

They all bear resemblance to my case in the fact of the implication of the bulb and differ in most of them in the non-extension of the degeneration to the cord or the implication of the pyramidal tracts. Charcot, Joffroy and Marie are presumably not wrong in their showing that the large ganglion cells of the anterior horns and the pyramidal tracts can be simultaneously affected, or one be grafted on the other. We probably all accept amyotrophic lateral sclerosis as a distinctive clinical classification from progressive poliomyelitis and, in lieu of a better pathological knowledge, that amyotrophic lateral sclerosis may consist of a lesion of the cortico-muscular tract, traceable from ganglionic cells of the central convolution and bulb to the end of the cord, and *vice versa.*

According to Marie, amyotrophic lateral sclerosis is a muscular atrophy with extremely well-marked fibrillary contractions; the latter may exist before the atrophy is marked. This is seen in our case. Accord-

ing to this author, the disease does not occur in youth or childhood.

The muscular atrophy, again, is gradual muscle by muscle, fascicilus by fascicilus, fibre by fibre.

Lower limbs are affected very late. Electric reactions vary. Degeneration may be present if muscles are completely implicated. The reactions are, however, never well-marked, and often absent.

According to this author the resumé of symptoms are as follows:

Age, paralysis, atrophies, contractures, retraction of tendons, exaggeration of reflexes, fibrillary twitchings. Marked spasticity is not positively essential, but the exaggeration of reflexes of both upper and lower extremities is necessary. Early psychic disturbances are frequent and must be considered essential. A lessened intellect, childishness and especial proneness to weeping and laughter, steadily progressive and death most frequently by respiratory troubles.

The age in my case precludes its classification among the ordinary ones of amyotrophic lateral sclerosis. The case as well has not taken the usually progressive course; it progressed rapidly up to six months ago, since which time it has been more or less in *statu quo.*

In all other respects the resumé of symptoms is alike. I consider the case, however, as one of the family type of infantile progressive bulbar paralysis, plus the same implication that we find engrafted upon the progressive poliomyelitis of adults which gives us amyotrophic lateral sclerosis. My case certainly presents more extensive clinical features than the cases on record of progressive infantile bulbar paralysis, and it is just as logical to affirm amyotrophic lateral sclerosis as in the adult cases.

To classify the case as a dystrophy, especially in the class of either scapulo-humeral or facio-scapulo-humeral dystrophies, is out of the question. The very fact that, although emaciated and weak, yet the scapulo-humeral group of muscles and the back muscles gener-

ally are the best ones he has, what muscles he possesses are active, the exaggeration of reflexes, the fibrillary twitchings and the retraction of tendons, though slight, precludes peripheral origin.

To argue on any electrical reactions is illogical, for as long as a muscle fibre is in healthful contact with its trophic centre a reply will be obtained by the faradic current. On an entire loss or entire implication by disease of trophic influence only, can one depend upon obtaining marked degenerative reactions. Electrical reactions are certainly very unsatisfactory in all forms of partial muscular atrophies, and to study them aright is a most laborious, tedious and unsatisfactory task.

In muscular myopathies the respiration is unaffected. The upper facial muscles are usually intact, and the "tapir" mouth is not observed. When atrophy takes place it is more irregular and apt to be more bilateral, that is, worse on one side.

It is becoming more and more the belief that amyotrophic lateral sclerosis originally emanates from the bulb. This is seen in the reports of cases and in the writings upon the subject. Authors unconsciously bring out the facts that even though the paralysis and atrophy may first be noticed in the hand, often important symptoms precede these, and throat, and tongue and lip symptoms are often primarily noted.

Senator has lately reported an autopsy where the pathological findings show that the pyramidal tracts were intact.

Londe has considered the question of infantile bulbar paralysis at length, draws attention to the inability to wink and the sluggish opening of the lids as a diagnostic feature. Remak draws attention to this clinical feature as well, in the report of his case.

Though in my case the ability to wink was preserved, there was an inability to close the eyes. All the other superior histrionic muscles were affected.

In adult bulbar paralysis the upper facial muscles are rarely affected and more extensive paralysis occurs down-

ward. In most all the cases of infantile bulbar paralysis reported, the neck muscles or below were not involved, never the arms or legs, except in one case on record of this type, and this seems to more nearly agree in most of its clinical features to my case.

Hoffman's case, boy of eleven years.—There was marked glosso-labio-laryngeal paralysis and atrophy with great emaciation of upper extremities and body and extending down to below hips. Exaggerated reflexes in lower extremities and lessened in the upper extremities. In this case the atrophy was too extreme probably in the upper extremities to illicit much response. In all the cases reported respiration was greatly impaired. The case I present differs from them all in the extent of its atrophy, general fibrillary twitchings, brightened reflexes and retracted tendons. None show a more general nuclea affection. The affection of the auditory nerve is not noted in other cases. This case, however, deserves to stand apart, though linked to this class of cases, as amyotrophic lateral sclerosis is in the adult linked with progressive muscular atrophy and nuclea disease of the bulb.

BIBLIOGRAPHY.

Prof. J. Hoffman. Ueber chronische spinale muskel atrophie im Kindesalt. auf familiärer Basis. *Deut. Zeitschrift für Nervenheilkunde*, p. 427, 1893.

M. Pierre Marie. Sur la localisation des lessons medullares dans la sclèrose laterale amyotrophique. *Bulletine et Mém. de la Soc. Med. des Hosp. de Paris*, 1893.

Rovighi and J. Levi, *Revista Sper. di freniatria.*, xvii, 4, p. 492.

Prof. H. Senator. A case of so-called Amyotrophic Lateral Sclerosis. *Deut. Medicin. Wochenshr.*

A. Schüli. Deutsch Zeit. für Nervenheilkunde, vol 4, p. 161, 1893.

Strumpell. Ditto.

Eupine. Revue Méd. de la Suisse romandi. Geneva 1893.

L. Newmark. Am. Jour. Med. Sciences, April, 1893.

Erben, of Vienna. Internat. Clinish. Rundschau, 1893.
Oppenheim. Archiv. für Psychiat. und Nervenkrank. 1893.
Marie. Diseases of the Bulb.
P. Londe. Paralysie Bulbaire, Progressive Infantile et Familiale Revue de Medecine, December, 1893; also *La Medicine Modern*, vol. 4, pp. 806–7, 1893.
F. Fazio. Reforma Medica, Nov. 1892.
E. Remak. Zur Pathologie du Bulbär Paralyse. Arch. für Psychiatrie and Nervenkr., 1892.
J. Hoffman. Deut. Zeitschrift für Nervenheilkunde, p. 169, 1891.
Bernhardt. Virchow's Archiv., p. 187, 1889.

Neuritis with Tabetic Symptoms Due to Mercurial Poisoning.— In *Médicine Modérne*, July 5, 1893, a letter from Berlin gives an interesting account, reported by Leyden, of a man, twenty-five years of age, who presented grave symptoms of locomotor ataxia. Mercurial inunctions had been used earlier for recent syphilis. Three days after the cessation of this treatment the patient began to have pains in the arms, then in the lower limbs, together with difficulty in walking and diminished sensibility. When first seen, there was no evidence of syphilis, though there were many signs of ataxia, as lancinating pains, loss of tendon reflex, characteristic gait, etc. In a few weeks the patient's condition was much improved and the tendon reflexes reappeared. The treatment consisted in small doses of iodide of potassium, in rest, careful and abundant feeding, with antipyrin for pain, and in the administration, when necessary, of chloral and morphine. The earlier existence of syphilis naturally suggests a specific ataxia. But a most painstaking examination of the case revealed the existence of a neuritis with tabetic symptoms, such as Leyden and Déjerine have already described under the name of acute ataxia. L. F. B.

Primitive Progressive Myopathy.— Rovighi and V. Levi, in *Rivista Sper di Freniatria*, xvii., 4, p. 492, records three cases of primitive progressive myopathy. There were violent cramps in the calves and lower limbs generally. The authors ask if there may not be some direct relation between primitive progressive myopathy and Thomsen's disease, which is also hereditary.

L. F. B.