ON SOME RECENT ADVANCES IN OUR KNOWLEDGE OF THOMSEN'S DISEASE.

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- (1.) 'A Consideration of the Thomsen Symptom complex, with reference to a new form of Paralysis Agitans.' By ALLAN McLANE HAMILTON, M.D. *The Medical Record*, New York, January 23, 1886.
- (2.) 'Klinisches und Pathologisch-Anatomisches von der Thomsen'schen Krankheit.' Von Prof. W. ERB. *Neurologisches Centralblatt*, No. 13, 1885.
- (3.) 'Thomsen'schen Krankheit bei vier Geschwistern.' Von Prof. EULENBURG und Dr. MELCHERT. *Berliner Klinische Wochenschrift*, No. 38, 1885.
- (4.) 'Une Observation de Maladie de Thomsen.' Par MM. PITRES et DALLEDET. *Archives de Neurologie*, No. 29, 1885.
- (5.) 'Ein Fall von Thomsen'schen Krankheit.' Von DR. G. FISCHER. *Neurologisches Centralblatt*, 15th February, 1886.
- (6.) 'Maladie de Thomsen et Paralyse pseudohypotrophique.' Par VIGOUROUX. *Arch. de Neurologie*, vol. 8, No. 24, Nov. 1884.
- (7.) 'Observation d'un cas de Maladie de Thomsen.' Par L. DELIGNY. *L'Union Médicale*, 1885, No. 5.
- (8.) 'La Maladie de Thomsen (Dysmyotonie Congénitale).' Par LONGUET. *L'Union Médicale*, 1883, November 25.
- (9.) 'Contribution à l'histoire de la Maladie de Thomsen.' Par P. MARIE. *Revue de Médecine*, 1883, p. 1064.
- (10.) 'Fem Tilfælde af den Thomsen'ske Sygdom. Af KNUD PONTOPPIDAN' (*Hosp.-Tidende*, 1884, 3 R. II., 34).
- (11.) 'Ueber eine dem Bilde der cerebros spinalen grauen Degeneration ähnliche Erkrankung des centralen Nervensystems ohne anatomischen Befund, nebst einigen Bemerkungen über Paradoxe Contraction.' WESTPHAL, *Archiv für Psych.*, Bd. xiv., p. 87 (especially p. 130).
- (12.) 'Malattia di Thomsen. Sua Analogia coi fenomeni muscolari dell'ipnotismo,' Sepilli, *Arch. Ital. per le malattie nervose ec.* Fasc v. 1885.
- (13.) 'La Malattia di Thomsen.' PETRONE, *Lo Sperimentale*. July, 1884.
- (14.) 'Handbuch der Kinderkrankheiten.' GERHARDT, t. v., p. 175.
- (15.) 'A Case of Thomsen's Disease; a form of Paresis of Motion, accompanied by muscular hypertrophy.' ENGEL, *Philadelphia Medical Times*, September 8, 1883.
- (16.) 'Thomsen's Disease.' By PAUL CHAPMAN, M.D. *Brain*, vol. vi.

In the article last mentioned there is an almost complete list of the literature of the subject up to that date, and I think the fifteen other references that I have enumerated made the whole of the literature very nearly, if not quite complete up to the present date. The first five of the above articles will be discussed in detail, for not only are they the most recent, but also the most important. The sixth article does not call for comment, and the seventh relates an example of the disease in which the patient was a man 25 years old; his father had had the disease, but his general health had in no way been affected by it. The patient was the only child; the cramps caused great pain, and came on several times a day, and lasted a long while; the reflexes were normal. The peculiarity of the case seems to be that the contractions lasted so long, and there was so much pain with them.

The eighth article gives a very good account of the literature of the subject up to date, but makes confusion worse confounded by using another term, viz. *dysmyotonie congénitale*, almost adopting Strumpell's term, *congenital myotonia*. The exact nature of the case in the ninth article is probably rather doubtful, for the cramps were strictly one-sided, and the patient also suffered from epileptiform seizures. The tenth article records five cases, two in one family, and three in another. Portions of muscle were examined microscopically, and are stated to have been healthy. This paper is to be found abstracted in the '*Neurologisches Centralblatt*,' 1884, p. 520.

In the eleventh of the above-mentioned papers Westphal discusses briefly the nature of paradoxical contraction, and comes to the conclusion, that the contraction of Thomsen's disease is allied to it, but that our knowledge about both of them is at present very elementary.

Petrone's account of the disease is good, brief, and to the point, but does not contain anything new about it.

The first of the above articles begins with a historical account of the malady. It is pointed out, as is well known, that Sir Charles Bell, in 1836, mentioned the existence of a disease which was probably the same as the one now under discussion, but gave it no distinct name. The next person to call attention to it was Thomsen, who in 1876 described the disease as occurring in members of his own family. Since that time many examples of the disease have been recorded, chiefly in Germany, together with a few in each of France, Sweden, and Italy, and two in America. Altogether about twenty to thirty cases have been recorded. Hamilton gives the clinical expressions of the disorder as follow:—(1) a sudden spasmodic muscular seizure which attends the performance of a voluntary act; (2) its subsidence after the repetition of the act; (3) an apparent increase in bulk in several muscles which are chiefly and most frequently the subjects of cramp; (4) an increase in electro-muscular irritability; (5) a normal or slightly excited state of tendinous reflex activity; (6) these symptoms are often of congenital origin, and progressive as regards intensity.

I shall show later on that all these symptoms are in the main

correct, but that by the light of our present knowledge they can be much more definitely described. Hamilton points out that the lower extremities are most often implicated, and that hence when the patient essays to walk, there is sudden flexion of the thigh and leg, in which position, owing to the stiffness in contraction of the muscles, they remain in this condition from five to ten seconds, so that the patient often falls. It may be some time after a volitional impulse that the muscle contracts, when it does there is this stiffness [Muskelssteifigkeit]. The muscles of the face may be affected. Cold and mental excitement increase the liability to the disease. In only one case were the reflexes exaggerated; a congenital history is not always the rule. Hamilton's object in writing the paper is to throw doubt upon the question whether Thomsen's disease is to be looked upon as a distinct malady, for he points out that this muscle-stiffness on contraction is a frequent symptom in a variety of maladies; in most cases it is purely psychic; it is an occasional symptom of the third stage of locomotor ataxy; it is common in hystero-organic disorders of the nervous system, and may be associated with coarse cerebral lesions, or may be present in Parkinson's disease. No mention is made of the recent article by Erb and others, or of the microscopical appearance of the muscles, nor are the electrical reactions and mechanical irritability reactions referred to in much detail; and although the question is still under discussion, and awaiting a promised article by Erb on the subject, I think we shall see that whilst it is by no means deniable that cramps may take place in a number of diseases, still this group of symptoms taken with the changes in the muscle, points strongly to the fact, that Thomsen's disease must be looked upon as a genuine and distinct malady. In further proof of this it may be observed that the hitherto recorded cases have not been suffering from paralysis agitans, nor any other disease mentioned by Hamilton. The author then gives an account of a boy, aged 10 years, who had always suffered from cramps, which were worse under mental excitement, and caused him to walk in a squat position. No change could be detected in the portions of muscle removed, and the electrical reactions were normal. Both these facts are against the case being one of Thomsen's disease, and no diagnosis seems to have been made. Another case is given in which a boy, 9 years old, who had suffered from headache and attacks of vomiting, had muscle-stiffness on contraction in the voluntary muscles; muscular enlargement and loss of power; the mouth was drawn slightly to the left side; the tongue pointed to the right; there was a prominence of the eyes and a slight lateral nystagmus. The headache and vomiting do not necessarily preclude Thomsen's disease, as the cases of Eulenburg and Melchert show; still the other symptoms probably do, and so we have here another example of cramps occurring in other diseases. For the convenience of those who wish to look up the subject, I may mention that Thomsen's original article is to be found in the 'Archiv für Psychiatrie und Nervenkrankheiten,' 1876. Hamilton, at the heading of his article, uses the phrase "Thomsen symptom complex." This is a pity, for not only does it lead to con-

fusion, because the phrase "symptom complex" has a wider significance, as we see in an article by Westphal, "Ueber einen eigenthümlichen Symptomencomplex bei Erkrankung der Hinterstränge des Rückenmarks,"¹ where the "Symptomencomplexes" apparently included paralysis, paradoxical contraction, rigidity and cramp in certain movements; but a much better phrase, viz., myotonic contraction, has been proposed, which is shorter and less cumbrous than "Thomsen symptom complex."

Erb begins his article by pointing out, that although there has never been a post-mortem examination on a case of Thomsen's disease, yet the five observers who have excised portions of the muscle have failed hitherto to find any change in it. He gives an account of two brothers, aged respectively 14 and 15 years, who had it typically, and in whom it had been present some time. The voluntary power of the muscles was slightly diminished, especially considering their size. On movement of the voluntary muscles there was the usual trouble, viz. a slowness and difficulty of contraction; then a stiffness and tension, which suddenly passed off; and then there was free movement. With a very powerful voluntary movement the duration of this tonus was from 10-25 seconds. The motor nerves with mechanical stimulation showed diminished excitability. With the faradic and galvanic currents the reactions of the nerves are practically normal, unless the stimulations follow very quickly, when there is a slight lasting contraction. In the muscles the mechanical excitability is increased, and after a strong stimulus remains from 12 to 30 seconds. A strong faradic current gives a contraction which remains for 20 or more seconds; but on opening, there is only a momentary contraction with even the strongest current. With the galvanic current the muscles usually give only closing contractions. K and A are usually alike, but in some muscles Ka S Z may be best, in others An S Z. It is with Ka S Z that one gets the best tonic contraction, which is often extremely good, and lasts 5-30 seconds. With the stimulations rhythmically following one another, we get wave like contractions also following each other, and going along the muscle from the kathode to the anode. This is well seen in the flexors of the fingers, the vastus internus and externus. All the above reactions are best shown in muscles which are under the influence of the will. The essentials are the slowness and duration of the contraction; a good name for it is myotonic contraction or myotonic disturbance of muscles. It shows itself on direct stimulation of the muscle, or, in the case of the nerve, when one stimulus quickly follows another.

On microscopical examination of a piece of excised muscle in the fresh state, the only thing that could be made out was that the fibres were a little broader than natural; but with Müller's fluid preparations they appeared many times wider than they should be, and not polygonal, but rounded. The interstitial tissue and vessels were normal; there was considerable thickening of the sarcolemma.

¹ 'Archiv für Psych.,' etc., 1885, Bd. xvi.

Eulenburg and Melchert have given a most interesting account in which the disease affected four brothers and sisters. The family history was unimportant; the mother suffered from cramp in the stomach and headache. Of the six children, all suffered from cramps and convulsions during teething, and during their school-days from hemicrania and vomiting. One daughter had migraine; another used to wake up with attacks of cardiac pain and palpitation. The three sons and the eldest daughter were typical examples of Thomsen's disease. In all of them the voluntary muscles were affected, and there was the usual contraction. The following are the details of each of the four cases.

W. K., 22 years old; he was first attacked when 7 years old, and then he found stiffness on attempting to run; this continued off and on, and from 14 to 18 he suffered very badly, so that he would often fall in the street; after 18 he became a little better; but during his service in the army the disease gave rise to so much trouble, that he had great difficulty in carrying out correctly the exercise orders; in the same way it was difficult, in shooting, to raise the musket to the shoulder, and he had much difficulty during his apprenticeship as a dentist. On examination, the peculiarities of contraction of voluntary muscles, as described in the two previous articles, were most apparent, and, as already mentioned, after a few contractions the myotatic contractions passed off. The peculiarity was present in the forearms, lowers, body, face, deglutition muscles, tongue and eye muscles. The myotatic contraction was most marked the longer the muscles had been at rest, worse after cold and inanition, but better after meals. On direct mechanical stimulation, after a short time the myotatic contraction appeared, lasting 3-5 seconds. With faradic stimulus to muscle and nerve there was nothing noteworthy. With the galvanic current to the muscles, K S Z appeared sooner than A S Z and A O Z > K O Z, but the contractions came slowly, and lasted 2-5 seconds; with galvanic current to the nerve, K S Z appeared sooner than A S Z, but the contraction was momentary. Patellar reflex weak.

George K., 21 years old; as a child had headache and vomiting. When a boy, had slight difficulty of movement, which showed itself in gymnastics. His muscles were, when he performed voluntary acts, in the same myotatic condition as those of his brother, but worse. Mechanical stimulation to muscles gave the same results, but they were more pronounced. Faradism as in brother; galvanism to nerve K S Z > A S Z, with no myotonic contraction; this and muscular galvanism also being the same as the brother. There was a prolonged latent period; there was in succession a prolonged contraction, and then a sudden relaxation, best seen after prolonged rest; frequent slight tremors of the muscles were to be seen, and at times there were very strong opening contractions with the kathode.

Carl K., 21 years old. Headache and vomiting when young. The trouble with the muscles came on at 12 years, when he was found to be bad at gymnastics. The symptoms were not so

marked in his case; the mechanical myotatic contraction was present in slight degree. Faradism gave no lasting contractions; the galvanism results were just the same, but weaker. Patellar reflex weak.

Louise A., 29 years old. The disease first appeared in childhood, and got worse at puberty. She has great difficulty in going upstairs, and in executing complicated movements, as dancing. All the symptoms were pretty much the same as in the brothers. The author directs attention to the fact, that the two children who have not had Thomsen's disease have anomalies of their nervous system; also that the symptoms increase after cold and exhaustion, and the daughter was better when she lived in more comfortable circumstances. The four cases showed slight qualitative differences to their electrical reactions, and Ebb's wave contractions with strong current were not observed, but there was a tendency to tremulous oscillations with faradic stimulation.

MM. A. Pitres et Dalledet have given a very full account of a patient suffering from this disease; they also give several myographic tracings. The grandparents and the father were healthy. The mother at the age of 15 suffered from muscular stiffness, like that which her son, the patient, suffers from now. This difficulty disappeared in two or three years; then there were cramps, chiefly in the sacro-lumbar region, the pains coming on day or night, without any relation to movement. She and her husband had six children; the three first being like the father in all points, the three younger, on the contrary, suffering from muscular stiffness on voluntary movement. Of these, the fifth is the patient, and will be described in detail; the fourth, a man of 26 years, is exactly like the patient in all the symptoms, but not so bad; and the sixth, a girl of 19 years, had a difficulty of voluntary movements when 15 and 16, but that has now disappeared. The difficulty of voluntary movement of which the patient complains began when he was a child. Up to the age of 22 it increased but slowly; since then it has gone on very quickly. He is now 24.

The first thing that strikes one is the hypertrophy of the muscles, which is well seen in the arm and forearm, and the thenar and hyperthenar eminences. The sterno-mastoids, triceps of thigh, and sacro-lumbar muscles are all hypertrophied. During contraction they are irregularly bossed, and as hard as wood. The power of the muscles is not so great as their hypertrophy would lead one to expect. The electrical reactions are normal. The difficulty of voluntary movement consisted in a restraint and stiffness on the attempt, which gradually passed off as the movement was frequently repeated. This is very well shown by a myographic tracing, which shows that the latent period is increased considerably, and the muscle remains contracted much longer than it should. The authors also insist on a point which their tracings show, but which has not been noted in other cases, namely, that not only is there a difficulty of contraction, but also of the first *décontractions*; in fact, in the tracings the down-stroke

often extends over a longer period than either the up-stroke or the crest. With a strong and rapidly repeated faradic current, the same thing is observed in the early contractions. This, it will be observed, brings this case somewhat in harmony with Erb's electrical myotatic contractions, and perhaps when the authors here speak of the electrical reactions normal, they did not look for the myotatic reactions. The peculiarities of contraction were increased by cold. The muscles of the face would sometimes remain in an emotional expression so long, that the hand had to be used to aid their return to normal, hence, sometimes there was difficulty of speech. The fear of the peculiarity being observed increased it. The tongue, deglutition, phonation, respiration and ocular muscles were not affected. Passive movements were easy, and sensibility was intact; reflexes normal, sensation of position in space normal. All other functions of body healthy.

The fifth paper, mentioned at the beginning of this notice, has only just been published. The author observes that in this case the skin and tendon reflexes were normal, and that the strength of the muscles was not so much increased as would have been expected from the increase of volume. Sometimes the myotonic contraction remained from 25-27 seconds after voluntary contraction, but after repeated movements it disappeared. On the whole, this case agrees with others, and hardly differs more from them than they do among themselves. The myotatic reaction, in response to direct muscle stimulation, was not so easily obtained, and no K O Z could be obtained on stimulation of the nerve. The rhythmic wave-contractions of Erb were shown very well indeed, and with a current of a strength of 15-20 M A, they were well formed, slow, and about 15-20 seconds apart, passing from the kathode and the anode. With a strong faradic current there were rapid oscillations of the muscles. On microscopic examination of the fibres they are found to be thickened. In no parts was the cross striping well marked. The contour of the bundles is uneven, and has here and there little tumefaction. The author raises the question whether these appearances are due to the mechanical stimulation of excision.

In conclusion, we may, I think, gather that the following are the chief characteristics of Thomsen's disease:

1. It occurs in young subjects, coming on usually in youth or childhood.
2. It lasts a long while; is never fatal; and may pass off.
3. The muscular difficulty is much increased by cold and mental excitement.
4. It occurs in members of the same family, and is slightly hereditary.
5. Other neurotic disturbances in the family are by no means always well marked.
6. Voluntary muscles only are affected, the legs perhaps oftenest.
7. There is an increase of bulk in the muscles.

8. But considering their increase in size, they are not so powerful as might be expected.

9. On attempted voluntary movement the latent period is usually prolonged; then comes a severe contraction, lasting from five to thirty seconds; then in some cases a sudden, and in others a slow relaxation. This peculiarity constitutes a myotonic contraction.

10. The above is more marked, the longer has been the interval of rest. After repeated contractions the difficulty becomes less, and soon passes off, the contractions being ordinary.

11. Mechanical stimulation of nerves shows a diminished excitability. Faradic or galvanic stimulation of nerves gives slight myotonic contractions on being rapidly repeated.

12. Mechanical stimulation of muscles shows increased excitability, with long myotonic contractions. Galvanic stimulation usually gives closing contractions; K S Z is generally the best, giving a good myotonic contraction, but the exact order is variable. Strong faradic current on closing gives a good myotonic contraction; on opening, a very slight contraction.

13. With stimulations rhythmically following each other, we get wave-like contractions passing along the muscle from the kathode to the anode.

14. The reflexes are normal.

15. There is increased width of the muscle fibres, which lose some of their striation, are rounded instead of polygonal, and have protuberances on their contour. The sarcolemma is thick.