(With tracings)

# Thomsen's Disease
## (Congenital Myotonia)

being a graduation thesis

for the degree of Doctor of Medicine

presented to the Senatus

of the University of Edinburgh.

by

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# On Thomsen's Disease
## (Myotonia Congenita)

The advent of a new disease is always of interest and since 1876 when Dr. J. Thomsen of Kappeln, Schleswig described graphically and established as a separate entity the affection which has since then deservedly borne his name, much attention has from time to time been given to it by different Continental observers. In Britain however, owing either to the rarity of cases or to failure to recognize them, no instance of the disease was shewn, so far as I can ascertain, until 1887 (Buzzard); and with the notable Exception of Hale Whites' masterly summaries in the Guy's Hosp. Reports - to be referred to later - British contributions to its literature were rather scanty.

I propose to describe a case which I have had very exceptional opportunities of observing, and of which the family history is well known to me, prefacing

the account with a résumé of the literature of the subject up to the present time.

Survey of literature

As far back as 1830 Sir* Charles Bell hints very strongly at this remarkable affection, and mentions that some people, who can lift heavy weights &c., have not the proper control of their limbs, & that excitement intensifies the defect. He describes symptoms since recognized as typical e.g. the knee stiffness; the danger of falling & the staggering gait on sudden movement & its wearing off on repeated movement; the freedom from pain, but there is no suggestion of rigidity or muscular spasm.

Chas Bell

Benedict

In Benedict's Nervous Pathology a similar condition is described; but, for all practical purposes, the history of the disease begins with Thomsen's classical & account, which was entitled "Tonische Krämpfe in willkürlich beweglichen Muskeln in folge von Ererbter psychischer dispositionen".

Thomsen

Dr Thomsen & several members of his family suffered from the complaint, which had existed in the family for five generations.

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Nervous System of the Human Body - Chas Bell. (1st Ed. 1830 P. 164)

J. Thomsen - Arch von psych und Nervenkrankheiten Berlin - 1876 Bd. VI. S. 702

Benedict - Nervon Pathologie und Electrotherapie 1st Ed 1864
2nd " 1874

The characteristic symptoms were (1) A peculiar tonic cramp-like contraction of the muscles without pain. coming on, on attempted voluntary movement after a state of rest, or on changing one type of movement for another (2) the disappearance of this tonic contraction after repeated similar movements eg. as in walking (3) the involvement of voluntary muscles only and those of the limbs more especially the facial, ocular, tongue & laryngeal muscles being rarely implicated.
(4) the presence of a greater or less degree of hypertrophy of the affected muscular groups (5) the aggravation of the condition by Emotion ~~&~~ Cold. (6) Its association with other Neuroses (7) Its hereditary and practically incurable character.
For reasons which will I think be evident on perusal of my final note to this paper I need not go into further details of Thomsen's paper merely drawing attention to the fact that he believed the disease to be a Neurosis, and the primary defect to be in the originating of motor impulses in the Cerebrum.

It should be noted that examination of muscle fragments from Thomsen's sons gave negative results (possibly due to defective methods of examination).

Seeligmüller

In 1876. Seeligmüller published a case in which there was hypertrophy of groups of muscles with skin & panniculus normal, a subjective sense of the limbs being asleep, and considerable opposition to passive movement especially in the knee joint. This observer inclined to the view that the defect was in the lateral columns of the cord - a congenital defect in development.

Erb

Two years later Erb in an article entitled "Tonic Spasms in muscles capable of voluntary movement", quotes the above mentioned case, and says that this and similar ones depend probably on some congenital anomaly of the nervous system occurring repeatedly in the same family. Two other articles by this authority have since appeared adding considerably to our knowledge of the electrical reactions and histology of the affected muscles, to which reference will be made later.

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Seeligmüller - Deutsche Med. Wochenschrift 1876. P. 389.

Erb. - Ziemssen's Cyclopaedia Vol. XIII trans New York 1878. P. 808.

Bernhardt.

In 1879 Bernhardt[1] recorded a case due to injury, & stated that the Electrical reactions were normal, but that the Muscular Contraction persists after the cessation of Electrical Stimulation. He also noted the wave like contractions passing from the ~~Anode~~ Kathode to ~~Cathode~~ Anode when the muscles were faradised & their increased Excitability to mechanical Stimulation - observations amplified by Erb in his later articles.

Peters.

In this same year the phenomena were well described by Peters[2], and in 1881 Strümpell

Strümpell

had a descriptive article entitled "Tonische Krämpfe in willkürlich bewegten muskeln (Myotonia congenita), and in Italy Petrone[4]

Petrone

followed suit.

Westphal & Moeli

The next important contribution I can find was that of Westphal[5] & Moeli, being an account of their case shewn Mar 9th 1881 before the Society of Psychology, Berlin.

The Symptoms were ably grouped as follows:-

1. Continued immobility of muscles on standing up after sitting - e.g. if the patient have been seated along time, on attempting to rise, he can only with apparent difficulty bend

(1) "Muskelsteifigkeit und muskelhypertrophie." Virch. Archiv. Berlin 1879. Art XXIX t. LXXVI p. 515.
(2) 'Deutsche milit ärztl Ztsch' 1879 t. VIII. p. 101.
(3) 'Berlin Klin. Woch.' 9. 1881.
(4) 'Revista sperim di Frenatria' 1881. t. VIII. p. 101.
(5) 'Archiv. de Neurologie' Sept. 1892.

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bend his joints, through the rigidity of his muscles.

2. Persistent contraction after more violent muscular efforts or after a single powerful effort e.g. sudden flexion of forearm. or

3. Inconvenient contractions under certain complicated movements e.g. Children will be seized in the midst of playing & suddenly brought to a standstill.

4. The tonic cramp will come on so suddenly that the somatic & physical phenomena can scarcely be separated - for instance one patient if he ran quickly & caught his foot against a stone would fall flat on face with arms extended and rigidity of whole muscular system.

5. That, as noted by Thomsen, the sudden thought of the affection or imagination will greatly Exaggerate the symptoms, as in some cases will cold & Exposure (all depressants of the Nervous System). On the other hand Continuous passive movement favours the restoration to normal for the moment.

6. It affects the muscles of the trunk and Extremities - the Sphincters being free

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The muscles supplied by the cerebral nerves are however subject to the same affection though in a less degree. Eg. there may be stiffness of the tongue in talking, of the masseters in eating, and of the eyelids in movement.

6. The muscles supplied by the spinal nerves have shewn an increase in bulk, and may be inordinately strong, though the increase of power is not usually proportionate to their size.

7. The mechanical and Electrical Excitability of the muscles is augmented

8. In 2 cases the Excised muscular fragments have appeared fairly normal.

(N.B. This latter statement must be changed in the light of our present knowledge.)

Vizioli

1882 In this year the leading article was that of Vizioli[1]. describing two cases of the affection. In one of which there was pain over the lower spinal vertebrae, radiating to the iliac bones + worse after walking - not increased however by pressure or passive movements.

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(1) F. Vizioli – "Due casi di rigidità muscolaire con lieve ~~ipertrophia~~" – 'Gior. di neuropatologia' napoli Luglio-Agost 1882 – Sett-Ott. 1882.

1883. saw several interesting Communications on this subject, notably that of Ballet[1] & Marie who gave a full report of a case in an Exhaustive article entitled "Spasme musculaire au début des mouvements volontaires (Étude d'un trouble fonctionnel jusqu'à ce jour non décrit En France) Amongst other points they called attention to the prolongation of the muscular contractions. Schönfeld[2] in Germany & Engel[3] in America also described cases arising from fright & nervous shock (a point which would militate strongly against the muscular theory of the disease referred to later) (No 13)
Westphal also shewed 2 cases at the sitting of the Berlin medical society in this year.
1884 The only reported case appears to be that of Vigouroux[4] entitled "Maladie de Thomsen et Paralysie pseudo-hypertrophique".
1885 In this year Erb[5] contributed another able article in which he called attention to the scantiness of pathological data up to that time, and described the electrical reactions of the muscles. He described also the histological appearance of Excised

. Ballet & P. Marie

Schönfeld & Engel.

ases from Shock to N.S.

igouroux

Erb.

(1) G. Ballet et P. Marie – 'Arch. de neurologie' Janv. 1883.
(2) Schönfeld – 'Berl. klin. Wochenschrift' July 1883
(3) Engel – 'Phil.a med. times' Sept 8. 1883
(4) Vigouroux – 'Arch. de neurologie': Vol 8. No 24. Nov. 1884
(5) Erb – 'Klinisches und pathologisch anatomisches von der Thomsen'schen Krankheit'
Neurolog. Centralblatt No 13. 1885.

muscular fragments. He points he observed were:- (1) the fibres were much wider than normal (2) they were rounded, not polygonal on cross-section (3) the interstitial tissue & vessels were normal, but there was thickening of the sarcolemma. He also noted and described in the vasti & other muscles the wave-like contractions (which are characteristic) passing from the Kathode to anode when the galvanic current is allowed to flow some time, or the shocks are repeated at rhythmical intervals. To the prolonged tonic muscular contraction with lengthened relaxation, readily obtained on cathodal closure & the associated neuro-muscular phenomena he gave the name Myotonic Reaction (MyR)

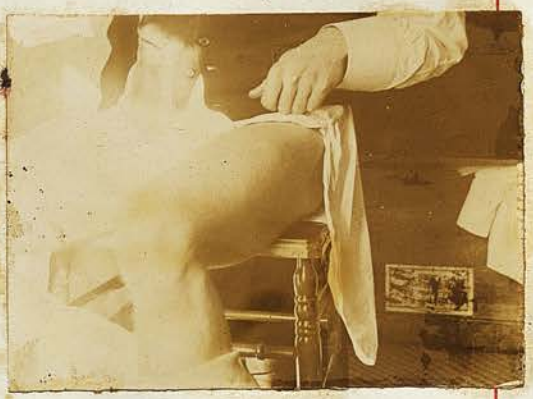
Erb's Myotonic Reaction MyR.

Eulenberg[1] & Melchert now attracted attention by publishing the account of another family group of 4 cases. The nervous encumbrance was marked. Two of six children all had cramps & convulsions during teething, & hemicrania & vomiting during their schooldays. One daughter had migraine, another, anginous attacks with vomiting. The electrical reactions differed a little quantitively.

(1) Eulenberg & Melchert - 'Thomsen'schen Krankheit bei vier geschwistern' - Berl. Klin. Woch. No 38. 1885

# Illustrating the muscular condition in Thomsen's disease

(1) (2)



(1) Forearm shewing the marked muscular development on slender bones.

(2) Shewing prolonged relaxation of Vastus Ext. some seconds after removal of tibia phore (My C.) (the relaxing muscle to upper + outer part of thigh)

(3) (should be turned sideways) (4)

(3) Lateral view of thigh relaxed - muscles flabby + inconspicuous.

(4) Front view of thigh with knee suddenly Extended, shewing iron-hard tonically contracted Extensors.

Photographs
illustrating the muscular condition

Other observers who published cases or articles in '85 were Pitres[1] & Dalledet, Deligny[2], & Bernhardt[3] the latter mainly on the pathological aspects of the disease.

1886. Hamilton[4] of New York had an important article in this year on "The Thomsen Symptom Complex". He noted the distribution of the recorded cases, and summarized the symptoms. He tried apparently to throw doubt on the Entity of T.D, but this is now put beyond Controversy.

More important still was an able summary by Hale White[5] of our knowledge up to that date "On some recent advances in our knowledge of Thomsen's disease". This observer after summarizing the literature; recorded cases & symptoms, gave a clear and able account of the Electrical reactions of the muscles & the microscopic appearances of the fibres. That part of his summary relating to the muscles may be here given.

(1) Muscles are increased in bulk.

(2) No corresponding increase in strength.

(3) On attempted voluntary movement, the

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(1) 'Arch de Neurologie' no 29. 1885

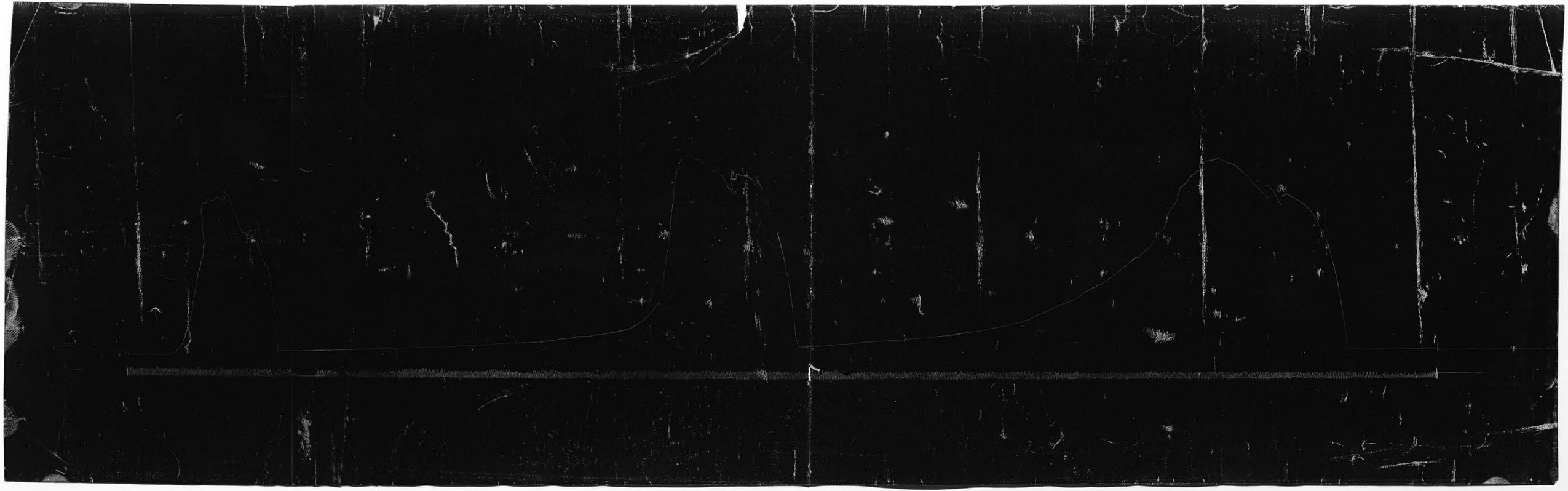
(2) 'Union Méd.' Paris 1885 xxxix 50-52

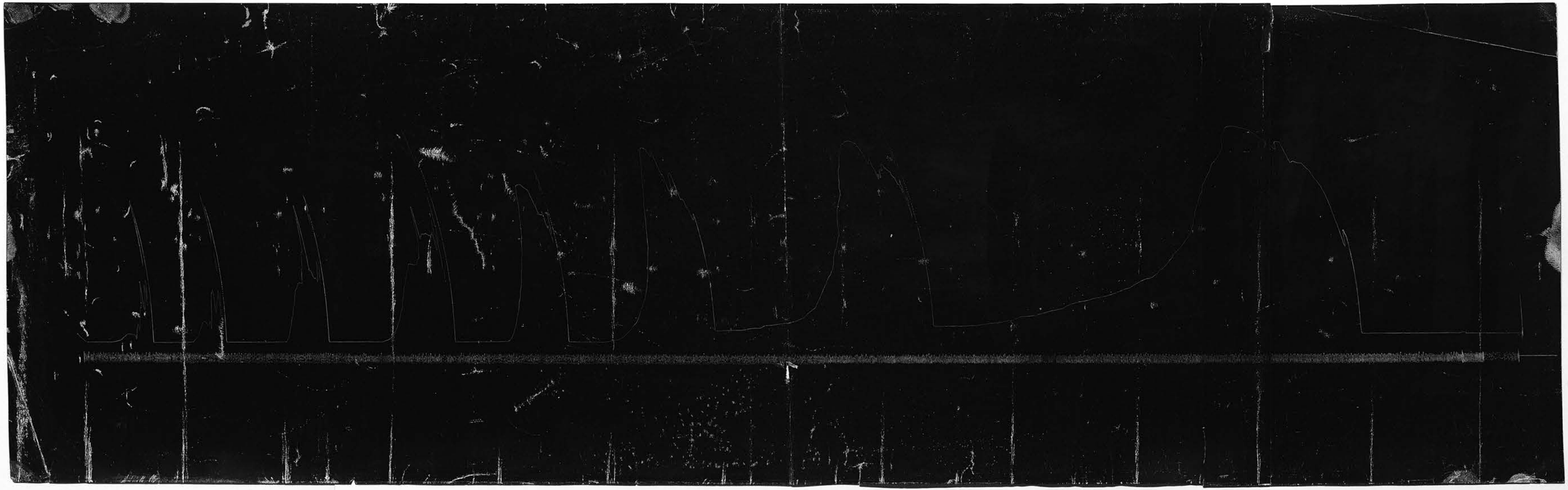
(3) 'Centralbl f. Nervenh.' Leipz. 1885. viii-122-126

(4) 'Medical Record', New York. Jan 23. 1886

(5) ~~'Guys Hosp. Reports' T. 46.~~

H. White - 'Brain'. part xxxiii. Ap. '86. p. 112.





latent period usually prolonged (this be modified later. N.B.) then comes a severe contraction lasting 5–30 seconds., then sometimes a slow sometimes a rapid (?) relaxation. This peculiarity constitutes a <u>myotonic contraction</u>

(4) The above is more marked the longer the interval of rest. & after repeated contractions the difficulty gradually wears off.

(5) Mechanical stimulation of nerves = diminished Excitability (cf. my case N.B.) Faradic or galvanic stimulation of nerve = slight Myotonic contractions or shocks being rapidly repeated.

(6) Mechanical Stimulation of muscle = increased Excitability – with long myotonic Contraction. Galvanic stimulation usually gives Closing contractions, KSZ is generally the best giving a good myotonic reaction but the Exact order is variable.
Strong Faradic current, on closing, gives a good MyR. On opening gives a very slight Contraction.

(7) Stimulations rhythmically succeeding each other we get the wave-like Contract$^{n}$ from Kathode to Anode.

Other cases by Hammond[1] of New York and Fischer[2] also appeared this year, and an article by Delmas[3]. "Maladie de Thomsen", which is of interest. Erb also issued another monograph 1887. Saw the first 2 cases exhibited in England Buzzard[4] shewing two brothers - typical cases along with Dr. Geo. Herschell he has the distinction of first demonstrating the disease in Britain (The diagnosis of my own case was made by myself and confirmed by medical friends before these cases were shewn). They were described in the Lancet. N.B.

In Germany an article appeared by Mibelcisen[5] "Zur Casuistik der Myotonia congenita oder Thomsen'schen Krankheit"

1888. Jacoby[6] of N.Y. in this year wrote an able article on the disease. he inclined to the "Muscular theory" as regards its pathology a point which will be discussed later.

Blumenau[7] shewed one case before the psychical society of St. Petersburg. in which the disability was greatly diminished under warmth & alcohol. He found lengthening of latent period (?) in his tracings)

(7) Blumenau - 'Neurolog Centralblatt' Dec 13. 1888. p. 679.
(6) Jacoby - Journ. nerv. & ment. diseases Vol XIV. no 3. p. 129.
(1) Hammond - 'Thomsen's disease' - Gaillard's Med. Journ. N.Y. 1886. XLI. 614-617.
(2) Fischer - "Ein Fall von Thomsen'scher Krankheit" 'Neurol. Centralblatt' Leipz 1886. V. 73-78.
(3) Delmas - 'Journ. de Méd. de Bordeaux' 1886-87 XVI - 97-100.
(4) Buzzard - Two cases of Thomsens disease, Lancet, Lond. 1887 I. 972-974.
(5) Mibelcisen - Munchen Med Wochenschr 1887 - XXXIV. 438.

1888. Bernhardt[1] one of the original workers at T. D. published a paper on its pathology in this year.

1889-90. The leading papers of this year were those of Hale White[2] in the Guy's Hospital Reports. - A very complete description of a case, with myographic tracings, electrical reactions, and a general discussion of all the points of interest - in fact a most able & careful study of the disease.

As I have already referred at some length to Dr. Whites previous article on this affection I need hardly go into details.

His account of the electrical condition agrees very closely with that of my case, and represents probably the most typical state of affairs. The rarity of the disease is shewn by his statement that only Buzzards' cases had been shewn at meetings in England up to Jan 27th 90, and that Erb[3] in an appendix to his monograph stated that 46 cases only had been recorded up to that date. Another article that attracted attention was that of Wising[4] - who described a Swedish case.

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(1) Bernhardt - "Zur pathologie der Thomsen'schen Krankheit" Centralblatt f. Nervenh. Leipz. 1887. pp 673-681.

(2) Hale White 'Guys Hosp. Reports' T. 46 - 1889.

(3) Erb - 'Deutsch archiv f. Klin. Med' 90 Bd XLV. S. 529.

(4) Wising P. J. 'Hygiea' - Stockholm 1889.

1890. In this year Cook & Sweeten[1] described a case in England, and Dreschfeld[2] another which was shewn at Manchester to the medical Society. The cases appear to have been pretty typical, and Hughes[3] & others published American cases some typical & some not. Renner called attention to two cases in which the Eye muscles were affected (this is well shewn in my own case NB)

Although other papers might be mentioned they do not chronicle any advance in our knowledge of the affection - indeed to Thomsen himself; Westphal, Marie & Especially Erb we are indebted for the gist of all we know about this remarkable disorder, which their researches have established as a separate Entity in the list of Diseases.

I shall only refer to one other article viz that of Déjérine[4] & Psottas describing an autopsy (So far as I am aware the Only autopsy) on a case of T.D. It was a man of 32. who died of Acute Nephritis. There was no abnormality

(1) Cook & Sweeten. Brit. med. Jol. 1890 - I. 73.
(2) Dreschfeld. do. 1890 I. 429.
(3) Hughes - 'Alienist & Neurologist' St. Louis. 1890. XI. 62-74
(4) Déjérine & Psottas - 'Revue de Médecine'. Mar 1895

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of the Nervous System detected. They gave a careful account of the histological appearance of the muscles - and described the vacuolation & interstitial growth of connective tissue as being a later degenerative stage of the affection. They found that the hypertrophy commences & is most marked in the muscles that are most exercised and came to the conclusion that it was a primary myopathy of functional origin.

Since this date I have been unable to find anything of real importance except that one of the Paris theses (1) for 1897 was on this subject. I regret that I was unable to obtain the article for reference & that even the name of the Author has escaped me. Some interesting ~~references~~ points about its relationship to other Nervous diseases were brought forward.

M.B.

(1) Paris - University of. Theses. 1897

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Description of Present case.

My own case, a man of 34 - 5ft 8 in in height and 150 lbs weight has the following history. The muscular stiffness was first noticed at the age of 7, when it was supposed to be due to rheumatism and precautions in the shape of heavy underclothing &c. accordingly taken.
Even as a young schoolboy he was noted amongst his fellows for physical strength associated with a degree of clumsiness of movement on initiating any voluntary action after a state of rest.
He found himself handicapped at cricket & football by the muscular spasm, & often suffered heavy falls & slight accidents as a result of it; e.g. when starting in a race he could take the first step or two & would then be pulled up by the strong spasm in the leg muscles, after a little the stiffness would wear off on continuing to run, and when fairly "limbered up" he was in no whit inferior to his schoolfellows in Speed or Endurance. Jf whilst running he

happened to strike his foot against an obstacle, he was very apt to fall headlong without being able to save himself and with all his muscles stiff & rigid.
The arms were equally affected and were early noticed to be more muscular than those of other boys of the same age.
On turning the head suddenly as in looking over the shoulder there would be marked stiffness of the neck muscles, and he has always worn a larger collar than most people. The jaw muscles frequently shewed the same tendency e.g. after taking one bite, he would sometimes be unable for a few moments to take another until the spasm in the masseters went off.
The tongue muscles & those of the eyeball at times shewed the characteristic prolonged contraction in marked fashion. giving rise to thickness & difficulty in articulation, and to fixing of the eyes in kind of "glare". These symptoms passing off rapidly. the latter one after stroking the eyelids a little. There appears also to have always been a similar affection

in the voluntary muscles concerned in the act of micturition. (This is interesting in view of the statement, frequently made, that these are never affected).

The other facial muscles would at times show traces of the stiffness, but this was only noticeable in cold weather.

My patient naturally found these muscular vagaries very annoying, and there can be no doubt, as Gowers remarks, that the complaint casts a gloom over the life of the sufferer.

The symptoms were noticeably improved by warmth & moderate exercise, but were greatly aggravated by any mental excitement by fatigue or by prolonged cold.

He describes the spasm as having been always preceded by a * dreadful sense of numb powerlessness, and has also a vivid recollection of many awkward & even ludicrous situations in which his then unrecognised complaint landed him in past years.

The patient has always been very "nervous" & highly strung, easily exhausted, and never

* A subjective feeling of the limbs being "asleep."

NB.

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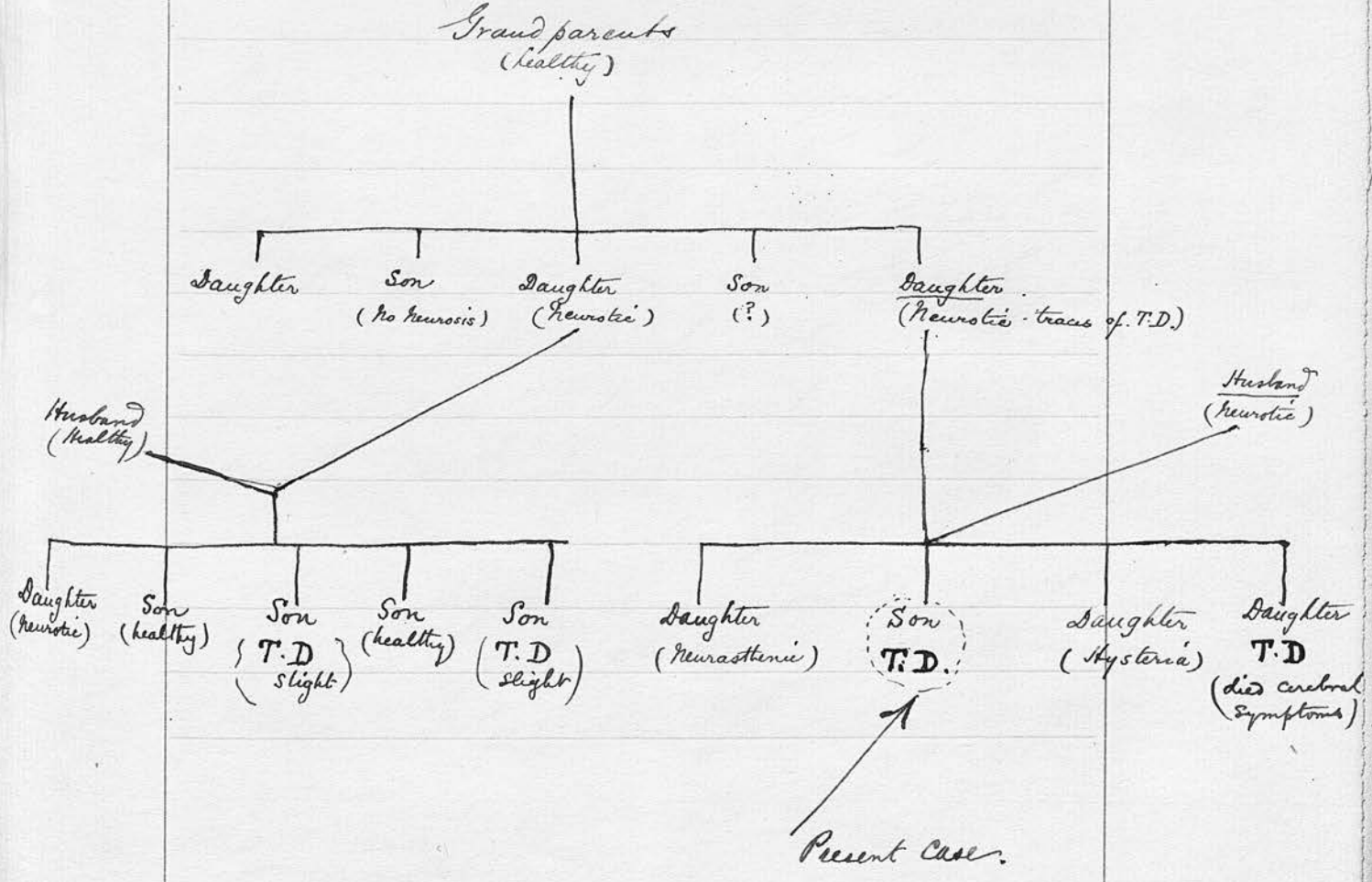
robust, but has been on the whole remarkably free from serious illness. I should note that Excessive Sweating seems to have always been present on anything like serious exertion, a Symptom I do not recollect seeing mentioned in other cases. It was most marked in head & neck.

Family History. Inquiry into the family history shews that the mother, and the only married maternal aunt transmitted neurotic tendencies to their children - in the one instance strongly, in the other only feebly.
In the case of the strong inheritance, the son (my case) and one daughter were well marked instances of Thomsen's Disease, the one highly "nervous" & the other partially imbecile (death at 18 with obscure Cerebral Symptoms - no autopsy) Of the other two daughters, one suffered from repeated attacks of Neurasthenia & one from an aggravated form of Hysteria for some time.
Amongst the children of the maternal aunt (four sons & a daughter), two sons were affected with a slight form of T. D. Shewing itself mainly in the lower limbs

and accompanied by some degree of the characteristic muscular hypertrophy.
No cases were traced on the paternal side of the family, but my patient's father was of neurotic type, whilst there was no such tendency in the Aunts' husband - this fact perhaps accounting for the more severe incidence of nervous symptoms in the children of the former.

Family tree 3 generations shewing incidence of T.D + other neuroses in patient's family.



Peculiarity of movement & appearance of the muscles. *

On stripping the patient it is at once apparent that he is very muscular; this is the more noticeable since the bones are rather slender and he carries not an ounce of superfluous fat.

The muscular hypertrophy is rather irregular in its distribution, being most marked in the muscles of forearm neck & thigh. In standing there is marked lordosis, and a slight degree of flexion of the elbow & knee due no doubt to the ~~greater~~ enlargement of the flexor & hamstring groups being more marked than in the case of their opponents. In the upper limb the deltoid, biceps & triceps are very well developed; but in the lower limb the calf muscles although standing out sharply, when contracted are not nearly as much hypertrophied as the vasti, hamstrings & glutei. The intrinsic muscles of the thumb & little finger are unusually well developed, and at the wrist the muscular fibres run far down on the tendons on the ulnar side though the joint is slender.

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Measurements

Neck = 16¾ inch, 17 in. with muscles contracted
Wrist = 6½ in.
Forearm = 11 relaxed 12 in contracted.
Biceps = 11¼ inch.
Calf = 13 "
Thigh = 19½ "
Chest = 35½ "

Examining the trunk muscles: the pectorals, when the arms are firmly crossed, stand out in bold relief; and on making the patient lunge forward with extended arm a beautiful demonstration of the Serratus with digitations is obtained.
The lumbo-sacral muscles are very massive, and the intermuscular spinal groove exceptionally deep. Glutei very powerful
The neck is thick & powerful with a remarkable development of the sterno-mastoids, especially on the right side.
The recti abdominis can be made to shew the ~~inte~~ segments very sharply by causing the patient to contract them.
The muscles at rest feel rather flabby than otherwise; but on sudden voluntary contraction. Eg. flexion of forearm those concerned in the movement are felt for a few seconds to be of almost iron hardness, which rapidly dies down leaving the muscles soft & rather fatigued

Strength of the muscles

Tested by passive resistence, the dynamometer and various other strength trials

N.B. It appears that the patient has never gone in for athletics to any extent and that the muscles "keep about the same" whether used much or little. It is however the ones most used that shew the hypertrophy. Best.

there is found great capability for short efforts of strength, but fatigue in the muscles exercised rapidly supervenes. The strength is really proportionate to the bulk of the muscles, but it is more quickly exhausted than natural. (i.e. it only lasts as long as the first few powerful tonic contractions)

Effect of Mechanical Stimulation of the Muscles.

Measurement of Neck = 16¾ in. of Forearm flexed = 12 inch. Of Upper thigh = 19½ inch.

Peculiarity of Movement

The peculiarity of movement is very noticeable. If the patient be made to sit for a few minutes, and then told to get up & walk across the room - he rises stiffly & clumsily & takes a step forward but on attempting to follow it up he is seen to be hampered by the prolonged contraction of the calf muscles, and it is only after walking a few yards that the spasm wears off. In the same way he can sharply flex the forearm at the word of command, but cannot for a few moments extend it fully again. Every attempt however is done more easily & at last a normal range &

and rapidity of movement is attained.
It is to be noted that the whole of the
muscular system shews a degree of
rigidity on setting up after a state of rest.
Change of movement also induces it. It is not only in initiating a movement
after rest that induces the prolonged
muscular contractions; a change from one
movement to another, thus introducing
a new set of muscles: Thus the patient
waltzes perfectly in the ordinary way;
but, when directed to "reverse", although
understanding perfectly what is required
he is unable to do so at first, being
brought up short by the muscular
spasm which at once supervenes.
Again, when directed to grasp the hand
of the Examiner strongly & then unclasp,
he cannot for a few seconds relax the
grip, a phenomenon which has naturally
proved very embarrassing both to himself
& others on more than one occasion.
The fixing of the eyeballs on suddenly
turning them sideways, and the resulting
stare only passing off on stroking
the eyelids to relax the spasm can be

readily demonstrated, as can also the thickness of speech on suddenly attempting phonation after a long silence. The interossei appear to be healthy, writing not being interfered with.

The accompanying myographic tracings shew well the characteristics of the contraction in Thomsen's disease. They represent the voluntary flexors of the fingers contractions repeated at the word of command as soon as the preceding relaxation was complete. (as indicated on the tambour).

It will be seen that the 1st Contraction is slow, powerful & much prolonged (lasting from 10 to 30 secs or even longer) that it is regular in character, while the period of relaxation is much prolonged and the relaxation irregular in character. The second contraction shews the same points but in a less marked degree and the approach to the normal muscular contraction becomes closer at each attempt until the Myotonic (Ed.) character is completely lost. It should be noted that if complete relaxation be

Neuro-Muscular reactions.

not waited for, the patient being told to flex & Extend the fingers repeatedly as quickly as possible that the second ~~relax~~ Contraction begins before the first has completely relaxed. & this, to a less extent, holds good with the second & third. The "latent period" did not seem prolonged.

Effect of Mechanical Stimulation of Motor Nerves
E.g. Rolling Ulnar N. between finger & thumb
Contraction = more ready and relaxation slower than normal. = somewhat increased irritability.

Effect of mechanical Stimulation of Muscle
If the Vastus Externus be struck several sharp blows in succession with a percussion hammer, a lumpy contraction is produced which persists sometimes 25 secs before slowly dying away = increased irritability
(Here compare photo indicating this slow relaxation in the Vastus Ext.)

Effect of Faradic Stimulation of Motor Nerves. (α) Quantitively = Normal
(β) Qualitatively = Contractions persist a long time, and relax very slowly; and the stronger the current the better this is marked.
No Opening Contractions. (My C. of Erb.)

N.B. MyR = Myotonic Reaction of Erb.
MyC = Myotonic Contraction as shewn in Myographs.

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Effect of Faradic Stimulation of Muscles.

Quantitively = normal., unless the current Qualitative. =is strong enough to produce any contractions when they persist & relax slowly as before. = (My C)

The stronger the current the easier these reactions are obtained. There are only short lightning like contractions on opening the Current.

Effect of Galvanic Stimulation of Nerves

In a momentary application of the current nothing abnormal; but if the electrodes are left in contact for a short time the resulting contraction persists some seconds and relaxes with abnormal sluggishness (My C.) as before. And as before, the stronger the current the more marked this phenomena. No opening contractions are obtained.

Effect of Galvanic Stimulation of Muscles.

Quantitively increased. ACC is quite as easily obtained as KCC in thumb muscles. flexors of fingers &c. With weak Currents contraction & relaxation is almost momentary. but with strong ones the Contraction is prolonged & relaxation also (My C.) often lasting many seconds. This is more

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marked if the current be allowed to run a short time. When the shocks were given at Regular intervals wave like flickerings were at times noticed passing from the ~~f~~ Kathode to the Anode, as has been described by Erb & Ther observers.

On two occasions a second very marked Contraction wave was noticed after the Rheophore was removed.*

The main point Elicited by this Examination is, that to both a Sudden Single Stimulation & a tetanizing current, the period of Contraction & relaxation of the muscles (Especially the last) is much prolonged in Thomsen's disease. Whether the Stimulus is Voluntary or Electrical does not matter. The Contraction is regular, the relaxation irregular.

Histology of Muscles.

Dr. Swayne of Bristol kindly excised a fragment from the Vastus Externus, and an Examination of this gave the following results:- The Muscle fibres were considerably larger than normal, appearing as if two or more had been fused together. They measured from 1/150 to 1/400 inch. in ~~Size~~ diameter (N= 1/750-1/400 Quain's Anat) They were rounded & not

* When the current (galvanic) was allowed to run for some time the same phenomenon was at times noted. MB

polygonal on cross-section.

The Nuclei of Sarcolemma were more numerous than in normal muscle, but not out of proportion to the size of the fibres. The striation was not quite distinct, but no excess of interstitial tissue or fat was noted (this as observed by Erb in one case, is probably a later degenerative stage) The "Cohnheim's fields" were also more distinct than usual, due to increased in the interfibrillar protoplasm.

These characteristics of the muscles, electrical and histological, agree closely with the descriptions of Erb, Hale White, Hamilton, Déjerine & Psottas & other observers, so that the case is quite a typical one in these respects.

Other Neuro. Muscular Symptoms. &c.

The Muscles are free from pain either on handling or during the tonic spasm but a cramp-like feeling is experienced during relaxation. There is no hyperæsthesia or anæsthesia. No delayed sensation - Muscular sense appears normal - The knee jerks are Equal & ready. Superficial reflexes natural. Special Senses shew nothing abnormal, Except that, along with a moderate degree of *Myopic astigmatism, there is the occasional Spasm

*N.B. The eyeballs are very prominent & shew the "fixing" well

of the extrinsic muscles of the eye + orbicularis previously referred to. Colour sense & vision fields are normal. The Profuse sweating on slight exertion or under emotion has been already noted. The viscera seem fairly healthy. Pulse is large & soft. Occasionally palpitation occurs under trifling causes. Patient has suffered for years from a degree of neurasthenia and his hyper-sensitive nervous system reveals itself in many little ways.

Profuse Sweating

N.B. There is a certain degree of fine tremor of hands - this has always been noted.

Theories of the Disease.

Having now described my case I shall attempt to shortly review the theories of the disease put forward by various Observers. It must be confessed at the start that the pathological material is of the scantiest. The only autopsy I know of is that of Déjerine & Problas already referred to; besides which in about 20 or 30 cases fragments of muscle have been Examined histologically.

The four chief theories are these:-

(1) That it is a psychosis, interchangeable with other neuroses e.g. hysteria & Epilepsy. the muscular hypertrophy being secondary. (Thomsen, Hamilton, Déjerine &c)

This was Thomsen's own view, and appears to be much the same as that of Hamilton[1] of New York who regards it as of hysterical nature dependent partly on insufficient nervous inhibition and partly upon an unstable emotional state interfering with volition. He explains the myotonia as due to a repeated reflex spasm made possible by a diminution of the inhibitive influence of the "upper level" over the multipolar cells of the cord.

This view undoubtedly derives strong support from the fact that T.D is often (even generally) associated with other neuroses in the same family or in the patient. That emotion and other depressants of the nervous system affect the symptoms so markedly: and (that cases have arisen from shock to N. Sys.) may here state that after most careful investigation of my own case I have come to the conclusion that this is the most tenable position.

(3) That it is a primary myopathy (Leyden)

This observer considers it an altered functional condition of the muscular tissue congenital in origin. The points put forward for this view are (1) the frequent alteration in the electrical excitability. (2) Ringer & Saundbury's experiments

(1) Hamilton - 'Med. Record' New York 1886. Vol. I. p. 85.

of frogs. Heg found that certain salts such as Sodium phosphate are capable of producing a similar muscular spasm in the frog & that the spasm persists both after the division of the nerve & after the intramuscular nerve endings have been paralysed by Curare. This however does not prove that the muscular derangement is the only factor. That it is not so is suggested by the facts that it has arisen in adults from shock[1] to N. S injury[1] (Bernhardt & Seligmüller & Engel)

Other modifications of this muscular theory are those of Jacoby, who assumes that there are too many sarcous elements in each fibre, of which there is no proof; and of Bernhardt[2], who thinks it is due to a faulty evolution of muscular fibres. He quotes Soltmann's[3] observations on new-born animals, in which it was shewn that the ascending limb of the myographic curve is prolonged & the descent of the curve very slow. An analogy was also drawn with the condition in fish and rabbits, in which there are two kinds of muscular fibre - red & white with

(1) Schönfeld - 'Med. Record' London 1883. P. 406.

(1) "A case of Myotonia arising from a fall" - Allen J. Smith. 'Intern. Med. mag.' Phil. Vol III. 1895. 129 et seq.

(2) Bernhardt - 'Virch. Archiv' Bd LXXV S. 576.

(3) Soltmann - Jahrbuch f. Kinderheilkunde 1877.

differences in their contraction curves.
Westphal, Ballet & Marie & others consider
it a congenital Error in muscular tonicity,
judging from (1) the Extension of motor troubles
to almost all the voluntary muscles.
(2) their transitory character — (3) the phenomena
produced by Electrical stimulation.
Septilli Kesteven (1) thinks it more probably due
to an inhibition of the Sympathetic or Vaso-motor
influence on the vessels of the muscles
arresting the removal of the chemical products
of muscular action & thus interfering with
nutrition & muscular movements: that the
muscles in fact are in a state approaching
Rigor mortis wherein myosin is developed.
This ingenious view has not (of course) any
proof adduced in support of it; and, along
with all the others which attempt to fix
the primary fault on the muscles, seems
to take far too little cognizance of the associated
Mental condition, & the now proven fact that
other Neuroses can almost invariably be traced
in the family. Again in my own & in several
other recorded cases it has been Expressly noted

(2) cf. Virchow u Hirsch - 'Jahresbericht' 1884
(1) Kesteven - 'London Med. Record' 1883 - p 189.

that the muscles shew no increased firmness, but are on the contrary rather flabbier than normal between ~~contra~~ when at rest.

(γ) That it is due to a developmental defect in the pyramidal tracts of the cord (Seeligmüller)[1] + Sharkey. Sepelli (heightened muscle tonus the result of the increased excitability of the innervation centres in the cord) + other observers: but the Experimental fact + the observed facts of T.D. both shew rigidity apart from obvious nerve centre lesions. This point (cf. negative results of Examination of Nervous System by Déjérine + Bottas) argues strongly against the Cord theory. There is also no Exaggerated Knee jerk or other "Spastic" signs.

(δ) That it due to an abnormal functional state of both Nervous System + muscles. Congenital in both. Overaction of the muscles being accompanied by overaction of Ganglion cells of the cord + perhaps also of the pyramidal cells of the cortex. (Gowers and others)

Cook + Sweeten[2] offer the following Explanation:- After a period of rest during which the motor cells have acquired their maximum force

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(1) Sharkey.- Gulstonian Lect. 'Lancet. Vol. I. 1896. p. 175.
(2) Cook + Sweeten - 'Brit. Med. Journal' Vol I. '90 p. 73.

the voluntary impulse causes an excessive discharge of motor energy which of itself sets up a second and succeeding discharges in the same cells, & thus the muscle is again stimulated to contraction before it has had time to relax from the first contraction, & that after the cells have been thus relieved from their excess of energy, they become more stable & are then able to work smoothly, or a similar theory might be applied to the muscles. To this it has been objected that the rigidity in T.D is not a tetanus (which results when there is a continued flow of stimuli to the muscle) for in this case it is with the cessation of the stimulus that the chief Effect comes out.

After a careful survey of the question and a close examination of my own case I have fixed on the psychical theory as the most tenable one, and regard it as a Disturbance of the psycho-motor areas of the brain, ~~by virtue of which Voluntary impulses~~ Congenital & often hereditary - closely allied to hysteria & other neuroses. that here is a defect either in the origination or the

transmission (or in both) of voluntary impulses, and that the muscular hypertrophy is a secondary condition. I think that some such explanation as that of Cook & Sweeten would explain the muscular condition as well as the psychical state. The pathological material is so very scanty however that it is hardly worth while adding to the already numerous explanations theories of the disease.

Diagnosis. This is to be made from (a) the history (β) the myotonic reactions of the muscles (Especially the Cathodal closing contractions) in typical cases. (γ) The fact that Spasm clears off. It may be confounded with Spasmodic Tabes dorsalis. which is also congenital & characterized by rigidity of lower limbs; but the rigidity does not pass off on movement. & this disease shews Exaggerated knee jerk & the symptoms of Spinal Epilepsy. The upper limbs, tongue &c are unaffected. T.D. may also be mistaken for Pseudo-hypertrophic paralysis by a careless observer which disease shews (1) true paralysis (2) Very large, but very weak muscles, side by

side with others quite atrophied

From Lateral Sclerosis it is distinguished by the facts that there is no loss of power, & that the rigidity wears off with Exercise.

Erb. it should be remembered, called attention to the fact that there are cases not typical in which the myotonic reactions are associated with other nervous conditions & symptoms. These appear to be very rare however.

Importance of making diagnosis early.

It is important for the sufferer & his friends that the condition should be recognized early. It disqualifies in Germany for the army & navy. In England the Indian & other public services (in which riding is an essential accomplishment) would also be out of the question. In fact any pursuit in which the free use of the muscular system is an essential is barred to the unfortunate subject of Thomsen's disease. It does, indeed, "cast a shadow over life" in Gowers' words.

Treatment

The life of open air muscular exercise* recommended by Thomsen is the best plan. Exercise short of fatigue and

* A warm & dry climate is a huge advantage. These patients suffer much more than ordinary folks from cold & damp. – the spasms being more marked.

carefully regulated. In both boys & girls (it is very rare in girls) games such as tennis, cricket, rowing &c may be freely Encouraged; but riding, swimming in the open & skating may subject the patient to the risk of serious injury, from his inability to save himself from the effects of sudden attacks of the spasm. brought on during the Exercise. Thus on one occasion my patient had a narrow escape from drowning on plunging into a lake from an anchored boat — the cold water & effort of the dive causing a prolonged rigid Contraction of the muscular system. altogether preventing the use of the limbs.

Those in care of such cases (schoolmasters &) should have the nature of the trouble Explained to them, that the effects of over-pressure physical & mental (which are most injurious) may be avoided carefully. The Nervous System should be kept braced up to the highest point of health & occasional courses of hot baths & massage may help the muscles a little at times of extra stiffness. No hopes of cure should be held out, but

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the patient may be told that no change for the worse is to be expected after puberty, and every effort used to combat the shyness, slack of confidence which is often Experienced by the Sufferer. For the Excessive Sweating a most annoying Symptom of the Complaint I have found nothing of any avail.

Vide Summary last page.

Note by the writer.

The above is an account of my own case, which was diagnosed by myself in 1887 (before any case had been shewn in England). The diagnosis has since then been confirmed by Dr. R. Caton of Liverpool & others. After considerable hesitation, I now submit it in the shape of a Thesis, thinking it might be of some interest from the fact that this is (so far as I am aware) the first time, since Thomsen's classical paper, that a physician has been in the unfortunate position of being able to describe this complaint as occurring in his own person. For obvious reasons I have refrained from recording the case in the medical journals.

Arthur Bird

Definition of Thomsen's disease

In the light of this & other typical cases described we may now attempt a comprehensive definition of this affection :—

It is then, an hereditary (rarely acquired) functional disease of those parts of the neuro-muscular system which are concerned in the origination and translation of voluntary motor impulses, of indefinite duration & somewhat progressive tendency, with its incidence chiefly in family groups & on the male sex: characterized by (1) a condition of general cerebral instability (2) a sudden spasmodic muscular seizure attending the performance of a voluntary act (3) ~~An apparent increase~~ (3) Its subsidence after the repetition of the act (3) An apparent increase in the bulk of various groups of muscles, which are chiefly & most frequently the subjects of the cramp-like contractions. (4) A marked increase in Electro-muscular irritability.

(5) A normal or only slightly Excited state of tendinous reflex activity (Sometimes sub-normal MB)

(6) An occasional involvment of the certain of the cranial nerves concerned in voluntary movement.

(7) An absence of any gross change (Demonstrable) in the N. S., but true overdevelopment of the muscular fibres.

MB.