

DYSTONIA MUSCULORUM DEFORMANS— OPPENHEIM'S NEW DISEASE OF CHILDREN AND YOUNG ADULTS.*

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IN 1911, Oppenheim¹ described a new and peculiar disease occurring in young people of Russian or Galician Jewish parentage, which he termed dysbasia lordotica progressiva or dystonia musculorum deformans. On the basis of four cases he attempted to separate the disorder from the tics, athetoses and the various muscular spasmodic states occurring in hysteria. At first the racial predilection of the condition suggested a comparison with amaurotic family idiocy, in fact so much so, that in 1912 Bergman² was enabled to make the rather positive statement that "the disorder has up till now been observed only in patients of Jewish extraction." More recent studies have shown that while the disease is pre-eminent among young Hebrews, yet it is not absolutely limited to them, as a few cases have been reported in Christian subjects. Since Oppenheim's original contribution, cases have been published by Belling,³ Fraenkel,⁴ Ziehen,⁵ Schwalbe,⁶ Flautau,⁷ Haenisch,⁸ Abrahamson,⁹ Bernstein,¹⁰ and Bergman.¹¹ Nearly all of these cases have occurred in young Jewish children.

The disorder presents many striking features which have hitherto been undescribed. The diagnosis is rather difficult to make, and can be determined with certainty only after long observation. In fact, all who have observed the disease emphasize the diagnostic difficulties. The clinical picture of the condition seems to lie between hysterical scoliosis or dysbasia, idiopathic bilateral athetosis and infantile pseudobulbar palsy. The Merzbacher-Pelizaeus syndrome (aplasia axialis extra-corticalis congenita) may also occasion some confusion, but the strongly organic nature of this disorder and the hereditary predisposition ought to clear up the diagnosis.†

The real nature of the disease is doubtful. Two of my cases, for instance, resembled hysteria, while the third, which I had the opportunity to study over a long period of time, possessed certain features which suggested an organic basis for the disorder. Oppenheim is inclined to believe that the condition is organic rather than functional, principally, it would appear, on the basis of his previous work on infantile bulbar paralysis with athetosis, and

on the practically negative results of psychotherapy. However, since hypnosis was the only psychotherapeutic procedure utilized, the failure of hypnotic suggestion does not militate against the functional nature of the condition, since, up to the present, no case has been submitted to a complete psycho-analysis. Déjerine¹² classifies the condition under the athetoses. I am inclined to believe that we may be dealing with two separate groups of disease pictures, fundamentally different, but identical clinically, and which further observation and study may perhaps separate.

The fact that the disease appears in the children of unaffected parents shows, that like amaurotic family idiocy, a recessive factor is at work. Although no specially strong familial trait can be demonstrated, yet one of my cases had a brother who seemed to possess an abortive form of the disease, but which has not progressed for the last two years. In Schwalbe's description of a similar disturbance, three of the cases were sisters, which would suggest a familial tendency to the disorder. The disease usually occurs in children between the ages of eight and fourteen, although since Oppenheim's original paper, the condition has been reported in young adults, one of my cases being twenty-eight years of age.

The clinical picture is strange and peculiar. The onset is slow, involving first either the upper or lower extremities, and appearing either as tremor of the arms, spasticity of the legs or some disturbance in gait. The gait is stiff and hypertonic; there may be a cross-legged progression or it may resemble that of a muscular dystrophy. One of my patients waddled like a duck. The gait may be similar also to the dysbasia lordotica of hysteria, and it was this circumstance which led to the condition being interpreted by Ziehen as a tonic torsion neurosis or as a pure hysterical disturbance. Twisting of the spine, tilting of the pelvis, lordosis and lardo-scoliosis are marked. Characteristic of the condition is the fact that the muscular disturbances tend to diminish or cease when the patient is at rest, but reappear very rapidly as soon as there is any attempt at voluntary movement.

There is a very rapid muscular fatigue in walking. Some of the muscles, particularly those of the neck, show a marked hypotonicity, causing the head to fall forward on the chest. While the patient is at rest, peculiar movements of the limbs may take place, not particularly athetoid or choreiform, but partaking of the nature of both. Such movements are inconstant, however. A combination of rigidity and tremor may also be found. There is no paralysis, atrophy, electrical changes, sensory disturbances, involvement of the sphincters, cranial nerve paralysis, speech abnormalities or mental disturbance. The condition of knee jerks is inconstant; they may be diminished, normal, or

* Read before the Forty-third Meeting of the New England Pediatric Society, March 31, 1916.

† For a minute study of this disease see Merzbacher's original monograph published in 1910. Also in English "An Unusual Type of Hereditary Disease of the Nervous System," by F. E. Matten and D. Wilkinson, *Brain*, 1914.

even exaggerated. Facial grimacing has also been observed, and in one of my cases, attempts at voluntary movements of the upper extremities brought out a condition suggestive of a cerebellar disorder.

That the condition had been observed for some time before it became elevated to the dignity of a nosologic entity, is shown by the following passage from the fifth edition of Oppenheim's text book, which was published several years before the appearance of his description of the disease. The reference is to a disturbance of the gait in hysteria, and it will be observed that this description bears a striking resemblance to his later account of dystonia musculorum deformans. "In one of my cases, which followed influenza, the waddling gait was exactly like that of dystrophy; the patient would not keep the trunk erect in sitting, but had to support it with her hands. The paralysis was localized in the lumbar-pelvic thigh muscles, but there was no atrophy or pseudohypertrophy, and the influence of suggestion made its hysterical nature evident."¹³ Thus it will be observed that, even before the symptoms had been studied in a group of cases, the condition was considered a pure neurosis, pre-eminently of an hysterical nature.

For the present, the true nature of the disease must remain in the dark, partly because of insufficient clinical material and partly because neither pathological reports nor complete psycho-analyses are available. Evidently the disorder is not purely hysterical, since suggestion has been ineffective in ameliorating the symptoms, and none of the cases seemed to follow an emotional shock. One of my cases, however, showed certain characteristics of a traumatic neurosis, while the other occurred in a highly neurotic Hebrew patient. If the condition is hysterical, future investigations will probably clear up its psychogenesis; if organic, the lesion is probably in the putamen and the caudate nucleus, the same as Oppenheim and Vogt¹⁴ found in certain cases of infantile bulbar paralysis with athetosis. The precipitating causes of the disorder are somewhat clearer, and suggest a certain analogy with hysteria. In Flatau's two cases, for instance, the condition followed an injury to the leg; in one of Schwalbe's cases, it appeared after an emotional shock; while in two of my cases emotional factors in the etiology could be incriminated to a certain extent.

The following three cases came under personal observation:*

CASE 1. A., age 9, a Russian Jewess, was the eldest child of parents free from any neurotic tendency. She was always well with the exception of measles. A week following a fall down two steps, in which she did not complain of any injury at the time, she began to walk "crooked" but without any

* Case 1 is from the Neurological Department of the Boston City Hospital (Service of Dr. Thomas). Cases 2 and 3 were seen privately.

complaint of pain. At first the right foot was affected, but under the application of a plaster-of-Paris bandage, in a few days she walked without any further evidence of a limp. At the time the diagnosis of hysteria was made. Shortly after the application of the plaster bandage was made, she began to walk lame again. A neurological examination at that time revealed the following symptoms:

Grasps good, strength and movements of arms normal. Movements of leg joints free. She walks with a marked contraction of the muscles of the right calf, but when lying down the contraction disappears. The knee jerks are lively and equal and there is a patellar twitch on both sides. Plantar reflexes normal. No Babinski. Testing of the knee jerks or sometimes manipulating of the leg at the ankle joint, produces a contraction of the calf muscles. Pupils normal. There is no disturbance of sensation to touch or pain. Tenderness in both iliac fossae, particularly on the left. No tenderness along spine.

The condition remained about the same for nearly six weeks. A tenotomy of the hamstring muscles was ineffective in altering the muscular spasm. An examination at this time revealed a tendency to hyperextend the right leg at the knee and to rotate the right leg outward in walking. Voluntary movements at the ankle, knee and hip joints can be performed, but rotation of the leg brings on a muscular spasm. Shortly afterwards the condition became decidedly worse. For several weeks, she was unable to sit down, since when, placed in a normal sitting posture, the body assumed the position of an opisthotonic arc de cerce, with jerky movements of the buttocks. In walking, she repeatedly jerked the body backwards and walked in a waddling duck-like manner, with the knee and hip semi-flexed. The right knee joint was particularly the seat of a strong contraction and she was unable dorsally to extend the foot. Unfortunately, this patient disappeared from observation and could not again be traced.

CASE 2. B., male, age 29, Russian Hebrew, upholsterer. The family history is negative except that the father had been a sufferer from chronic bronchitis for a number of years. He had worked steadily at his trade and there had never been any serious physical disease. Married six years, three healthy children, the youngest of whom was ten months old. There was a history of coitus interruptus for years on account of poverty. For four years there had been a complaint of a sensation of numbness of the left side of the body, without, however, in any way interfering with his work.

Several days previous to being seen, without any definite precipitating cause which could be ascertained, he suddenly began to suffer from peculiar attacks of trembling and a disturbance of gait. These attacks were not constant, but took place perhaps every fifteen minutes or so. In the normal interval, any sudden noises, even music, sudden changes of position, or the swallowing of food would produce an attack. The voice became weak and hoarse, he appeared easily frightened, with a facial appearance of extreme anxiety, and he was constantly looking to his family for sympathy, which, needless to state, made the attacks more severe and frequent. There were no gastric disturbances and with the exception of over-anxiety, the mental condition was normal.

Examination. Trembling of the entire body with

a facial appearance of distress, weakness and anxiety. He whimpers a great deal, speaks in a weak voice, which at times becomes a hoarse whisper, resembling a partial hysterical aphonia. Observation shows peculiar attacks of muscular spasm influencing the position of the body and the gait. These attacks strongly resemble an hysterical dysbasia lordotica. The attacks begin with a look of distress and anxiety, then a trembling of the entire body, and in an attempt to walk he throws the head backwards, walks very slowly with a marked lordosis of the spine, particularly in the lumbar region, bending of the knees and balancing movements of the arms. Under these conditions the gait becomes waddling, like that of a duck, and at times it is strongly suggestive of a muscular dystrophy. In the attack after an initial tremor of the trunk and limbs, the arms and legs become rigid and spastic, the spasticity being of the tonic variety. The muscles of the spine also became spastic and it is this spasticity that produces the lordosis. The contracted and spastic muscles become rapidly fatigued, the spasm then slowly relaxes and the attack ceases after five to ten minutes. Even between the attacks, however, there remains a certain amount of a lordotica dysbasia with rigidity of the spinal muscles and a tendency to bring the edges of the scapulae close together. It seems as if the muscular fatigue were responsible for the cessation of the individual attacks and as the fatigue disappears, a new attack takes place. The attacks can also be produced experimentally by sudden noises or by sudden changes of position of the body and tend at times to cease when they are ignored by indifferent questions, even at the height of the attack and before the onset of the muscular fatigue. No difficulty in swallowing and no globus. No tachycardia in the attacks. The condition is most liable to take place on attempts at walking, since when he lies down the muscular spasm relaxes.

The pupillary reactions and the ocular movements were normal and there is no nystagmus. Visual field not contracted. Tongue central and tremulous. No tremor of the outstretched hands. Knee jerks and Achilles jerks much exaggerated. Double spurious ankle clonus. No sensory disturbances to pain or touch. Sphincters normal. Heart negative. No paralysis.

Under the administration of bromides, rest and suggestion, with purposeful neglect of the symptoms, the dysbasia disappeared within a week's time and has not since returned after more than six months.

CASE 3. C., age 7, a Russian Jewess.

Family History. All the ancestors were born in Russia and it is interesting to note that the family had the same social background of poverty, mental anxiety and persecution, as has been observed in my studies on amaurotic idiocy. Grandparents healthy. On the maternal side an uncle has tuberculosis, an aunt is neurotic, another uncle suffers from chronic lead poisoning, while a paternal aunt has been hemiplegic since childhood. When the mother was six years old she had to work hard, and up to eleven years of age had scarcely enough to eat. At eleven, she sewed as a dressmaker's apprentice and during her entire life she has been under a severe physical and mental strain under conditions of extreme poverty. The father has also been under a severe strain all his life, yet both father and mother show no evidence of any neurosis and are physically strong and healthy. There are five children in the

family. The mother has had no miscarriages. The two children who are younger than the patient, likewise a sister who is one year older, are strong and healthy. The eldest brother, aet. 13, shows a tendency to nystagmus, and has slightly unequal pupils and knee jerks. He has no muscular spasm or tremor. The patient is the third child. Since the descent of the disorder is through similarly unaffected stock, the condition must be due to a recessive factor according to the scheme of Mendelian inheritance.

Personal History. The child walked and talked at a normal age and was well and strong up to shortly after she was five years old. Following a tonsillectomy, tremor of the right hand and a stiffness of the left foot was noticed. Then she began to stagger in walking and running, locomotion became cross-legged, the head bent to the left and began to shake and a marked curvature of the spine developed. No incontinence of urine or faeces. No headache, vomiting, abnormal salivation or blindness. No complaint of pain and no hyperacusis. No evidence of mental deterioration.

Examination. The child is well nourished and of normal size for her age. No signs of any mental deterioration. As the child sits up, which she can do only with assistance, the head falls to the left and shows a fine tremor, there is a marked kyphosis of the dorsal spine and also a lumbar lordosis. While the lower spinal muscles appear to be spastic, yet the hypotonia of the neck and cervical muscles is so marked that the head falls forward on the chest. The thigh and hamstring muscles are in a condition of hypertonia, likewise the anterior tibial group, thus keeping the big toe of each foot in a condition of hyperextension. She can walk only with assistance and then the gait is of the cross-legged projection type. The adductors of the thighs are in a state of spasm. Right thigh rotated outwards and left thigh rotated inward, thus tipping the pelvis to a certain extent. There is no muscular or fibrillary twitching.

As the child is at rest, but becoming greatly increased on voluntary movement, there are observed constant, restless and slow choreiform movements of the arms. In attempting to grasp objects, the choreiform movements of the arms became jerky, discontinuous, awkward, and associated with a coarse tremor and ataxia, resembling the dysmetria of cerebellar disease. The tremor is absent during repose, but the choreiform movements are constant, though less marked than on attempts at voluntary movement. All attempts at movement produce a rapid fatigue. Thus the peculiar restlessness is much reduced when the patient is quiet, and becomes markedly visible only on voluntary movement.

A further analysis of the movements of the arms revealed the following: Constant restless and slow choreiform movements of both arms. On account of the muscular spasm, she is unable fully to open the left hand, which remains semi-flexed. On attempting to grasp a tumbler, the movements of the right arm became jerky, discontinuous, excessive, and likewise very awkward, resembling the condition seen in hereditary ataxia or after destruction of the cerebellum in monkeys.

Pupils equal and react promptly to light and accommodation, both directly and consensually. No nystagmus. Fundi normal. No muscular atrophy. Speech normal. No facial paresis. Tongue central, without tremor. Knee jerks exaggerated and equal. No Babinski or Oppenheim reflexes, although on

stimulation of the right plantar surface, there is an inconstant and apparently voluntary extension of the big toe. Achilles jerks absent. No ankle clonus. No sensory disturbances to pain or touch could be demonstrated, even on the most careful testing with von Frey's hair esthesiometer. On attempts at exertion the child perspired profusely and became rapidly fatigued. No hyperacusis.

The patient was kept under observation for about a year. During this time the peculiar movements of the arms on voluntary movement became more marked and the hypertonicity of the muscles, particularly of the legs, increased, finally rendering walking impossible. With all this, there was no muscular atrophy, abnormal reflexes or mental deterioration. The neck muscles, however, remained hypotonic. Speech became somewhat bulbar and indistinct and there was a moderate degree of drooling of saliva, without, however, any atrophy of the tongue or difficulty in swallowing. None of the muscular incoördination was increased when the eyes were closed. Massage, motor re-education and the use of sedatives, were ineffective in ameliorating the condition.

In discussing this material, which, unfortunately, is small on account of the rarity of the condition, several factors stand out prominently. All the patients were of Russian Jewish origin, and their ancestors had been subjected to physical strain and persecution, the same ancestral history which we have found in our investigations of amaurotic idiocy. In two of the cases the family history was negative; in the third, certain neurotic tendencies were clearly defined. In all the cases, the disorder appeared in the descendants of unaffected stock, thus corresponding to a recessive condition, according to the Mendelian laws of heredity. Here again the comparison with amaurotic idiocy is rather striking.

The first two cases strongly resembled an hysterical dysbasia, but without any corresponding sensory disturbances; in the third case there was evidence of some organic affection of the nervous system as shown by the spasticity, the gait, the bulbar symptoms and the variations in the muscular tonicity, yet lacking the usual pathological reflexes of the spastic group of diseases. This latter case possessed certain features which were strongly suggestive of a Friedreich's ataxia, but after prolonged observation, it seemed warranted to place it in the group of the dystonias. In one case the condition followed a slight trauma, in another it appeared after a tonsillectomy, while in the third, it was engrafted on a highly neurotic and probably latent hysterical individual.

The prominent features in all the cases were the peculiar gait, the changes in the muscular tonicity, and the rapid onset of the disease without any pathological reflexes or changes in sensation. All reported cases of the disease show that it begins in an extremity; in our first case in the right foot, in the second case in the leg, in the third case in the right hand and left foot. The onset in this latter

patient exactly resembled the beginning of the disease in Flautau's two cases, and on which he lays considerable diagnostic emphasis.

The disorder presents certain difficulties of diagnosis because of its obscure relationships to hysteria, on one hand, and to organic diseases of the nervous system on the other. For this reason it is difficult to place the condition in any definite nosological entity at present. Possibly we may be dealing, as suggested at the beginning of this paper, with a varying pathological condition, but with an identical clinical symptomatology.

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Society Report.

NEW ENGLAND PEDIATRIC SOCIETY.

A MEETING OF THE NEW ENGLAND PEDIATRIC SOCIETY WAS HELD AT THE BOSTON MEDICAL LIBRARY ON MAR. 31, 1916, THE PRESIDENT, DR. A. C. EASTMAN OF SPRINGFIELD, IN THE CHAIR.

The following papers were read:

- I. DYSTONIA MUSCULORUM DEFORMANS, OPPENHEIM'S NEW DISEASE OF CHILDREN AND YOUNG ADULTS.*

By ISADOR H. CORIAT, M.D., Boston.

- II. FURTHER EXPERIENCES WITH HOMOGENIZED OLIVE OIL MIXTURES.

By MAYNARD LADD, M.D., Boston.

- III. A STUDY OF FIRST EXAMINATION, WITH A REPORT OF FIFTY CONSECUTIVE PHYSICAL AND MENTAL EXAMINATIONS OF SO-CALLED WELL CHILDREN.

By WILLIAM R. P. EMERSON, M.D., Boston.

DISCUSSION.

DR. BARRON (Dr. Emerson's paper): I have a few statistics along the line of Dr. Emerson's investigations which I would like to present to the Society.

* See JOURNAL, page 383.