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TORSION-SPASM (DYSTONIA LENTICULARIS):

WITH CASE REPORT.

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FROM the large group of diseases and clinical syndromes which may show evidence of striatal dysfunction a well-defined group can be separated; these cases were first described in 1910 by Ziehen who gave them the title of torsion-neurosis, though the earliest is possibly that of Gowers⁷, which he called 'tetanoid chorea.' Since then the condition has been seen frequently, especially in Germany and America, and various names have been applied to it, including 'dysbasia lordotica progressiva' or 'dystonia musculorum deformans' (Oppenheim¹²), 'torti-pelvis' (Fränkel⁵), 'torsion dystonia' (Mendel¹¹), 'torsion-spasm,' 'myastasia' and 'dystonia lenticularis' (Thomalla¹³). It forms a fairly clear-cut clinical syndrome, but its exact nosological position is still a matter of some doubt.

The original cases all occurred in Jews, especially in Russian or Polish Jews, and in 1920 only five out of forty cases had occurred in Gentiles; in that year Blandy³ recorded the first case in England, the sixth Gentile. In his monograph of 1919, Mendel¹¹ considered the following to be the characteristics of the condition. The onset, in childhood or adolescence, is insidious, the patients usually being males. Involuntary movements form the most striking manifestation and they affect the trunk and proximal portions of the limbs; these movements are the result of an irregular increase and decrease of muscle tone occurring in a haphazard manner but producing twisting movements of the trunk with ultimate lordosis and scoliosis. There is no true paralysis or evidence of pyramidal disease, and the tendon reflexes are normal or diminished. The face conspicuously escapes and though the disease runs a chronic progressive course there is no involvement of speech, nor is

there any intellectual impairment, muscle atrophy, sphincter disturbance or sensory change. The involuntary movements are always more marked when the patient is standing or walking and they disappear during sleep; the gait becomes grotesque, resembling that of quadrupeds, so that Oppenheim applied the term 'dromedary' or 'monkey' gait; some patients have found backward progression easier than forward.

The following personal case appears to be unique in some of its manifestations and will therefore be reported in some detail.

CASE REPORT.

Carl C., aged 17, is a Jew who was admitted to the Leeds General Infirmary on November 6, 1930, under the care of Dr. Maxwell Telling; he complained of progressive weakness of the right arm and leg with involuntary movements and difficulty in speaking.

His father died of 'melancholia' 13 years ago; his mother, sister and two brothers are alive and well; the mother has had no miscarriages. There is no history of familial disorder of any kind. The patient was quite normal during the first five years of life; he suffered from measles when 18 months old, and from chickenpox and pertussis at the age of two.

The present condition dates from an acute illness which began on March 18, 1918, when he was five years old, and during which, after three days of malaise, tremor of the right hand suddenly developed; it lasted for about an hour and for this period he was unable to use his hand. This was followed by difficulty in standing, for which he was admitted to the Leeds General Infirmary on the third day; he was found to have some dragging of the right leg and there was slight drawling dysarthria; all the tendon jerks were present, there was no clonus and the plantar responses were flexor in type. No definite conclusions were reached and he was sent home. During the following year there was steady improvement in his condition but there was slight residual weakness of the right foot. At the end of the second year (1920) the weakness began to increase and with it weakness of the right arm returned and progressed; soon after this the arm and leg were noticed to be getting thinner, and on January 5, 1921, he was again brought to the Infirmary where he was examined by Dr. C. W. Vining; there was tremor and unsteadiness of the right leg and well-marked dysarthria; the right calf was wasted; the tendon-jerks were sluggish and the plantar responses were flexor; the tongue was protruded to the right and there was slight lack of facial balance; there was tremor of the right hand and right foot was dragged. A diagnosis of infantile hemiplegia was made and it was thought to be possibly postencephalitic in origin.

In March 1922 his right arm was not being used as much as before and the spinal curvature was first noticed; about this time he was found to have weakness of dorsiflexion of the right foot, and in June 1922 the right ankle was put into plaster following a tenotomy of the tendo Achillis and he was supplied with a spinal jacket for his increasing scoliosis. In March 1924 the wasting of the right arm and leg had increased considerably and the arm was assuming the hemiplegic posture (fig. 1) though there was no alteration of muscle-tone and the plantar response was flexor. According to the mother it was about this time (age 11) that the involuntary movements began, and there was a period of nausea and headache which have not recurred. Since 1924 the disability has increased and he has been unable to walk without

assistance since 1925; he was able to sit at table up to 1926, about which time the severe involuntary spasms, which are still present, first began. Throughout the illness he has been able to dress himself and he is still able to move about the house and go upstairs without assistance, although he cannot move more than two or three yards without holding on to the furniture.

FIG. 1.

During the whole period he has grown steadily though the spinal deformity has increased. There has been a progressive increase in the dysarthria and for some years he has made a minimum of spontaneous remarks. His education has of necessity been neglected but he always has been very fond of reading and has an excellent memory; he has never been able to write properly. At no time have there been any sphincter disturbance, loss of consciousness, hallucinations, visual disorder, dysphagia, deafness or involvement of the left arm or leg. Except for the period of nausea he has had no gastro-intestinal symptoms, and he has enjoyed good general health.

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Condition on Examination. Higher cerebral functions.—Making allowances for the severe dysarthria and for his limited education, the youth is intelligent and co-operative; memory is good and he reads well; he was originally right-handed and has developed the faculty of writing with his left hand (see specimen). There is no evidence of aphasia. Speech is very defective and all

answers tend to be monosyllabic; he speaks in a monotone and with considerable hesitation, though after a variable latent period he will speak rather quickly; all syllables are slurred and the voice tends to fade away at the end of a sentence.

Cranial Nerves.—There is no involvement of the olfactory nerves. Visual acuity is 6/36 on the right and 6/18 on the left; the visual fields are full and the discs normal; slit-lamp examination was impossible owing to the severity of the involuntary movements, but the iris appears normal to the naked eye. The pupils are slightly unequal, the left being the larger, and they react normally to light and on accommodation; there is no diplopia or strabismus

FIG. 2.—Position of choice.

and the eye movements are full except for a little weakness of the left eye in convergence; there is a little irregular nystagmus on looking to the left. The corneal reflexes are brisk and equal and there is no sensory loss over the face. There is marked weakness of the lower half of the right side of the face during voluntary movements; this is less marked when he is smiling; there is a mild degree of bilateral ptosis more marked on the right (fig. 2); blinking is normal and there is ill-sustained blepharoclonus; the facial expression changes very little. Hearing is good on the two sides. The palatal reflex is present and the uvula moves in the mid-line, but the range of movement is small. There is no impairment of taste over the tongue. The sternomastoids show no involuntary movements and are equally strong, but there is marked weakness

of the right trapezius. There are frequent spasmodic contractions of the left trapezius but none of the right. The tongue is protruded to the right and there is some flattening of the right half, but there is no fibrillation or notching of the edges.

Motor system.—After allowing for the deformity the patient is definitely under-sized and is not more than 57" in height. He prefers to lie in the prone position with the right leg under the left so that he is half on his right side (figs. 2 and 3). He lies with both legs slightly flexed at the hips and the knees. When in this position intermittent contractions of the left erector spinae muscles are seen at an average rate of about 50 per minute; there is contraction of the muscles of the abdominal wall which can be felt to harden and relax synchronously with the back muscles. In this position there are no involuntary movements of the limbs. The right arm is semi-flexed at the elbow and flexed to a right angle at the wrist; it shows considerable wasting, and at corresponding points it is 1" to 1½" smaller in circumference than the left arm. There is a full range of passive movements at the shoulder and

FIG. 3.

active movements though impaired in range are quite strong. Active movement at the elbow is very much impaired, extension being stronger than flexion; it is impossible to extend the arm fully owing to contracture of the biceps, which is painful on stretching. The forearm is held in a position mid-way between pronation and supination, and it is impossible to supinate passively beyond this position; there is no voluntary pronation or supination. He can extend the wrist but all movements at this joint are weak. Grip is very weak and he is quite unable to relax it although there is only slight weakness of the extensors of the fingers; he says that this phenomenon has been present for some years and he has had to unfasten his grasp passively with the left hand. The thumb can only be approximated to the first and second fingers. There is increase in muscle tone in all the muscles of the right arm, the rigidity being of the cog-wheel type and being most marked at the elbow and wrist; it is more marked in extension than in flexion. There appears to be no incoordination though tests are limited by the

small range of voluntary movements. There are no involuntary movements of the right upper limb except at those moments when he is having a generalised spasm. The abdominal muscles are strong and equal on the two sides. There is a full range of movement at the right hip, but flexion and extension at the knees are both weak and there is considerable adductor spasm; there is marked footdrop on the right, with no voluntary movement at the ankle or toe joints, and marked contracture of the tendo Achillis

FIG. 4.

(fig. 3). Muscle tone is increased in the right leg but to a less extent than in the arm; the cog-wheel element is only present at the knee-joint. The right leg is wasted to the same degree as the arm: the left leg is in every way normal. There is an extreme degree of lumbar lordosis and dorsal kyphoscoliosis, the convexity being to the right (figs. 3 and 4). Involuntary muscular spasms involving the whole body occur frequently. These spasms depend largely on the position of the body both for their frequency and their

degree; they are maximal when he is standing up, being then continuous. During a spasm the trunk is always bent towards the left (figs. 4 and 8) and it may be also extended (fig. 7) or, more commonly, flexed (fig. 5); the flexion is often so marked that his head nearly touches the ground. The movements are slow and snake-like until the attack passes off when the trunk is straightened rather suddenly. During one of these spasms he is quite unable to speak, breathing is stertorous and he makes expiratory grunting noises; the flexion of the right arm at the elbow increases and as the upper arm is also adducted the hand is pressed into the face; to prevent this he holds the right hand with the left during a spasm (figs. 4, 5 and 6). The flexion of the

FIG. 5.

right leg becomes more marked. When the flexion of the trunk is maximal he remains motionless for five to ten seconds. The eyes are not affected.

These attacks are quite irregular except when he is standing, each spasm being then followed by another immediately. They do not occur when he is lying on his right side or when he is in the prone position, nor do they occur when he is lying back in a chair, but they are practically continuous if he is sitting on the edge of a table; he has learned to assume a posture in which movements are minimal. The spasms disappear during sleep. The involuntary movements are not affected by passive alteration of the position of the head in space.

Gait.—He is able to move a few yards without assistance, but this is a very grotesque and difficult performance as he has to rely almost entirely on his left foot to support his weight; in spite of this he never falls backwards and rarely falls forwards, but if he is entirely unsupported for more than a few seconds he has to sink to the ground. He says that on some days he is able to stand for several minutes without having an involuntary spasm, but this has not been witnessed while he has been in hospital.

There have been no sensory phenomena of any kind, and there is no objective sensory change. There is a marked tendency to sweating which varies from day to day; the skin of the face is very greasy.

The tendon-jerks are all present, brisk and equal on the two sides; there is no clonus at the knee- or ankle-joints. The abdominal reflexes are all present but are slightly diminished on the right side. The cremasteric reflex is not obtained on the left and is sluggish on the right. The plantar response on the

FIG. 6.

left side is flexor in type but that on the right side is equivocal. Sexual development is normal. There is no evidence of disease of the gastro-intestinal tract, the cardiovascular or respiratory systems. The blood-pressure is 116/75 mm. There is an oblique oval patch of brown pigmentation of the skin on the back of the left thigh, about 3" × 1" (fig. 3).

Special investigations.—The electrical reactions of all the muscles are normal. The blood Wassermann reaction is negative. It was impossible to examine the cerebrospinal fluid. The laevulose tolerance test shows no abnormality of liver function, and the Van den Bergh reaction is negative. The daily output of urine is rather small, but the urine contains no abnormal constituents.

Progress.—The patient was seen again on November 2, 1931, when he complained of soon becoming tired and he thought that the involuntary movements were becoming more frequent; he still has them partially under

voluntary control. He has developed no new symptoms except some pain in the back. The right facial weakness has increased and there is now practically no movement of the palate. Grip in the right hand is still very weak but is much stronger if the wrist is passively dorsiflexed. The inability to relax the grip in the right hand has increased and he is now unable to approximate

FIG. 7.

FIG. 8.

the thumb and first finger; the arm-jerks are brisker on the right side. The abdominal reflexes have now disappeared and the right ankle-jerk is brisker than the left. The right plantar response now appears to be extensor in type; the left is still flexor but the movement is poor. The involuntary spasms have not increased in degree.

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COMMENTARY.

This case appears to be a typical example of dystonia lenticularis with certain unusual and additional features. It seems to be the first case in which there has been an involvement of the pyramidal system, and this is more remarkable as the pyramidal involvement appears to have been the first manifestation of the disease. Although there are no involuntary movements in the hemiplegic limbs, there is undoubtedly an increase in muscle tone in them at those moments when the trunk is involved in a major spasm. The hemiplegic limbs are wasted, though wasting without electrical changes has been noted in cases of torsion spasm by Hunt⁸ and Keschner¹⁰. Affection of speech has also been seen by Keschner¹⁰, Wimmer²⁰ and others, and inequality of the pupils with nystagmoid jerks occurred in one of Taylor's cases¹⁴. Spasmodic breathing occurred during generalised spasms in Blandy's case³ and facial involvement was noted by Bregman. Contractures of the feet occurred in Goodhart and Kraus cases⁶ and in Hunt's case, resulting in the 'ventral' foot; the 'semilunar' foot occurs in the more marked cases (Hunt⁸). The 'reverse' or 'paradoxical phenomenon' described by Hunt⁸, in which an attempt further to flex the partially flexed foot is followed by extension and attempted extension results in increased flexion, is not shown by this patient, but his inability to release the grip of the right hand (which appears to have occurred in Abrahamson's case¹) may be a related phenomenon, though Hunt has not seen a true example of myotonia in torsion spasm. Oppenheim¹³ was the first to describe alternating hypo- and hypertonia, but in this case, as in the cases described by Bregman, Flatau and Sterling and Hunt⁸ no hypotonia was demonstrable. Excessive sweating was seen in Keschner's case¹⁰ and in one of Taylor's cases¹⁴ the cremasteric reflexes were absent.

ETIOLOGY OF DYSTONIA LENTICULARIS.

The original cases of dystonia lenticularis were thought to be hysterical, and the first diagnosis of Wimmer's case²⁰ was hysteria, but since Oppenheim's review in 1911 it has been grouped among the striatal group of disorders. The Vogts¹⁶ recognise four types of striatal disease, viz., status marmoratus (a developmental defect), status fibrosus, total necrosis (Wilson's disease) and acute lesions, both inflammatory and vascular; the corpus striatum may also be involved in widespread lesions of the cerebrum such as tuberosc sclerosis, pseudosclerosis (Westphal-Strümpell) and dementia paralytica. Unfortunately, as Wilson¹⁸ has pointed out, the two cases of dystonia lenticularis, viz., those of Wimmer²⁰ and Thomalla¹⁵, which have come to autopsy, have shown widespread lesions, and indeed Mendel doubted whether Thomalla's case really belonged to this nosological group. Mendel considers torsion-spasm to be a disease sui generis with

specific etiology; on the other hand Taylor considers it to be a syndrome probably of congenital origin and points out that transitional forms are outnumbering those cases which conform to the assumed definite type. Certainly both Wimmer's and Thomalla's cases had cirrhosis of the liver, which brings them into line with Wilson's disease, and Thomalla himself preferred to include torsion-spasm, Wilson's disease, double athetosis and pseudosclerosis under the single heading of dystonia lenticularis, as do Jelliffe and White⁹; Westphal¹⁷ agrees that it is to be assigned to the corpus striatum together with paralysis agitans, pseudosclerosis and Wilson's disease.

Again, typical cases have followed encephalitis lethargica; the first of these was described by Mourgue¹² in 1922; others have been reported by Goodhart and Kraus⁶ and cases are referred to by Economo⁴ in his monograph on encephalitis lethargica. This feature is interesting in regard to the present case which began rather suddenly in March 1918, the time at which encephalitis lethargica was making its appearance, and it is possible that a postencephalitic basis might be responsible for the atypical features of this case.

But the striking frequency of Jewish cases and the occasional occurrence of two affected members of the same family (Taylor¹⁴) suggest very strongly that torsion-spasm is an inherited condition. In view of the small number of cases on record it is impossible to elucidate any simple type of inheritance though it is quite possible that the condition is inherited as a Mendelian recessive, the incidence of affected persons being perhaps due to inter-marriage; the marriage of cousins and other near blood-relations is more strictly forbidden by the Jewish religion than by others, but as few people are able to trace their ancestry backwards through more than two or three generations, it is very easy for such marriage to take place. Bielschowsky² classifies torsion-spasm with the heredodegenerations of the central nervous system, including it, together with Wilson's disease, in the group of abiotrophies with local necrosis of the parenchyma.

The involuntary movements of torsion spasm are tic-like, resembling neither chorea nor athetosis; they are not rhythmical, but show a certain monotony, and they have been described as tonico-clonic, but the clonic element is not obvious in the present case; nevertheless tremor, athetosis and chorea may be associated with torsion-spasm (Hunt). Certain elements of the involuntary postures may resemble those of decerebrate rigidity (Blandy³), in which condition respiratory disturbances are frequent (Wilson¹⁹). Whether the lesion is localised to the lenticular nuclei, or whether it is more diffuse, it certainly gives rise to gradual loss of a tone-controlling mechanism resulting in motor activity of a non-cortical type. In this case it is interesting that minor involuntary movements of the trunk muscles can be seen at all times, including the abdominal muscles on the hemiplegic side, although the Vogts¹⁶ have seen no example of striatal disease in spite of pyramidal disease.

In conclusion, this case appears to be typical as regards the Jewish incidence, progression and the type of involuntary movements, but there is an associated progressive hemiplegia; the patient shows no evidence of cirrhosis of the liver or corneal pigmentation and the possibility of the condition being postencephalitic cannot be entirely excluded.

I wish to express my gratitude to Dr. Kinnier Wilson for his valuable assistance and to Dr. Maxwell Telling for permission to publish this case.

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