A Peculiar Form of Family Nervous Disease Resembling Multiple Sclerosis Occurring in Children

BY

ISADOR H. CORIAT, M.D.

Second Assistant Physician for Diseases of the Nervous System, Boston City Hospital; Neurologist to the Mt. Sinai Hospital

Reprinted from the Boston Medical and Surgical Journal,
Vol. clx, No. 16, pp. 506–508, April 22, 1909

BOSTON

D. C. HEATH & COMPANY

120 Boylston Street

1909
A PECULIAR FORM OF FAMILY NERVOUS DISEASE RESEMBLING MULTIPLE SCLEROSIS OCCURRING IN CHILDREN.*

BY ISADOR H. CORIAT, M.D.,
Second Assistant Physician for Diseases of the Nervous System, Boston City Hospital; Neurologist to the Mt. Sinai Hospital.

Hereditary or family diseases in children are always of interest, particularly when they depart from well-recognized clinical pictures. Atypical disease pictures are frequently seen in children, in whom conditions are often observed which seem to represent transitional forms between definite types of disease. Without committing myself to any definite conclusions, because of the limited time in which the cases have been under observation, I wish to describe a peculiar type of nervous disease, observed so far only in children and with the earliest symptoms, dating from before the period of puberty. The cases observed occurred exclusively in children of Russian Hebrew parentage, but whether or not this is a mere coincidence can only be determined by further observation of identical cases. The disorder seemed to represent a transitional form of nervous disease, related on the one hand to Friedrich's ataxia and on the other to multiple sclerosis, although the symptoms of the latter condition largely predominated. There was a strong tendency for the disease to run in families, either in its complete form or as a group of isolated and discrete symptoms. The fact that in two of our cases no other distinctly allied disorder could be

found in any other member of the family does not militate against the family nature of the affection, because the same tendency to isolated cases has been found in amaurotic family idiocy, Friedrich's disease and in some forms of muscular dystrophy.

The occurrence of the cases in the families of Russian Hebrews would suggest a comparison with amaurotic family idiocy. The symptom-complex in all four cases was characterized briefly by a tremor of the hands, increased on voluntary movements, ataxia and marked changes in the reflexes. The knee jerks were never normal, but were either diminished, absent or exaggerated. Ankle clonus was obtained once and atypical Babinski reflex twice. In two cases the ankle jerks were slight, in a third they were absent. One case showed a transitory spasticity, in another there was an atrophy of the small intrinsic hand muscles with a reaction of degeneration pointing to a bilateral affection of the anterior horn cells in the eighth cervical and first dorsal segments. There was no optic atrophy in any of the patients, and in only one case was a true nystagmus demonstrable. None of the cases showed any staggering or a tendency towards a progressive dementia or loss in weight. In two of the patients there was a moderate degree of feeble-mindedness, but even in these cases we cannot interpret the tremor and ataxia as a form of motor incoordination which is sometimes found in feeble-minded individuals, because the degree of mental weakness was too slight to permit of any such interpretation.

The onset in all the cases was a gradual one, a fact that is perfectly compatible with the assumption that the anatomical basis of the disorder was probably of the nature of the slow formation of disseminated sclerotic foci (disappearance of the myelin sheaths with persistence of the axones) in various parts of the central nervous system. In none of the cases was there a history of an immediately preceding infectious illness, so that we may safely assume that the symptoms were not of the nature of the acute ataxia which is occasionally seen in children after infectious diseases. In this latter condition the lesion is of the nature of either a diffuse or focal encephalitis, or there may be a primary toxic cell degeneration, usually of the cerebellar cortex.

A search through the literature failed to disclose any cases that were exactly identical with ours. The diseases grouped by Freud under the name of family or hereditary types of cerebral diplegia showed clinically, however, many symptoms that resembled our cases. Whether or not these cases occurred in Russian Hebrew families is not mentioned in any of the communications. As the condition is a comparatively rare one, it might not be unprofitable to briefly review the essential features of this group.

Pelizaeus has reported a rare form of family disease, attacking only the male members of several generations, and transmitted only through the mother of the family. All the cases showed the following identical symptoms: Normal pupillary reaction and normal optic disks, but with a pale fundus, disturbances of co-ordination of the arms, spastic paralysis of the upper and lower extremities without atrophy, bilateral horizontal nystagmus and increased reflexes. The electrical reactions were normal and there was no disturbance of sensation. The nystagmus, muscular spasticity and inadequacy appeared early in life. All the cases showed mental weakness.
Two of the patients were living at the time the article was written (1885). Three had died, at the ages of twenty-five, thirty-two and twenty-three years respectively. Although Pelizaeus regarded these cases as hereditary types of multiple sclerosis, associated with a spastic paraplegia, yet they certainly were not pure types of the disease. Heredity is rare in real multiple sclerosis, although the disorder may develop in the children of neurotic parents.

Under the name of the family type of cerebral diplegia, Freud described two brothers, who developed in early life horizontal nystagmus, convergent strabismus, optic atrophy, monotonous slow speech, ataxia, tremor of the arms, spastic rigidity and weakness of the legs, increased reflexes and Romberg's symptom. In the elder brother the symptoms dated from birth; in the younger they appeared at the end of the second year. A fifth brother was paralyzed and idiotic from birth and died at the age of ten months from rachitis. Freud interpreted these cases as representing transitional forms between Friedreich's ataxia and multiple sclerosis.

Some closely allied cases occurring in four sisters are reported by Higier. Birth and development were normal, but at ages varying in each patient from the seventh to the twelfth year there developed weakness and stiffness in the legs, disturbances of gait, contractures and deformities of the feet (hyperextension and pes equinovarus), trophic disturbances in the legs, exaggerated reflexes, bulbar symptoms, optic atrophy, monotonous slow speech, strabismus, nystagmus, ataxia and intention tremor. One of the cases showed an atrophy of the upper extremity, including the small muscles of the hand.

Gardener had recently described similar cases occurring in a Russian Hebrew family. He considers the condition as a form of Friedreich's ataxia, but they all bear a remarkably close resemblance to our cases and, therefore, must be interpreted as belonging to the type of multiple sclerosis. In the mother, who was thirty-eight years of age, the symptoms had appeared five years previously and consisted of intention tremor, ataxia, exaggerated reflexes and nystagmus. In a seventeen-year-old daughter the condition dated from three years of age. She presented the same symptoms as her mother, but in addition showed slow speech, spastic paraplegia, Babinski reflex and talipes equinovarus. Three other children had absent knee jerks. None of the cases showed any mental weakness.

There are a number of hereditary and family nervous diseases of children, in which we are dealing, not with isolated disease forms, but with a distinct overlapping of types. The closest resemblance is found in those conditions of idiocy or dementia associated with a degeneration of the peripheral optic neurones, such as amaurotic family idiocy (Tay-Sachs) with the typical macular changes, the juvenile type of amaurotic family idiocy (Spiekmeyer) with a retinal atrophy of the type of a retinitis pigmentosa and the amaurotic-paralytic form of idiocy (Schaffer) which is distinct from the juvenile form. Tumors of the interpeduncular space may resemble amaurotic idiocy (Williams), whereas tumors of the basal ganglia in children may simulate the juvenile form of this disease, as in a case which I recently had the opportunity of observing. To this group can also be added the hereditary cases described by Freud, Higier, Gardener and Pelizaeus, all of which resembled multiple sclerosis, although the disease described by Pelizaeus was free from optic
atrophy or changes in the macula. Dementia, idiocy or mental weakness are not an absolute accompaniment of any of these conditions, but a mental change seems to occur, particularly in those cases associated with optic atrophy or disease of the macula. To these hereditary types resembling multiple sclerosis, although in many ways distinct from these, partly because of the racial tendency and the characteristic symptoms, belong those cases which I wish to describe. There are allied types of a family disorder resembling multiple sclerosis, in the same manner that there exist disorders allied to amaurotic family idiocy. Raymond in an interesting communication has shown that the clinical types which constitute family diseases are not identical in all cases, because each family tends to work out its own disease, and family diseases are often met with as isolated cases.

All my patients were observed at the neurological clinic of the Mt. Sinai Hospital. The first two cases were two sisters, age ten and thirteen years respectively, when they first came under observation. Both parents are Russian Hebrews. The mother and three other sisters are healthy. The father has suffered from pain and weakness in the legs for years, but has no tremor or awkwardness of the hands. A female cousin fourteen years of age, on the father's side, is said to have been clumsy with the hands for several years, the handwriting is tremulous, she is unable to execute the finer movements of the fingers, such as tying her shoestrings, but there is no staggering or other disorder of gait.

The first patient, M. K., had always been in good health, except for an attack of diphtheria when quite a small child. She is fairly bright mentally and is in the fourth grade at school. When nine years of age a certain clumsiness of the hands was noted, and there was inability to play children's games requiring any amount of muscular co-ordination, such as jackstones. She would spill liquids on carrying a glass or cup to the mouth and later in sewing or attempting fancy work she was decidedly clumsy in the manipulation of the needle. Although she complained of some vertigo, she has never been any loss of consciousness or staggering. These symptoms gradually increased in intensity and about three months before she first came under observation she began to complain of the pain in the left leg, but an x-ray examination revealed nothing abnormal. An examination of the patient showed the following: Moderate ataxia of arms and legs, increased when the eyes were closed and more marked in the left hand. Some swaying in Romberg's position. No spinal curvature, nystagmus or disorder of speech. No staggering. Sensation to touch and pain was normal in all parts of the body. The pupils were equal and reacted normally to light and distance. The optic disks were normal. There was a fine vibratory tremor of the outstretched hands, increased on voluntary movements. The knee jerks were diminished and obtained only on reinforcement. The Achilles jerks were absent and there was no ankle clonus. A moderate Babinski and Oppenheim reflex was obtained on the right, but not on the left. There was no muscular atrophy. Although the patient was given Frænkel exercises, yet the tremor and ataxia of the hands remained the same and the knee jerks progressively diminished, although they have never been entirely absent. With the exception of a transitory spastic condition of the left leg, the condition has remained the same.

The other sister, A. K., is also bright and intelligent. At the age of twelve she developed a jerky tremor of the right hand, but could always sew well and carry liquids to the mouth without spilling them. Up to the time of her present trouble she was a healthy girl. The examination showed fine vibratory tremor of the outstretched hands without ataxia, but the tremor was
increased on active movements. It was more marked in the left hand.

The knee jerks and Achilles jerks were slight. There was no ankle clonus, Babinski or Oppenheim. Sensation was normal, likewise the gait and station. There was no nystagmus and the pupils reacted normally to light and accommodation. Spasticity and muscular atrophy were absent. The grasps were steady and strong. The tongue was protruded in the median line, but was unsteady. There has been some improvement in the condition under Frankeil exercises. Neither of the patients showed any defect of vision or hysterical stigmata.

The third patient, J. G., was a boy eighteen years of age. Both parents are Russian Hebrews. The mother is nervous, afraid of everything, has had a trembling of speech for several years, but the hands are said to be steady. Three years ago the father had a tremor of the hands which persisted for a year and said to be due to the excessive use of tobacco. He denies syphilis. At present the father's hands are free from tremor, there is no nystagmus and the knee jerks are present and active. A sister is said to be nervous like the mother. Three other sisters and a brother are healthy. The patient was born at full term and the delivery was instrumental. He has never had any severe illness, except a slight convulsion when two years of age, said to be due to teething. When thirteen years of age a tremor of the right hand was noticed and he became awkward and clumsy with the movements of this hand. Six months later the left hand became affected. Because of this tremor of the hands he was compelled to leave school, and although in the sixth grade at the time he was never considered very bright. This tremor of the hands grew progressively worse, the handwriting became decidedly shaky, he spilled liquids on attempting to drink, and such actions as lacing the shoes or buttoning the clothes were performed in an awkward and blundering manner. There has been no headache, vertigo or further convulsive or fainting attacks. Mentally, he is rather dull and seclusive.

An examination of the patient showed a rather dull, apathetic and vacant expression. Cranial vault high and pointed, head narrow in the frontal region. Circumference of head, 57.5 cm. There was a marked jerky tremor of both hands, more marked on the left and greatly increased on voluntary movements. The tongue was central, without atrophy or tremor. The pupils were equal and reacted promptly to light and distance. There was no real nystagmus, but on extreme inward and outward rotation of the eyeball a little unsteadiness developed. The gait was normal, there was no staggering and no swaying in Romberg's position. There was a marked ataxia of the upper extremities, particularly increased when the eyes were closed and interfering with all movements requiring a fine co-ordination of the fingers, such as buttoning the clothes or lacing the shoes. There was moderate ataxia of the legs. The knee and Achilles jerks, elbow and wrist reflexes were absent. There was no Oppenheim or Babinski reflex. The plantars were normal but sluggish. There were no sensory disturbances. Speech normal. There was no muscular spasticity. The handwriting showed a decided tremor of the letters. The movements of the left hand were decidedly more ataxic and fumbling than the right. The optic disks were normal, and there was no atrophy or pallor of the temporal side. There was a moderate atrophy of the thenar and hypothenar groups of muscles and of the interossei, with a slowed reaction to faradism and galvanism, while the anodal closing contracture was greater than the cathodal closing contracture (reaction of degeneration). The shoulder girdle muscles were free from atrophy. Under the administration of small doses of potassium iodide and the use of Frankeil exercises, there resulted only a slight improvement in the tremor and ataxia within a period of five months. The physical signs remained unchanged.

The fourth case, M. K., was a girl eight years of age. Both parents were Russian Hebrews. The mother died of heart disease when twenty-five years old, the father and a ten-year-old brother are living and well. The patient was born full term. Delivery was without instruments. Neither walking nor talking were delayed.
She began to attend school at five, but backward in all her studies and very slow to obtain promotion. She has never had any illness excepting measles. The patient was brought because of her backward mental development, no other neurological symptoms having been noted. She was moderately feeble-minded. The head was symmetrical in shape, its circumference being 51.5 cm. The facial expression was rather stupid. The ears and palate were well formed. There was a rapid, vibratory tremor of the outstretched hands, increased on voluntary movements and more particularly marked on the left. Ataxia in the finer movements of the upper extremities was present, such as in touching the tip of the finger to the nose or approximating the individual fingers of each hand. The ataxia was increased when the eyes were closed. There was some asynergia of the legs, but no ataxia. Sensory disturbances were absent. The grasps were strong, no irregularity, no signs of chorea. The knee jerks were equally exaggerated, there was a double ankle clonus and a tendency to a double Babinski (flexion of the big toe, then extension). The ankle jerks and the abdominal reflexes were normal. The pupils were equal and reacted promptly to light and accommodation. Moderate horizontal nystagmus, particularly on looking toward the right. Vision was normal, likewise speech, no scanning, slowness or bulbar disturbance. No atrophy of intrinsic hand muscles. No tremor of the handwriting. Station and gait were normal; no staggering, no spasticity. The child was of normal size for her age. The toes of both feet are hyper extended, particularly the big toe on the right.

BIBLIOGRAPHY.

1 Pelizaeus, Fr.: Ueber eine Eigenthümliche Form spastischer Lähmung mit Cerebralscheinungen auf hereditä rer Grundlage (Multiple Sclerose), Arch. für Psychiat., Bd. xvi, 1885.
5 Raymond, F.: The relationship of the so-called family diseases to a premature physiological senescence localized to certain organic systems. Lancet, June 27, 1908.