AN ACCOUNT
OF
TWO FAMILIES, SEVERAL MEMBERS
OF WHICH ARE ATAXIC.

BY
J. A. ORMEROD, M.A., M.D. Oxon., F.R.C.P.,
ASSISTANT PHYSICIAN TO THE NATIONAL HOSPITAL FOR THE PARALYSED AND
EPILEPTIC, AND TO THE CITY OF LONDON HOSPITAL FOR DISEASES
OF THE CHEST, VICTORIA PARK; ASSISTANT MEDICAL
TUTOR TO ST. BARTHOLOMEW'S HOSPITAL.

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SOME years ago the late Professor Friedreich¹ described for the first time a form of ataxia which he believed essentially to resemble the classical type of locomotor ataxy, but which differed from it in several particulars. The resemblance consists in the main symptom of incoordination in movement, beginning in the legs and spreading at a variable rate upwards; the differences are as follows: (1) Friedreich's disease attacks many members of one family, usually brothers and sisters, and females no less than males; (2) it attacks at a comparatively early period of life, sometimes quite in childhood; (3) the disorder of gait, caused by the ataxy, is uncomplicated and at first the only symptom, whereas in ordinary tabes the ataxic stage may be preceded by a large variety of symptoms,

¹ 'Virchow's Archiv,' vols. xxvi, xxvii, lxviii, lxx.

motor, sensory or trophic, and above all, by that of pain ; (4) as the disease progresses, two symptoms may appear not usually seen in tabes, viz. disorder of articulation and nystagmus. These also Friedreich regarded as due to incoordination of movement. Sundry other symptoms, such as curvature of the spine and contractures of the feet, have been added. The pathology of the disease rests as yet on a few cases only ; but it seems to be established that there is always systematic sclerosis of the posterior columns of the cord, and often disease of other parts of the cord as well.

I will not now go into the literature of the subject,¹ but will merely mention that the cases exhibited by Dr. Carpenter at the Medical Society (1871), Dr. Gowers at the Clinical Society ('Transactions,' vol. xiv), those published by Dr. Dreschfeld in the 'Manchester and Liverpool Hospital Reports' (1876), and by Mr. D'Arcy Power in the 'Reports of St. Bartholomew's Hospital' (1882), comprise, I believe, all that have been reported in England.

The cases which form the subject of this paper are taken from two different families ; three from one, and two from another. In this second family there is a third ataxic patient whom I have not seen, and a fourth who will possibly become ataxic. None of the cases are far advanced. With respect to the upward spread of the disease, speech is affected in some only ; well-marked nystagmus has not yet appeared in any.

CASE 1.—Kate Woodcock, then *æt.* 14, was brought to me at the National Hospital for the Paralysed and Epileptic, on October 1st, 1880 ; the account given by her mother was that she had had measles three years previously, and soon after it a fever which the mother called rheumatic fever ; after this she became subject to fits in the night-time, in which fits she screamed and became

¹ See 'Brain,' No. 25, April, 1884 ; and for more recent cases—(α) Schmidt's 'Jahrbuch,' December, 1884,—cases by Wälle and others. (β) 'La Rivista Clinica,' October, 1884,—cases by Musso.

insensible when she was held ; next, she became unable to walk straight. She was also said to fidget with her hands and drop things. The only symptoms I could notice were that she had a twitching of the mouth and forehead just like that of chorea ; and, as regards the gait, that after walking several times up and down the room she became rather unsteady. I took the case to be probably chorea, and till June, 1881, she continued to attend, but remained in much the same condition. She came again during the last three months of 1882, when she seemed, if anything, more unsteady. In January, 1883, she was laid up with scarlet fever, but came back to me in the following April. The unsteadiness had then increased so much that it was evidently due to something else than chorea. She swayed from side to side as she walked, and had difficulty in turning ; she could not stand at all with her eyes shut. The patellar tendon reflex was absent on both sides. Dr. Buzzard, who was present, kindly verified this for me. The contractility of the quadriceps muscles (to faradism and to percussion) and their nutrition, were normal. Till August her condition remained stationary. It may be summarised as follows :—Gait unsteady, the left foot turns in as she walks ; patellar tendon reflexes absent, plantar reflexes present, tactile sense in feet and toes normal ; muscular sense normal (*i. e.* she knows the position of her limbs when her eyes are shut) ; occasional pain in the head, but in the limbs no pain, numbness, or paræsthesia, except a slight occasional pricking in the feet ; slight awkwardness in the hands, but nothing really noticeable, speech normal, no nystagmus, pupils contracting sluggishly to light, readily for accommodation ; fundus of eyes, except for some tortuosity of the retinal vessels, normal. Still some twitching of the forehead and of the left upper limb. Extensor tendons of both great toes prominent ; the mother thought that the feet had become humped.

In August she became worse. She often went down on her knees in the streets, the ankles seeming to give ; she

had another fit, apparently a kind of fainting fit. On August 25th I saw her at her mother's cottage, looking pale and thin, and only able to get about by holding on to the banisters and furniture; she had difficulty in directing the movements of the legs, *e.g.* in crossing and uncrossing them, or in describing a circle in the air with her foot; though she still knew how they were placed. In addition there seemed to be some actual muscular weakness and the thigh muscles were rather flabby. The Achilles tendons were becoming tight, and the feet looked short and stumpy.

She went to the Convalescent Home of the Hospital at Finchley; and since then has been at the Holborn Union Infirmary, where, by the kind permission of Dr. McLearn, I have twice seen her during the present year (1884). In March, she was in bed, unable to get about alone; there was slight general stiffness in the legs. In October, though out of bed, she could not walk; she had difficulty in managing the legs even when sitting; some awkwardness of the hands; handwriting bad, she says her head shakes when she tries to write; in reading, she separates the syllables and sometimes drops one. The legs are cold, their muscles small, some rigidity at the ankles, though none at knees; the feet more deformed than before, being in a position of equino-varus, the plantar arch high, the toes, especially the great toes, tending to turn up, and the toe-balls projecting on the plantar surface. Sensibility in feet and legs still normal, except that a prick with a needle hurts her very little.

She is a poorly-developed girl; though she is now 18, menstruation has not yet appeared.

CASE 2.—John Woodcock, younger brother of the last patient, came to me first in April, 1883 (age at that time 13). The mother said that during the last two years she had noticed a "catching" in his legs, which seemed to give when he walked and to make him unsteady. This had come on gradually; she ascribed it to a fright from

an operation for removal of his tonsils. She had noticed no twitching of his face or limbs, though she said he was fidgety in his ways. Two or three months ago he had complained of pains in his limbs, apparently of an aching character. He had convulsions when a baby, measles with his sister Kate, and also the so-called rheumatic fever. When seen, he walked rather unsteadily, swaying somewhat from side to side, though not nearly so badly as his sister. With eyes shut and feet placed together he swayed about but did not fall. Patellar tendon reflexes absent (the quadriceps muscles being normal). Plantar reflexes slight, sensibility in feet normal ; speech plain, no nystagmus, action of the pupils normal.

During the time of his attendance (from April to October, 1883), he did not improve. Twice he had an attack of giddiness and fell off a chair ; from time to time slight irregular movements of the head were noticed ; he became unable to stand with his eyes shut.

In March, 1884, I saw him with Dr. McLearn at the Infirmary ; the walk was more unsteady, he lurched from side to side and, when he turned, his feet seemed to get crossed ; Romberg's symptom was well marked. The nurse said he did not always know where his hands were. She had also noticed a peculiarity in his speech, not exactly a stammer, but a slowness in bringing out his words. He paused between words and syllables as he read aloud to me.

In October there was little change. I noticed that the toes had a tendency to turn up.

CASE 3.—Hannah Woodcock, an elder sister of the last two patients, present age 20. In 1877, she had the so-called rheumatic fever with the others. Afterwards she used to stoop as she walked. She went to service, but her health failed. She was under Dr. Sturge at the Royal Free Hospital from October 1880, to June, 1881. She had been knocked down by a drunken woman in the street, came home all starting and trembling, and was

subject afterwards to the same "startings" and "nervousness." The mother says that she was told her daughter had the St. Vitus's dance (I mention this to show the probable similarity of onset in her own and her sister's case). Curvature of the spine was discovered, and treated at the Orthopædic Hospital, with a spinal support. This made her much better able to walk.

I have seen her on various occasions from April of last year till the present time. At first there was nothing noticeably wrong with her walking, but she has since acquired an unsteadiness of gait and occasional sideward lurch. Romberg's symptom, variable at first, is now well marked; she can neither stand with her eyes shut nor walk in the dark. Patellar tendon reflexes absent. During the present year some weakness, or rather awkwardness, of the left hand has shown itself. There is usually a droop of the left eyelid, and sometimes an internal squint of the left eye; otherwise nothing abnormal about the eyes or pupils. There is considerable lateral curvature of the spine. There is a deformity of the feet, not unlike that in her sister's case, but no well-marked equino-varus. An aching and tenderness of the feet, which she sometimes has, may be due to this deformity; over and above it I have found no sensory abnormality, nor any true paralysis of movement, nor loss of electro-contractility in the muscles. Menstruation has been regular and natural for the last two or three years.

There are four other members of this family (not including a child that died of scarlet fever); one an elder brother, age between twenty and thirty, three younger children, ages from nine to six. All these are in good health,¹ but I may note that in some of them I have been unable to obtain any patellar tendon reflex, and in others it seems to be uncertain. In the father and mother it is normal. The nervous inheritance seems to come from the mother's side. She had fits till the age of twelve, another at seventeen, and another when pregnant with

¹ Two have since shown symptoms of disease. See note on p. 157.

the first of these patients (Kate). She has no brothers and but one sister, living at Hailsham. This sister is insane, but has nevertheless a large family. This family seemed to me quite healthy, but I had difficulty in obtaining tendon reflexes in the two youngest members.

As to the maternal grandfather of my patients, there seems good reason for thinking he was ataxic. He was taken ill at the age of fifty, had severe pains in his legs, for which he had to take opium; walked with unsteadiness as if he were drunk; finally took to crutches, and died at the age of seventy-five of bronchitis, having had two or three fits.

In the next family there are three cases of ataxy, and one girl who will probably become ataxic.

CASE 4.—Ruth Harriet Harmer, æt. 20. I have seen her at her mother's cottage on two or three occasions. Her condition is as follows:—She sits in a humped-up position, but there is, I am told, no spinal curvature; her head sometimes sways a little. When she stands there are constant balancing movements of the body, and she becomes more unsteady when she shuts her eyes. She can only walk by supporting herself against the wall and furniture, and seems as if she would tumble to one side. The patellar tendon reflexes are absent. There is, I think, some actual muscular weakness of the lower limbs, but they are well nourished and the electro-contractility of the muscles is normal. When she tries to describe a circle with her foot she makes a very irregular figure. The cutaneous sensibility of the feet, in all its modes, and the muscular sense are normal. The skin of the feet and legs is cold and mottled. The plantar arch is high and the foot stumpy looking. Her speech sometimes appears to be slow and drawling; her mother says it becomes thick if she reads for long. There is a certain tremulousness of the eyeballs as she follows an object, but no definite nystagmus. The pupils act normally to light. The upper limbs are but slightly affected, if at all.

The unsteadiness has been noticed ever since she was thirteen, but she has been bad for two or three years, since an attack of "low fever." (Her mother and another sister had this same fever; the symptoms were sorethroat, bad cold, and feverishness, with much debility afterwards.) In October, 1882, she became subject to cramp in the limbs if they were long in one position. A year later sharp shooting pains in the legs began, and tearing pains in the loins. These improved during the recent hot summer. Her menses began at seventeen, and have been regular and natural since.

CASE 5.—Alice Rose Harmer, æt. 16, sister of the last patient. As in her sister's case it was noticed that from the age of thirteen she was apt to fall and tumble about, but her walking has been worse the last two or three years. She now walks unsteadily, and occasionally reels from side to side; she is slightly unsteady when she stands with her eyes shut. Patellar tendon reflexes absent; muscular power unimpaired; farado-contractility of muscles normal. Sensory functions normal in every way. No pain. There is the same chilliness and mottling of the legs as in her sister's case, and the same shape of foot. She has had during the present year a small but rather deep ulcer on each middle finger. No marked ataxy of upper limbs. Speech not noticeably affected. No nystagmus. She is pallid, and subject to severe headaches, which often end with vomiting. Menses regular since the age of fourteen or fifteen; twice during the present year they have been profuse and accompanied with epistaxis.

There is an elder brother (æt. 21) in New Zealand; since he went there he has become hump-backed and unable to walk about; he, like his sisters, used to fall about when he tried to run from the age of thirteen onwards.

A third sister, æt. 11 or 12, has been brought to me

because the mother thinks she is beginning to walk like the other two girls. I cannot corroborate this myself, but the patellar tendon reflexes are absent, or nearly so.

There are five others, all said to be healthy. Two have died of convulsions and there have been two stillborn. The father appears to be not strictly sober ; but neither in the parents nor grandparents can I make out any definite history of nervous disease.

The chief interest in this class of cases attaches to the ætiology. There is a family predisposition, not merely in the sense of a neurotic diathesis manifesting itself in various forms, such as epilepsy, insanity, migraine, &c., but predisposition to organic disease of a particular part, viz. sclerosis of the posterior columns, with possibly other parts of the cord. The repetition of the disease takes place usually in the members of one generation. Rutimeyer observed no less than four collateral branches of the same stock simultaneously affected. There are several other nervous diseases which attack brothers and sisters ; for instance, pseudo-hypertrophic muscular paralysis, progressive muscular atrophy, Thomsen's disease, colour-blindness, and a disease of the yellow spot lately observed by Mr. Waren Tay. Most of these are also transmissible, hereditary, that is, in the strict sense, and in some of them a curious mode of propagation has been traced, viz. that the disease manifests itself in males but is perpetuated through their female descendants. Thus, a man is diseased, his sons and their descendants are exempt, his daughters are themselves exempt, but bear diseased sons. I would refer in this connection to two interesting family trees of colour-blindness and of hæmophilia, published by Dr. Wickham Legg in 'St. Bartholomew's Hospital Reports,' vol. xvii. Dr. Gee, in vol. xiii of the same 'Reports,' gives an account of a family where diabetes insipidus was propagated through the daughters. But no such definite mode of propagation has been shown to exist in Friedreich's disease. Indeed, with the exception of Carré's family, Rutimeyer's family, and the first of my

own, I do not think that ataxia has been shown to exist among the ancestry at all. Neither are females exempt from the disease ; indeed, Friedreich thought they were particularly prone to it, but this may have been accidental to his observations, for amongst later cases there is a good proportion of males.

It is worth noting that these ataxic families are generally numerically large. One is tempted to think that the rapid production of children may have caused imperfect development in some of them.

Lastly, since the disease does not appear at birth, what other factors besides the family predisposition assist in its development? Friedreich held that it was connected with puberty, and doubtless this is often the case, but not invariably. I believe that the influence of acute disease is another possible factor. In two at least of my three first cases the unsteadiness followed on a feverish attack. This was described to me as rheumatic fever, but as there was no redness or swelling of the joints and no heart affection was left, it can scarcely have been rheumatic fever. There was sorethroat, feverishness and aching of the limbs, and several of the family had it at the same time. Supposing it to have been diphtheria, it would be likely enough that a complaint, which under ordinary circumstances may be followed by temporary ataxia, should, in the presence of a family predisposition, prove the starting-point of permanent spinal disease. Again, in Case 4, the symptoms were said to have become much worse after a "low fever" with sorethroat, &c., from which the mother and another sister also suffered. In Case 1, too, it was after an attack of scarlet fever that the ataxia of gait first became so marked as to be unmistakable.

- [A. Woodcock family.] — Adams, drowned, æt. 40, a pilot.
 Henry Adams, *probably ataxic*. Had a brother and sisters healthy (?)
- Mrs. Woodcock (has had fits occasionally). Mrs. Gravitt, *insane*.
- Has children— Has children—
1. Son, æt. 23, healthy.
 2. Daughter, æt. 20 (Case 3), *ataxic*.
 3. Daughter (Case 1), *ataxic*.
 4. Son (Case 2), *ataxic*.
 5. Daughter, æt. 9, healthy,¹ but *tendon reflex absent*.
 6. Son, æt. 7, healthy,¹ but *tendon reflex absent or doubtful*.
 7. Daughter, æt. 6, healthy; *tendon reflex not always obtainable*.
 8. Son, died young of scarlet fever.
1. Daughter, æt. 29, married, healthy.
 2. Daughter, æt. 24, healthy.
 3. Daughter, æt. 21, married, healthy.
 4. Son, reported to be healthy.
 5. Daughter, æt. 16, healthy.
 6. Daughter, æt. 13, healthy, but *tendon reflex uncertain*.
 7. Son, æt. 10, healthy, but *tendon reflex uncertain*.
- (Besides these, three or four either dead or stillborn.)

B. Harmer family.

No nervous disease in father or mother, or in their parents, so far as was known.

Their children are—

1. Daughter, æt. 28, married, healthy.
2. Son, æt. 24, healthy so far as is known.
3. Son, æt. 21, *ataxic*.
4. Daughter, æt. 19 (Case 4), *ataxic*.
5. Daughter, æt. 17 (Case 5), *ataxic*.
6. Son, æt. 14, reported healthy.
7. Daughter, æt. 11, *tendon reflex absent*.
8. Son, æt. 9, healthy.
9. Daughter, æt. 7, healthy, but *tendon reflex feeble*.

(The first two children that were born died of convulsions at two months and two years respectively.)

¹ Since this paper was written both these children have shown symptoms similar to those with which the disease began in their elder relatives (July, 1885).

(For report of the discussion on this paper see 'Proceedings of the Royal Medical and Chirurgical Society,' New Series, vol. i, p. 346.)