Oculomotor Abnormalities in Friedreich’s Ataxia

T. H. KIRKHAM, D. GUPTON, A. KATSARKAS, L. B. KLINE AND E. ANDERMANN

SUMMARY: A clinical neuro-ophthalmological and electro-oculographic study was made on fourteen patients with Friedreich's ataxia. None had evidence of optic neuropathy. Ten patient complained of oscillopsia although all had ocular motor dysfunctions of varying degrees, which appeared to be related to the severity of the general manifestations of the disease. The defects comprised square wave jerks, jerky pursuit with inability to maintain eccentric gaze resulting in gaze paretic nystagmus and rebound nystagmus. There was failure to suppress by fixation the vestibulo-ocular reflex. The slow phase velocity of caloric nystagmus was always of reduced velocity. There was inability to augment the slow phase velocity of optokinetic nystagmus with increasing stimulus velocity. Abnormalities of the saccadic system were manifest particularly as hypermetria. These signs in combination are suggestive of disease involving the cerebellar folium and vermis or their brain stem connections. No abnormalities were found in 17 parents or siblings.

INTRODUCTION

Friedreich's ataxia, despite its original detailed description between 1861 and 1877, has resisted precise clinical or biochemical classification (Andermann, 1976). The object of the present report is to outline the clinical neuro-ophthalmic and electro-oculographic findings in 14 patients with "typical" Friedreich's ataxia as defined in The Quebec Co-operative Study of Friedreich's Ataxia (Geoffroy et al, 1976; Barbeau, 1978). Of the 14 patients, the 8 who were examined by electromyography showed normal motor but absent sensory action potentials in the digital and sural nerves. The disease is inherited as an autosomal recessive trait (Andermann et al, 1976).

MATERIAL AND METHODS

Clinical neuro-ophthalmic examinations were made on 14 patients. There were 7 males and 7 females and their ages ranged from 15 years to 42 years with a mean of 29 years. Five patients were examined more than once, with an interval of 12 to 26 months between examinations. Electro-oculography was performed on 11 patients to document and supplement the clinical findings. In addition, 17 siblings and parents were examined in order to detect minor abnormalities in heterozygotes or early manifestations of possible Friedreich's ataxia in younger siblings. Visual acuity for distance, near, and color vision was determined in all patients. Pupillary reactions were assessed. Visual field examination on the Goldmann perimeter was possible in 9 cases. Fundus examination was routinely performed after dilatation of the pupils. Ocular motility was examined clinically with respect to ocular alignment and pursuit capabili-
ties. The presence of nystagmus was sought in horizontal and vertical planes and rebound nystagmus was carefully looked for on return to the midline after eccentric gaze had been maintained for 20 seconds. Optokinetic responses to a hand-held rotating drum were studied. The ability to suppress the vestibulo-ocular reflex in the horizontal and vertical planes was noted (Zee, 1977). Saccadic accuracy and velocity was studied. Following the clinical examination, electro-oculography was performed.

The eye movements were recorded using Beckman miniature silver-silver chloride skin electrodes. The horizontal eye movements were recorded separately for each eye and vertical movements from the right eye were also recorded. Rectilinear recordings were made on a standard chart recorder after AC amplification with a time constant of 3 seconds and a band width of 30 Hz. Calibration of the recording, which also gave the performance of the eyes during horizontal saccadic movements, was performed by asking the patient to make re-fixation saccades to targets of 10° to the right and left of the midline. We measured the velocities and accuracy of 10° and 30° saccades. Pursuit movements were studied by asking the patient to follow a periodically moving target at 18°/second. Optokinetic stimulation was produced using a projector (model 3400, L.T. Instruments, Houston, Texas) and the induced nystagmus was recorded in both horizontal directions for three increasing velocity stimuli (25, 50 and 70 degrees per second). The optokinetic stimulus was projected onto a screen in front of the patient and occupied approximately 35° of the visual field. Caloric irrigation of 20 seconds duration was given using water at 30°C and then 44°C from a bath with automatic temperature control. The responses were recorded with the eyes closed. The slow phase nystagmus velocity was measured at the peak of the response. The ability to visually suppress the post-caloric nystagmus was tested about 100 seconds from the beginning of the irrigation by asking the patient to open his eyes and fixate a target. None of the patients were taking medication and all were asked not to use alcohol for 48 hours before the examination.

RESULTS
No neuro-ophthalmic abnormalities were detected in the clinical or electro-oculographic examinations of parents or siblings of the Friedreich's ataxia patients.
In the patients with Friedreich's ataxia the severity of the disease was variable. Some patients were bedridden and severely dysarthric by the age of 15 years and others were relatively normal at 35 years. No abnormalities in visual acuity, color vision, pupillary reactions, or visual fields were discovered in any patients who were able to co-operate fully in the tests. The fundi were normal in all cases. All the patients demonstrated abnormalities of ocular motility of varying degrees which correlated more with the severity of the general manifestations of the disease rather than with the age of the patient. The findings are given in Table 1.

**Fixation and Pursuit System**
Fixation instability was clinically manifest in 3 patients as square wave jerks and electro-oculography showed the occurrence of square wave jerks in 6 of the 11 patients examined. The square wave jerks occurred at irregular intervals and often in small bursts. They were as frequent on the recordings with the eyes open as well as with the eyes closed. They occurred

<table>
<thead>
<tr>
<th>Case; Sex Age (yr)</th>
<th>Square Wave Jerks</th>
<th>Pursuit System</th>
<th>Optokinetic Nystagmus</th>
<th>Gaze Paretic Nystagmus</th>
<th>Rebound Nystagmus</th>
<th>F.F.S.</th>
<th>Caloric Responses</th>
<th>Saccades</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. M 15</td>
<td>no</td>
<td>normal</td>
<td>22,32,27</td>
<td>no</td>
<td>no</td>
<td>no</td>
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<td>normal</td>
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<td>2. M 17</td>
<td>no</td>
<td>normal</td>
<td>16,21,25</td>
<td>no</td>
<td>no</td>
<td>no</td>
<td>7</td>
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<tr>
<td>3. F 18</td>
<td>no</td>
<td>+</td>
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<tr>
<td>4. F 19</td>
<td>no</td>
<td>+</td>
<td>21,20,20</td>
<td>yes</td>
<td>yes</td>
<td>yes</td>
<td>5</td>
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<tr>
<td>5. M 37</td>
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<td>20,20,18</td>
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<td>yes</td>
<td>yes</td>
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<tr>
<td>6. M 21</td>
<td>yes</td>
<td>+</td>
<td>23,27,27</td>
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<td>no</td>
<td>yes</td>
<td>4</td>
<td>dysmetria</td>
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<tr>
<td>7. M 34</td>
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<td>+</td>
<td>20,26,24</td>
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<td>no</td>
<td>no</td>
<td>10</td>
<td>dysmetria</td>
</tr>
<tr>
<td>8. F 27</td>
<td>yes</td>
<td>++</td>
<td>10,12,12</td>
<td>yes</td>
<td>yes</td>
<td>yes</td>
<td>4</td>
<td>dysmetria</td>
</tr>
<tr>
<td>9. F 25</td>
<td>yes</td>
<td>+++</td>
<td>4,3,2</td>
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<td>yes</td>
<td>yes</td>
<td>5</td>
<td>dysmetria</td>
</tr>
<tr>
<td>10. F 39</td>
<td>no</td>
<td>+++</td>
<td>4,3,4</td>
<td>yes</td>
<td>yes</td>
<td>yes</td>
<td>4</td>
<td>dysmetria</td>
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<tr>
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<td>yes</td>
<td>+++</td>
<td>8,8,4</td>
<td>yes</td>
<td>yes</td>
<td>yes</td>
<td>6</td>
<td>dysmetria</td>
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<tr>
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<tr>
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<td>+++</td>
<td>no responses</td>
<td>yes</td>
<td>yes</td>
<td>not tested</td>
<td>yes</td>
<td>not tested</td>
</tr>
</tbody>
</table>

The symbols +, ++, and +++ indicate the increasing degree of severity of defect in the pursuit system. The figures for Optokinetic Nystagmus represent the maximum nystagmus slow phase velocity (degrees/second) obtained for target velocities of 25, 50 and 70°/sec. F.F.S. = failure of fixation suppression of the vestibulo-ocular reflex. The figures given for Caloric Responses are the maximum slow phase velocities obtained for post-caloric nystagmus in degrees per second.
randomly to either side of the midline. The square wave jerks occurred for the most part in those patients whose pursuit system was relatively severely deranged (Table I).

Clinical testing of pursuit capabilities showed 2 patients who were considered to be normal (Cases 1,2). Two others (Cases 3,4) were normal when examined one year previously but were now found to have jerky pursuit. The remaining 10 patients all had jerky pursuit both horizontally and vertically. Electro-oculography confirmed the clinical impression. The patients with pursuit defects manifested marked inability to pursue targets moving at velocities of 18°/sec without the use of multiple small, "catch-up" saccades.

**Optokinetic Nystagmus**

Clinical optokinetic responses were tested in all patients using a hand-held rotating drum. Eight patients (Cases 1-8) were judged to have normal optokinetic responses which appeared to be symmetrical in both horizontal and vertical planes; 4 others showed diminished responses, and no responses were obtained in 2 cases (Cases 13,14).

The horizontal optokinetic responses were recorded by electro-oculography at three different velocities. The responses obtained were variable and appeared to depend on the degree of the severity of the systemic manifestations of the disease. Three patients (Cases 9,10,11), who were severely affected, had very low initial slow phase velocities of between 4°/sec and 8°/sec for a target velocity of 25°/sec, and when the stripe speed was increased to 50°/sec and 70°/sec they were completely unable to augment the slow phase velocity of their nystagmus. These 3 patients had marked defects of the pursuit system and were among the patients with clinically marked gaze paretic and rebound nystagmus. The remaining patients who were tested showed a marked variability in the range of optokinetic nystagmus slow phase velocities initially. None of them was able to increase significantly the slow phase velocity with increasing stimulus velocity. Some increased slightly their slow phase velocity for the stimulus at 50°/sec from that obtained at stimulus velocity of 25°/sec (Cases 1,2,3 and 7), others remained at their original velocity and still others reduced their velocity. When the target velocity was increased to 70°/sec, no patient was able to increase the slow phase velocity of the optokinetic response. Some maintained the velocity obtained at 50°/sec and the majority lowered the response velocity. The one patient (Case 1) who showed the best optokinetic responses, for example, had slow phase velocities of 22 ± 3°/sec, 32 ± 2°/sec and 27 ± 5°/sec for three stimulus velocities. All the other patients had significantly poorer responses than this for the 50°/sec and 70°/sec velocities. The ranges obtained from our normal subjects were 25 ± 4°/sec, 34 ± 6°/sec and 40 ± 9°/sec.

**Gaze Paretic and Rebound Nystagmus**

Marked horizontal gaze paretic nystagmus was clinically observed in 6 patients. They also had upbeat nystagmus on upward gaze but no nystagmus on downward gaze. The nystagmus was most pronounced in the more severely disabled patients. In 3 other patients (Cases 5, 6 and 8), a few beats of poorly sustained gaze paretic nystagmus was seen when the eyes were held in eccentric gaze at 45° from the midline. Some of the electro-oculographic tracings confirmed the clinical observations and showed the slow phase of the nystagmus to have a decreasing exponential velocity characteristic of gaze paretic nystagmus (Zee et al., 1976).

Rebound nystagmus was sought clinically in all patients by having them first maintain eccentric gaze at 45° from the midline for 20 seconds and then asking them to return the eyes to the midline. Prominent rebound nystagmus was seen in the 6 patients who showed marked gaze paretic nystagmus, but it could not be clinically detected in the other patients. One electro-oculographic tracing showed the slow phase of the rebound nystagmus to have a decreasing velocity exponential slope. A few jerks of rebound nystagmus were detected on the tracings of 2 of the other patients who had minimal gaze paretic nystagmus (Cases 5,8). The rebound nystagmus lasted from 3 to 15 seconds in different individuals.

**Vestibulo-ocular reflex**

The ability of the patient to suppress vestibular nystagmus by fixation was tested by having the patient fixate on an object rotating at the same velocity as his head. A pointer with a fixation object was held on the patient's head and the examiner moved the head from side to side and then up and down. When fixation suppression is normal no nystagmus is observed, but when it is inadequate the eyes are carried constantly off the target under the influence of the vestibulo-ocular reflex so that repeated saccades are needed to refixate the object and these appear as nystagmus (Zee, 1977). This test showed failure of fixation suppression of the vestibulo-ocular reflex in all 12 patients who had clinical impairment of the pursuit system. The test was positive in both horizontal and vertical planes.

Caloric testing was undertaken in conjunction with electro-oculography in 11 cases. The caloric responses were always bilaterally reduced and fairly symmetrical and were poor for both hot and cold irrigations and the maximum slow phase velocity obtained in our patients varied from 4°/sec to 10°/sec. The degree of fixation suppression of the vestibular nystagmus resulting from the caloric stimulation could not be accurately assessed from most of the tracings because of the very poor, irregular, low amplitude responses obtained, but in 9 cases fixation did not appear to significantly alter the nystagmus responses. In the 2 patients (Cases 1,2) who had normal pursuit, there did seem to be total suppression of the nystagmus by fixation despite the low slow phase velocities obtained with the eyes closed. In our laboratory the normal range of slow phase velocities obtained for post-caloric nystagmus is 28 ± 9°/sec.

**Saccades**

Clinical testing of saccadic eye movements was performed for 30° saccades to and away from the midline. Most saccades were judged to be accurate but a varying number were either hypometric or hypermetric.
Hypometria appeared to be more frequent away from and hypermetria more frequent towards the midline.

A number of horizontal saccades of 10° and 30° were measured for each patient towards and away from the midline to right and left by electro-oculography. All patients had saccadic velocities which fell within the normal range for saccades in our laboratory (265 ± 38°/sec for 10° saccades for both abducting and adducting saccades and 408 ± 56°/sec for abducting and 436 ± 65°/sec for adducting saccades). Four patients (Cases 1,2,3,4) made very precise normometric saccades and these were the 4 patients who were found to have good pursuit systems on their initial clinical examinations. The remaining 7 patients who were examined by electro-oculography all showed dysmetria, either hypometria or hypermetria, both towards or away from the midline, equally on rightward or leftward movements, between 13% and 28% of saccades measured for 10° saccades and 30% and 44% for 30° saccades. The hypometric saccades and hypermetric saccades were irregularly interspersed with normometric saccades. There was no significant difference in the number of hypometric or hypermetric saccades or in their direction towards or away from the midline in contrast to the clinical suspicion. The hypometric saccades were corrected by a single saccade to place the eyes on the target (dual step saccades) in over 95% of cases. The velocities of the second step of the saccades were within the range of normal. In the case of multiple step saccades it was not possible to determine accurately the velocities of the low amplitude third step from our recordings. All the hypermetric saccades were corrected by a single step.

DISCUSSION

The neuro-ophthalmic literature on Friedrich's ataxia is confused by the inexact diagnosis of most of the reported cases. Friedrich emphasized nystagmus as the only consistent ocular finding and, although nystagmus has been noted in several subsequent reports, many of the cases were far from typical (Whyte, 1898; Walsh and Hoyt, 1969; Tyrer, 1975). Periodic alternating nystagmus was said to be present in one case but the report of the clinical neurological features renders the diagnosis difficult to accept (Gorman and Brock, 1950). Horizontal gaze paresis (Kreindler et al, 1963) and pursuit difficulty and nystagmus (Spiller, 1910) in more typical cases have been described. An electro-oculographic study of 5 patients with Friedreich's ataxia (Balogh et al, 1975) showed the pursuit system to be universally deranged; 4 of the patients had gaze paretic nystagmus, 3 had hypoactive or reduced caloric responses and abnormal optokinetic responses and 3 had abnormal saccadic movements. A study of vestibular function in Friedreich's ataxia (Monday et al, 1978) showed most to have reduced caloric responses and several had abnormal pursuit and gaze paretic nystagmus.

Fixation instability, manifest as square wave jerks, was detected in 6 of our patients. Square wave jerks are conjugate saccadic movements of less than 3° amplitude by which the eyes move off the fixation point and then, after a latent period of about 200 msec, return by a saccade to the fixation point. Such square wave jerks have a frequency of about 2 Hz (Dell'Osso et al, 1975). The occurrence of square wave jerks, with eyes open during fixation, is suggestive, but not diagnostic, of cerebellar disease (Zee et al, 1976; Troost and Daroff, 1977); they cannot be considered a specific sign of Friedreich's ataxia although they have been noted previously (Balogh et al, 1975; Dale et al, 1978; Monday et al, 1978).

Twelve of our 14 patients had pursuit defects manifest as “saccadic” or “cogwheel” pursuit, so-called because the movements resulting from the attempt to follow a slowly-moving target requires the addition of numerous small “catch-up” saccades in order to stay on the target. The patients were unable to make pursuit velocity match the target velocity of 18°/sec. In our study we used sinusoidally-moving targets at a frequency of 0.72 Hz with an amplitude of 25°; normal subjects are able to perform excellent smooth pursuit with this type of predictable movement up to about 1 Hz (Young, 1971).

Smooth pursuit appears to be closely related, although not identical, to the slow phase velocity of optokinetic nystagmus. It is interesting, therefore, to compare the pursuit and optokinetic capabilities of our patients. The optokinetic system comprises two subsystems driven respectively by stimulation of central (foveal) and peripheral retina (Dichgans, 1977). The function of the peripheral optokinetic system is probably to complement the vestibulo-ocular reflex and to stabilize the eyes during movement of the external world (Robinson, 1977). During testing with a 35° area of visual field both the fovea and a portion of the peripheral retina are stimulated by the stripe motion; consequently eye movements due to stimulation of both pursuit and peripheral optokinetic systems are present, the stripes being of such a large diameter that they could not be considered as a foveal optokinetic stimulus (Dichgans, 1977).

Our electro-oculographic recordings showed significant abnormalities in optokinetic nystagmus in all patients tested when more than one stimulus velocity was used, there being inability to augment normally the slow phase velocity of the response with increasing stimulus velocity. Five of the patients with abnormal pursuit (Cases 3-7) who were unable to match a pursuit target velocity of 18°/sec were able, under optokinetic stimulation, to immediately produce a slow phase velocity beyond this and also to slightly augment the slow phase velocity with increasing stimulus velocity, although less than in normal subjects. If the patients' slow phase velocity had remained at their pursuit level it would not have been possible, with steady state testing, to distinguish their optokinetic response from their smooth pursuit response. The face that they were able to augment, albeit slightly, their slow phase velocities beyond 18°/sec indicates that the peripheral optokinetic system contributed to the response (Robinson, 1977). Four other patients (Cases 8-11), who had very poor pursuit, gave
very low abnormal optokinetic responses suggesting complete disruption of their optokinetic systems.

The slow phase velocity of optokinetic nystagmus can be increased by normal subjects as the velocity of the stimulus increases (Morissette et al., 1974) and may be fairly linear up to 80°/sec (Mizukoshi et al., 1977), the linearity depending largely on the area of the horizontal visual field being stimulated (Dichgans, 1977). With a suboptimal (35°) field of optokinetic stimulation as was used in our experiments, we found the normal patients gave responses decaying along the curve indicated by Dichgans (1977). The responses obtained from our patients were significantly below the normals except for the 25°/sec stimulus. An inability to augment the slow phase velocity of optokinetic nystagmus correlates experimentally (Takemori and Cohen, 1974) and clinically (Nemet and Ron, 1977) with lesions of the cerebellar flocculus and vermis. Abnormally low slow phase velocities of optokinetic nystagmus have been previously reported in Friedreich's ataxia, but only one stimulus velocity was tested (Balogh et al., 1975).

All of the patients who had deficient ocular pursuit were clinically unable to suppress by fixation their vestibulo-ocular reflex (Zee, 1977). Probably it is the pursuit system which overrides the vestibulo-ocular reflex during fixation of a target rotating at the same velocity as the head and, fixation may be considered as pursuit at zero velocity (Troost et al., 1976). Caloric testing in conjunction with electro-oculography in 9 patients showed apparent inability to suppress by fixation the caloric induced nystagmus. Only Cases 1 and 2 who had good pursuit systems had normal visual suppression. Such failure of fixation suppression of vestibular nystagmus clinically correlates with cerebellar and brain stem disease (Kato et al., 1977; Zee et al., 1976) and experimentally with floccular and nodular lesions (Hassul et al., 1976; Takemori and Cohen, 1974). All our patients had significantly reduced caloric responses and this has been noted previously in Friedreich's ataxia (Balogh et al., 1975; Monday et al., 1978). None of the patients had decreased hearing and the reason for the reduced caloric responses is not understood.

Gaze paretic nystagmus was seen in our patients when the eyes were held in horizontal eccentric gaze at 45° from the midline. The eyes tended to drift off the target towards the primary position with a decreasing velocity exponential wave form so that repetitive saccades were required to return the eyes to the target resulting in a gaze paretic nystagmus. The neural network responsible for maintaining the eyes in an eccentric position is a neural integrator which integrates a pulse of unit activity and provides a constant innervation to the ocular motor neurones (Robinson, 1974). This integrator, located in the brain stem, appears to need a cerebellar input to maintain its function, so that either cerebellar or brain stem disease could result in its deficient action when gaze paretic or "integrator" nystagmus occurs.

We found rebound nystagmus clinically in 6 of our patients and in 2 others by electro-oculography. Interestingly, it was not seen in a previous electro-oculographic study of Friedreich's ataxia (Monday et al., 1978) but we have noted that it is usually difficult to detect unless the eyes are held in an eccentric position for at least 20 seconds before return to the midline. The duration and slow phase velocity of rebound nystagmus has been noted to increase with the duration and degree of eccentricity of attempted lateral gaze (Zee et al., 1976). The existence of rebound nystagmus in patients who show gaze paretic nystagmus may suggest the involvement of adaptive mechanisms (plasticity) attempting to repair the faulty brain stem integrator. It is possible that after maintained eccentric gaze, a compensating bias develops which assists in the maintenance of the eccentric ocular position. When the eyes return to the midline rebound nystagmus occurs due to the persistent bias and, as the bias decays, the rebound nystagmus disappears. Rebound nystagmus has been considered a sign of cerebellar disease (Hood et al., 1973).

Saccadic dysmetria, either hypermetria or hypometria and reduced saccadic velocity (Balogh et al., 1975; Monday et al., 1978), have been previously recorded in patients with Friedreich's ataxia. We did not observe reduction in saccadic velocities either for 10° or 30° saccades in our patients. Dysmetria was frequently observed, however, particularly for the larger amplitude saccades. Dual step hypometric saccades were common but did not occur predominantly either to right or left or towards or away from the midline and occurred in less than 50% of all saccades, so that this phenomenon could well be normal (Troost et al., 1974). Multiple step hypometric saccades occurred in less than 5% of our patients and since the initial step velocities were within the normal range this may also be a normal finding (Troost et al., 1974). Hypermetric saccades have received less study: they were frequent in our patients and could well be pathological since they are rare in normal subjects. The hypermetric saccades occurred equally towards or away from the midline. Experimental lesions of the vermis (Ritchie, 1976) and surgical lesions of the human vermis (Ron and Nemet, 1977) result in dysmetric saccades.

The oculomotor defects present in our patients with Friedreich's ataxia cannot be considered as specific manifestations of Friedreich's ataxia since they have all been noted previously in patients with cerebellar and brain stem disease (Troost and Daroff, 1977; Zee et al., 1976). In a spinocerebellar degenerative disease such as Friedreich's ataxia, the cerebellum and its connections with the brain stem ocular motor control complex are likely to be involved so that widespread abnormalities of eye movements are commonly seen and were present in all our patients. The cerebellar flocculus and vermis have been implicated in the generation of pursuit movements (Miles and Fuller, 1975) and optokinetic movements (Nemet and Ron, 1977). Disease involving their connections could explain the pursuit defect, gaze paretic nystagmus, rebound nystagmus, failure of fixation suppression of the vestibulo-ocular reflex, and inability to augment the slow phase velocity of...
optokinetic responses seen in our patients. The vermis is important in the control of saccadic movements (Optican and Robinson, 1978) and disease of its connections could be responsible for the saccadic hypermetria found in our patients with Friedreich’s ataxia.

The prominence of individual ocular motility disorders in patients with Friedreich’s ataxia obviously depends on the extent of the pathological process involved and they can be expected to appear at some stage of the disease. We have not undertaken a prolonged longitudinal study of our patients, but those who have been followed for more than a year do appear to have shown a deterioration in the ocular motor manifestations of their disease.

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