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Short report

Reading difficulty as a presenting symptom in Wilson's disease

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SUMMARY A case of Wilson's disease is reported in which the major symptom before treatment was a reading disability. This was caused by ocular dysmetria, and the abnormal eye movements were recorded by electronystagmography and optokinetic drum testing.

In Wilson's disease the most common ophthalmological finding is the Kayser-Fleischer corneal ring. Nystagmus and diplopia rarely occur (Walsh and Hoyt, 1969). We report a case with abnormal eye movements which was particularly unusual as the patient was not severely disabled by other aspects of the movement disorder.

Case report

In August 1977 a 27 year old woman was admitted to hospital for investigation. She had enjoyed excellent health until March the same year but now complained of a progressive difficulty in reading print. She could see the individual words clearly but could not find the beginning of a fresh line having completed the previous one. There was no diplopia or oscillopsia. In addition, she complained of unsteadiness on walking, hand shaking, slurred speech, and a tendency to giggle inappropriately. There was no history of liver disease and no known familial neurological disease.

She was anxious with staring eyes, sweaty hands, dysarthria, and Kayser-Fleischer corneal rings. Pupillary reactions and convergence were normal. Uncorrected visual acuities were right J4 and left J2. The visual fields were full, and there was no diplopia or nystagmus. On rapid pursuit movements of the eyes in both the horizontal and vertical directions she appeared to overshoot but quickly fixated. This was interpreted as ocular dysmetria. Her gait was ataxic with a coarse postural tremor in both hands exacerbated by finger-nose testing. Heel-shin testing was normal. There was slight cogwheel rigidity in the arms. All tendon jerks were very brisk but the plantar responses were flexor.

The serum caeruloplasmin was 159 mg/l, the plasma copper was 6 μmol/l, and the urine copper 4.25 μmol/24 hours.

Electronystagmography (Watkins-Victor Cardiotrace ENG Recorder) calibrated at 30° eye movement for a 30 mm deflection and optokinetic drum testing with a 200 mm drum and 25 mm black stripes were both performed. Horizontal fixation was normal but overshoot was recorded on rapid movements to the right and left. Overshoot of the fast return phase was also seen with horizontal drum rotation to the left and right. There was a marked overshoot of the fast return phase on vertical drum testing with drum rotation both upwards and downwards (Figure, a).

She was treated with penicillamine, a maintenance dosage of 1000 mg daily being achieved. After two months speech and gait were normal, and after three months reading was again possible but very slow. After five months her reading ability was normal, and at that time her eye movements showed considerable improvement on both electronystagmography and drum testing. Slight abnormalities were still present on vertical drum testing (Figure, b).

Discussion

Ocular dysmetria is said to be present when a patient with normal visual fields and normal visual acuity shows consistently an overshoot on changes of fixation from one object to another (Cogan, 1954).

Wilson described eye movement abnormalities in two of his original four cases (Wilson, 1912). In case 1, ". . . when left to herself her eyes 'danced' slightly before her gaze came to rest on a given

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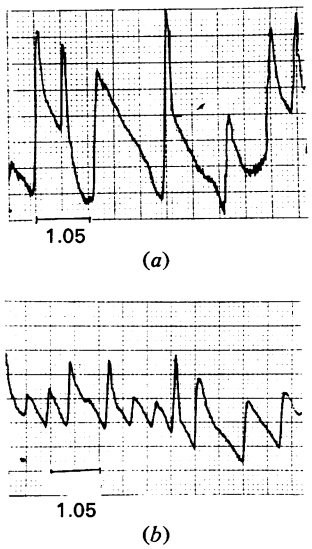

Figure *Vertical drum testing with drum rotation downwards, (a) before and (b) after treatment. (Time marker on illustrations: 10 mm=1 second).*

object." In case 3 he wrote, "when the patient moved his eyes about of his own accord and not to command it was noted that there was occasionally some unsteadiness in fixing an object; this however, was only momentary." In both cases it is possible that ocular dysmetria is being described. Twenty-nine years earlier in 1883 Westphal described a 27 year old man who in retrospect probably suffered from Wilson's disease (Westphal, 1883). He wrote that there was "slowness of all movements, including those of the eyes, and disturbance of speech." "Stiffness of ocular movement" and "punctuate" movements were mentioned but not described in detail in Wilson's later writings (Wilson, 1954) but neither of these phrases, nor the description by Westphal appear consistent with ocular dysmetria.

Several authors (Wilson, 1954; Goldberg and von Noorden, 1966; Walsh and Hoyt, 1969) have commented on the surprising dearth of literature concerning abnormal eye movements in Wilson's disease in view of the widespread involvement of other muscle groups. Goldberg and von Noorden (1966) investigated ocular motility in eight patients. Optokinetic nystagmus was normal in all and no pathological nystagmus was found. In one patient (case 8) they describe irregular and jerky pursuit movements with a "staircase" pattern. However, the patient was severely ill and weak and they suggest that inattention could have caused the anomalous eye movements. Kirkham and Kamin (1974) reported excessively slow saccadic eye movements in a patient with very severe neurological signs. In contrast, our patient was not severely disabled by other aspects of her movement disorder, and the reading disability was one of her earliest and most disabling symptoms, and one of the last symptoms to clear on treatment.

When Cogan (1954) described ocular dysmetria he used six illustrative cases. Three had invasive tumours arising from the cerebellar hemispheres, one had a malignant tumour in the roof of the fourth ventricle, one had "primary cerebellar atrophy," and the last had presumed neurosyphilis. He concluded that the sign reflected damage to either the cerebellum or the cerebellar pathways, that it was analogous to cerebellar dysmetria of the limbs and that it was of "great localising value." The nature of the lesions described suggests that none of the patients had pure cerebellar hemisphere disease.

Histologically in Wilson's disease involvement of the cerebellum is frequent although not often grossly apparent (Schulman, 1968). The dentate nucleus is the most regularly affected structure although the superior vermis and the lateral and posterior parts of the hemispheres may be involved. Within the brainstem, foci of degeneration can involve the superior cerebellar peduncles, and deep blue-staining droplets in the walls of small vessels near the floor of the fourth ventricle have been reported.

In the present patient both electronystagmography and optokinetic drum testing suggested that saccadic movements were involved in the overshoot. Widespread lesions in the frontal cortex and basal ganglia can destroy the relevant bilateral frontobulbar pathways, and produce saccadic paralysis (Kirkham and Kamin, 1974). However, our patient's abnormal eye movements, while involving the saccadic pathways, were not caused by saccadic paralysis, and we suggest that the ocular dysmetria was the result of small, discrete lesions in the upper brainstem, perhaps involving the decussation of the superior cerebellar peduncles.

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