Sir,

Ocular myasthenia presenting in an 11-month-old boy

Ocular myasthenia is uncommon in childhood. A case of an 11-month-old boy with ocular myasthenia confirmed by a positive Tensilon test and positive serology for acetylcholine receptor antibodies (AChR-ab) is described. Autoimmune ocular myasthenia has not been previously described in a child of this age.

Case report

An 11-month-old boy presented to the eye casualty department at the Bradford Royal Infirmary after his parents noticed that he had developed bilateral droopy eyelids over the previous 2 weeks. He was able to open his eyes immediately on waking but had increasing difficulty in doing so after several hours. This pattern was repeated after periods of sleep.

He was born by normal vaginal delivery at term and was otherwise fit and well. He had normal motor development and had no difficulty in feeding, swallowing or breathing. His parents were fit and well, as were his two sisters and a brother aged 7 years, 2 years and 6 weeks respectively. The parents were unaware of any first-degree relatives with similar symptoms, although it was difficult to be certain as most of their relatives lived abroad.

On examination, he was noted to have bilateral ptosis with poor orbicularis tone (Fig. 1). He also had limitation of adduction and elevation of both eyes. Both pupils were equal and reactive and fundoscopy was normal.

Myasthenia gravis was suspected clinically and a Tensilon test was performed in theatre with an anaesthetist present; 1 mg of edrophonium (100 μg/kg) was administered intravenously. This was strongly positive (Fig. 2).

He was subsequently referred to the paediatricians for further assessment and investigations. Full general medical examination was normal. In particular, there was no demonstrable fatiguability of limbs, neck flexors or respiratory muscles. A positive serology for AChR-ab of 45×10^{-10} M (normal $< 2 \times 10^{-10}$M) was found on two separate blood tests. Consultation with regional immunology and paediatric neurology services at Leeds confirmed the diagnosis of autoimmune myasthenia gravis. Further investigations including thyroid stimulating hormone levels, antinuclear and antithyroid antibodies were negative. A chest radiograph and CT scan of the mediastinum demonstrated a normal thymus. Neither electromyography nor muscle biopsy was performed as it was felt that the diagnosis of autoimmune myasthenia was already established. The patient was commenced on pyridostigmine 30 mg once daily and showed a good response to treatment. He remains well at 6 months follow-up.

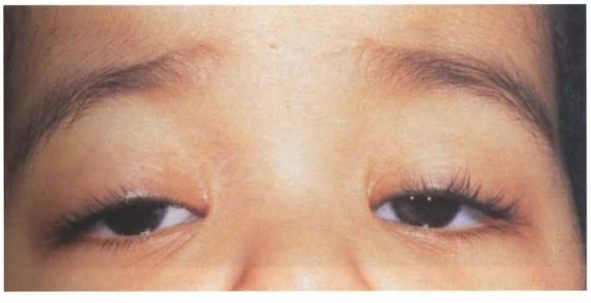

Fig. 1. *Before administration of edrophonium.*

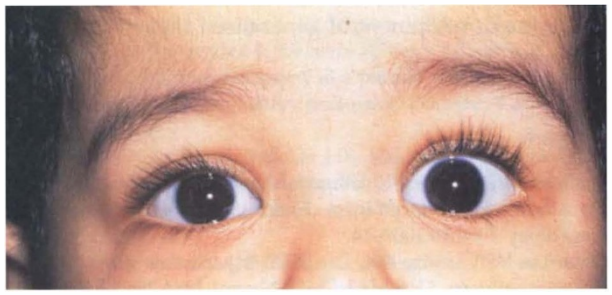

Fig. 2. *After administration of 1 mg edrophonium.*

Discussion

Myasthenia gravis is primarily a disease of adulthood. It is very rare before the first year of life. A review of 447 cases by Millichap and Dodge[1] reported only 16 cases occurring at or soon after birth. However, 10 patients had transient disease and were born to mothers with known myasthenia gravis; only 6 of these patients were born to mothers without the disease (1.3%). None of these patients had pure ocular myasthenia although ptosis was a common sign. The age of onset of myasthenia in oriental children is earlier than amongst Caucasians and pure ocular myasthenia seems to predominate.[2]

Childhood myasthenia can be broadly divided into two groups.[3–5] Firstly, there is hereditary myasthenia, which usually presents before the age of 2 years. This is not an autoimmune disorder but represents a genetic defect in neuromuscular transmission. An autosomal recessive pattern of inheritance is thought to be most likely, with other siblings often affected. The second group is autoimmune myasthenia. Onset of symptoms usually occurs between the ages of 2 and 20 years and clinically resembles adult myasthenia. This is associated with a positive serology for AChR-ab in 87% of patients and increased incidence of thyroid dysfunction. There is no recognisable pattern of inheritance and most patients have no affected siblings.

This case appears to be an autoimmune ocular myasthenia and is virtually unique because of its early age of presentation at 11 months. Seybold *et al.*[5] described 1 infant who presented with ocular myasthenia between the ages of 6 and 12 months with positive AChR-ab serology who then subsequently developed generalised myasthenia at the age of 4 years. Two further cases of generalised myasthenia with positive AChR-ab serology presenting at age 1 year and 14 months have been described.[5] This confirms the presence of autoimmune myasthenia below the age of 2 years. Although at present it appears that this case may be unique, long-term follow-up is obviously essential to

exclude the development of generalised myasthenia in the future.

References

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Sir,

Asymptomatic vaso-occlusive retinopathy in Hughes' syndrome

The antiphospholipid (APL) syndrome, first described in 1983,[1] is a disorder associated with specific antibodies directed against phospholipids, particularly cardiolipin and phosphatidyl serine. Present also are elevated levels of the lupus anticoagulant. Although originally studied in patients with systemic lupus erythematosus (secondary APL), APL is now known to occur as a primary (Hughes') syndrome.[2] The main clinical feature is thrombosis, both venous and arterial, and the major feature of the syndrome in women is recurrent miscarriages.[3] The eye may be involved symptomatically as a feature of transient ischaemia,[4,5] or as a direct, local, ischaemic event.[6–9]

We report here a patient who presented with severe vaso-occlusive retinal disease in the absence of any ocular symptoms.

Case report

A 48-year-old white man was referred to us by his optician who noted several 'haemorrhages' in both retinae with new vessels at the left optic disc. He had no ocular symptoms other than presbyopia. Eleven months earlier, he had been investigated by the physicians for fatigue, sleeplessness and weight loss, associated with hypertension.

The ESR was 30 mm in the first hour (Westergren). Fasting blood sugar was normal. The platelet count was 79×10^9/l (normal 120-400 $\times 10^9$/l). Prothrombin time, fibrinogen and fibrinogen degradation products were normal but the augmented partial thromboplastin test (APTT) was raised at 47 s (normal 29–37 s). The Dilute Russell Viper Venom Time Ratio was 1.50 (normal 0.8–1.12). The following tests were normal/negative: autoantibodies to glomerular basement membrane, anti-double-stranded DNA, complement C3 and C4, c-ANCA and p-ANCA, serum electrophoresis, and Bence-Jones protein. He had a positive antinuclear antibody and raised titre of anticardiolipin antibody (IgG, 23 GPL u/ml; and IgM, 3 MPL/ml). With a diagnosis of accelerated hypertension secondary to the antiphospholipid syndrome, antihypertensive medication was started.

At presentation to the eye clinic, the patient was well and his blood pressure was 155/85 mmHg. Visual acuity was 6/6 and N5 either eye. There was no afferent pupillary defect. Anterior segments were normal, and intraocular pressures were 14 mmHg in both eyes. Fundus examination showed occluded and sheathed arterioles in the mid-periphery of both eyes, together with frank neovascularisation at the left optic disc (Fig. 1). Fluorescein angiography showed peripheral arteriolar attenuation with frank occlusion at some points (Fig. 2). We performed left panretinal argon laser photocoagulation. Simultaneously, the patient was referred to the haematologist who commenced him on warfarin 5 mg o.d. to prevent further vaso-occlusion,

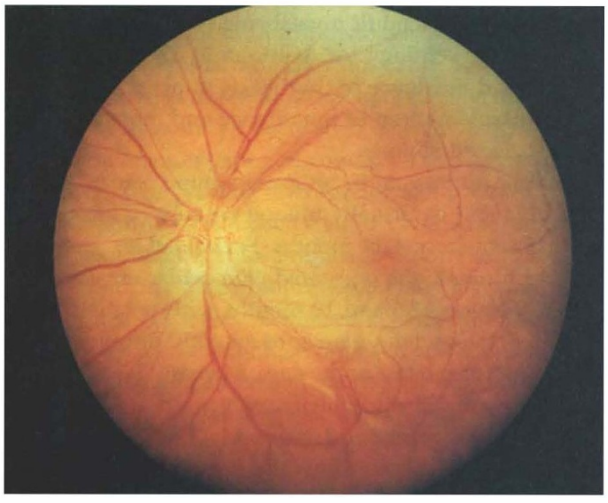

Fig. 1. *Fundus photograph of left eye at presentation showing disc neovascularisation and occlusion of an arteriole superior to the fovea.*

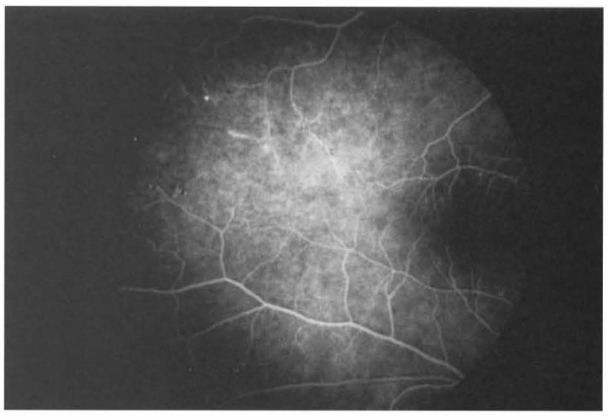

Fig. 2. *Fluorescein angiogram of the right eye showing a clipped arteriole temporal to the macula.*