THE OCULAR MYOPATHIES

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There is a group of diseases which affect the workings of the muscles of the body. The muscles affected may be those in the limbs, for example, as in the disease known as muscular dystrophies; or they may be the extra-ocular muscles, when the disease is called an ocular myopathy. One or several extra-ocular muscles may be affected, and there may be other associated eye disorders. An ocular myopathy and a muscular dystrophy may occur together.

In these myopathies and muscular dystrophies there is some evidence that the fault may lie in the biochemical mechanism within the muscle fibres, at the junction between the nerve and the muscle it drives, or even in the nerve itself.

The ocular myopathies are probably more common than is realised. They are often diagnosed in retrospect, only when review of ocular motility findings reveals a pattern too bizarre to be otherwise explained.

Two case histories will serve to illustrate what happens:

1. James C. first presented at age seven, with increasing diplopia from a left lateral rectus paresis and a history of minor head injury. There were no other neurological signs and a year later, with no change in his Hess screen chart, surgery corrected his diplopia.

   At the age of ten, he presented again with a new diplopia, this time from a left superior rectus palsy. Again the neurologist pronounced him normal, and a subsequent resection of the affected muscle cured him.

   At the age of thirteen, he had further diplopia, this time from weaknesses of the right lateral rectus and left superior oblique muscles. He had a left ptosis. Inspection of family photographs suggested a minor degree of right ptosis had been present, unrecognised, from the age of five. His father was now attending the neurological clinic because of multiple sclerosis. The diagnosis of a classical case of ocular myopathy was now very clear.

2. Andrew M. was referred at age five, because at his school entry test he read only 6/60 in each eye. He had a fully concomitant alternating convergent squint. Visual acuity and binocularity were restored by +10.00 dioptre spheres for each eye.

   Two years later, vision in his right eye dropped, as he developed a $+15^{\circ}$ RCS with some L/R. He was unenthusiastic about constant light-tight occlusion, but recovered 6/6 vision and had a right recession and resection for this squint.

   At the age of ten and a half, he had some vertical diplopia which increased, and his grandmother pointed out his left ptosis. His inconstant Hess screen charts caused great distress to his orthoptist. He mentioned he wasn't very good at football, although he seemed to be very sturdy. The diagnosis was now clear. He was carefully fasted and rested overnight, so that a blood test for muscular dystrophy could be taken. It confirmed the clinical suspicion of muscular dystrophy with ocular myopathy.

   Diagnosis is relatively easy in a classical case showing the following features, as in the first patient:

   Onset in childhood

   Slowly progressive

   Ptosis usually precedes ocular motility upset

   Males are involved more frequently than females

   There is commonly some neurological upset in closer relatives

   Pupil reactions are not changed from normal

   Gaze palsies do not occur.

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There may be a very long interval between the onset of ptosis and ocular motility problems. A familial incidence is sometimes found. The exasperation and depression of wildly discrepant synoptophore or Hess screen readings at successive orthoptic visits becomes replaced by something approaching elation when the reason is suddenly clear.

Some readers may have seen patients with ocular myopathy, but felt it was something else, because it was given a different name; for example, progressive external ophthalmoplegia, or abiotrophic ophthalmoplegia externa. Ocular myopathy is the term preferred, as it fits best with our current knowledge of the disease involving primarily the extra-ocular muscles. Some recent evidence, though, tends to confirm a suggestion made by Von Graefe over a hundred years ago that there may be some neural involvement also.

The obvious conditions which have to be differentiated are:

Myasthenia gravis

Endocrine myopathy

Myositis

Myotonic syndrome

Drug effects (for example, Atropine, Vincristine)

The most important one to distinguish is myasthenia gravis. Electro-myogram of the extra-ocular muscles, if available, can help clinch the diagnosis and can help distinguish ocular myasthenia. This test also offers the most satisfactory method of carrying out the Tensilon test for myasthenia. Alternative methods of doing a Tensilon test are worth knowing about. In a myasthenic, an injection of Tensilon intravenously, briefly raises the intra-ocular pressure by over 5mm. Hg. within a minute of the injection being given (usually 20-40 seconds). This can be recorded by continuous applanation tonometry, or better, by a brief tonography.

A new method of assessing extra-ocular muscle power has recently been described from Sheffield. It has considerable promise and is within the capabilities of the average orthoptist. A contact lens on the eye, is attached to a strain gauge which feeds into an oscillograph. The patient sits in front of a Hess screen and has merely to quickly look in the direction of the suspected affected muscle.

Some associated conditions are worth noting:

(a) Muscular dystrophy. This was present in one quarter of the patients in the original Kiloh and Nevin series. My second case is in this group. Biopsy of skeletal and extra-ocular muscle may help clarify a doubtful diagnosis in these patients, but only few centres can do the necessary elaborate work well. Serological tests can help. The two enzymes most checked are the serum creatine phosphokinase and serum aldolase.

(b) Atypical retinitis pigmentosa. This is commonest in females with ptosis early and ophthalmoplegia late. The fundus picture does not show the classical "bone spicules" of true retinitis pigmentosa, and electro-diagnostic tests of retinal function are normal. Night blindness is also not a feature.

(c) Hereditary ataxias such as Friedreich's ataxia and Marie's ataxia. Nystagmus is common in the first, and optic atrophy in the second.

(d) There is a group of patients with ocular myopathy, cardiac muscle myopathy and retinal degeneration. These patients are liable to sudden death. They are deaf, dwarfed, and have weak trunk and limb muscles.

(e) A miscellaneous group includes such conditions reported rarely as the Bassen-Kornzweig syndrome, Refsum's syndrome and choroideraemia.

Treatment

If the cause is myasthenia gravis, the standard therapies for this also relieve the diplopia. Competent surgery can help most of the patients, thought the good results sometimes last for a disappointingly short time. Fresnel prisms are an important advance, because of the ease with which they can be changed. For the underlying causes however, there is no known remedy. You may wish to read a little more widely. The following references should help.

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