Diagnosis of ocular myopathy

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Hutchinson (1897) described a disorder in which ptosis was associated with paralysis of the other extraocular muscles to which he gave the name "external ophthalmoplegia". Fuchs (1890) reported five patients with ptosis, in two of whom the disorder was familial, and Dutil (1892) and Delord (1903) published two other cases of an unusual familial form of dysphagia associated with ptosis. The prevailing opinion at the time was that external ophthalmoplegia was due to a degeneration of cranial nerve nuclei. The idea that ptosis was but a fragment of a progressive dystrophy of all striated ocular muscles was put forward by Treacher Collins (1922) and by Sandifer (1946). This opinion has gained acceptance only since the critical study of Kilch and Nevin (1951), in which five patients with progressive muscular dystrophy with early and predominant affection of the external ocular muscles were described; they suggested that the term "ocular myopathy" be applied to these cases.

Lees and Liversedge (1962) described three examples of the descending form of this disease. In their cases the ptosis and ophthalmoplegia preceded by many years the involvement of the face, pharynx, larynx, neck, shoulder, and pelvic girdles, and the course of the illness was slow and relatively benign. Victor, Hayes, and Adams (1962) called attention to a form of restricted myopathy of cranial muscles characterized by dysphagia and ptosis which they called oculopharyngeal myopathy; both familial and sporadic cases were described and firm evidence was presented that the disorder was myopathic. Roberts and Bamforth (1968) analysed 26 further patients diagnosed as cases of ocular myopathy; six of their patients had dysphagia with motility and radiographic changes suggesting that the dystrophic process also involved the smooth muscle of the oesophagus.

Because of the apparent rarity and lack of general recognition of ocular myopathy, we thought it of interest to describe four cases seen over the course of one year.

Case reports

Case 1, a 22-year-old single woman, developed drooping of the eyelids at the age of 11 years. She had two operations to correct this at the ages of 11 and 15 years. Her relatives noticed that her eye movements were limited but she had not complained of double vision. She stated that if she talked or read aloud for some length of time her voice became weak. There was no family history of myopathy.

Examination

There was severe bilateral ptosis, all eye movements were markedly restricted, those of the left eye being less affected; there was 10° of side-to-side movement, no movement above the horizontal, and only 5° downward movement. The pupils were equal and the reactions were normal. This patient also had bilateral weakness of the orbicularis oculi and of the lower facial muscles leading to a mournful expression. There was considerable wasting of the muscles of the neck and she was unable to lift her head up from the pillow. Her tongue was small but there was no myotonia; her voice was weak. Palatal movements appeared to be normal. There was considerable wasting of the muscles of the forearms on both sides. The upper limbs showed asymmetrical weakness, more marked on the right side and affecting especially the trapezii and triceps. There was also weakness of the trunk muscles and of hip flexors. All the tendon reflexes were absent in the upper limbs, but normal in the lower limbs; the plantar responses were flexor.

Investigations

Electromyography of the left lateral rectus was consistent with myopathy, the units on voluntary deviation being of extremely low amplitude (about $20 \mu\text{V}$) and rather scanty. Serum creatinine phosphokinase was 116 mU/ml . (normal range for women $5\text{--}50 \text{ mU/ml}$.) but the serum aldolase at 0.9 mU/ml was within the normal range $0\text{--}6 \text{ mU/ml}$.

Muscle biopsy of the deltoid (Fig. 1) showed some variation in the size and staining properties of the muscle fibres with minimal degenerative changes affecting single segments rather than the whole fibre. The changes were slight but myopathic in type. It should be noted that the muscle used for biopsy was not clinically affected.

FIG. 1 Biopsy of deltoid muscle in Case 1. $\times 90$

Case 2, a 34-year-old married woman, developed drooping of the right eyelid at the age of 25 years. For a year she had noticed blurring of vision but no true diplopia on lifting the eyelid, and she had had mild pain in the right eye, with difficulty in reading. There was no family history of myopathy.

Examination

She was a pale, thin, anxious woman. There was an obvious right ptosis but the pupils were equal and reacted normally. The upward and downward movements of both eyes were full. There was moderate weakness of abduction of the right eye and slight weakness of the left and considerable weakness of adduction of the right eye. There was a little weakness of both orbicularis oculi muscles and definite weakness of the neck muscles and of the right triceps. The tendon reflexes were brisk and equal. Plantar responses were flexor.

Investigations

A tensilon test was negative. Serum creatinine phosphokinase was normal at 39 mU/ml . and the aldolase was within normal limits. The electromyogram was compatible with a mild myopathy.

Muscle biopsy of levator palpebrae (Figs 2 and 3) revealed that the muscles were almost completely destroyed and contained only a few surviving, apparently normal, fibres. The bulk of the specimen consisted of fibres and fatty tissue containing numerous nerve bundles. The good preservation of the latter suggested that the lesion was a myopathy.

FIG. 2 *Biopsy of levator palpebrae muscle in Case 2.*
× 120

FIG. 3 *Biopsy of levator palpebrae muscle in Case 2.* × 120

Treatment

A surgical repair of the ptosis was made when the muscle biopsy was performed. The patient was satisfied with the result of this operation.

Case 3, a 46-year-old married woman, developed drooping of the right eyelid at the age of 41 years. For the previous 6 months she had complained of aching of the right eye but there was no definite diplopia. There was no family history of myopathy.

Examination

There was moderate right ptosis but the eyelid did not cover the pupil. There was also mild proptosis of the right eye. There was limitation of abduction of both eyes, and of adduction of the right. There was bilateral weakness of the orbicularis oculi and the voice was weak. The neck muscles were also weak and the patient was unable to lift her head from the pillow. The right triceps muscle was slightly weak. The reflexes were brisk and equal; the plantar responses were flexor.

Investigations

A tensilon test was negative. Creatinine phosphokinase was raised 63 mU/ml. Electromyography of the sternomastoid showed only a few short motor units in the right. The motor unit pattern was otherwise normal. Radioactive iodine uptake and serum protein bound iodine were both within normal limits. Muscle biopsy was not done.

Case 4, a 71-year-old man, had developed drooping of the eyelids at 50 years of age. This had been much more marked during the last year, when he had noticed double vision on reading, images being vertically separated. He had also noticed slight weakness of the neck and shoulders. He had been diagnosed as suffering from myxoedema 10 years earlier, and had since taken thyroid tablets (120 mg. a day). There was a family history of ptosis and of haemophilia, as shown in Fig. 4.

FIG. 4 Case 4. Family tree

Examination

He had severe bilateral ptosis (Fig. 5), two-thirds of each pupil being covered by the upper lid. All eye movements were severely restricted; there was less than 5° movement up and down and less than 10° side-to-side. The pupils were equal in size and reacted to light and accommodation. The forehead was constantly wrinkled and the eyebrows were raised above the level of the supraorbital ridges. Both orbicularis oculi muscles were weak and there was slight weakness of the lower face and of both sternomastoids and trapezii. The tendon reflexes were brisk and equal; plantar responses were flexor. Muscle biopsy of the sternomastoid is shown in Fig. 6.

FIG. 5 Case 4. Bilateral ptosis

FIG. 6 Biopsy of sternomastoid muscle in Case 4. $\times 100$

Investigations

A tensilon test was negative. Electromyogram of the right sternomastoid showed patches of small, short disintegrated myopathic units throughout the muscle. The right deltoid and right quadriceps showed normal activity. Serum creatinine phosphokinase was raised at 82 i.u./litre. Muscle biopsy of the sternomastoid (Fig. 6) showed a considerable increase in the amount of fibrous tissue, which contained a few remaining muscle fibres. Outside the areas of fibrosis the muscles showed no abnormality. These appearances were those of a low grade myopathy.

Treatment

This patient had a bilateral repair of his ptosis and was pleased with the improvement in his ability to see and in his appearance.

Methods of investigation

Investigation of patients with ocular myopathy is reasonably simple and the estimation of serum muscle enzymes is a useful confirmatory test, although these may not always be abnormal if the disease is restricted to a small group of muscles. Muscle biopsy of the external ocular muscles is not easy to perform and not a minor procedure, although it can be combined with correction of the ptosis. It was useful in two of these patients to perform a plastic operation on the eyelid and to take a biopsy of the levator palpebrae simultaneously. Unfortunately the state of this muscle was not entirely satisfactory from the pathological point of view, as the disease process was found to be far advanced by the time of the biopsy. In another patient, in whom a limb girdle muscle was sampled, this showed early changes of muscular dystrophy.

Discussion

Cases 1 to 3 illustrate the descending ocular myopathy of the type described by Lees and Liversedge (1962). As in their cases, there was no family history of any similar condition. In Case 1 the disease was more or less symmetrical but in Cases 2 and 3 it was markedly asymmetrical. Case 2 at first glance resembled a unilateral third nerve palsy and it is of interest that this patient had been investigated as such in a Neuro-surgical Unit by means of carotid arteriograms and air studies. Case 3 had also been misdiagnosed as one of exophthalmic ophthalmoplegia because of slight unilateral proptosis. However, the weakness of the neck and shoulder girdle muscles is out of keeping with this diagnosis unless there is coexistent thyrotoxic myopathy and a definite unilateral ptosis would also be most unusual in this disease. There was no oedema of the eyelids or injection of the conjunctivae and major symptoms of ocular discomfort characteristic of exophthalmic ophthalmoplegia were absent.

Case 4 illustrates a slowly progressive, familial form of this disease. The family tree suggests a dominant mode of inheritance, although the patient's parents were allegedly normal. The disease had progressed slowly over two decades to affect the face, neck, and shoulder muscles as in the other patients.

The most important differential diagnosis in these patients is that of myasthenia gravis. The tensilon test usually distinguishes this condition (edrophonium chloride 10 mg. intravenously). While there may not be much improvement in the power of the external ocular muscles with tensilon, the drooping of the eyelids almost invariably improves with an injection of this substance.

Ophthalmologists should always be on the lookout for this neurological disorder in patients presenting to them with ptosis.

Summary

The literature of ocular myopathy is reviewed. Four cases of this condition are presented and the differential diagnosis discussed.

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