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Oculopharyngeal myopathy with sensorineural hearing loss

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Abstract
A case is reported of a 59-year-old Caucasian male with oculopharyngeal myopathy and sensorineural hearing loss. He presented with progressive ptosis, sensorineural hearing loss over several years and symptoms of mild dysphagia. Further enquiry into his family history revealed that every male member in his family that lived beyond the age of 60 exhibited identical symptoms. Symptoms of ptosis and dysphagia are consistent with the rare autosomal dominant condition of oculopharyngeal myopathy, believed to be due to mitochondrial disease. The combination of ptosis, dysphagia and sensorineural hearing loss with normal distant muscle group biopsy has not been described before.

Key words: Hearing loss, sensorineural; Myopathy, oculopharyngeal

Introduction

Oculopharyngeal myopathy is an autosomal dominant disorder developing in later life and characterized by dysphagia and progressive ptosis of the eyelids (Victor *et al.*, 1962). It is considered to be due to mitochondrial disease. The diagnosis of mitochondrial disease is made on the finding of 'ragged red fibres' on muscle biopsy. Mitochondrial disease has a multi-system involvement, but nearly always includes muscles. There are proven and postulated defects of aerobic oxidative metabolism (McKusick, 1988). Various defects result in different clinical expression of the disease (Nishizawa *et al.*, 1987).

Case report

A 59-year-old Caucasian male thought to have oculopharyngeal myopathy was referred to the ENT outpatient department with deafness. He had become aware of progressive deafness over several years. He sought medical advice as his eyelids had slowly and progressively begun to droop over the preceding three years, his right eye being more severely affected than his left (Figure 1). He gave a history of difficulty swallowing which had progressively worsened over the last few years. He had no history of diplopia and had no symptoms of weakness in his arms or legs. His speech was normal. He had no cutaneous sensory symptoms.

Further enquiry into his family history revealed that his father and both parental uncles were affected with similar progressive ptosis, difficulty swallowing and deafness (Figure 2, and family tree, Figure 3). His father's third brother had died young during the first world war, but his sister who lived to the age of 86 was asymptomatic. His paternal grandfather had also suffered with progressive ptosis and deafness. His two sisters, one of whom died at the age of 62, and the other who is now aged 57, were both asymptomatic. His 54-year-old male cousin has developed deafness, over the last few years. His son, aged 34, has recently become aware of bilateral deafness becoming progressively worse but as yet has no ptosis or difficulty in

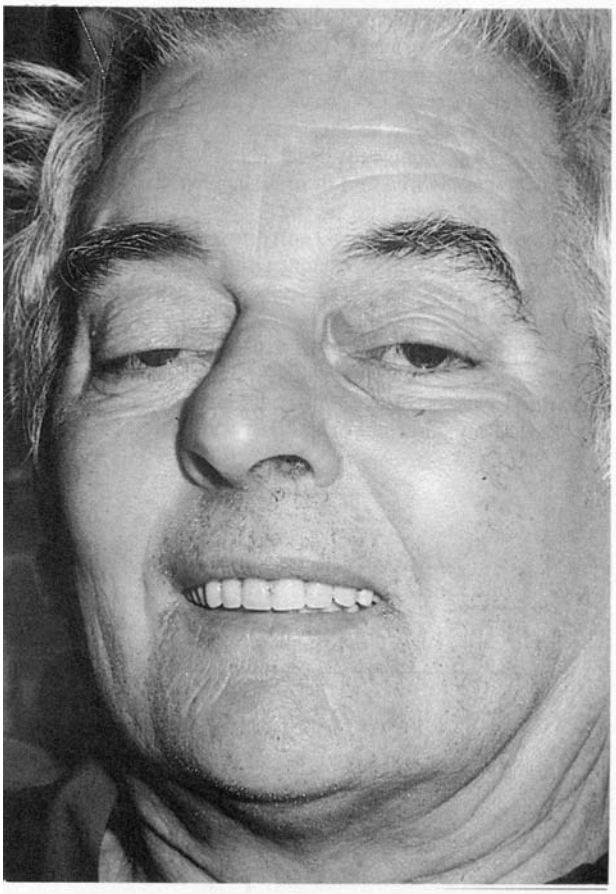

Fig. 1
Patient with ptosis.

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FIG. 2

Family wedding photograph showing both the patient's father (on bride's left side) and the patient's uncle (far left of picture), both demonstrating ptosis.

swallowing. The onset of this patient's symptoms started in the latter half of the fifth decade as indeed was the case with his father, uncle and grandfather.

Examination showed bilateral ptosis, more marked on the right than the left. The fundi were normal. The range of external ocular movements of the right eye was slightly impaired in upward gaze and lateral movements. Strength was normal in all skeletal muscle groups except for a slight weakness of right shoulder abduction but he was left handed. Reflexes were present and symmetrical and he had a flexor plantar response. There was no ataxia and sensory examination was normal. Cardiovascular and respiratory systems were normal. Otoscopic appearance of his ears was normal, the Rinne test was positive bilaterally, and the Weber test was not lateralized. Examination of the nose and throat was normal and indirect laryngoscopy revealed a normal hypopharynx and larynx.

Haematological investigations revealed a normal full blood count with an ESR of 3 mm/hr. Urea and electrolytes, liver and thyroid function tests were all normal. The Tensilon test with 10 mg edrophonium was negative. Auto-antibody screening was negative. Chest X-ray was normal as was his ECG. A vastus lateralis muscle biopsy was performed which showed no evidence of mitochondrial disease and no other abnormalities were detected.

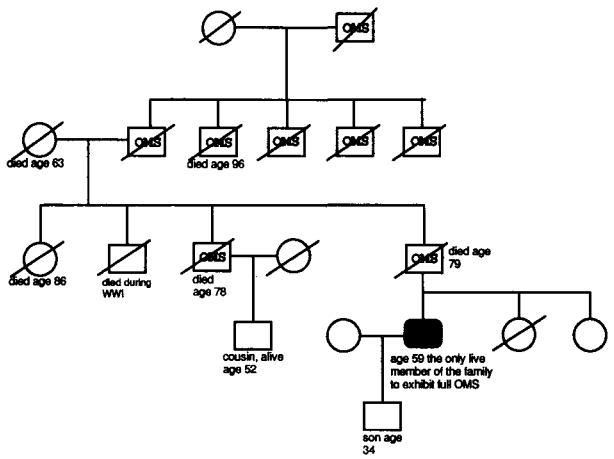

FIG. 3

Family tree shows all the males above 60 years to be affected with oculopharyngeal myopathy with sensorineural hearing loss (OMS).

Pure tone audiometry showed bilateral symmetrical sensorineural hearing loss with a ski slope pattern. Hearing deficit ranged from 40 dBHL in the lower frequencies to 90 dB in the high frequency range (Figure 4). Stapedial reflexes were absent and electric response audiometry testing at 95 dB failed to produce repeatable responses, probably because of the severity of the hearing loss. 'Site of lesion' testing showed a recruiting deafness with absence of tone decay, and a speech discrimination score, suggestive of a cochlear hearing loss. Caloric testing revealed no labyrinthine dysfunction. Video fluoroscopy showed evidence of some pooling of contrast in the piriform fossae, with no evidence of aspiration.

Discussion

The combination of ptosis and dysphagia in this patient with a family history of similar symptoms points to a diagnosis of oculopharyngeal myopathy. This is somewhat at odds with the muscle biopsy findings which did not show evidence of mitochondrial disease. Although the diagnosis of mitochondrial disease is usually made by the finding of ragged red fibres on muscle biopsy, it is increasingly unlikely that this feature is always present (Baraister, 1990). There is also a consistent finding in affected family members of deafness. This has not been previously reported. The patient's 54-year-old cousin has not developed ptosis or dysphagia yet, although he has sensorineural hearing loss, pure tone audiometry showing a similar 40–60 dB sensorineural hearing loss with a ski slope pattern. The patient's 34-year-old son was found to have a 20 dB sensorineural hearing loss in the high frequencies bilaterally. All the nine male members of the family who have lived beyond 60 years of age have been affected with ptosis, dysphagia and deafness.

This condition was first recognized in a French-Canadian family with ptosis and progressive dysphagia by Taylor in 1915, who also noted the familial nature of the syndrome. Victor *et al.* (1962) described a family in which nine members of three generations were known to be affected. One member had also total external ophthalmoplegia and weakness of the limb-girdle muscles.

Many of the subsequent families reported were from Canada and the United States (McKusick, 1988). Barbeau

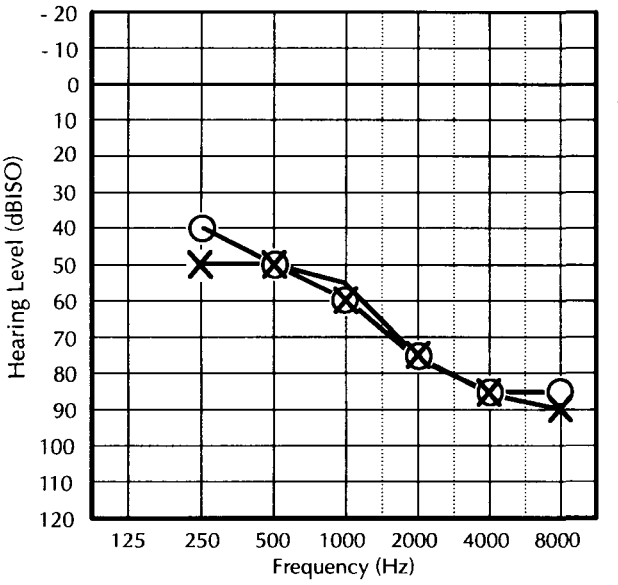

FIG. 4

The patient's pure tone audiogram showing bilateral symmetrical sensorineural hearing loss.

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(1966) showed that all of the numerous reported French-Canadian cases could be traced back to a single ancestor who emigrated from France in the 1600s. Morgan-Hughes and Mair (1973) studied four patients with oculoskeletal myopathy. All complained of generalized muscle weakness and fatigability. All showed bilateral ptosis with external ophthalmoplegia, facial and sternocleidomastoid weakness and diffuse wasting in the limbs. Two patients were dysphagic and one had pigmentary retinal degeneration. Triceps biopsies revealed certain isolated or clustered muscle fibres to contain accumulations of sarcoplasmic matter. Electron microscopy showed degenerative muscle fibre changes in all biopsy samples as well as striking abnormalities of muscle cell mitochondria. The mitochondria were seen to have laminated crystalline inclusions between the cristae. Sometimes the intercristal spaces were wide and electron dense. The authors stated that similar types of mitochondrial abnormalities have been described in other forms of myopathy. Knoblauch and Koppel (1984) described a family from eastern Switzerland with seven affected persons in three generations. Bilateral ptosis and dysphagia began in the fourth decade (McKusick, 1988). It seems likely that many of the so-called ocular myopathies described in older literature would now be found to be mitochondrial myopathies, although whether this is always the case remains uncertain (Baraister, 1990).

As there is male to male transmission, the mode of inheritance cannot be X-linked and must be either autosomal dominant or Y-linked. It is extremely unlikely that the mode of inheritance is Y-linked as this is extremely rare and the only confirmed Y-linked condition is the 'hairy ear' syndrome. If the present condition was inherited as an autosomal dominant, then it must be purely coincidental that no female members of the family over three generations have been affected. The chance of an autosomal dominant condition being inherited in this fashion is in the order of 1:16.

The association of oculopharyngeal myopathy with sensorineural hearing loss has not been previously reported.

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