



### Oculopharyngeal muscular dystrophy (OPMD)

The term muscular dystrophy is used to cover a wide range of conditions which have in common progressive muscle weakness due to an inherited genetic defect (mutation). There is huge variation in severity between the different conditions. Some present in very early childhood and progress to severe disability, whereas others can be extremely mild and not problematic even in old age. Each type relates to a specific genetic abnormality and if more than one family member is affected they each have the same type of muscular dystrophy.

It has been recognised for many years that some patients with muscle disease have particular problems with the muscles around the eyes, although other parts of the body can also be involved. Whilst research is continuing, it appears that most of these patients have either oculopharyngeal muscular dystrophy (OPMD), the subject of this factsheet, or mitochondrial chronic progressive external ophthalmoplegia (CPEO). Often, when patients first present, it may not be clear whether the patient has OPMD or CPEO but specific investigations can differentiate between the two.

The medical terms relating to these conditions may cause the lay person some confusion. They include: ptosis, external ophthalmoplegia, diplopia and dysphagia.

**Ptosis** - this describes drooping of the eyelids due to weakness of the muscle that normally lifts up the eyelid.

**External ophthalmoplegia** - this means weakness and restriction of muscle movement around the eye (external to the eye). It shows as slowness and incomplete range of movement of the eyes, and includes the eyelid muscle weakness that causes ptosis. These problems typically progress very slowly, hence the term 'chronic progressive external ophthalmoplegia'.

**Diplopia** - this simply means double vision and occurs when the eye muscles on each side are not affected equally, so that the eyes point in slightly different directions.

**Dysphagia** - this means difficulty in swallowing. When mild, it may simply be a feeling of food sticking in the throat, but patients with severe dysphagia may not be able to swallow at all and can even choke on their own saliva.

#### Symptoms and signs

Although the abnormal gene causing OPMD is present from birth, patients do not usually develop symptoms until the fifth or sixth decade of life. The first sign of the disorder is usually **ptosis**, but occasionally it is **dysphagia**. Very slowly, over many years, these problems progress. There is progressive restriction of eye movements and in rare cases this can lead to **diplopia**. The increasing ptosis may lead to the eyelid covering the pupil and impairing vision, and in an effort to compensate for this the forehead muscle becomes overactive, trying to help to lift up the eyelids, giving a frowning appearance, and the patient adopts a rather characteristic posture with the head tilted backwards.

Dysphagia, which is initially mainly for solid and dry foods, progresses slowly and eventually even swallowing fluids, including saliva, may become a problem. If dysphagia is severe there is a danger of aspiration (food, drink or saliva "going down the wrong way" - into the chest rather than stomach) which greatly increases the risk of a chest infection. After many years the patient may become aware of limb weakness, first around the shoulders and later around the hips. This is usually relatively mild

but can occasionally be severe and disabling, many years after the first onset of symptoms. Facial weakness may develop, and be commented upon by the specialist, but rarely causes any particular problems. Life expectancy is little, if at all, altered.

### **Management**

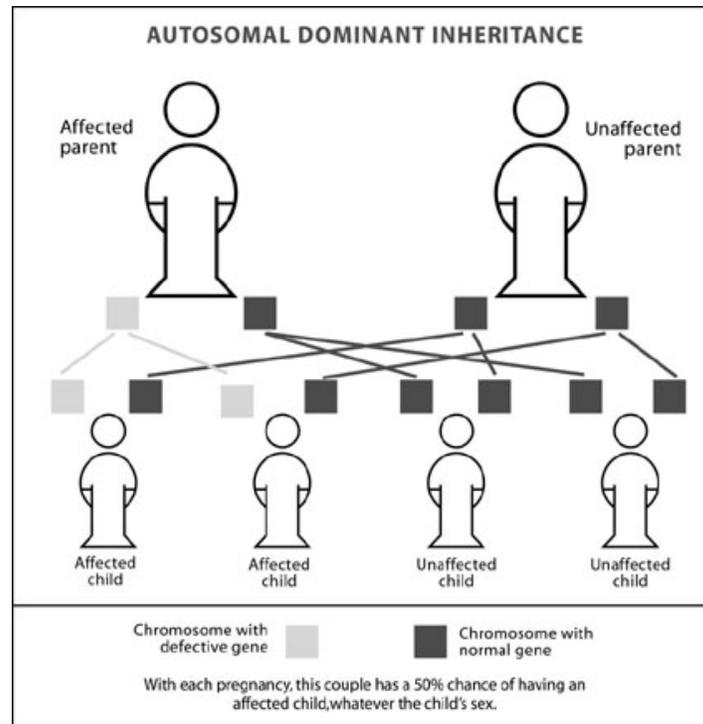
There is no specific treatment for OPMD, but much can be done to help the main symptoms of ptosis and dysphagia. Glasses can be fitted with fine metal bars (ptosis props) that lift up the drooping eyelids. If these are unacceptable, and if the ptosis is severe, surgical elevation of the eyelids can be very successful – several procedures are possible and should be tailored to the individual patient.

Mild dysphagia can be helped by suitable attention to the consistency of the diet (with a dietician's advice) and by exercises taught by a speech therapist. In more severe cases, a relatively minor operation called cricopharyngeal myotomy, which cuts one of the throat muscles internally, can be valuable. Another approach that is sometimes helpful is to inflate a balloon to dilate the gullet. But as for all surgical procedures there are potential hazards and the final choice of treatment depends upon many individual factors. Recently there have been a few reports of the use of botulinum toxin injections; rather than cut or stretch a muscle, the toxin relaxes the muscle and that can aid swallowing. However, further studies are needed to see whether this will prove to be a useful long-term treatment. If the dysphagia is preventing adequate nutrition or there is a risk of aspiration pneumonia, then alternative methods of feeding can be used. The most acceptable, in the long term, is gastrostomy. A minor operation is used to pass a tube through the front of the abdomen directly into the stomach. Patients and their relatives find this easy to manage at home. If the normal diet is compromised, then a dietitian can offer advice with respect to supplements which can help to maintain adequate nutrition.

Physiotherapy may be useful to help patients cope with limb weakness, although this is usually mild, and to reduce the risk of chest problems.

### **How is OPMD inherited?**

In almost all cases the condition is inherited as an autosomal dominant disorder (see figure below), which means that each child of an affected individual has a 50% risk of inheriting the same condition. It is now possible, through a blood test, to determine whether somebody has inherited the abnormal gene (called PABPN1) but that is not always terribly helpful. Even if somebody has inherited the abnormal gene, it is impossible to predict when, if ever, they will develop symptoms. Such testing should only be performed after detailed discussion with a suitably experienced neurologist or genetic counsellor.



### Diagnosis

The diagnosis can be confirmed by a blood test that identifies the underlying genetic abnormality. Electrical tests (EMG) and muscle biopsy are now rarely necessary.

### Is there any research being conducted into OPMD?

The genetic fault that causes OPMD was identified in 1998. Although this was a very important discovery, which has given us a simple diagnostic test, it is likely to be some considerable time before the research allows us to identify a specific treatment for this condition. In the meantime, there is a great deal of research trying to identify how the genetic fault causes the physical problem. Since 1998 over 100 research papers on OPMD have been published. Whilst none of these have yet led to a major change in management, they should be regarded as the building blocks for progress in the future.

### Mitochondrial chronic external ophthalmoplegia (CPEO)

As mentioned above, this condition can be confused with OPMD. It is often sporadic (i.e. occurs in an individual with no family history of a similar condition) but occasionally is inherited so that there is a history of similarly affected relatives.

In CPEO the restriction of eye movements tends to be much more severe than in OPMD, but diplopia is still uncommon. As in OPMD, ptosis can be marked. Dysphagia is less common in CPEO than OPMD. Limb muscle weakness can be similar to that in OPMD, but may be more severe, and associated with exercise-intolerance.

Mitochondrial disorders can also affect other organs giving rise to deafness, diabetes, heart problems, and brain problems including epilepsy and dementia.

### Other factsheets that may be useful

- Gastrostomy
- Mitochondrial myopathy (for more information on CPEO)

MC35

Published: 01/06

Updated: 10/09

Author: Dr D.Hilton-Jones, Consultant Neurologist, Oxford.

**Disclaimer**

*Whilst every reasonable effort is made to ensure that the information in this document is complete, correct and up-to-date, this cannot be guaranteed and the Muscular Dystrophy Campaign shall not be liable whatsoever for any damages incurred as a result of its use. The Muscular Dystrophy Campaign does not necessarily endorse the services provided by the organisations listed in our factsheets.*

