Oculopharyngeal muscular dystrophy (OPMD)

Oculopharyngeal muscular dystrophy (OPMD) refers to a genetic condition that causes muscle weakness in a characteristic pattern. It is useful to explain the words that make up OPMD:

- **oculo** refers to the fact that the eye muscles (specifically the eyelids) are affected, causing eyelid drooping – also called ‘ptosis’

- **pharyngeal** refers to the fact that the throat muscles, in particular those related to swallowing, are affected. Difficulty in swallowing is called dysphagia

- **muscular dystrophy** is a term which usually means the condition is genetic, and causes progressive muscle-wasting and weakness, both of which are true in OPMD.

Symptoms

People with OPMD do not usually develop symptoms until after the age of 40, with an average age of onset of around 50. The first sign of the condition is either ptosis, or dysphagia (difficulty swallowing). Very slowly, over many years, these problems worsen.

The increasing ptosis may lead to the eyelid covering the pupil and so obstructing vision. In an effort to compensate for this, the forehead muscles become overactive, trying to help lift up the eyelids. People with OPMD often adopt a rather characteristic posture with the head tilted backwards to see clearly. However, the eye itself is not affected, and even when the condition is advanced, people with OPMD have normal vision if the eyelids are raised.

Dysphagia, which initially mainly affects swallowing solid and dry foods, progresses slowly. 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 increases the risk of a chest infection (aspiration pneumonia).

Many years after the onset of the condition, people with OPMD may become aware of limb weakness, often first around the shoulders and later around the hips. This is often relatively mild but can progress to be more severe and disabling, many years after the first onset of symptoms. Life-expectancy, with modern management of the condition, is close to normal.
Diagnosis

If OPMD is suspected, it can be confirmed genetically by a blood test. This test identifies the underlying genetic abnormality in the mutated gene (called PABPN1). Electrical tests of the muscle – electromyography (EMG) and muscle biopsy are now rarely necessary.

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 percent risk of inheriting the condition.

If someone in a family with known OPMD develops likely symptoms, then a diagnostic genetic test will often be advised.

![Inheritance pattern in autosomal dominant disorders](image)

**Figure 1: Inheritance pattern in autosomal dominant disorders**
Sometimes, people with a parent with OPMD wish to know whether they are likely to develop the condition. They may wish to consider a predictive test (i.e. prior to symptoms). After suitable discussion/counselling of the advantages and disadvantages of having this knowledge, a genetic test may be offered. In this situation, however, when a family member has inherited the mutated gene, it remains impossible to predict precisely when they will develop symptoms.

**Condition management**

There is no overall curative treatment for OPMD. However, the two most prominent problems - ptosis and dysphagia – can be helped by medical management.

**Management of ptosis**
Surgical elevation of the eyelids can be very successful – several procedures are possible and should be tailored to the individual. None of the procedures is major, but quite delicate and fiddly. The aim is to preserve symmetry and allow restored vision while still allowing eye closure. Like many procedures, these are best performed by a surgeon with an interest in this form of surgery, often an oculoplastic surgeon.

**Management of dysphagia**
Mild dysphagia can be helped by suitable attention to the consistency of the diet and by strategies taught by a speech therapist (often called a speech and language therapist – SALT). Food supplements may be advised by a dietician if there is weight loss. In more troublesome dysphagia, because the upper oesophageal sphincter can obstruct weak swallowing, stretching or cutting this muscle often helps, at least for a while. This is achieved either by stretching the muscle with a dilator, or cutting it in an operation called cricopharyngeal myotomy. A similar but temporary effect can be produced with botulinum toxin (‘Botox’) injection. The choice of procedure is often dictated by local expertise and facilities as much as patient preference.

If dysphagia over many years fails to respond to such measures, and progresses to 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 a gastrostomy. This is a minor operation to pass a tube through the front of the abdomen directly into the stomach. These days this is often achieved by a PEG (percutaneous gastrostomy), or a RIG (radiographically inserted gastrostomy). These relatively minor procedures can sometimes be done as a day case.

A gastrostomy (PEG or RIG) has several advantages: it ensures adequate nutrition and hydration, it lessens the chance of aspiration pneumonia, while still allowing people with OPMD to swallow small quantities of food or drink if they are able. A PEG or RIG is unobtrusive and can be concealed under clothes.
Experienced physicians advise that a gastrostomy be performed before swallowing becomes very impaired. Problems may arise if it is delayed until dysphagia is severe. A dietician can offer advice with respect to PEG or RIG feeding and supplements to help maintain adequate nutrition.

Physiotherapy and assistive aids may be useful to help cope if limb weakness occurs. Occupational therapy is often helpful, particularly if activities of daily living are affected by limb weakness.

Further suggestions to reduce problems with OPMD include an annual flu vaccination, especially for older people. In patients with significant dysphagia, a prompt evaluation of a productive cough is advised.

Is any research being conducted into OPMD?

The genetic mutation that causes OPMD was identified in 1998. This has allowed productive research into different aspects of the condition. The molecular mechanism of how the mutation causes the symptoms is complex and is not yet completely clarified. Nevertheless, increased understanding has already led to several initiatives of different treatment strategies, but there are no proven specific medical treatments so far.

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The friendly staff in the care and support team at the Muscular Dystrophy UK’s London office are available on 0800 652 6352 or info@musculardystrophyuk.org from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can’t help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

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