

OCULOPHARYNGEAL MUSCULAR DYSTROPHY



What is Oculopharyngeal Muscular Dystrophy (OPMD)?

OPMD is a form of muscular dystrophy which affects the ability to swallow, and the muscles around the eyes. It is an inherited condition with an increased occurrence among people of French Canadian descent. OPMD is due to an abnormality in the PABPN1 gene. The condition is one of the milder forms of muscular dystrophy.

What are the symptoms of OPMD?

Although the abnormal gene causing OPMD is present from birth, people do not typically develop symptoms until after age 40. Early signs of the disease include trouble swallowing (dysphagia), and/or droopy eyelids (ptosis) due to weakened muscles in the eyelids. Muscles around the eyes can also be affected (external ophthalmoplegia). This additional muscle weakness leads to a decreased range of motion of the eyes causing problems such as difficulty gazing upwards and double vision (diplopia). Diplopia occurs when the eyes point in slightly different directions.

How is OPMD inherited?

The vast majority of OPMD is inherited as an autosomal dominant trait. This means a person can inherit the defective gene which causes the disease from just one parent. Each child of an affected individual has a 50% risk of inheriting the same condition. Autosomal dominant diseases generally have a strong family history. In some rare cases, OPMD is inherited as an autosomal recessive trait. Here, the defective gene needs to be inherited from both parents. People with a family history of the disease may choose to have genetic counseling to help understand the risks of passing the gene on to their children.

How is OPMD diagnosed?

Thanks to research, a blood test is now available which can detect the presence of the abnormal PABPN1 gene and can confirm a diagnosis of OPMD.

How progressive is OPMD?

Compared to other muscular dystrophies, OPMD has slow progression. OPMD generally does not affect life expectancy.

Dysphagia generally starts with difficulty swallowing solid and dry foods. With progression, weakened muscles lead to trouble swallowing fluids – including saliva. Mild dysphagia may simply present as a feeling of food sticking in the throat.

However, patients with severe dysphagia may not be able to swallow at all and can even choke on their own saliva.

Some people with OPMD develop weakness in facial muscles, lower extremity muscles, and shoulders and hips.

What treatments are available for OPMD?

There is no cure for OPMD, but there are various treatments that help manage the symptoms. For patients where ptosis interferes with vision, surgery can be performed to lift droopy eyelids.

Speech pathologists can monitor and assess the progression of dysphagia. Assessments of the ability to swallow can help identify people who are at risk for aspiration pneumonia -- lung infection due to the 'breathing in' of food or liquids. Aspiration pneumonia and its complications can be prevented with careful monitoring. Surgery may also help relieve problems due to dysphagia, and should be

considered if there is marked weight loss, near-fatal choking, or recurrent pneumonia.

Upper arm and shoulder weakness that limits function can be addressed with adaptive techniques through occupational therapy.

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