

Oculopharyngeal Muscular Dystrophy

What is Oculopharyngeal Muscular Dystrophy?

Oculopharyngeal Muscular Dystrophy (OPMD) is the name of a type of muscular dystrophy. The term oculopharyngeal comes from several Greek words:

- ‘oculo’ means eyes
- ‘pharyngeal’ meaning windpipe or throat

What are the signs and symptoms of OPMD?

Muscle weakness

- OPMD commonly affects the muscles of the legs (thighs) and sometimes the arms.
 - People with OPMD may have progressive tongue weakness.
 - Weakness of the muscles around the eyes results in droopiness of the eyelids (ptosis).
 - Onset of symptoms around 40 to 60 years of age.
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Other common signs or symptoms include:

- limitation of upward gaze
- wet voice due to pooling saliva
- trouble swallowing/choking (dysphagia)
- unintentional weight loss

What causes OPMD?

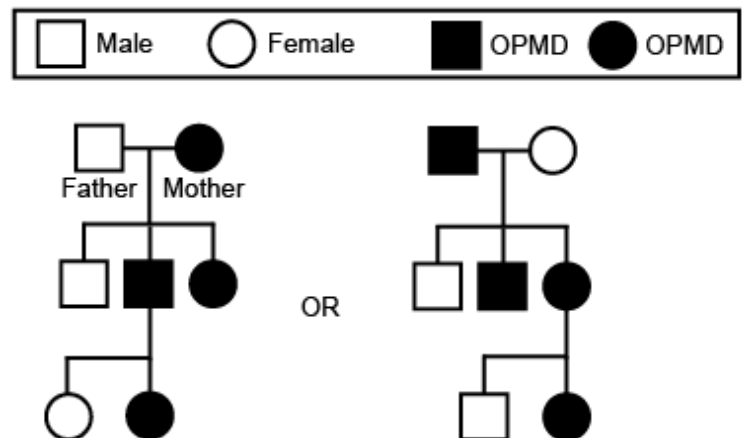
OPMD is a genetic condition that is inherited from your parents. The abnormal gene can be inherited from a mother or a father. Having the affected gene means that you have the condition. This is called “autosomal dominant inheritance”.

Autosomal dominant inheritance: this is when it is seen in several relatives, such as your parents, aunts, uncles, children.

How do you know I have OPMD?

Your doctor or specialist can tell if you have OPMD from:

- talking with you about your symptoms and family history
- reviewing the results of a genetic test for OPMD



Genetic tests

Most people with OPMD have an expansion (extra information) of genetic material near a gene known as PABPN1. The extra material is 3 letters, GCN, that are repeated over and over. More than 10 GCN repeats is a diagnosis of OPMD. This can be measured using a special genetic test (bloodwork) that is available in Ontario.

How is OPMD managed?

OPMD is usually managed with a combination of methods and your management plan will be designed to meet your needs. The goal of management is to prevent or lessen your symptoms and help you retain the strength in your muscles.

Throat

- For swallowing difficulties, Botox injections can help relax the tight muscles in the throat.
- Surgery may also be an option to cut and open these tight muscles.

Eyes

- Special glasses with fine metal bars (ptosis props) can help to lift up drooping eyelids. If these are unacceptable, and the ptosis is severe, surgery may be done to lift the eyelids.

Exercise

- We provide strength monitoring and exercise advice with a DVD and exercise hand-outs.
- We recommend slowly increasing exercise tolerance starting off slowly and gradually to eventually attain 3 to 4 sessions each week of mixed endurance (such as biking, rowing, walk, jog, elliptical) and strength (such as weights, elastic bands, pilates) exercises.

Nutrition

- Your diet is also important to your treatment. Refer to Eating Well with Canada's Food Guide to make sure you have enough protein and calories in your diet.
 - We recommend that you take 2000 IU of Vitamin D.
 - A supplement of Creatine Monohydrate can also help improve strength along with exercise. Take ½ tsp (2 ml) in the morning and take ½ tsp (2 ml) in the evening with food. Do not take more than this amount. You can add it to a glass of juice or sprinkle it on cereal or yogurt.
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How will having OPMD affect my life?

Each person responds differently to OPMD. Outcome depends on the severity of your condition. Living with a chronic condition can be challenging and emotional. Here are some ideas for living well with a chronic disease.

Educate yourself

- Learn as much as you can about the disease and how to manage symptoms. If you have questions, ask your health care providers. This will help you make informed decisions about your care.

Take part in your care

- Work closely with your health care providers, as a team. Follow your treatment plan. Keep track of your symptoms and your response to treatment.

Take care of yourself

- Learn to listen to your body. Rest and conserve your energy when you feel tired. When you are active, go at your own pace.

Talk about your feelings

- It is normal to have mixed emotions when you are living with a chronic illness. At times, you may feel sad, angry or helpless. It can help to talk about your feelings with your partner, someone close to you or another person who has faced a similar situation. Remember that your health care providers can also help.