

# Chronic Progressive External Ophthalmoplegia (CPEO)

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## What is CPEO?

Chronic Progressive External Ophthalmoplegia or CPEO is the medical term for a disease that affects the muscles of the eye lids and eye ball. CPEO is progressive, which means it gradually gets worse over time.

## What causes CPEO?

CPEO is a genetic disease where there is a piece of DNA material missing (deletion) from the mitochondrial DNA. This may be inherited (passed down from one or both parents), but usually this is often caused by a spontaneous DNA mutation (85%). Spontaneous means that the deletion occurs at conception for no known reason, despite parents' healthy genes and no family history of the disease. The more rare (15%) inherited forms can either be autosomal dominant, meaning that a person with the disease has a 50% risk of passing it on to each child, or maternally inherited where all offspring could be affected.

The abnormal gene affects mitochondria, tiny structures inside almost every cell in your body. Their main job is to use the food and oxygen that enter the cells to make energy. Almost all of the energy your body needs for daily life and growth comes from mitochondria.

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## What are the characteristics of CPEO?

Each person responds differently to the disease. The most common characteristics of CPEO are:

- Droopy eyelids (ptosis). This can make the muscles of the forehead overactive, as they try and do the work of the eyelids.
- Limited eye movements. As the disease progresses, all eye movements can be lost. The eye can become fixed in the mid-position and the person has to turn his or her head to see in different directions.
- Hearing loss – especially at high sound frequencies.
- Mild muscle weakness of the arms and legs.
- Difficulty with swallowing (dysphagia).
- Cataracts.

## How do I know I have CPEO?

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

- Talking with you about your symptoms and family history.
- Checking for the physical characteristics of CPEO with a complete neurological exam.
- Reviewing the results of the following tests.

### Muscle biopsy

- A biopsy is a procedure to take a tiny sample of muscle to be examined under a microscope.
- This procedure is done in the clinic and takes about 20 minutes. The sample is usually taken from your thigh muscle (quadriceps). First the area is “frozen” or numbed with a local anesthetic. Then the doctor makes a tiny cut (incision) and removes a sample of muscle with a needle. The incision is closed with a stitch that will need to be removed in a few days.

### Blood and urine tests

- Several tests may be abnormal in patients with CPEO including amino acids (elevated alanine), lactate, CK activity, and urine organic acids.
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## Genetic tests

- Blood tests to check for mutations in the genetic material (DNA) of your chromosomes and the mitochondria's own DNA.
- CPEO usually requires that the genetic test be done on a muscle biopsy sample.

## How is CPEO treated?

There is no cure for CPEO, but treatment can control your symptoms.

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.

Some patients find that hearing aids may improve hearing. If hearing loss is severe, a special surgical procedure (cochlear implant) may be needed.

Muscle weakness is best prevented and treated with resistance exercise. The staff at the Neuromuscular and Neurometabolic Clinic can provide you with a DVD that describes the exercise program. A supplement of Creatine Monohydrate can also help improve strength in conjunction with exercise. The amount you should take is based on your weight. You can add it to a glass of juice, milk, or a smoothie – or sprinkle it on cereal, applesauce, or yogurt.

We have studied the effects of anti-oxidants such as alpha lipoic acid, coenzyme Q10, and vitamin E in a large group of individuals with various types of mitochondrial diseases. There was evidence of lower oxidative stress, but these effects were less apparent in individuals with CPEO.

If you smoke, we strongly recommend that you quit, because the mitochondria are very sensitive to smoke. There is some evidence that the anti-oxidants discussed above may actually INCREASE cancer risks in smokers, so we strongly advise against taking alpha lipoic acid, coenzyme Q10, and vitamin E if you smoke.

A Speech-Language Pathologist can help you with swallowing problems by ordering a swallowing study. Your treatment plan may include special exercises and following a diet with thickened foods.

## How will having CPEO affect my life?

The health care team at the Neuromuscular and Neurometabolic Centre can give you the help, information and support you need to live well with CPEO.

CPEO progresses slowly, and you can expect the symptoms to gradually worsen over time.

It is rare, but possible that you may pass this disease on to your children. If you want to have children, you can meet with a Genetic Counsellor for more information and advice.

**If you have any questions about CPEO,  
please speak with your doctor or nurse  
at the Neuromuscular and Neurometabolic Centre.**