Facioscapulohumeral Muscular Dystrophy

What is Facioscapulohumeral Muscular Dystrophy?

Facioscapulohumeral (FSH) Muscular Dystrophy is the medical term for a disorder of the muscles. FSH most often affects the muscles of the face, shoulders and upper arms. It causes muscle weakness and wasting. FSH is progressive, which means it gradually gets worse over time.

The term ‘facioscapulohumeral’ comes from several words in Latin:

- ‘facio’ means face
- ‘scapulo’ means shoulders
- ‘humerus’ means the upper arm

FSH is one of several types of muscular dystrophy.

What are the signs and symptoms of FSH?

Each person responds differently to this disorder. Symptoms often do not appear until age 10 to 26, but they can also appear in childhood or much later in life.

Most people have mild symptoms that slowly get worse. They are usually able to function well and the disorder does not affect their life span. Some people never develop symptoms in spite of inheriting the gene.

The most common signs of FSH are:

- Weak facial muscles, which may cause droopy eyelids, less facial expression and difficulty using muscles around the mouth (to smile, use a straw or whistle).
- Weak shoulder muscles, which may cause the shoulder blades to stick out (scapular winging), sloping shoulders and difficulty raising the arms.

As the disorder progresses, it is possible for the muscle weakness to spread to the lower legs (foot drop), and affect walking. The lower abdominal muscles are often affected, resulting in a bulge in the abdomen that resembles a hernia. There is often a “sway” in the lower back that leads to a waddling type of walk. The weakness in most of the muscles is often asymmetric.

How do you know I have FSH?

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

- talking with you about your symptoms and family history
- completing a full neurological examination
- reviewing the results of the following tests

Blood tests

- CK Level: If your muscles are breaking down (called muscle wasting or atrophy) they release an enzyme (creatine kinase) into the blood.

Genetic tests

- In over 90% of patients, this disease is due to a genetic mutation (a deletion or missing piece) in a region of the chromosome called 4q33. We refer to this as FSHD1. This can be measured using a special genetic test available in Ontario.

- Some patients do not have the usual 4q33 deletion. Most have had a chemical change (methylation) of the 4q33 region due to a mutation in a gene called SMCHD1. Testing for this gene is not currently available in Ontario. However, our team can arrange for this testing through an Ontario government program.

Electromyograph (EMG)

- This test measures the electrical activity in your muscles and nerves.

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.

How will having FSH affect my life?

Having FSH is very unlikely to change your life expectancy. Most patients live productive lives and very few ever require the use of a wheelchair.

Although there is no cure for FSH, the goal of treatment is to control your symptoms and help you have the best quality of 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 FSH.

How is FSH treated?

Exercise

Exercise is very important to keep muscles strong and remain healthy. Our team can give you a DVD that will help you with exercise therapy.

We recommend a combination of:

1. **Resistance exercise** to improve, maintain or slow the loss of muscle strength, and
2. **Endurance exercise** to lower your risk of diabetes, obesity and heart disease

Strenuous exercise is not recommended as it could worsen damage and make your muscles weaken more rapidly. Inactivity, such as bedrest, can make symptoms worse.

Physiotherapy and Occupational Therapy

Depending on your needs, a physiotherapist may help with your exercise program. Physiotherapy can also help if you have problems caused by weak muscles, such as joint contractures or shoulder and/or back pain.

A physiotherapist or occupational therapist may prescribe a brace to support weak muscles. The most common ones are ankle-foot orthotics (AFO), back and shoulder braces.

For severe muscle weakness, an occupational therapist can assess:

- your need for assistive devices such as a cane, walker or wheelchair
- the accessibility of your home

Nutrition therapy

	Research findings	Our recommendations
Vitamin D	• 80% of our patients do not have enough Vitamin D	• We recommend that all patients take 2,000 iu of Vitamin D a day.
Vitamin B12	• 13% of our patients do not have enough Vitamin B12	• If a blood test shows that you lack this vitamin, we will recommend a supplement.
Multi-vitamin	• Patients who are not very active or eat less may lack other vitamins.	• If you are inactive or not eating well, we may recommend a multivitamin.
Testosterone	• 50% adult men with muscular dystrophy have low testosterone	• If a blood test shows that you lack testosterone, we will discuss testosterone replacement.
Creatine monohydrate	• This safe, dietary supplement may increase muscle strength by about 8%.	• If you are interested, we can give you more information about this supplement and how to take it.
Anti-oxidants	• An antioxidant 'cocktail' (coenzyme Q10, alpha lipoic acid, and vitamin E) has been helpful for patients with other muscle disorders	• We will discuss the current research and whether you may benefit from taking this antioxidant cocktail twice a day (at breakfast and supper).

Can I pass this disorder to my children?

Yes, you may pass the gene for FSH to any children that you have. This is an autosomal dominant disorder, which means that you will pass the gene on to 50% of your children. However, your children may not be affected in the same way that you are.

If you want to have children, you can meet with a Genetic Counsellor for more information and advice.

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

Telephone: 905-521-7933

Notes and questions

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