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Diagnosis and treatment of facioscapulohumeral muscular dystrophy

2015 guidelines

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WHAT DID THE AUTHORS STUDY? Dr. Tawil led a committee of doctors who specialize in diagnosing and treating facioscapulohumeral muscular dystrophy (FSHD). Together, they reviewed published articles and research in FSHD and similar muscular dystrophies. They assembled detailed recommendations about the diagnosis and treatment of people with FSHD.[1]

HOW IS FSHD DIAGNOSED? The initial step to the diagnosis of FSHD is taking a careful medical history. This starts in the doctor's office. The doctor will ask many questions about the person's weakness: how it started, where it is most noticeable, how quickly it is worsening, and whether there is a family history of the same kind of problem. If there is a family history of FSHD in a first-degree relative (mom, dad, sibling), no genetic testing is required. If there is no family history or if the illness starts in an atypical way, genetic testing to look for the number of copies of the D4Z4 gene is recommended to confirm the diagnosis.

HOW SEVERE CAN FSHD GET? Patients often ask, "How bad can my FSHD get?" In short, "What should I prepare for?" One way to predict this is by counting the number of copies of D4Z4. A smaller number often means more severe illness. The earlier in life that the weakness begins often correlates with more severe disease.

WHAT ARE OTHER POSSIBLE COMPLICATIONS OF FSHD? The weakness can sometimes affect the muscles needed for breathing. Although uncommon, some people may need a device to help them breathe more easily. The weakness starts very gradually. Often, people with mild breathing problems are unaware that there is a problem. They may not feel "short of breath." Instead, because of the reduced lung function, they may have trouble sleeping well at night. A person with FSHD should have a pulmonary function test, a measure of how well the lungs are working.

Although the heart is made of muscle, it is a different kind of muscle called cardiac muscle. FSHD does not affect the heart muscle. It does not cause the heart to beat irregularly. Routine heart testing is not needed in people with FSHD. However, a person with FSHD could develop heart problems unrelated to FSHD. If a person with FSHD developed heart problems, he or she would need to see a doctor for an evaluation and treatment.

Although rare, patients with a low number of copies of D4Z4 may develop problems with their vision. They develop Coats disease, which can be detected by an ophthalmologist using special equipment called indirect ophthalmoscopy. In short, a person who has a low number of copies should be screened and evaluated for this possibility by a trained eye specialist.

Pain is common in people with FSHD. The pain occurs in the muscles and bones. It often responds to several medications and physical therapy. The neurologist often helps to choose the best combination of treatments.

HOW IS FSHD TREATED? Currently, there are no medications that can improve or stop FSHD. However, research trials are ongoing and may find a helpful medication in the future.

Although there are no medications, aerobic exercise may help. Often, the exercise routine must be tailored to the person's specific needs. Involving a physical therapist might be needed to organize exercises that will help the most.

A small number of people with FSHD experience very limited shoulder movements. This is due to the weakness of the shoulder muscles. In some patients a surgical procedure called scapular fixation can help. A detailed assessment of the shoulder weakness is needed to decide whether surgery of this type can help.

WHAT IS THE FUTURE OF FSHD? FSHD affects 1 out of every 15,000 to 20,000 people. It causes a specific pattern of muscle weakness. It is genetic: in some people, a genetic test can be helpful for diagnosis. In others, the genetic test may help to predict the severity of their disease. People with FSHD are at risk for certain complications like pain, breathing problems, and vision problems. They may need to be assessed for these complications and followed routinely. Although no medication has yet been found to reduce or stop FSHD, research is ongoing. In the meantime, treating the muscle weakness and

the complications of FSHD are critical to reaching the best quality of life for people with FSHD.

REFERENCE

1. Tawil R, Kissel JT, Heatwole C, Pandya S, Gronseth G, Benatar M. Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy: report of the Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology 2015;85:357–364.

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About facioscapulohumeral muscular dystrophy

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Facioscapulohumeral muscular dystrophy (FSHD) is a genetic illness. It is the third most common kind of muscular dystrophy. In about 70% of people with FSHD there is a family history of the same problems. In 30%, the disorder occurs "at random" or spontaneously.[1] FSHD affects 1 out of every 15,000 to 20,000 people. FSHD symptoms can occur at almost any time in life. However, in most people (more than 95%),[2] the symptoms begin in adolescence.

Although the term "facioscapulohumeral" is long, it describes the muscles that are affected. In FSHD, both sides of the body may be affected, although one side may have more weakness than the other. "Facio" means face: most often the weakness begins in the face muscles. It occurs very gradually, and at first the person may not notice the weakness.[2] Later, the upper back ("scapula," which refers to the scapula) and upper arm ("humeral" refers to the upper arm bone, which is called the humerus) muscles become weak. In some people, the lower leg may become weak as well, causing a "foot drop." In most people with FSHD, the weakness gets worse over time. It is a very gradual process, although there can be times when the weakness quickly worsens in a particular muscle. Generally speaking, the earlier in life the FSHD begins, the greater the muscle weakness. About 20% of people with FSHD become wheelchair dependent later in life.[2]

FSHD is a genetic disease. Although more than one kind of FSHD has been discovered, the problem is on chromosome 4 in all types. On this chromosome, there is a place known as the D4Z4 repeats. A person without FSHD has 11–100 of these repeats. However, a person with FSHD has only 1–10. In fact, fewer repeats means more severe disease. Those who have 1–3 repeats become weaker than those with 4–10 repeats and are more likely to become wheelchair dependent.[1,2] Furthermore, those with fewer repeats often start having problems at a younger age.[1]

DO THOSE WITH FSHD JUST HAVE MUSCLE WEAKNESS? FSHD can affect other parts of the body as well. The most common symptom is pain. About 80% of people with FSHD describe lower back pain, leg pain, shoulder pain, and neck pain. Fifteen percent of people with FSHD will develop high-frequency hearing loss. The hearing loss occurs most often in people with the fewest number of repeats. One-fourth of patients will develop a tangle of blood vessels in the back of their eye (called retinal telangiectasia). Fortunately, only a small number of people (0.6%) with retinal telangiectasia will experience visual problems.[1,2] One in 10 people (9.7%) with FSHD have an irregular heart rate. One percent to 13% have breathing problems due to the severity of their muscle weakness.

HOW IS FSHD TREATED? The goal of treatment is to improve muscle strength and function. Although several types of medications have been tried, none has been shown to help improve muscle strength. Although surgery does not improve muscle strength, in some instances shoulder surgery can help to improve shoulder function. One study showed that weight-lifting exercise does not improve strength in FSHD. However, another study suggested that low-intensity aerobic exercise might help improve strength by 17%.[1]

REFERENCES

1. Tawil R, Kissel JT, Heatwole C, Pandya S, Gronseth G, Benatar M. Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy: report of the Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine. Neurology 2015;85:357–364.
2. Tawil R, Van Der Maarel SM. Facioscapulohumeral muscular dystrophy. Muscle Nerve 2006;34:1–15.

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