New Hope for FSHD Patients

Facioscapulohumeral muscular dystrophy (FSHD) is one of the most common types of muscular dystrophy, affecting about 1 in 8,000 people. It causes weakness in the face, shoulders, arms, and legs. Over time, lifting the arms over the head and walking can become difficult, impacting daily activities.

FSHD is inherited in an autosomal dominant pattern. If one parent has the genetic mutation, each child has a 50% chance of inheriting it.

Current care

FSHD is not usually fatal, but it can cause serious challenges. Many people eventually need mobility assistance, but losing the use of the arms is often more troubling, according to Nicholas Johnson, MD, associate professor of Neurology and Human and Molecular Genetics and vice chair of research in Neurology at Virginia Commonwealth University. “We can provide support to get around, but once you lose the ability to use your arms, we’re very limited in the number of assistive devices that are available,” he says.

Breathing problems and pain — similar to fibromyalgia — are also common in FSHD. “A multidisciplinary approach in an MDA-supported clinic is important,” Dr. Johnson says.

New treatments in progress

It was only recently that scientists identified that unwinding part of chromosome 4 turns on a gene called DUX4 that is toxic to muscles. This allowed researchers to begin developing FSHD treatments targeting the root cause of the disease. For instance, Fulcrum Therapeutics’ experimental drug losmapimod, which aimed to reduce DUX4, advanced to phase 3 clinical trials. Unfortunately, the phase 3 trial results were disappointing, and Fulcrum halted development of losmapimod in 2024.

However, companies including Avidity Biosciences, Dyne Therapeutics, Arrowhead Pharmaceuticals, and miRecule are pursuing different approaches that knock down DUX4. Avidity’s phase 1/2 trial of del-brax is showing promising early results — improvements in function and strength, and a significant reduction in biomarkers.

The role of registries

Because FSHD is a slow-moving condition, long-term data is key. Registries collect information on symptoms, quality of life, and disease progression.

MDA and the FSHD Society both support the FSHD Clinical Trials Research Network, an academic network of clinicians seeking to understand the disease and prepare for therapies.

“Disease registries provide the opportunity to understand what’s important to patients and, in the event that a treatment is successful, what do we expect the long-term benefit to be,” Dr. Johnson says.

Resources

- MDA’s Facioscapulohumeral Muscular Dystrophy (FSHD) webpage
- MDA’s Facioscapulohumeral Muscular Dystrophy fact sheet
- MDA Quest Media: Simply Stated: Updates in Facioscapulohumeral muscular dystrophy (FSHD)
- MDA Virtual Learning: FSHD Updates in Research
- FSHD Clinical Trial Research Network
- FSHD Society’s Research & Clinical Care webpage