

Investigating Possible FSHD Treatments

In recent years, researchers have gained a deeper understanding of the genetic mechanisms behind facioscapulohumeral muscular dystrophy (FSHD).

“That has attracted a lot of pharmaceutical companies to start working on FSHD,” says Rabi Tawil, MD, Professor of Neurology at the University of Rochester Medical Center.

Currently, no treatments can stop FSHD from progressing, but pharmaceutical companies have begun investigating therapies that may eventually treat the condition.

Research highlights

Researchers at Fulcrum Therapeutics are investigating the experimental oral medication losmapimod in a [phase 3 trial](#). In a lab setting, it has been shown to block the abnormal expression of DUX4 protein, which is believed to cause FSHD.

“The trial is based on some evidence of either stabilization or slight improvement of patients who got the active drug in the initial phase 1/2 studies,” Dr. Tawil says. “The big question on the part of the patients is: Will this stop progression, or stop progression and make me stronger? We won’t know until the trials are done.”

RNA-based treatments used to treat myotonic dystrophy are also being studied for their ability to block DUX4 production in patients with FSHD. A [phase 1/2 trial](#) of AOC 1020, an RNA-based intravenous treatment from Avidity Biosciences, will begin soon.

Roche Pharmaceuticals will soon launch a [phase 2 trial](#) to investigate RO7204239, a subcutaneous myostatin inhibitor, which may improve muscle growth.

Building better clinical trials

While telehealth can be an essential tool to increase access to healthcare, it does have limitations, especially when it comes to physical examinations.

When pharmaceutical companies launch clinical trials for FSHD, they rely on the data collected by the ReSolve study, the largest FSHD natural history study, conducted by the [FSHD Clinical Trial Research Network \(CTRN\)](#), composed of 12 American and 3 European sites that Dr. Tawil developed with Jeffrey Statland, MD, at the University of Kansas Medical Center. The study was intended to facilitate the conduct of clinical trials in FSHD.

“The data from the ReSolve study is now being used by multiple pharmaceutical companies in planning upcoming FSHD clinical trials,” Dr. Tawil says.

The network’s goal is to make new FSHD therapies available sooner. This aligns with MDA’s research goals, which include accelerating the delivery of treatments and cures to people with muscular dystrophy and other neuromuscular diseases.