FRED HUTCH
CURES START HERE®

BIOMARKERS FOR FSHD

Gene Expression Biomarkers to Track Facioscapulohumeral Dystrophy Disease Progression

Brief Description of Technology

DUX4-dependent gene expression is a major molecular signature of Facioscapulohumeral dystrophy (FSHD).

BUSINESS OPPORTUNITY
Exclusive license
Non-exclusive license

TECHNOLOGY TYPE
Diagnostic
Nucleic acid
Orphan
Epigenetic

STAGE OF DEVELOPMENT
Preclinical *in vitro*
Clinical validation

PATENT INFORMATION
EP 3119909

INVESTIGATOR
Stephen Tapscott, MD, PhD
Human Biology and Clinical Research Divisions

LEARN MORE
Tech ID: 14-029
partnering@fredhutch.org
206-667-4304

Technology Overview

Facioscapulohumeral dystrophy (FSHD) is a human muscular dystrophy that initially affects the muscles of the face and upper extremities but can progress to affect most skeletal muscles. Despite genetic evidence suggesting that DUX4 mRNA expression is necessary for FSHD, due to its low abundance in muscle biopsies, its critical role in disease pathogenesis has been questioned. Researchers in the Tapscott lab have studied biopsies from FSHD affected and normal patients and have identified a panel of DUX4-dependent biomarkers that associate with clinical severity. These biomarkers could be used to monitor patients and also to assess the efficaciousness of new therapeutic entities that are currently in the clinic for FSHD.

Applications

- Monitor disease progression of FSHD
- Stratify patient populations for clinical studies

Advantages

- Four genes as a core set of the biomarker panel
- Biomarker panel can be detected in FSHD1 and FSHD2 patient cohorts
- Identifies patients prior to the manifestation of clinical symptoms

Market Overview

The estimated prevalence for FSHD is about 1 in 15,000 to 1 in 20,000 people with approximately 95% of the cases being Type 1 FSHD. Currently, there are no approved therapeutics to treat FSHD and no clinical endpoint to serve as an optimal biomarker in clinical trials. Indicators commonly used to test clinical utility of an investigational therapeutic are MRI, motor function tests, quantitative muscle testing, and quality of life index questionnaires. Many recent clinical trials for FSHD therapeutics have failed on their primary endpoints. Thus, there is an unmet need for molecular diagnostic biomarkers that can provide insights to disease progression before the manifestations of clinical symptoms and allow for patient monitoring during clinical trials.