

Biomarker-Based Diagnostic Tools for Facioscapulohumeral Muscular Dystrophy (FSHD)

A diagnostic method that uses specific immune system markers in blood and other biological samples to detect and monitor Facioscapulohumeral Muscular Dystrophy (FSHD), a progressive muscle-wasting disease.

What is the Problem?

Facioscapulohumeral Muscular Dystrophy (FSHD) is one of the most common forms of muscular dystrophy, yet it remains difficult to diagnose and monitor. FSHD is a progressive muscle-wasting disorder caused by a genetic mutation that leads to abnormal expression of the DUX4 gene in adult muscle tissue. It primarily affects the face, shoulders, and upper arms, with symptoms usually beginning before the age of 20. Current diagnostic methods often rely on genetic testing and clinical evaluation, which can be time-consuming, expensive, and inconclusive in early stages. There is a lack of accessible, non-invasive tools that can reliably detect disease presence or progression. This creates challenges for both clinical management and the development of new therapies.

What is the Solution?

This technology offers a method to detect FSHD using biomarkers found in blood and other bodily fluids. The approach focuses on measuring levels of neutrophil extracellular traps (NETs)—a type of immune system activity—as well as mitochondrial proteins that are elevated in individuals with FSHD. These biomarkers can be detected using a multiplexed antibody Enzyme-Linked Immunosorbent Assay (ELISA), offering a less invasive and more scalable alternative to genetic testing. The method can be applied to a variety of sample types, including blood plasma, saliva, and spinal fluid, making it adaptable to different clinical settings.

What is the Competitive Advantage?

Non-invasive and accessible: Uses blood or saliva samples instead of muscle biopsies or complex genetic tests.

Early detection potential: Biomarkers may indicate disease activity before clinical symptoms become apparent.

Scalable for clinical and research use: Compatible with standard lab assays, enabling broader adoption.

Technology ID

BDP 8808

Category

Selection of Available
Technologies
Diagnostic

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Supports therapeutic development: May serve as a measurable indicator of disease activity for use in clinical trials.

Applicable to multiple sample types: Could offer flexibility for patient monitoring and research applications.

Patent Information:

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