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MedGenome Labs unveils FSHD1 genetic screening test for rare disease diagnosis

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Bengaluru-based MedGenome Labs has announced the launch of a ground-breaking first-ever Facioscapulohumeral Muscular Dystrophy Type 1 (FSHD1) test in India. MedGenome Labs is the first commercial lab to offer this new genetic test that will help individuals with FSHD1 to get an early and accurate diagnosis, leading to improved disease management options.

The FSHD1 Optical Genome Mapping Test (OGM) is a highly advanced diagnostic tool that uses cutting-edge technology to provide a comprehensive and accurate assessment of genetic changes in patients with FSHD1. The test can detect large-scale insertions and duplications, as well as more subtle changes in DNA that can cause FSHD1.

Facioscapulohumeral Muscular Dystrophy (FSHD) is a common form of muscular dystrophy with an extremely complex genotype. It is progressive myopathy which accounts for 2 to 3% of the muscular dystrophy cases in India. FSHD1 is a rare genetic disorder that affects the muscles of the face, shoulder blades, and upper arms. The disease is caused by the deletion of genetic material from the D4Z4 repeat region on chromosome 4, leading to the loss of muscle tissue and weakness in affected areas.

FSHD1 affects approximately 1 in 20,000 individuals worldwide, with symptoms typically starting in early adulthood. FSHD1 can have a significant impact on quality of life, as it can result in difficulty with daily activities such as lifting objects or raising arms. The disease is also progressive and can lead to severe disability in some cases. In the present day, there is no known cure for FSHD1, and treatment options are limited. However, many clinical trials are being conducted at an accelerated rate.