

# PAIN & DISABILITY<sup>SM</sup>

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## Muscular Dystrophy - GENERAL

### Disease characteristics:

Fascioscapulohumeral muscular dystrophy (FSHD) typically presents before age 20 years with weakness of the facial muscles and the stabilizers of the scapula or the dorsiflexors of the foot.

### Genetic counseling:

FSHD is inherited in an autosomal dominant manner. Offspring of an affected individual have a 50% chance of inheriting the mutant allele.

### Clinical Diagnosis:

The diagnosis of FSHD is suspected in the presence of bilateral facial weakness and weakness of either the scapular stabilizers and/or foot dorsiflexors and the absence of: a) ptosis, b) extraocular muscle weakness, c) sensory loss, d) skin rash, e) neurogenic changes on muscle biopsy, and f) myotonia, fasciculations, or neurogenic potentials on EMG.

### Treatment:

- Physical Medicine and Rehabilitation
- Surgical Management