



POSNA

The Core Curriculum

Fascioscapulothoracic dystrophies

Objectives

1. Describe presenting symptoms and natural history of fascioscapulothoracic dystrophy
2. Describe the genetic transmission of fascioscapulothoracic dystrophy
3. Discuss useful orthopaedic intervention for fascioscapulothoracic dystrophy

Discussion

Fascioscapulothoracic dystrophy (FSTD) is an unusual form of muscular dystrophy which was one of the types described by Walton and Nattrass in their classic 1954 work on the muscular dystrophies. It is transmitted as an autosomal dominant with significantly higher penetrance in males than in females. The genetic defect was located in 1990 located on chromosome 4, and it has been documented that in normal individuals, genetic probing detects an EcoRI fragment > 35 Kb, while in FSTD families, a shorter fragment, usually between 14-35 Kb is found. FSTD affects the facial and shoulder girdle musculature in a highly variable fashion, both in age of onset and severity of disease. The deltoid is spared. Some individuals with the condition can only be detected with careful exam techniques, while about 10% become wheelchair bound. About 85% are affected by age 20. In a recent study, the penetrance was 95% for males and 69% for females.

The major orthopaedic problem with FSTD is weakness of shoulder abduction and flexion. Posterior scapulothoracic fusion stabilizes the scapula, and affords better mechanical advantage to the unaffected rotator cuff and deltoid.

References

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