



POSNA

The Core Curriculum

Limb-girdle muscular dystrophy

Objectives

1. Describe clinical symptoms, mode of genetic transmission, and age of onset of limb-girdle muscular dystrophy
2. Describe the usual patterns of weakness in the limb-girdle dystrophies

Discussion point

1. Why is there so much research interest in this type of muscular dystrophy?

Discussion

If one can understand the limb-girdle dystrophies, one can understand all the dystrophies. Walton, who devised the original classification of dystrophies, felt at the outset that the limb-girdle dystrophies were a heterogeneous group even though characterized by proximal weakness. However, the distribution of weakness in the limb-girdle dystrophies may be very similar to that observed in Duchenne or Becker dystrophy, management is thus similar. Testing for the dystrophin gene or protein has become a routine part of the evaluation of patients with progressive proximal weakness, elevated serum creatine phosphokinase, and biopsy evidence of a dystrophic process. Patients with no dystrophin abnormalities are assumed to have an autosomal recessive type of dystrophy, in most cases a limb-girdle dystrophy. There are a number of glycoproteins that span the muscle cell membrane, which are now known as sarcoglycans. As more sarcoglycans were discovered, a problem arose with nomenclature. An international congress in 1995 deemed that the rare, mild autosomal dominant type of limb-girdle dystrophy would be called limb-girdle muscular dystrophy 1 (LGMD1) and the much more common autosomal recessive types of this dystrophy would be LGMD2. LGMD2 has been further subdivided into LGMD2a, LGMD2b, etc depending on the chronologic discovery of the location of the responsible gene. Duggan et al discovered that some patients with a primary sarcoglycinopathy had a phenotype indistinguishable from Duchenne dystrophy, although all patients with sarcoglycinopathies had normal cognition. Study of the glycoproteins of the cell membrane has shed better understanding of the complex molecular biology of this structure, which may be the clue to developing a better treatment than what is presently available.

References

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