Condition: Duchenne/Becker muscular dystrophy

Inheritance:

Duchenne/Becker muscular dystrophy

Genetic etiology:

Mutation in *DMD* gene on X chromosome, which encodes the muscle membrane protein dystrophin. Duchenne muscular dystrophy results from mutations that lead to lack of dystrophin production, whereas Becker dystrophy results from mutations that lead to deficient or defective dystrophin. Most mutations are deletions that juxtapose out-of-frame (Duchenne) or in-frame (Becker) exons. Other mutations include duplications, point mutations, and splicing mutations.

Frequency:

Duchenne: approximately 1/5,000 males; Becker: approximately 1/18,000 males.

Clinical features:

Both Duchenne and Becker dystrophy are characterized by progressive degeneration of skeletal muscle cells, leading to atrophy and weakness. There is often an associated cardiomyopathy and there may be cognitive dysfunction. Duchenne presents in the early years of life, with most affected boys being wheelchair-bound by the teen years, and most not surviving beyond the second decade due to cardiopulmonary insufficiency. The age of onset and rate of progression of Becker dystrophy tends to be more variable, but usually is later and slower.

Management:

Supportive care; treatment with steroids used to improve muscle strength.

Genetic counseling:

Carrier females transmit the disorder to half their sons; 2/3 of mothers of sporadically-affected males with Duchenne dystrophy are carriers, often of a new mutation allele.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.