Muscular Dystrophies – Quick Proff-Oriented Notes

Definition

- Inherited group of myopathies
- Progressive muscle weakness
- Caused by defects in muscle structural proteins

Duchenne Muscular Dystrophy (DMD)

- Most common and most severe
- X-linked recessive, dystrophin gene (Xp21)
- Frameshift mutation → absent dystrophin
- Onset < 5 years
- Gowers' sign, calf pseudohypertrophy
- Very high CK (10–100x)
- Death due to cardiomyopathy or respiratory failure

Becker Muscular Dystrophy (BMD)

- X-linked recessive
- Non-frameshift mutation → reduced dystrophin
- Onset in adolescence or adulthood
- Milder course than DMD
- CK elevated but less than DMD

Limb Girdle Muscular Dystrophy (LGMD)

- Autosomal dominant or recessive
- Shoulder and pelvic girdle involvement
- Difficulty climbing stairs or lifting arms
- No facial muscle involvement

Facioscapulohumeral Dystrophy (FSHD)

- Autosomal dominant
- Chromosome 4q35
- Facial weakness, scapular winging
- Normal life span

Myotonic Dystrophy

- Most common adult muscular dystrophy
- Autosomal dominant (CTG repeat expansion)
- Myotonia, distal muscle weakness
- Cataracts, cardiac conduction defects, endocrine abnormalities

Important Exam One-Liners

- Most common childhood MD: Duchenne
- Most common adult MD: Myotonic dystrophy
- Pseudohypertrophy of calves: Duchenne
- Facial weakness + scapular winging: FSHD
- X-linked MD: Duchenne, Becker