MUSCULAR DYSTROPHY

DYSTROPHY – Degeneration of tissue due to disease or malnutrition

- inherited disorders - x-linked recessive
- progressive muscle weakening and wasting (often eventual replacement by fibrous tissue and fat)
- dilated cardiomyopathy and respiratory failure (due to weakness of muscles of respiration and kyphoscoliosis) can occur by late teens
- Duchenne and Becker muscular dystrophies are caused by mutations in the dystrophin gene, which codes for a large sarcolemmal protein essential for muscle membrane structure and functional integrity. In Duchenne muscular dystrophy, no dystrophin is produced. In Becker muscular dystrophy, there is abnormal dystrophin.

DUCHENNE MUSCULAR DYSTROPHY

KEY CLUES:

Boys < 5 years old
Progressive symmetrical weakness
Delayed motor milestones
Gower's sign (weakness of hip and knee extension)
Waddling gait (weakness of hip abductors)
Accentuated lumbar lordosis (weakness of hip extensors)

- one of most common and severe muscular dystrophies
- 1/3 are due to de novo mutations
- affects boys in early childhood (<5 years) - NOTE: 10% of female carriers manifest symptoms due to skewed X-chromosome inactivation

- Presents with:
  - Progressive symmetrical weakness
  - Wasting of predominantly proximal muscles; spreading distally
  - Calf pseudohypertrophy and Achilles tendon contractures are common
  - Delayed motor milestones
  - Usually wheelchair-dependent by 13 years.
  - 20% have cognitive impairment

- life expectancy 20-30 years

BECKER MUSCULAR DYSTROPHY

- closely linked with and very similar to Duchenne muscular dystrophy, but more mild (most patients are able to walk through teenage years and early adult life, sometimes for much longer)
- later onset than Duchenne muscular dystrophy
- life expectancy better than Duchenne muscular dystrophy