Pediatrics Neuromuscular Disorders

Topics to Cover today

- Duchenne Muscular Dystrophy (DMD)
- Myasthenia Gravis (MG)
- Floppy Infant
- Spinal Muscular Atrophy (SMA)
- Guillain-Barre Syndrome (GBS)
- Bell's Palsy

Duchenne Muscular Dystrophy (DMD)

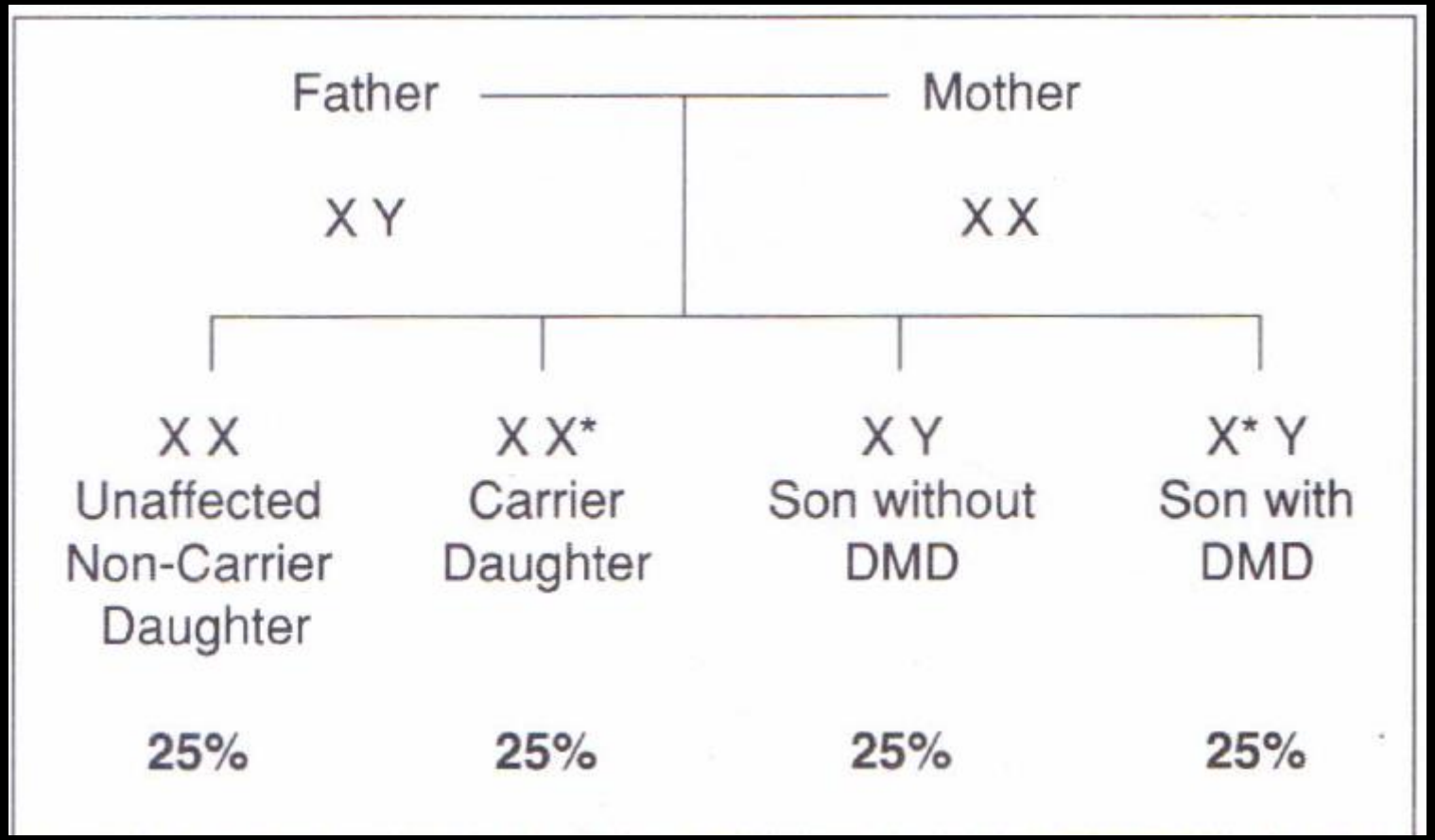
DMD-Induction

- It is inherited X-linked recessive condition. 1/3rd are new mutations.
- These patients do not produce dystrophin.
- Dystrophin is a protein localized to muscle cell membrane.

Cont….

- Incidence is 1:3500 male births.
- Female carriers are usually asymptomatic.

Duchenne Muscular Dystrophy Mode of Inheritance



DMD-Clinical Features

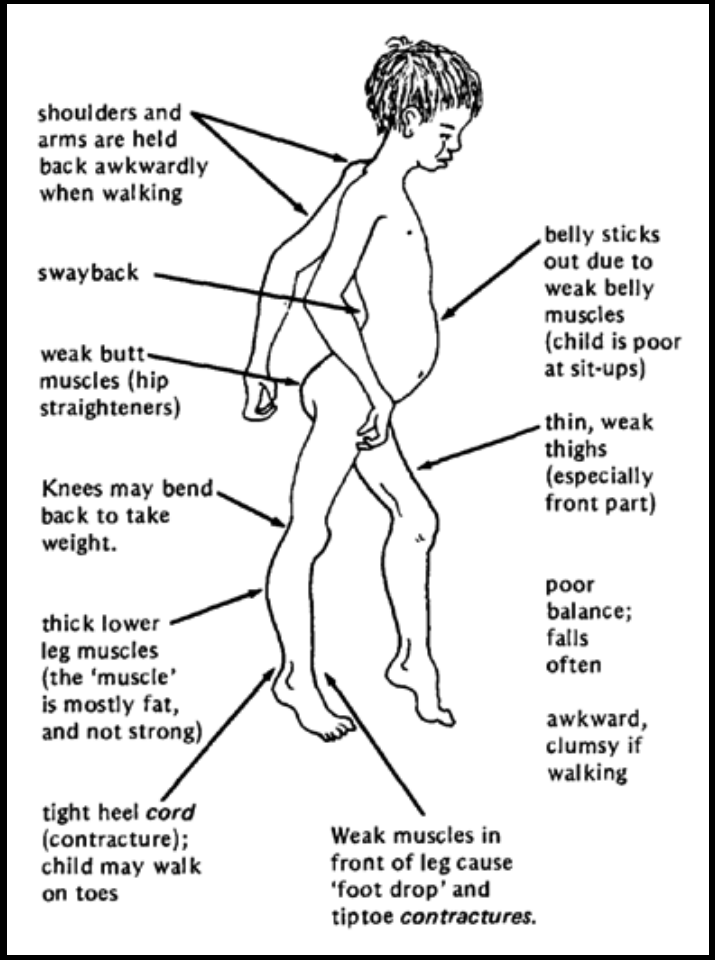
- Onset is in early years. There is delay in motor milestones. Typically, boys lose ability to walk between 8 and 12 years.

- There is difficulty in climbing stairs, and lordosis with waddling gait.

Cont…

- There is pseudo-hypertrophy of calves and progressive muscular weakness.
- Respiratory failure occurs.

Duchenne Muscular Dystrophy Clinical Features



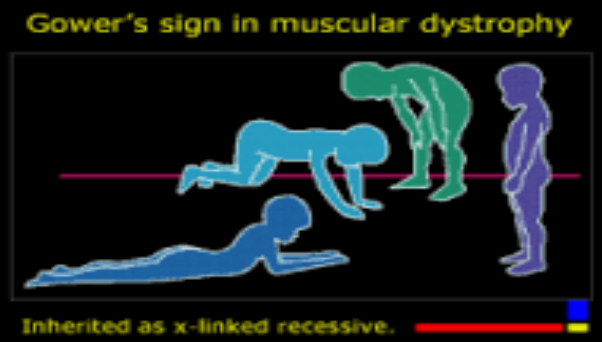
Duchenne Muscular Dystrophy
Pseudo-Hypertrophy

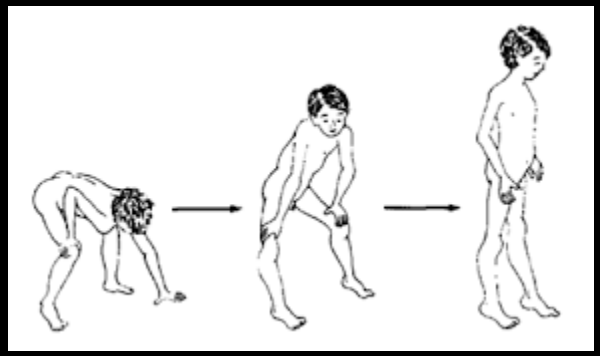
Duchenne Muscular Dystrophy

Diagnosis:

- High plasma creatine kinase (> 5000iu/L).
- Muscle biopsy shows absent dystrophin.
- Positive Gower's sign

Duchenne Muscular Dystrophy
Gower's sign





Treatment:

- There is no curative treatment for Duchenne muscular dystrophy.
- Physiotherapy and appropriate seating is needed.
- Steroids have some transient role.