Muscular Dystrophy
↓ Difficult ↘ Nourished
Muscle weakness (Skeletal)
→ Not in nerve or neuromuscular junction.
D/D Myasthenia etc.
→ It is a inherited disorder caused by genetic mutation, Progressive, degenerative.
① Dystrophinopathies (Mutation in Dystrophin gene).
X linked recessive → 1) Duchenne muscular dystrophy (P(21.2)
2) Becker muscular dystrophy. (Short arm 21)
② Other Gene Mutation (Protein complex with Dystrophin protein.
1) Autosomal dominant
i) Myotonic dystrophy DM1
ii) Proximal myotonic dystrophy DM2.
iii) Fascioscapulear humoral.
iv) Oculopharyngeal dystrophy.
② ~~X-linked~~ recessive Many gene mutation
→ Limb girdle (Same like Dystrophinopathy)
③ X linked recessive.
→ Emery - Dreifuss Syndrome
(Mutation in Emerin gene).
↳ Present in nuclear membr

Dystrophinopathis
(Dystrophin gene mutation)
↓
Which stabilizes the muscle cell membrane
↓ → Mother carrier
X linked recessive

↓
Loss of Dystrophin
↓
Duchenne muscular dystrophy

↓
Misshapen Dystrophin
↓
Beckers Muscular dystrophy

Pathophysiology

No dystrophin protein
↓
Sarcolemma breaks → Leaks Creatine phosphokinase and Intake more Calcium
↓
Cell death By proteolysis
↓
Hypertrophy by fat & nerves
↓
Muscle atrophy ← Skeletal

Clinical Features

→ Walk later in childhood.
→ Waddling gait → like a duck → Gluteal muscle
→ Calf Pseudohypertrophy enlarged by fat and fibrosis not muscle sign.
→ Gowers sign ⊕ Use arms to help stand up from lying flat; floor → ankle, leg, thigh, Back
Positive
→ Respiratory failure → Weak diaphragm.
→ Scoliosis, Lordosis → Due to trunk muscle
→ Cardiomyopathy and arrhythmia
→ Duchenne → < 5 years, so early death
Becker → Late adulthood; no transfer to offsprings; 5th decade

Investigation

- Electromyocardiography ↓ EMG
⇒ Muscle Biopsy ⇒ Dystrophin ↓ or absent.
⇒ Genotyping ⇒ Genetic testing.
⇒ ↑ Creatine Phosphokinase. CK-MB ↑
⇒ ABG Gas, Echomyocardiography

Complication

→ Foot drop ⇒ If anterior compartment.
Toe walking ⇒ If posterior compartement.
Knee Bent Back.
Ocular muscle → Diplopia.
Pharynx → Dysphagia. / Mental retardation.