Proximal weakness
Characteristics of muscle disorders

- Muscle disorders cause a LMN picture with low tone, decreased reflexes and atrophy.
- Muscle disorders cause proximal weakness, neuropathies distal weakness.
- CK levels are usually elevated in muscle disorders; the more active a myopathy the higher the CK levels.
Classification

- Inflammatory muscle disorders
  - Polymyositis
  - Dermatomyositis
  - Inclusion body myositis

- Muscle dystrophy
  - Duchenne/Becker
  - Facioscapulo humeral (FSH)
  - Limb-girdle
  - Oculopharyngeal
  - Myotonia dystrophica
  - Congenital

- Metabolic myopathies
  - Thyroid
  - Steroid
  - Storage disease
  - Carnitine deficiency
  - Mitochondrial

- Congenital myopathies
  - Central core
  - Nemaline rod
  - Centro nuclear, etc
INFLAMMATORY MYOPATHIES

- **POLYMYOSITIS**
  - Sub acute symmetrical proximal weakness
  - Develops over weeks to months
  - ♀ > ♂, 30-60 years (15 years)
  - **Clinical picture:** Proximal weakness
  - Neck weakness
  - Dysphagia/dysphonia
  - Muscle tenderness not on the foreground
  - **Dx:** ↑ CK
  - EMG
  - Muscle biopsy
  - **Etiology:** ? Autoimmune
  - T-cell disorder
  - **RX:** High dose Prednisone
  - Immunosuppression
NORMAL MUSCLE: H&E STAIN

- Polygonal shape
- Dark nuclei, peripherally

Overall assessment:
• Inflammatory infiltrates
• Necrosis, regeneration
• Atrophy, splitting
• Vessels, connective tissue
Polymyositis biopsy
Inflammatory myopathies

- **DERMATOMYOSITIS**
  - Weakness as in PM, with dermatological involvement.
  - ♀ > ♂, kids and adults
  - Clinical: Skin rash first, heliotropic, edema (eyes/mouth), elbows, knuckles, knees; Calcifications (esp. in kids); weakness as in PM.
  - **DX:** As in PM
  - **Etiology:** B-cell disorder
  - **RX:** Steroids
Dermatomyositis
Dermatomyositis
INCLUSION BODY MYOSITIS

- Slow progression.
- ♂ > ♀; . 50 years
- **Clinical:** Legs affected first with proximal weakness, CK normal or (↑)
- **Dx:** Filament like inclusion bodies on biopsy
- **Etiology:** ? T-cell disorder
- ?? Virus
- **RX:** None at present
MUSCLE DYSTROPHIES

- Progressive hereditary degenerative muscle disorder.

**DUCHENNE/BECKER:**

- X-linked; Xp 21 dystrophin gene abnormality.
- Duchenne starts at ± 3 years; Becker ± 11 years.

**Clinical picture:** Falls easily.
- Hip muscle often first: waddling gait.
- Pretibial muscle weak: toe-walking.
- Later pecs and upper limbs weak.
- Pseudohypertrophy: Calves, sometimes deltoids and quads.
- Duchenne: Cardiac involvement and mental retardation; die in adolescence.
- Becker: Benign course; wheelchair by 30 years.

**Dx:** Clinical; high CK’s, EMG active myopathy; muscle biopsy.
Duchenne MD
Muscle Dystrophies

- **FSHD (Facioscapulohumeral)**
  - Slowly progressive dystrophy involving face and shoulders predominantly.
  - Inherited in AD mode; starts before 20 years; chromosome 4.
  - **Clinical picture:**
    - Inability to elevate arms.
    - Winging of scapulae.
    - Orbicularis oculi and oris weak (can’t close eyes tightly; can’t whistle).
    - Brachioradial muscle atrophy.
    - Later: Hip weakness/ankle weakness.
  - **Dx:** Clinical; CK not very high; biopsy; genetics
Muscle Dystrophies

- **OCULOPHARYNGEAL**
- AD inherited; starts after 45 years.
- Ptosis, dysphagia/proximal weakness.
- **Dx:** Clinical; genetics;
- **NB:** Differentiate from Myasthenia gravis
MYOTONIA DYSTROPHICA

- Muscle atrophy, myotonia, dystrophic changes in other tissues.
- AD, chromosome 19.
- Clinical picture: “Only” muscle disorder with distal weakness.
- Small muscle atrophy of the hands; weak forearm extensors.
- Ptosis, masseter atrophy, sternocleidomastoid weak, foot drop.
- Associated: Frontal hair loss; pharynx, larynx weak; prolonged PR time, testicular atrophy, cataracts, mental retardation.
- Myotonia: Thumb, forearm and tongue.
METABOLIC MYOPATHIES

- **MITOCHONDRIAL:**
  - Muscle disorder as a result of abnormal mitochondria – energy deficiency.
  - Ragged red fibres on muscle biopsy.
  - **Examples:** Kearn-Sayre syndrome with progressive external ophthalmopathy, pigment retinopathy and heart block.