## Proximal weakness



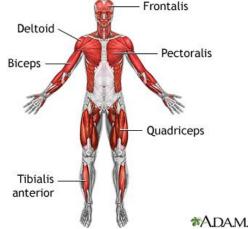




 Muscle disorders cause proximal weakness, neuropathies distal weakness.

 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
- 9 > 3, 30-60 years (15 years)
- Clinical picture: Proximal weakness
- Neck weakness
- Dysphagia/dysphonia
- Muscle tenderness not on the foreground
- Dx:

**↑** CK

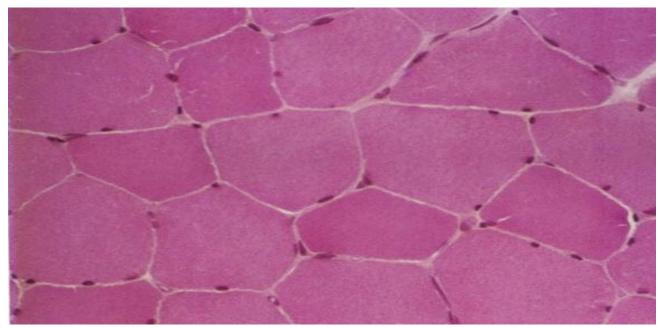
- EMG
- Muscle biopsy
- Etiology: ? Autoimmune
- T-cell disorder
- RX: High dose Prednisone
- Immunosuppression



#### **NORMAL MUSCLE: H&E STAIN**



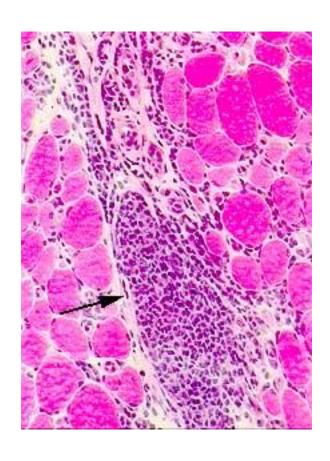
- Polygonal shape
- Dark nuclei, peripherally



### Overall assessment:

- Inflammatory infiltrates
- Necrosis, regeneration
- Atrophy, splitting
- Vessels, connective tissue

# Polymyositis biopsy



# Inflammatory myopathies

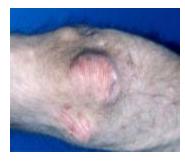
### DERMATOMYOSITIS

- Weakness as in PM, with dermatological involvement.
- $\Rightarrow$   $\Rightarrow$   $\Rightarrow$   $\Rightarrow$  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

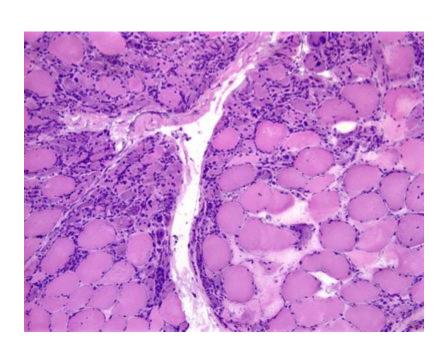








# Dermatomyositis









### INCLUSION BODY MYOSITIS

- Slow progression.
- $\emptyset$  >  $\emptyset$ ; . 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

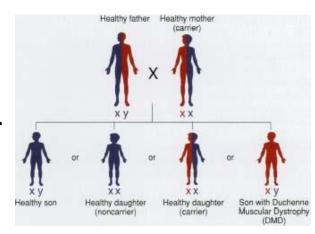


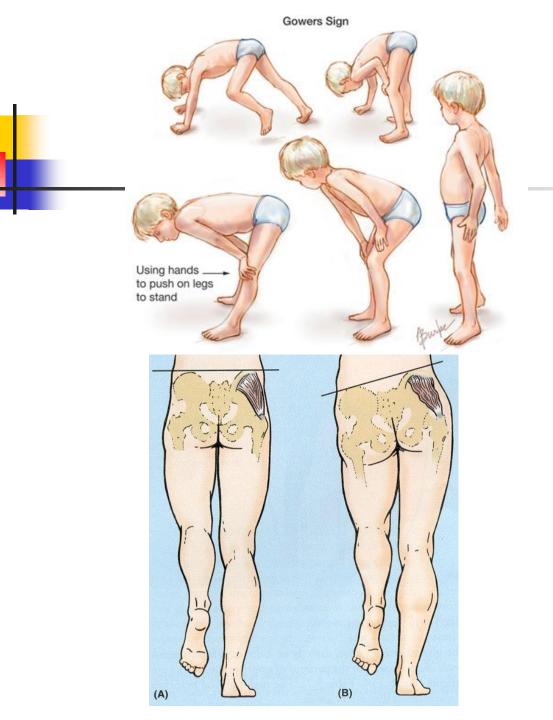
### MUSCLE DYSTROPHIES

Progressive hereditary degenerative muscle disorder.

#### DUCHENNE/BECKER:

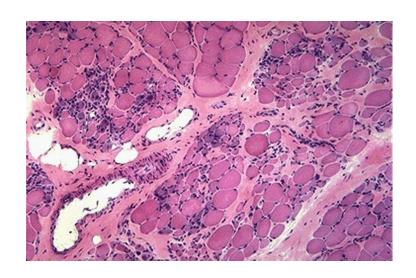
- X-linked; Xp 21 dystrophin gene abnormality.
- Duchenne starts at ± 3 years; Becker ± 11 years.
- Clinical picture: Falls easily.
- Hip muscle often first: waddling gait.
- Pretibial muscle weak: toe-walking.
- Later pecs and upper limbs weak.
- Pseudohypertrophy: Calves, sometimes deltoids and quads.
- 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.

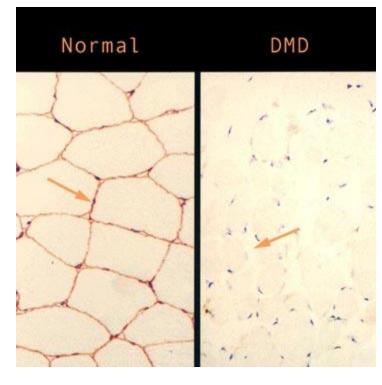






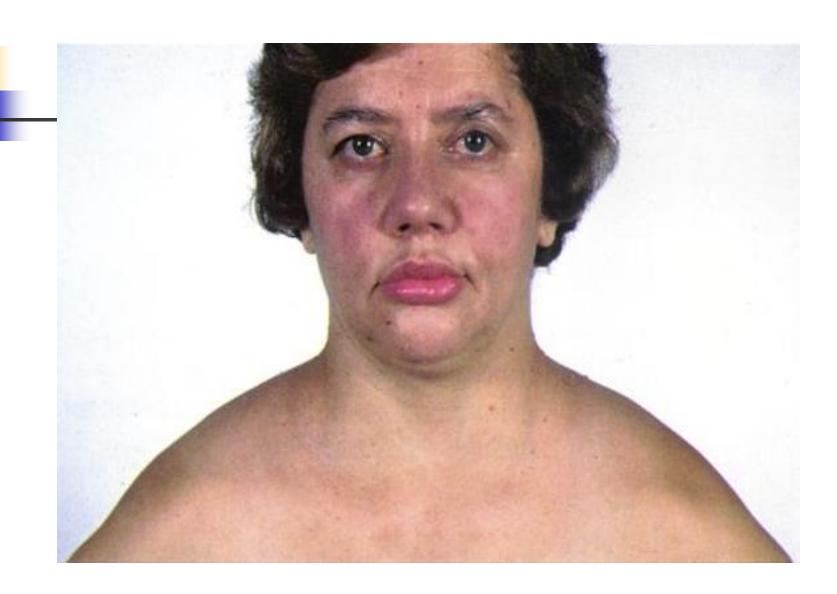
## **Duchenne MD**





# 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.
- <u>Dx:</u> Clinical; CK not very high; biopsy; genetics





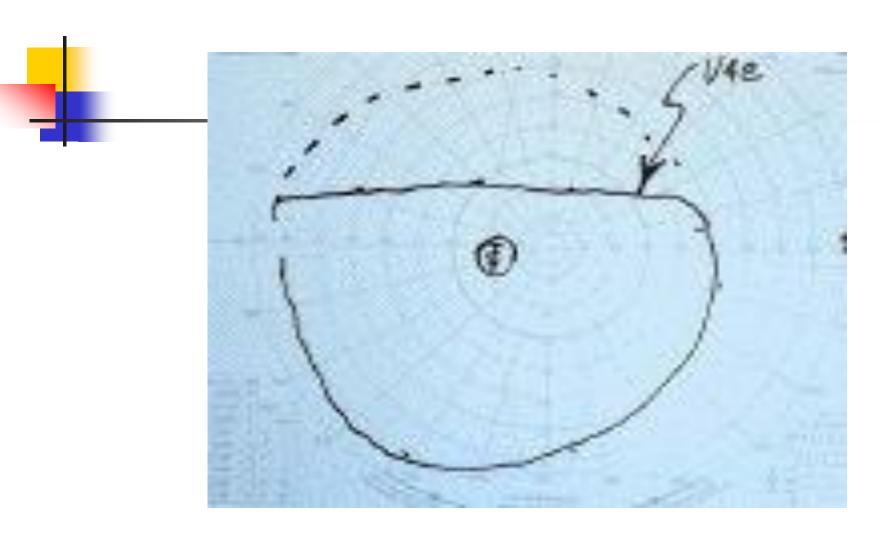


## Muscle Dystrophies

### OCULOPHARYNGEAL

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





## MYOTONIA DYSTROPHICA

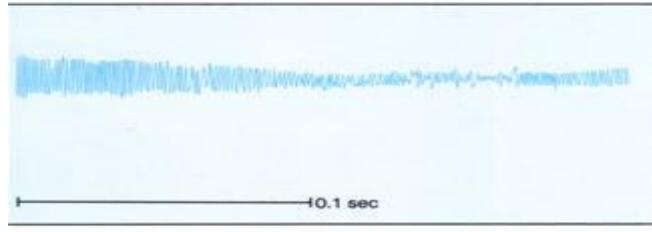
- Muscle atrophy, myotonia, dystrophic changes in other tissues.
- AD, chromosome 19.
- Clinical picture: "Only" muscle disorder with <u>distal</u> weakness.
- Small muscle atrophy of the hands; weak forearm extensors.
- Ptosis, masseter atrophy, sternocleidomastoid weak, foot drop.
- Associated: Frontal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





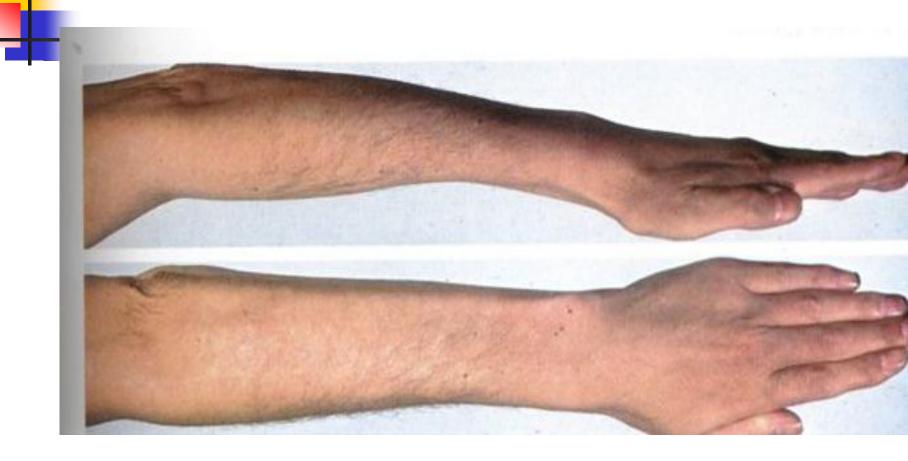














## METABOLIC MYOPATHIES

### MITOCHONDRIAL:

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

