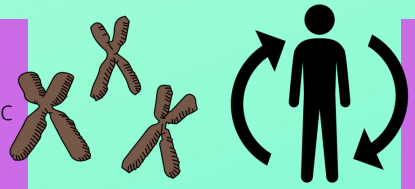
MYOPATHIES

Primary disease of muscle- classically symmetrical, proximal weakness. Genetic diagnoses relatively common.

Genetic

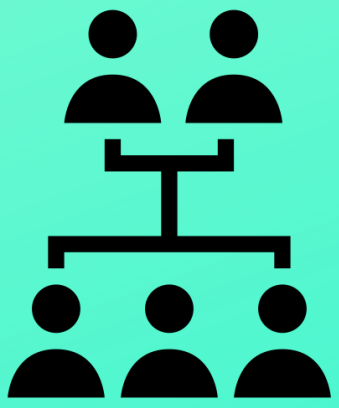


Muscular Dystrophies- degenerative genetic myopathies. Present at any age.

Duchenne Muscular Dystrophy- X-linked recessive dystrophinopathy (mutation in **dystrophin** gene)- therefore affects males more commonly. Commonest childhood muscular dystrophy. Onset by **age 5** (often delay in walking), usually unable to walk by 10. Die prematurely from **respiratory failure**. Signs= waddling gait, **calf pseudohypertrophy** (fat and fibrosis), **Gower's sign**, dilated cardiomyopathy, may have intellectual impairment. High CK, muscle biopsy, test for dystrophin mutation. **Steroids** slow degeneration, physical therapy improves QoL.

Becker Muscular Dystrophy -another **dystrophinopathy** (X-linked recessive) but less severe mutation>> **milder** disease. Later onset by **age 10-20**. Same ix and treatment as DMD.

Myotonic Dystrophy (DM1)- Commonest **adult** mucular dystrophy (age 20-30). **Autosomal dominant trinucelotide repeat** (CTG) in type DM1. SK, smooth and cardiac muscle affected. **Distal muscle wasting and weakness**, myotonic facies, frontal **balding**, bilateral ptosis, **cataracts**, **myotonia**, gonadal atrophy, mental impairment, cardiac involvement (heart block, cardiomyopathy).



Facioscapulohumeral Dystrophy

Metabolic

Due to **deficiencies in enzymes** involved in the use of carbohydrate, lipid, or mitochondrial phosphorylation. **Muscle weakness**, **myoglobinuria, rhabdomyolysis**.

McArdle's Disease- Autosomal recessive glycogen storage disease. Deficiency of **myophosphorylase**>> decreased muscle glycogenolysis>>painful muscle cramps and myoglobinuria after exercise, low lactate during exercise.

Pompe's Disease- Autosomal recessive alpha-glucosidase deficiency. Young onset myopathy, early resp involvement. Enzyme **replacement** therapy.

Mitochondrial Myopathies- due to mutations in mtDNA or nuclear genome coding mt proteins.

Inflammatory

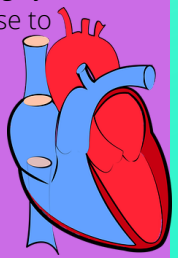
Polymyositis- subacute, proximal, symmetrical muscle weakness/myalgia. Often assc with **systemic autoimmune conds** and **autoantibodies**. Raised CK. Muscle biopsy diagnoses. **Prednisolone** +/- steroid-sparing agent.

Dermatomyositis- adults and children. Proximal and **resp muscle** weakness, dysphagia plus **skin features**: photosensitive, **heliotrope** rash periorbitally, erythematous rash face and trunk, **Gottron's** papules. Assc with **malignancy**- investigated for.

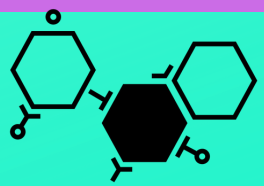
Inclusion Body Myositis- Most common **acquired** myopathy. After age 50. More common in **males**. Slowly progressive asymmetrical weakness. Especially deep finger flexor (affecting grip), quadriceps and dysphagia. **No response to immunosuppression.**

Acquired Myopathies and Ix

Endocrine and **drug-induced** toxicity are most common. Muscle involvement in hyperthyroidism and hypothyroidism (inc TED). Also in acromegaly and DM. **Statins** can give myopathy as a S/E- from mild myalgia and CK rise to rhabdomyolysis.



Genetic Tests
Bloods- CK
ECG- cardiomyopathy, arrhythmias
EMG- may show abnormal motor unit potentials
Muscle MRI- guide biopsy, identify other muscles involved
Muscle Biopsy- establish diagnosis. Partially involved muscle used.



Soton Brain Hub