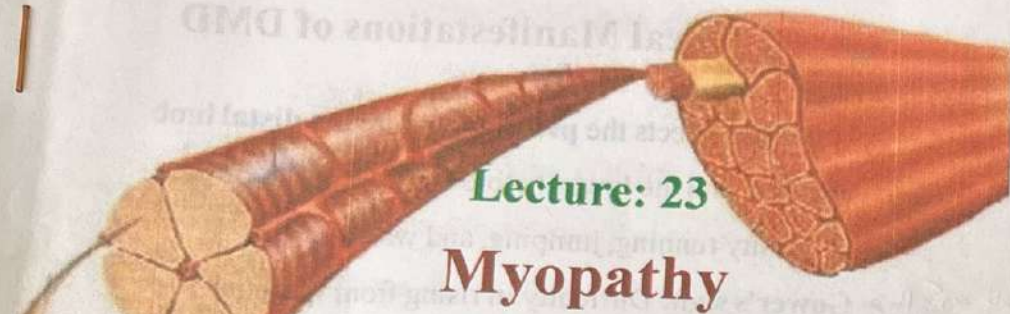




Lecture: 23

Myopathy

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Muscular Dystrophies

Genetic defect ← نوع من أنواع myopathy ، بس ناتجة عن

- ❖ The muscular dystrophies are **an inherited group of progressive myopathic disorders** resulting from defects in a number of genes required for normal muscle function.

- ❖ **Muscle weakness** is the **primary symptom**.



Myopathies Presenting in Adulthood

Muscular dystrophies

- Duchene
- Becker
- Facioscapulohumeral
- Limb-girdle

Inflammatory myopathies

- Polymyositis
- Dermatomyositis
- Inclusion body myositis
- Viral (HIV)

Metabolic myopathies

- diabetic

Endocrine myopathies

- Thyroid
- Parathyroid
- Adrenal
- Pituitary disorders

Toxic myopathies

- Alcohol
- Corticosteroids
- Local injections of narcotics

- Colchicine for gout

- Statins → يقلل الدهون

Myotonic dystrophy

Duchenne Muscular Dystrophy

الأشهر والأخف "DMD"

- The **most common** type of muscular dystrophy.
- DMD is caused by mutations of **the dystrophin gene**.
- Inheritance:** X-linked recessive.
- Affects males, females are carrier.
- Onset:** 2-5 years.
- DMD is associated with the **most severe clinical symptoms**.

كلما ازداد severe يتكون عجز

ما يكون قادر على الوقوف
بغير مساعدة
ليسند على ركبتيه
زنا العواجز

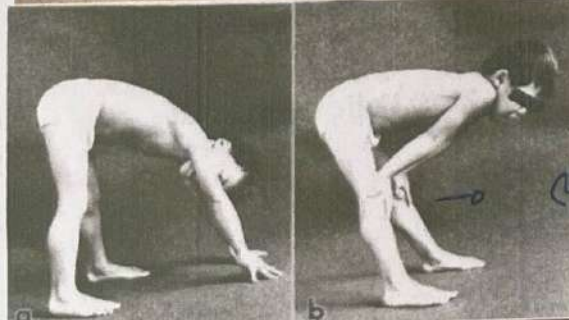
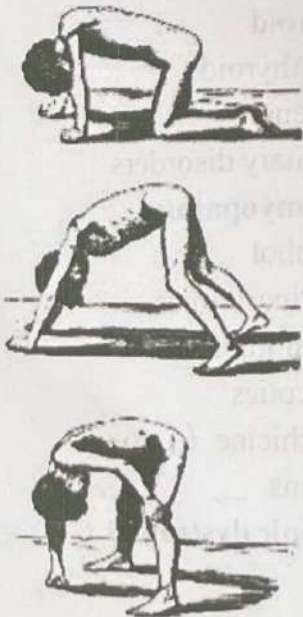
Clinical Manifestations of DMD

- Weakness affects the **proximal** before the **distal** limb muscles, and the **lower** before the **upper** extremities.
- Difficulty running, jumping, and walking.
- Gower's sign:** Difficulty in rising from the floor.
- Waddling gait. زعجة البطة
- Pseudohypertrophy of calf muscles. due to fat deposition
- Inability to walk by 9-12 years. → بجهد على كسبي متوكة

عيب LL قبل UL
distal < proximal



Gower's Manoeuvre



بسبب weakness of proximal m. of thigh

بند
نفسه



Pseudohypertrophy of calf muscles

المظهر الخارجي
صينية هنا فخمة
لكن هذا دهون
العضلة
فخمة

Complications of DMD

- Dilated cardiomyopathy (DCM). ✓ →
- Orthopedic complications: Fractures involving the arms and legs, lumbar lordosis, progressive scoliosis.
- Respiratory failure.
- Shortening of the achilles tendons.

بتموتوا بسببه أد
resp. failure

Becker Muscular Dystrophy "BMD"

- BMD is the **second most common type** of muscular dystrophy.
- BMD is caused by mutations of **the dystrophin gene**.
- **Inheritance:** X-linked recessive.
- Affects males, females are carrier.
- **Onset:** variable. →
- BMD has a similar presentation to DMD, but typically has **later onset** and **more benign course**.
- Muscle involvement is less severe than in DMD, cardiac involvement in BMD is often more evident.

يكون ما بين 5 إلى 10 سنوات

أقل مبطونة وأقل تدهور

Prognosis

- ❖ Patients with DMD are often confined to wheelchair by about age 12 years. Death by age 20-30 years.
- ❖ **Cause of death:** respiratory insufficiency or cardiomyopathy.

✓ بين 12 إلى 15 سنوات في عمر 20-30
موتوا في عمر 20-30

Becker's Muscular Dystrophy

benign
عمر متوسط 30-40، وهو يعني أنه دليل أنه

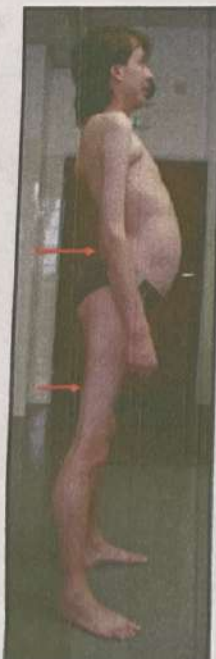
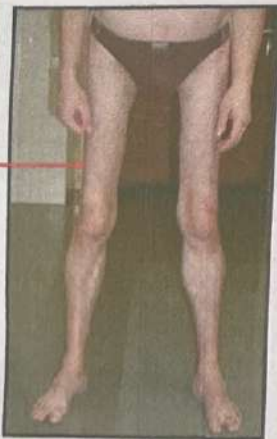


Facioscapulohumeral Muscular Dystrophy

"FSHD"

- Also known as **Landouzy-Dejerine disease**.
- FSHD is the **third most common type** of muscular dystrophy, post (**Duchenne and Becker muscular dystrophies**).
- **Autosomal dominant**.

Facioscapulohumeral Muscular Dystrophy



Clinical Manifestation of FSHD

more benign than DMD

- **Onset:** varies from infancy to middle age, but is usually in the second decade.
- Characterized by slowly progressive muscle weakness involving the:
 - ✓ **Facial.**
 - ✓ **Scapular.**
 - ✓ **Upper arm.**
 - ✓ **Lower leg.**
 - ✓ **Hip girdle muscles.**
- Usually with **asymmetric involvement**.

يبدأ بالوجه

Scapula - Facial

لا يمكن تجنب ضعف العضلات

Other Manifestations of the FSHD

- Pain. non skeletal symptom
- Kyphosis and lordosis. انحناء قوسا في الظهر
- Retinal vasculopathy. myopathy symptom
- Hearing loss. m. weakness
- Cardiac arrhythmia. اضطراب نظم القلب
- Cognitive impairment. الاضطراب المعرفي
- Epilepsy.

Limb-girdle Muscular Dystrophy "LGMD"

- A muscular dystrophy with **predominantly proximal distribution of weakness**.
- **Spare** distal muscles as well as facial and extraocular muscles.
- **Inheritance**: Usually **autosomal recessive** and **less frequently autosomal dominant**.
- **Onset**: Onset usually in the **late first or second** decade of life.
- Expression in either male or female sex.
- Weakness may affect the **shoulder girdle (scapulohumeral type)**, the **pelvic girdle (pelvifemoral type)**, or both.

يُصيب proximal، يمكن يبدأ بـ 15، 16 و أواخر الـ 20 و

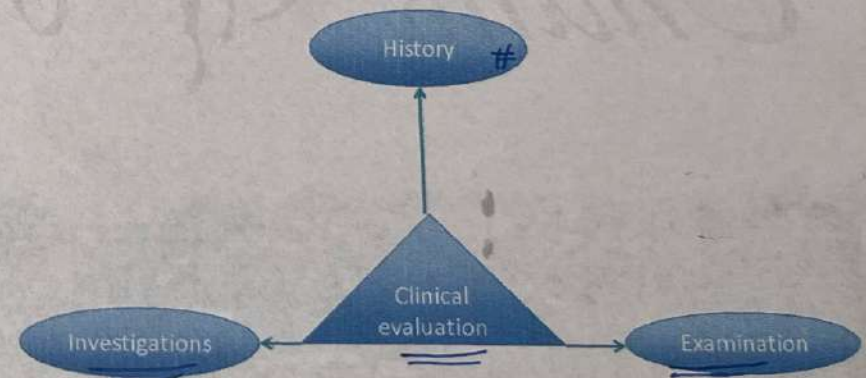
Secondary Myopathy

- **Endocrine**: **hypothyroidism**, **hyperthyroidism**, **Cushing's syndrome**, **adrenal insufficiency**, **hyperaldosteronism**, **hyperparathyroid**, **DM**.
- **Metabolic**: carbohydrate metabolism of lipid metabolism, vitamin E deficiency, hypokalemia, hyperkalemia. Carnitin deficiency.
- **Rheumatologic diseases**.
- **Drugs and Toxins**: cocaine, heroin, alcohol, corticosteroids, colchicine, **bezafibrate**, statin. مغذرين تأخذ هـ ستون
- **Viral infections**: influenza, HIV, cytomegalovirus, Epstein Barr virus.

لـ ما تفوق بـ تكون وكونها تقابله هذا هـ تأش
صوم الفايروس (2nd myopathy)

Limb-girdle Muscular Dystrophy

- **Autosomal recessive subtypes**: associated with:
 - ✓ Earlier age of onset, *more dangerous*
 - ✓ More rapid progression *لا الارتفاع هـ بـ*
 - ✓ High creatine kinase level. *CK عالي بـ د*
- **Autosomal dominant subtypes**: associated with: *degeneration muscle*
 - ✓ Later age of onset.
 - ✓ More gradual progression. *more benign course*
 - ✓ Minimal elevation of creatine kinase level. *لـ هـ يكون CK حتى نورال*



Muscular Dystrophy Investigations

- CBC, ESR.
- Blood chemistry: ^{liver} LFT, ^{kidney} KFT, serum electrolytes, creatinine kinase "CK".
- Metabolic screen.
- TFT, cortisol level. } 2nd إذا myopathy
- ECG. → arrhythmia إذا
- Echocardiography.
- Electromyography.
- Muscle biopsy. → to confirm dx
- Genetic study.

Treatment of Muscular Dystrophies

- dz progression ← low dose
- Glucocorticoid: prednisone "0.75 mg/kg per day" for DMD.
- Gene therapy. # فتن علاج
- Carnitin. كامل
- Multivitamins.
- Symptomatic therapy.
- Orthopedic management.
- Physical therapy and rehabilitation

* إذا 2nd هتعالج السبب ...

الدكتور عرفه منديو كد آخر المحاضرة

كلالة DMD ...

Thank You

