MEDICINE

432 Team

51 Myopathies

Done By:
Abdurahman Alakeel

Reviewed By:
Sarah Al-Essa

جامعة الملك سعود
King Saud University

COLOR GUIDE: • Females' Notes • Males' Notes • Important • Additional

Objectives

Not Given

Myopathies:

- Myo -is muscle, pathos is suffering in Greek
- Disorders in which there is a primary functional (like problems in ion channels) or structural (destruction of muscle tissue replaced by connective tissue or fat) impairment of skeletal muscle.

- **Helpful link:**
  https://www.youtube.com/watch?v=8wa04qYsaps

Approach:

- THREE STEPS:

1. Distinguishing true muscle weakness from other symptoms.
    - Distinguishing type of pain or weakness is the first step in evaluating patients with muscle-related complaints.
    - SOB (shortness of breath), joint pain (asthenia), fatigue, poor exercise tolerance, or paresthesia, rather than a true muscle weakness.
    - Heart failure, arthritis, depression…
2. Distinguishing CNS from PNS lesions. (PNS lesions present bilaterally)
3. Determining the etiology.

Asthenia:

Motor impairment do to pain or joint dysfunction

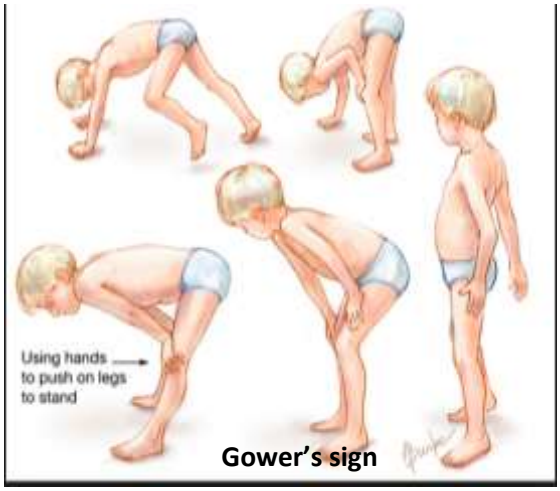
	Myopathic disease	Nonmyopathic disease
Symptoms	Acute to chronic Usually progressive Commonly associated with systemic symptoms[2-4]	Chronic Remitting and relapsing Nonprogressive Minimal systemic symptoms May have concomitant anxiety/ depression[1]
Physical	Vital signs may be abnormal Weakness is common Atrophy present in advanced disease[2-4]	Vital signs are normal Strength is preserved Atrophy is absent[1]
Lab results	Creatine kinase, aldolase are often increased Lab abnormalities are suggestive of a primary disorder, such as infection or an endocrine or electrolyte disturbance[2-4]	Lab work most often is normal[1]

SYMPTOMS:

- **Positive symptoms**: Myalgia ,myotonia ,cramps ,contractures ,myoglobinuria.
- **Negative symptoms**: Weakness, atrophy, exercise intolerance, periodic paralysis.

1- Weakness

- Weakness is the cardinal symptom
- The distribution of weakness is variable and may change over time .
- Most of the time in the proximal muscles; muscles of the shoulder girdle.
- Patients complain from difficulty arising from a chair or low toilet, difficulty climbing stairs, a waddling gait, difficulty lifting objects over the head, combing hair or brushing teeth.
- Distal weakness is less common.
- Patients with proximal leg weakness may rise from sitting on the floor by "climbing up their legs with their hands", This is termed Gower's sign.



Gower's sign

❖ Important points to ask:

- Onset. If hyper acute usually vascular.
- Course. Fixed course like in congenital myopathies or progressive like dystrophies.
- Limbs involved, all four limbs; either symmetrical or not. If in one side only think of CNS lesions.
- Muscles involved. Proximal. if distal think of neuropathies.
- Progression.
- Presence of sensory/autonomic symptoms, they are not caused by myopathies, but may be associated with them (e.g. mitochondrial myopathy).
- Patient's demographics

❖ Pattern of Weakness in Myopathies:

1. PROXIMAL LIMB GIRDLE
2. DISTAL DISTRIBUTION, like in inclusion body myositis.
3. SCAPULOPERONEAL DISTRIBUTION, proximal upper limbs (scapulo-) and distal lower limbs (-peroneal).
4. DISTAL ARM AND PROXIMAL LOWER LIMB, like in inclusion body myositis.
5. PTOSIS -/+ OPHTHALMOPLEGIA, as in occulopharngeal dystrophy

2- Exercise Intolerance

- A less reliable negative symptom, not a specific symptom.
- Often reflects the general level of conditioning and health
- In patients without any objective weakness depression should be considered.
- Exclude certain ***metabolic myopathies*** or ***mitochondrial cytopathies***.
- Ask if it is elicited by brief or long term exercise, which orients towards a disorder of carbohydrates or lipid metabolism, respectively.

3- Myalgias

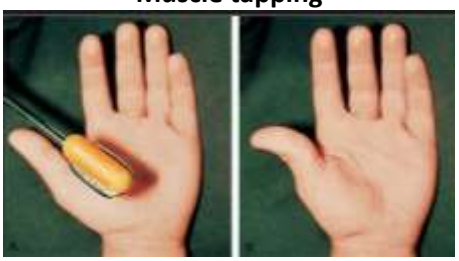
- An infrequent symptom, seen in inflammatory and metabolic myopathies.
- Orthopaedic or rheumatologic conditions are more frequent causes (we should rule them out).
- Constant proximal muscle pain often accompanies ***inflammatory myopathies.***
- Episodic myalgias after exercise point to ***metabolic myopathies***.
- In individuals with waxing and waning, diffuse myalgias, especially in neck and lower back anxiety should be ruled out.

4- Cramps

- Involuntary painful contractions of muscle that last for seconds to minutes, seen in metabolic myopathies.
- Most are **benign** and occur predominantly in calves .
- Risk factors are dehydration, old age, prolonged sitting, use of diuretics, hypothyroid state, DM
- They are most common in ***motor neuron disease*** ,and in ***chronic neuropathies*** rather than in myopathies
- Cramps are only common in ***metabolic myopathies*** such as myophosphorylase deficiency (McArdle's disease), and in ***hypothyroid myopathy***.

5- Myotonia

Muscle tapping



- Impaired relaxation after sustained voluntary contraction.
- A painless phenomenon
- Commonly involves intrinsic hand muscles and eyelids .
- It is due to repetitive depolarization of the muscle fibers
- It improves with repeated exercise.
- Clinically, myotonia can be seen by tapping the muscle (percussion myotonia) or by voluntary contractions of muscle groups (action myotonia).

Gripping examiner's hand

- Typical tests are squeezing the hand of the examiner or forceful closure of the eye.

6- Myoglobinuria

- Excess myoglobin in urine ,resulting in a cola coloured urine.
- It is an uncommon finding.
- Severe and relatively acute massive muscle fiber damage.
- CAUSES: idiopathic, unaccustomed strenuous exercise, after drugs (statin) or toxin intake, infections, heat stroke, etc .
- In case of recurrent myoglobinuria, **glycogenosis**, lipid storage myopathies or **central core** disease with malignant hyperthermia should be ruled out.

Lab Investigations:

CK could be high because of IM injection, strenuous exercise, trauma

- Muscle enzymes: CK ,aldolase, LDH and the aminotransferases
- ANA, ENA antibodies (anti-Ro/SSA ,anti-La/SSB, anti-Sm, and anti-RNP), done to detect inflammatory myopathies.
- Myositis specific Ab (e.g, anti-histidyl-t-RNA synthase"anti-Jo-1")
- Genetic testing.
- Electromyography: An electro-diagnostic technique for evaluating and recording the electrical activity produced by skeletal muscles, the signals can be analyzed to detect medical abnormalities.

* With EMG we can differentiate between myopathies and neuropathies.

- Nerve conduction study (NCS). Done before EMG to exclude neuropathy.
- MR, rarely done.
- Muscle biopsy, done for all inherited or congenital myopathies.

Classification:

- **Congenital Myopathies.**
- **Muscular dystrophies.**
- Channelopathies.
- Metabolic myopathies.
- Mitochondrial myopathies.
- **Inflammatory myopathies.**
- Acquired toxic, infectious, and metabolic myopathies.

Electrolyte abnormality	Signs/symptoms	Exam findings
Hyperkalemia	Generalized muscle weakness Impaired cardiac conduction	Weakness Paralysis
Hypokalemia	Generalized muscle weakness Ileus EKG abnormalities	Weakness Ascending paralysis (severe) Respiratory depression (severe)
Hypercalcemia	Impaired concentration Confusion Fatigue Polyuria Polydipsia Muscle weakness GI complaints Mood disorders Abdominal pain Constipation Anorexia	Hypertension Weakness
Hypocalcemia	Paresthesias Numbness Muscle cramps Muscle stiffness	Chvostek's sign Trousseau's sign Hypotension Dry, coarse skin
Hypermagnesemia	Hypotension Nausea Vomiting Facial flushing Urinary retention Ileus	Hypoactive deep tendon reflexes Paralysis (severe) Respiratory depression (severe)
Hypomagnesemia	Paresthesias Weakness	Hyperreflexia Fasciculation Tremors Seizures Altered mental status

These mimic myopathies, it is important to do urinalysis to exclude them.

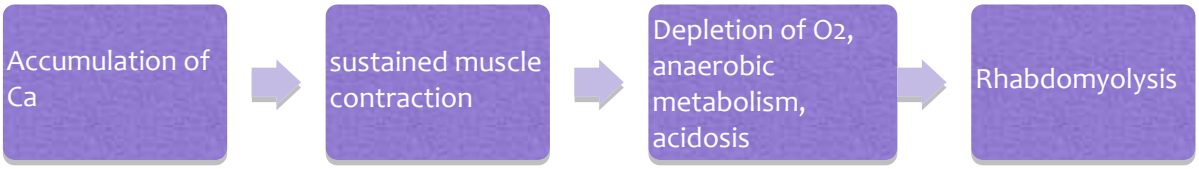
1) CONGENITAL MYOPATHIES:

- Clinical characteristics present from birth or prenatally.
- Prenatal: decreased fetal movements.

- Postnatal: Hypotonia ,poor respiratory effort ,difficulty feeding, reduced muscle bulk, weakness.
- First year and beyond: hypotonia, weakness, delayed milestones, failure to thrive, recurrent respiratory infections, flaccid speech.
- Slow or non-progressive course.
- No treatment.

- Types of congenital myopathies (not important):
    - Central core disease
    - Multicore (minicore) disease
    - Nemaline myopathy
    - Myotubular (centronuclear) myopathy
    - Myofibrillar (desmin related)
    - Congenital fiber type disproportion
- Management:
    a) Genetic counseling
    b) Detection and treatment of orthopedic complications
    c) Prevention of complications (e.g. general anesthesia)

- Malignant Hyperthermia (MH)
    - Hypermetabolic crisis.
    - MH-susceptible individual is exposed to a volatile anesthetic or succinylcholine.
    - Genetic skeletal muscle receptor abnormalities, allowing excessive calcium accumulation in the presence of certain anesthetic triggering agents.



    - Sign and symptoms:
        - Masseter spasm immediately following anesthetic induction.
        - Hypercarbia.
        - Sinus tachycardia.

- Generalized muscular rigidity.
- Tachypnea.
- Cyanosis.
- Rapidly increasing temperature is a *later sign* of MH and is typically absent when the diagnosis is initially suspected.
- Sweating.
- Cola-colored urine.
- Ventricular fibrillation.

2) MUSCULAR DYSTROPHIES (MD)

- Inherited myopathies.
- Variable age at onset.
- Progressive degeneration of the muscles with connective tissue replacing muscle fibers.
- Systemic involvement.
- Types of MD:
    a. **Dystrophinopathies** (Duchenne and Beker) most common.
    b. Emery-Dreifuss muscular dystrophy.
    c. Barths syndrome.
    d. Autosomal dominant dystrophies:
        - Facioscapulohumeral MD.
        - Oculopharyngeal MD.
        - Classic, Congenital and Proximal myotonic dystrophy.
    e. Limb girdle MD.

2\a. Dystrophinopathies:

- X linked recessive disorders, so it affects males.
- **Duchenne** & **Becker** (DMD, BD).
- Caused by mutation in the dystrophin gene.
- Dystrophin provides mechanical reinforcement to the sarcolemma and stabilizes the glycoprotein complex.
- Its absence causes digestion of the glycoprotein complex.
- This initiates degeneration of muscle fibers, resulting in muscle weakness.

A. Duchenne MD (DMD)

- Complete absence of Dystrophin.
- Present early in life.
- Weakness of upper and lower limbs that is more pronounced proximally.
- Scapuloperoneal distribution: TA (tibialis anterior) and peronei muscles more than gastrocnemius and tibialis posterior.
- Neck flexors, wrist and digit extensors.
- Respiratory muscles.
- Sparing of the cranial muscles.
- Motor developmental delay.
- Toe walking, as a compensation for the progressive weakness of the knee extensors.
- Difficulty rising from sitting position.
- Gower's sign.
- Lumbar lordosis, waddling gait ,pseudo-hypertrophy of the calves.
- 12 years: loss of ambulation ,marked wasting of muscles, contractures ,kypho-scoliosis, exaggerated lumbar lordosis.
- Death due to respiratory complications between 15-30 years.
- SYSTEMIC INVOVEMENT:
    - Cardiomyopathy: CHF and arrhythmias, we should do ECG.
    - Malignant Hyperthermia like reactions with rhabdomyolysis.
    - Intestinal pseudo-obstruction.
    - CNS involvement: Mental retardation, learning disabilities.
- INVESTIGATIONS:
    - CK is markedly elevated early in the disease.
    - Electromyography: myopathic potentials.
    - Muscle biopsy: necrosis ,replacement with connective tissue and fibrosis, variation in muscle fiber size, absent dystrophin.
- Management:
    - Early detection of systemic involvement:

- Assessing for evidence of cardiac dysfunction and treatment accordingly.
- Screening for orthopedic complications to maintain function and prevent contractures.
- dietary calcium and vitamin D supplementation, and yearly DXA scanning.
- Weight and growth monitoring.
- Avoidance of anesthesia and sedation if possible.

- Glucocorticoids are the mainstay:
  - For boys 5 years of age and older who are no longer gaining motor skills, or whose motor skills are declining.
  - It increases strength, muscle function, and pulmonary function.
  - Reduces cardiomyopathy and lowers mortality.
  - It has an anabolic action in contrast to its catabolic action on normal skeletal muscle in unaffected people .
  - Stabilizes sarcolemma
  - Side effects:
    - Weight gain
    - Cushingoid facial appearance, acne.
    - Short stature, compression fractures.
    - Delayed puberty.
    - Excessive hair growth.
    - Gastrointestinal bleeding.
    - Psychosis, and behavioral changes.

B. Becker Dystrophy

- Older age at onset.
- Less severe symptoms.
- Loss of ambulation is usually in the 4th decade.
- Muscle biopsy shows decreased staining patterns rather than complete absence of dystrophin.

2\b. Myotonic Dystrophy (MD):

- The most prevalent inherited neuromuscular disease in adults.
- Autosomal dominant.
- Age of onset average is 29 years.
- Myotonia.
- Weakness of the forearms and peroneal muscles (distal muscles).
- Ptosis and weakness of other facial muscles.
- Temporal wasting; patient will has narrow or thin face.
- Frontal bolding.
- Mild axonal neuropathy.
- Heart involvement.
- GIT dys-motility, constipation and diarrhea.
- Cataract.
- Endocrine abnormalities, insulin resistance may have DM, menstrual disturbances and difficulty to get pregnant.
- Low IQ.

3) INFLAMMATORY MYOPATHIES:

a. Polymyositis (PM)
b. Dermatomyositis (DM)
c. Inclusion body myositis (IBM)

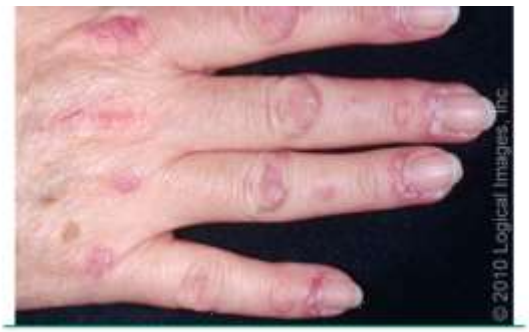
a + b: PM & DM

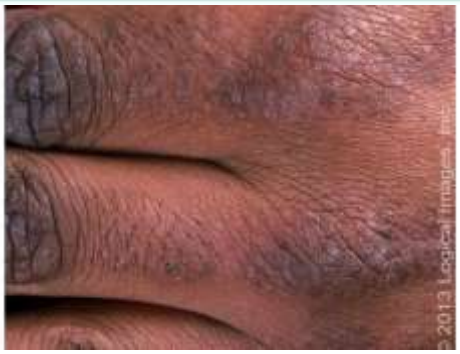
- Epidemiology:
  - The combined incidence is 2/100,000 annually
  - Female to male ratio of 2:1
  - DM affects children and adults

- PM affects mainly adults
- The peak incidence in adults occurs between the ages of50-40
- Skin is involved in dermatomyositis.

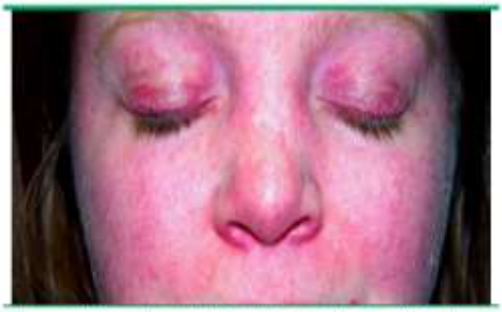
- Clinical Features:
  - Multi-system disorders.
  - Sub-acute progressive proximal skeletal muscle weakness.
  - Mild myalgias and muscle tenderness.
  - Neck flexors are commonly involved.
  - Facial muscles are usually spared.
  - Dysphagia due to pharyngeal and upper esophageal muscles involvement (30%), may lead to aspiration.
  - In advanced cases muscle wasting and hypo-reflexia.
  - Respiratory muscles weakness.
  - Interstitial lung disease 10% (anti-tRNA synthetase or Jo-1).
  - Cardiac arrhythmias, bundle branch block and ST changes.
  - Polyarthritis.
  - Raynaud phenomenon.
  - Cutaneous manifestations in DM that precedes or accompany weakness.
  - Malignancy:
    - Risk in DM is 40-30% (ovarian and lung)
    - Risk in PM is 15% (non-Hodgkin's lymphoma and lung)

- DM:





Gottron's papule: Multiple violaceous., scaly papules are present overlying the joints on the dorsal hand.



Violaceous erythema on the upper lids in a patient with dermtomyositis. Mid-facial erythema that does not spare the nasolabial folds is also present.

Eyelid edema in dermatomyositis

Edematous upper and lower eyelids in a patient with dermatomyositis.

Courtesy of Jeffrey Callen, MD, FACP, FAAD.

Nailfold changes in dermatomyositis

Dilated capillary loops at the proximal nailfold, cuticular overgrowth, and periungual erythema.

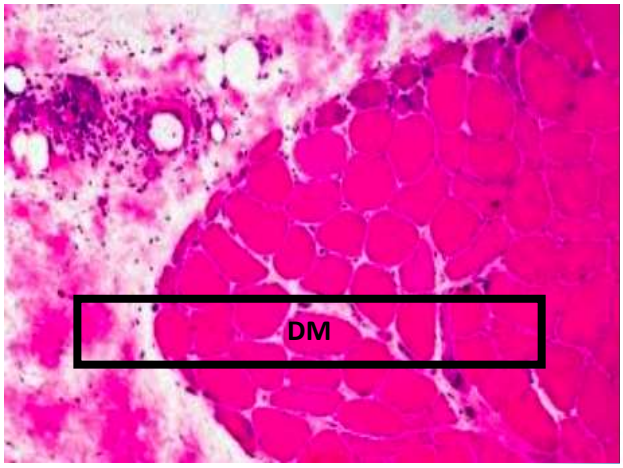
Courtesy of Jeffrey Callen, MD, FACP, FAAD.

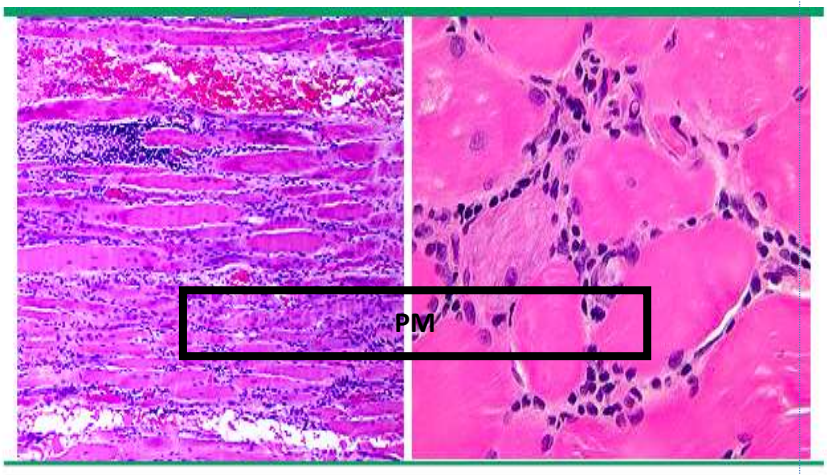
- *Cutaneous Manifestations:*
  - Gottron's papules and the heliotrope eruption are the hallmark and pathognomonic features.
- Generalized erythroderma.
- Psoriasiform changes in scalp.
- Calcinosis cutis.
- Mechanic's hands.

Heliotrope eruption involves nasolabial folds while SLE doesn't.

- DIAGNOSIS of PM & DM:
  - Elevated levels of muscle enzymes (CK -very high-, LD, aldolase, AST, and ALT).
  - Autoantibodies, ANA, in up to 80% of patients.
  - Antibodies associated with primary myositis syndromes: anti-Jo1 (20%), anti SRP (5%), anti-Mi-2 (15-35% of DM and 5-9% PM).
  - Antibodies associated overlap syndrome: anti-PM/Scl, anti-Ro, anti-La, anti-U1 snRNP (SLE, systemic scleroderma, RA or mixed CTD), anti-U2 snRNP (scleroderma).
  - Elevated levels of serum and urine myoglobin.
  - EMG.
  - MRI: inflammation, edema with active myositis, fibrosis, and calcification
  - DM MUSCLE BIOPSY:
    - Perifasicular atrophy.
    - Microvascular injury and deposition of membrane attack complex, endothelial microtubular inclusions and hyperplasia.
    - Perimysial inflammatory cells.
  - PM MUSCLE BIOPSY:
    - Endomysial, perimysial and perivascular inflammatory infiltrates.
    - Muscle destruction and regeneration, muscle fiber size variation.
    - No perifascicular atrophy, microvascular injury, endothelail hyperplasia and inclusions.

- TREATMENT





- Prednisone (corticosteroid) at 1mg/kg/day to be tapered gradually over 12 months
- Calcium and vitamin D
- glucocorticoid-sparing agent :azathioprine and methotrexate
- Physiotherapy and occupational therapy
- Prevention of aspiration :swallowing assessment, elevation of the head of the bed, nasopharyngeal or gastric tube, Semi thick diets
- Sun protection, especially in DM

c. Inclusion Body Myositis (IBM)

- Epidemiology:
    - Adults ***older than 50*** (third to the ninth decade).
    - More common in ***men***.
    - More common in whites.
- CLINICAL FEATURES:
    - Proximal lower extremity weakness is usually the first sign.
    - Chronic slowly progressive symmetric myopathy.
    - Asymmetric weakness could occur.
    - Myalgia 40%.
    - Wrist and Fingers flexors, hip flexors, quadriceps muscles.
    - Mild facial weakness in 60%.
    - Esophageal dismotility and dysphagial in 60%.
- DIAGNOSIS:
    - Muscle enzymes are **typically normal** or mildly elevated, CK is less than 10 times normal.
    - Myositis-specific autoantibodies are **typically absent** in patients with IBM.
    - EMG.
    - MRI.
    - BIOPSY:
        - Endomysial inflammation 90%
        - Basophilic-rimmed vacuoles 70%

- Eosinophilic inclusions adjacent to the basophilic-rimmed vacuoles 50%
- The definitive diagnostic feature is filamentous **inclusions** and vacuoles 90%

- TREATMENT
  - the response to therapy is generally very poor
  - Steroids and steroids sparing agents
  - IVIG for dysphagia
  - Physiotherapy, occupational therapy

4) ACQUIRED TOXIC MYOPATHIES:

- Alcohol ,Cocaine
- **Lipid lowering agents**
- Steroid
- Antimalarials ,Antiretrovirals
- Antipsychotic
- Chemotherapies
- Ipecac

STATINS INDUCED MYOPATHY

- The mechanism is not well understood, it may affect coenzyme q10.
- CLINICAL FEATURES:
  - Myalgia, 2-11%
  - Myopathic weakness, 2-11%
  - Myositis
  - Myonecrosis 0.5%
  - Rhabdomyolysis, 0.1%
- Prevention:
  - Pravastatin and fluvastatin, they are less likely to cause myopathy.
  - A baseline CK level prior to starting statin
  - Patients should be alerted to report the new onset of myalgias or weakness
  - Caution in patients with renal failure, hypothyroidism and liver failure.

Conclusion:

- H & PE are are of paramount importance for differentiation of weakness due to myopathy from that caused by other causes
- Myopathy could be inherited) congenital, MD..) or acquired (steroid induced, infections(..
- There is a wide variation in the age of onset of different types of myopathies but the in all the cardinal symptom is weakness
- Screening for systemic involvement (cardiac, pulmonary…) early on diagnosis affects long term prognosis

SUMMARY

Main differences between muscle diseases and nerve diseases (lower motor neuron):

- Neuropathies are more distal; myopathies are more proximal usually & symmetrical.
- Neuropathies have sensory symptoms; myopathies are not related to sensory symptoms.
- In neuropathies, reflexes are absent from the beginning; in myopathies the reflexes are found in the beginning of the disease (then they become reduced or absent).

Signs and symptoms of myopathies:

- **Positive symptoms**: Myalgia ,myotonia ,cramps ,contractures ,myoglobinuria.
- **Negative symptoms**: Weakness, atrophy, exercise intolerance, periodic paralysis.

Congenital myopathies:

Presentation: Early childhood presentation, affects growth. Malignant hyperthermia might occur in anaesthesia.

Duchenne Muscular Dystrophy:

Presentation: More in males, 3-5 year olds. - Caused by: dystrophin deficiency, progressive muscle loss.

Becker's Muscular Dystrophy:

- Amount of dystrophin is REDUCED, has more benign course.
- Muscle biopsy:
  - If dystrophin staining is absent: Duchenne.
  - If reduced: Becker's.

Myotonic Dystrophy:

- Difficulty in muscle relaxation after muscle contracted
- Symptoms: myotonia, progressive muscle weakness, cataracts, frontal balding, cardiac abnormalities, mental retardation, endocrine problems.

Inflammatory Myopathies:

- Presentation: More in females than males = 2:1
- Types: **polymyositis, dermatomyositis.**
  - Polymyositis: usually in adults * dermatomyositis: usually in younger people. Characterized by: proximal muscle weakness and myalgia.
  - Polymyositis symptoms: cardiac disturbances, respiratory involvement (interstitial fibrosis, pneumonitis), systemic symptoms (arthralgia, fever, malaise, Raynaud's phenomenon)
  - Dermatomyositis symptoms: Same as polymyositis symptoms + skin manifestation (Gorton's sign on fingers, Heliotrope rash on eyelid, Shawl sign on the back and shoulders).
  - Idiopathic inflammatory myopathies may be associated with malignancies: * Polymyositis: non-Hodgkin's lymphoma or lung cancers * Dermatomyositis: ovarian, lung cancer.

Inclusion body myositis:

- Muscle biopsy shows amyloid inclusions in muscles fibers.
- Usually in elderly (above 50's)

Statin-induced myopathy

Approach to Muscle Weakness

MYOPATHY

DIFFERENTIAL DIAGNOSIS

INFLAMMATORY MYOPATHY polymyositis, dermatomyositis, inclusion body myositis, juvenile dermatomyositis, vasculitis, overlap syndromes (SLE, scleroderma, rheumatoid arthritis, Sjogren's)

INFECTIOUS MYOPATHY

- *BACTERIAL* pyomyositis, Lyme myositis
- *VIRAL* influenza, parainfluenza, Coxsackie, HIV, CMV, echovirus, adenovirus, EBV
- *FUNGAL*
- *PARASITIC* trichinosis, toxoplasmosis

DRUG/TOXIC MYOPATHY steroid, alcohol, cocaine, heroin, colchicine, antimalarial, statins, fibrates, penicillamine, zidovudine

ENDOCRINE MYOPATHY hypothyroidism, hyperthyroidism, Cushing's, diabetes, acromegaly

METABOLIC MYOPATHY hypokalemia, hypocalcemia, hypophosphatemia, hyponatremia, hypernatremia, disorders of carbohydrate/lipid/purine metabolism

NEOPLASTIC MYOPATHY paraneoplastic

RHABDOMYOLYSIS

- *DRUGS* alcohol, cocaine, statins, neuroleptic malignant syndrome, malignant hyperthermia
- *HYPERACTIVITY* seizures, exertion
- *TRAUMA/OPERATION*
- *IMMOBILITY*

NEUROLOGIC

- **MOTOR CORTEX** stroke, multiple sclerosis, brain tumor, abscess
- **CORTICOSPINAL TRACT/ANTERIOR HORN CELLS** spinal cord injury, vitamin B12 deficiency, ALS, polio, lead
- **SPINAL NERVE ROOTS/PERIPHERAL NERVES** Guillain Barre, myeloma, amyloidosis, diabetes
- **NEUROMUSCULAR JUNCTION** myasthenia gravis, botulism, Eaton Lambert, organophosphate poisoning
- **MUSCLES**

CLINICAL FEATURES

APPROACH TO CLINICAL DIAGNOSIS

1. FUNCTIONAL VS. TRUE MUSCLE WEAKNESS?

- if functional, consider cardiopulmonary disease, arthritis, anemia, cachexia from malignancy or chronic disease, depression, deconditioning, fibromyalgia
- if true muscle weakness, proceed to 2

2. GENERALIZED VS. LOCALIZED MUSCLE WEAKNESS?

- ✓ if generalized, consider myasthenia gravis, long standing periodic paralysis, advanced disuse atrophy from prolonged bed rest, or advanced muscle wasting from malignancy
- ✓ if localized, proceed to 3

3. ASYMMETRIC VS. SYMMETRIC MUSCLE WEAKNESS?

- ➢ if asymmetric, consider disease of central or peripheral nervous system (stroke, spinal cord injury, demyelinating disorders, compression neuropathy, mononeuropathy/neuritis), disuse atrophy, myasthenia gravis
- ➢ if symmetric, proceed to 4

4. DISTAL VS. PROXIMAL MUSCLE WEAKNESS?

- if distal, consider peripheral neuropathy, myasthenia gravis, motor neuron disease
- if proximal, consider myopathies (see differential diagnosis), myasthenia gravis, Duchenne muscular dystrophy

MRC MUSCLE STRENGTH GRADING

0=no contraction
1=flicker
2=possible only with gravity eliminated
3=against gravity only
4=power decreased but muscle contraction possible against resistance
5=normal power resistance

MUSCLE STRENGTH preserved in patients with cachexia despite advanced generalized muscle atrophy. In contrast, patients with true muscle weakness due to myopathy generally have normal muscle bulk at time of presentation

MUSCLE TENDERNESS usually not associated with one of the causes of true muscle weakness, except for infectious myopathies, certain drug induced myopathies, thyroid myopathy, and inherited metabolic myopathies

Questions

1) A patient complains of a history of generalized muscle weakness. On examination, his facial muscles show marked atrophy, and when you ask him to shake your hand, he appears to be unable to relax his grip for an extended period. What is the likely diagnosis?

a. Myasthenia Gravis.
b. Inclusion body myositis.
c. Becker's dystrophy.
d. Myotonic Dystrophy.

2) The parents of a 3-year-old boy are concerned that he is not walking as well as other boys his age. Both parents are healthy, and there is no family history of neuromuscular disease. He has three older brothers who are healthy. Physical examination shows that he has large calf muscles and lower extremity proximal muscle weakness, as demonstrated by the need to use his arms and hands to assist in standing from a seated position. CK levels are elevated. If you are suspecting Duchenne dystrophy, what will the results of dystrophin staining show?

a. Reduced dystrophin staining.
b. Normal dystrophin staining.
c. Increased dystrophin staining.
d. Absent dystrophin staining.

*The scenarios of the questions were all taken from: USMLE Step 1 Secrets, 3rd Edition, by Thomas A. Brown & Sonali Shah

432 Medicine Team Leaders

Raghad Al mutlaq & Abdulrahman Al Zahrani
For mistakes or feedback: *medicine341@gmail.com*

Answers:

1st Questions: D

2nd Questions: D