

# Myopathy

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# Site of lesion

- Muscle structure
  - Muscle cell membrane: genetic
  - Muscle fiber destruction: inflammation, toxic
- Muscle channel
- Muscle metabolism
  - Metabolic myopathy
  - Mitochondrial myopathy

# MYOPATHY

## Acquired

Inflammatory  
Toxic

Endocrine

Electrolyte

Infection

CIM

## Inherited

Muscular  
dystrophy

Myotonic  
dystrophy

Channelopathy

Metabolic  
Mitochondrial

Congenital  
Myopathy,  
MD

# Acquired myopathy

## Inflammatory

- DM
- PM
- IBM

## Toxic

- statin
- steroid
- AZT
- amiodarone
- CQ, HCQ
- colchicine
- D-  
penicillamine

## Endocrine

- Cushing  
synd
- hyper/hypo  
thyroid
- hyper/hypo  
parathyroid

## E'lyte

- hypo/  
hyperK
- hypo/  
hyperNa
- hyperCa
- hypoP
- hyperMg

## Infectio n

- parasiti  
c

## CIM

# Inherited myopathy

## Muscular dystrophy

-XL:

DMD, BMD

-AD: FSH,

OPMD

-AR: LGMD

## Myotonic dystroph

y

-DM 1,2

## Channelopathy

-HypoKPP

-Thyrotoxic  
hypoKPP

-HyperKPP

## Metabolic Mitochondrial

## Congenit

al

Myopath

y/MD

# Clinical manifestations

## Negative

- weakness & atrophy
- exercise intolerance

## Positive

- Pain/myalgia
- Cramps
- Myotonia
- hypertrophy
- dark red urine

# Clinical manifestations

## Negative

- weakness & atrophy
- exercise intolerance
- Metabolic
- Mitochondrial

## Positive

- Pain/myalgia
- Cramps
- Myotonia
- hypertrophy
- dark red urine

# Pattern of weakness

- Proximal weakness
- Distal weakness
- Scapulo-peroneal pattern (prox arm/distal leg)
- Distal arm/proximal leg weakness
- Ptosis c/s ophthalmoplegia
- Prominent neck extensor weakness



# Pattern of weakness

- Proximal weakness
  - Inflammatory: PM,DM
  - Endocrine
  - Toxic
  - Muscular dystrophy (most)
  - Electrolyte
- Distal weakness
- Scapulo-peroneal pattern (prox arm/distal leg)
- Distal arm/proximal leg weakness
- Ptosis c/s ophthalmoplegia
- Prominent neck extensor weakness

# Pattern of weakness

- Proximal weakness
- Distal weakness
  - Myotonic dystrophy
  - IBM
- Scapulo-peroneal pattern (prox arm/distal leg)
- Distal arm/proximal leg weakness
- Ptosis c/s ophthalmoplegia
- Prominent neck extensor weakness

# Pattern of weakness

- Proximal weakness
- Distal weakness
- Scapulo-peroneal pattern (prox arm/distal leg)
  - FSHD
  - LGMD
- Distal arm/proximal leg weakness
- Ptosis c/s ophthalmoplegia
- Prominent neck extensor weakness

# Pattern of weakness

- Proximal weakness
- Distal weakness
- Scapulo-peroneal pattern (prox arm/distal leg)
- **Distal arm/proximal leg weakness**
  - IBM
- Ptosis c/s ophthalmoplegia
- Prominent neck extensor weakness

# Pattern of weakness

- Proximal weakness
- Distal weakness
- Scapulo-peroneal pattern (prox arm/distal leg)
- Distal arm/proximal leg weakness
- Ptosis
  - With ophthalmoplegia
    - OPMD
    - CPEO
  - Without ophthalmoplegia
    - Myotonic dystrophy
- Prominent neck extensor weakness

# Pattern of weakness

- Proximal weakness
- Distal weakness
- Scapulo-peroneal pattern (prox arm/distal leg)
- Distal arm/proximal leg weakness
- Ptosis c/s ophthalmoplegia
- **Prominent neck extensor weakness**
  - Inflammatory: PM,DM,IBM
  - Endocrine: hypothyroid, hyperparathyroid
  - Hypokalemia
  - Myotonic dystrophy

# Myalgia

- Inflammatory myopathy (<30%)
- Toxic
  - Statin, colchicine, CQ
- Hypothyroidism
- BMD
- Metabolic, mitochondrial
  - Pain on exercise

# Prox muscle weakness + highCK

≠ polymyositis

- Inflammatory myopathy (x1,000)
- Toxic
- Hypothyroidism
- Muscular dystrophy (x1,000)
  - BMD, FSH, LGMD 2B
- Metabolic myopathy



# Autoimmune inflammatory myositis

# Autoimmune inflammatory myopathy

- Primary autoimmune process
  - PM
  - DM: vasculopathy ( $\neq$  PM+skin)
- Primary degenerative process
  - IBM

# Pathology

## **DM**

- Vasculitis
- Perivascular inflam
- Perifascicular atrophy
- Fiber necrosis/regeneration
- CD4 T cells
- B cells

## **PM**

- Endomysial inflammation
- Infiltration of intact myofiber
- Fiber necrosis/regeneration
- CD8 T cells

# AutoAb

- Unspecific Ab → overlapping syndrome
  - ANA
  - Anti-RNP, anti-SSA, anti-PM-Scl
- Muscle specific autoAb
  - Antisynthetase
  - AntiSRP

## Associated conditions

- Autoimmune CTD
- Malignancy
- Retroviral infection

# Associated conditions

- Autoimmune CTD

## Overlapping syndrome

- MCTD
- Scleroderma
- Sjogren's syndrome
- Rheumatoid arthritis
- SLE
- Malignancy
- Retroviral infection

# Associated conditions

- Autoimmune CTD
- Malignancy
  - DM > PM
  - Before or after myositis
  - Male: prostate
  - Female: breast, OBGYN
  - Others: lung, NHL, germ cell, urinary bladder, GI, nasopharynx
- Retroviral infection

# Associated conditions

- Autoimmune CTD
- Malignancy
- Retroviral infection
  - HIV
  - HTLV-1



# Clinical manifestations

- Muscle weakness
- Myalgia <30%
- Dysphagia 30% (DM>PM)
- Skin
- Extramuscular manifestation
  - joint, heart, lung, GI
- Associated conditions
  - Overlap, CA, retrovirus

# Muscle weakness

- Proximal muscle weakness & axial weakness
- Dysphagia 30% (DM>PM)
- Spare facial & extraocular muscles
- Onset
  - DM: acute – subacute
  - PM: chronic

# Skin in DM

- Heliotrope
- Gottron's papules/sign
- Periungual telangiectasia
- V sign, shawl sign
- Holster's sign
- Mechanic hand
- Subcutaneous calcification
- Poikiloderma vasculare atrophican

# Joint

- Symmetrical polyarthritits
- Polyarthralgia
- Most overlap synd, anti-synthetase synd

# Heart

- Pericarditis
- Myocarditis/ myocardial fibrosis
- Conduction defect
- CM, CHF

# Lung

- Interstitial lung disease
- 10% of PM/DM
- Anti-Jo1

# GI

- Dysphagia
- GERD
- Intestinal pseudoobstruction
- Malabsorption
- Ulceration
- Hemorrhage

# Diagnosis

- Clinical
- CK
- EMG
- Muscle biopsy
- AutoAb



# EMG

- Spontaneous activity
  - F-wave, PSW
  - CRD
- Myopathic pattern
  - Small polyphasic units of short duration
  - Rapid recruitment
  - Full interference pattern

# Treatmen

t

- Corticosteroid
- Immunosuppressive agent
- IVIG
- Targeted therapy

# Corticosteroid

- Acute: IVMP 1 g x3days
- Non acute: oral pred 1-1.5 MKD x 4-6weeks

# Immunosuppressive agent

- No improvement from steroid in 4 mo
- Early combination
  - Severe case, systemic involvement
  - Late diagnosis, muscle atrophy
  - High risk of steroid: elderly
- AZA, MTX

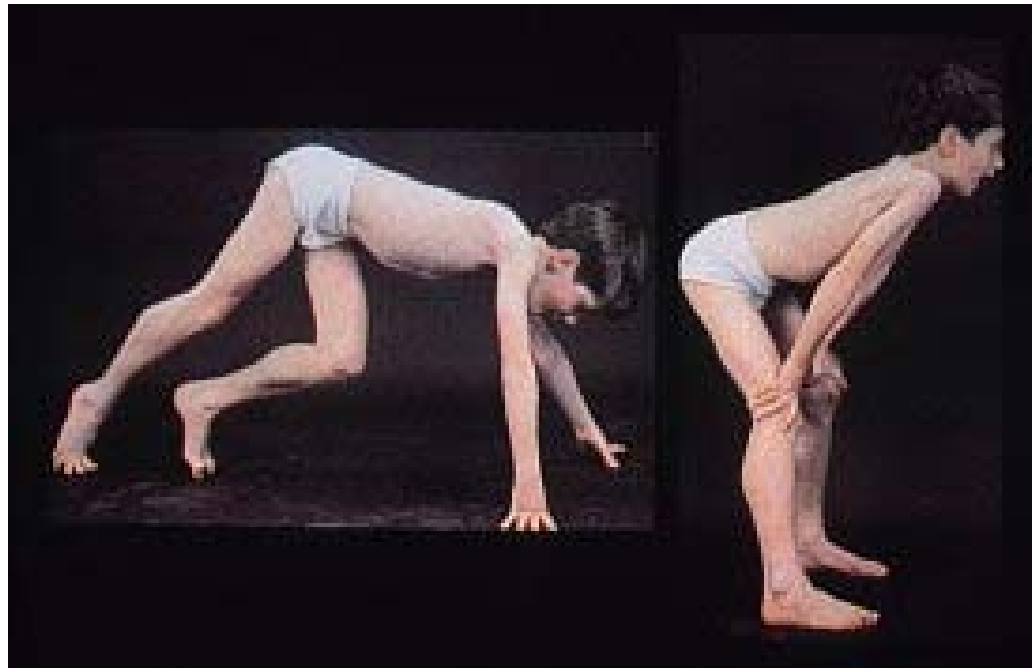
# IVIG

- Steroid resistance
- Immunosuppressive resistance
- Severe dysphagia or respiratory involvement
- Immunodeficiency case: HIV

# Dystrophinopathy

|                     | DMD                                         | BMD          |
|---------------------|---------------------------------------------|--------------|
| Onset               | 2-3 yr                                      | 5-15 yr      |
| Weakness            | UL before LL                                | LL > UL      |
| Calf hypertrophy    | +                                           | +            |
| Muscle pain         | -                                           | +            |
| Cardiomyopathy      | +                                           | +            |
| Respiratory failure | 18-21 yr<br>70% DNA<br>30% dystrophin study | Late<br>same |
| Diagnosis           |                                             |              |

# DMD



# Myotonic dystrophy

- AD
- Nucleotide repeated disorders
- Cl- channel
- Myotonia
- DM 1,2,3



# DM 1

- CTG repeated
- Muscle weakness
  - Distal hand > ankle
  - Facial & ptosis without ophthalmoplegia
  - Neck flexor
  - Sternocleidomastoid
- Grip myotonia
- Percussion myotonia: thenar, tongue

## Associated findings

- Cataract
- Premature balding
- Heart block
- DM/ glucose intolerance
- Testicular atrophy

# Grip myotonia



# DM 1



[http://commons.wikimedia.org/wiki/File:Myotonic\\_dystrophy\\_patient.JPG](http://commons.wikimedia.org/wiki/File:Myotonic_dystrophy_patient.JPG)

<http://www.duke.edu/~ema5/Golian/Slides/8/musculoskeletal5.html>

# Channelopathy

- PP
  - hypoK (Ca&Na)
  - Thyrotoxic PP (?)
  - hyperK (Na)
- Myotonia/paramyotonia
  - Myotonia congenita (Cl)
  - Paramyotonia congenita (Na)

# PP

|                       | HyperKPP                       | HypoKPP                   | TPP                               |
|-----------------------|--------------------------------|---------------------------|-----------------------------------|
| Inheritance           | AD                             | AD                        | ?                                 |
| Channel               | Na                             | Ca,Na                     | ?                                 |
| Onset                 | < 10                           | 10-30                     | 30                                |
| Attack duration       | Min-hr                         | Hr-day                    | Hr-day                            |
| K <sup>+</sup>        | high                           | low                       | low                               |
| Precipitating factors | +                              | +                         | +                                 |
| -Rest after exercise  | -                              | +                         | +                                 |
| -High carb meal       | +                              | +                         | -                                 |
| -Cold                 | +                              | -                         | -                                 |
| Myotonia              | Thiazide/<br>acetazolami<br>de | KCl/<br>acetazolami<br>de | KCl/B-<br>blocker/<br>antithyroid |
| Rx                    |                                |                           |                                   |