Diagnostic approach in Foot-drop

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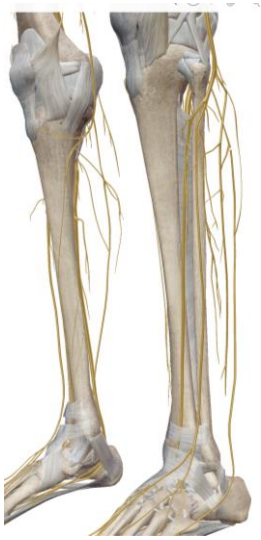
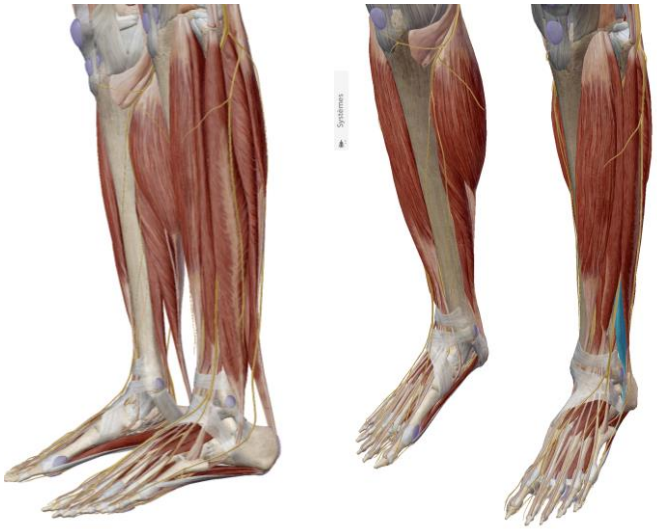
Marseille

Foot- drop

Steps regular and even, advancing foot hangs with toes pointing down , Leg lifted high so that foot clears floor, slapping noise as foot strikes floor.





Observation 1

- Patient de 35 ans
- ATCD : 0
- Déficit de dorsiflexion du pied G
- Brutal et au réveil
- Hypoesthésie dorsale pied gauche

Observation 2

- Patient de 47 ans
- ATCD : Thyroïdite de Hashimoto
- Déficit séquentiel de
  - IO gauche
  - ECD droit
  - dorsiflexion du pied G
  - Atrophie musculaire des territoires atteints
  - Atteinte sensitive même territoires
- Subaiguë /2 mois
- Hypoesthésie dorsale pied gauche
- AEG

Observation 3

- Patient de 44 ans
- ATCD : 0
- Déficit séquentiel de
  - ECD droit
  - IO droit
  - dorsiflexion du pied G
  - absence d'atrophie musculaire
  - absence d'atteinte sensitive
- 12 mois
- Absence d'AEG

Observation 4

- Patient de 53 ans
- ATCD : DIND découvert depuis 2 ans
- Troubles de l'équilibre depuis plusieurs mois
- A l'examen :
  - ataxie, Romberg 2/3 ; marche en tandem 3 pas
  - impossibilité de se mettre sur les talons
  - dorsiflexion du pied G
  - absence d'atrophie musculaire
  - atteinte sensitive en chaussette et les pulpes des doigts
  - Aréflexie diffuse

Observation 5

- Patient de 50 ans
- ATCD : 0
- Pied tombant D depuis plusieurs mois
- A l'examen :
  - impossibilité de se mettre sur les talon D
  - atrophie musculaire TA, les muscles pédieux conservés
  - absence d'atteinte sensitive
  - réflexes normaux
  - discret atteinte asymétrique des orbiculaires des yeux

Diagnostic approach

- Time course: acute, sub acute, chronic
- First sign
- Onset age
- Family history
- Clinical examination (Associated signs)
- Spinal cord
- Neuropathy
- Myopathy

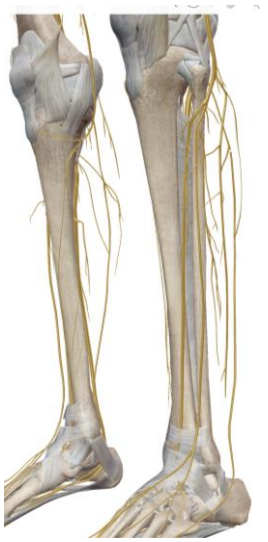
Ancillary investigations

- NCS, EMG
- CPK
- CSF
- MRI
- Muscular Biopsy
- Nerve Biopsy

Neuropathies

Sensory signs

- Lesion of L5, plexus, sciatic nerf, common or profound peroneal nerve)
- Peroneal nerve compression
- Vasculitis
- CIDP, GBS
- MMN
- CMT, HNPP
- Others NRP



Atteinte radiculo –plexique

- Atteinte L5
  - Mécanique, traumatique
- Atteinte plexique
  - traumatique
  - diabétique
  - dysimmune
  - néoplasique
- Atteinte du nerf fibulaire
  - mécanique, traumatique, HNPP, Kyste, neuroniome, …

Vasculitis

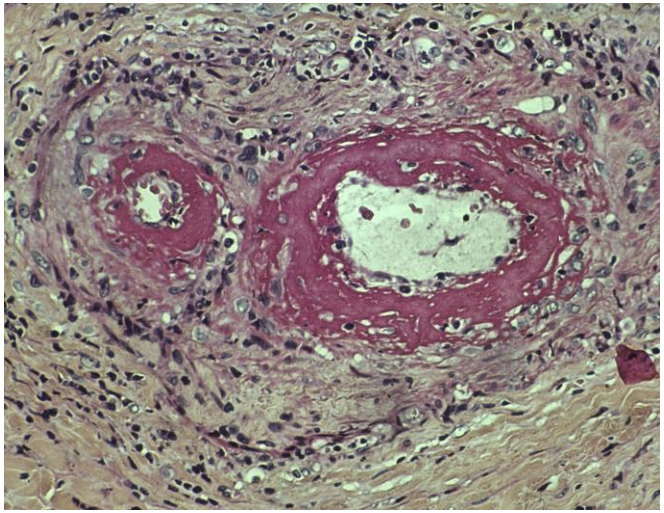
- Sub acute
- Onset age = adult, variable
- Sensory motor deficit
- Painful+++
- Distal
- Asymmetric and multifocal
- Reflex = ±

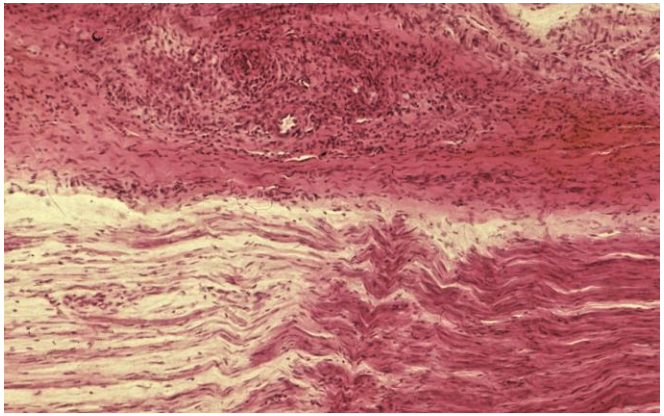
Vasculitis

SR: increased

NCS: Axonal NRP

Nerve biopsy





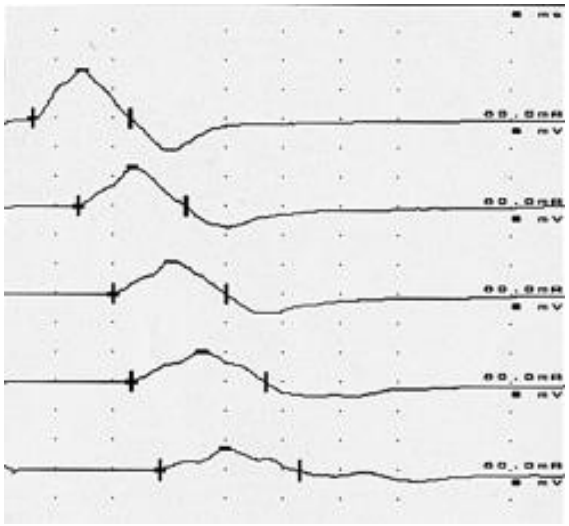
CIDP

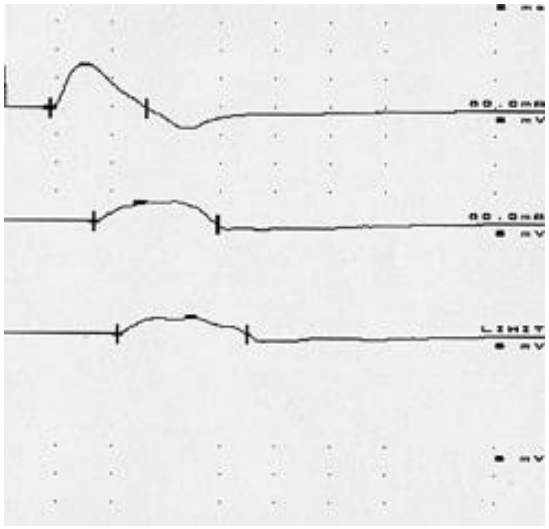
- Family history: -
- Onset age = adult (40)
- Sensory motor deficit
- Distal and proximal, predominantly proximal
- Symmetric
- Diffused areflexia

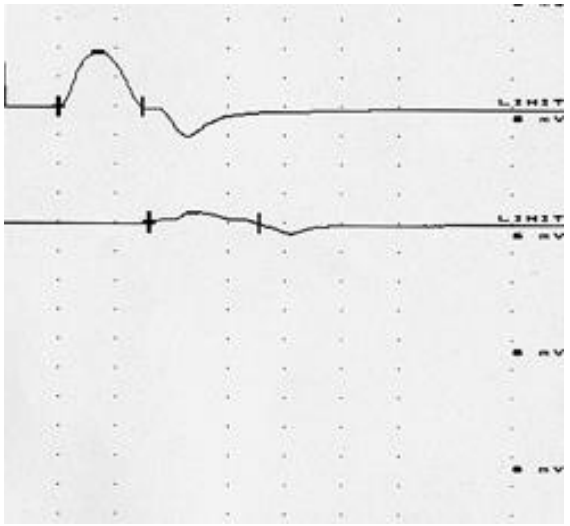
MMN

- Onset age = adult (40)
- Sub acute
- Pure motor deficit
- No muscle atrophy
- Distal, hands
- Asymmetric and multifocal
- Reflex = +

MMN



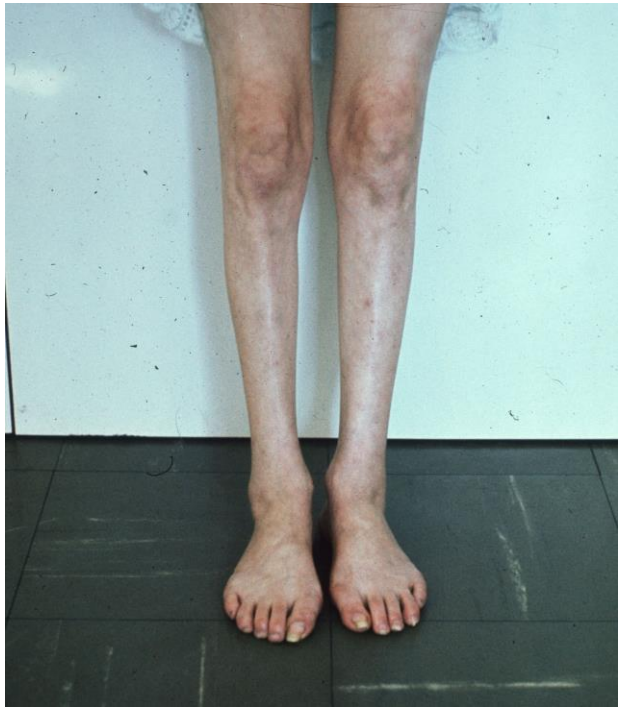


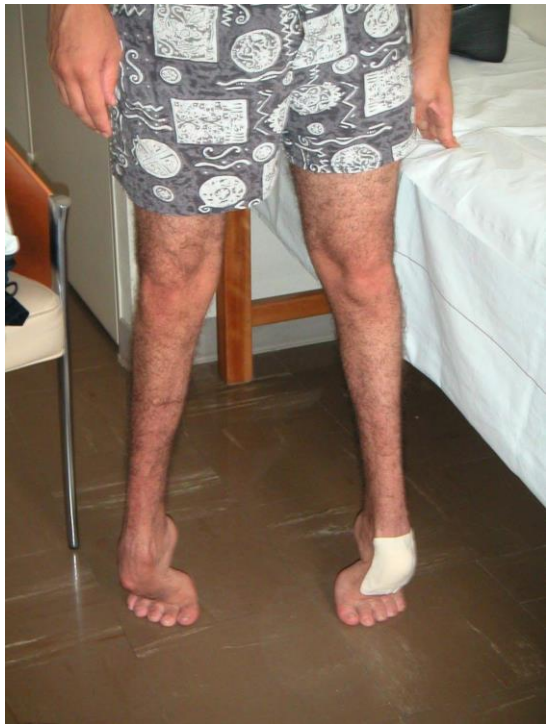


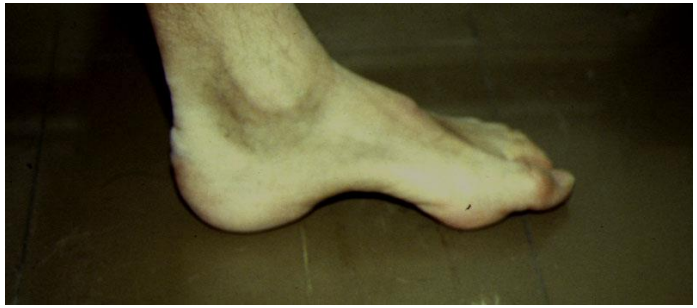
CMT

- Family history
- Onset age = infancy
- Sensory & motor deficit
- Distal limbs distribution
- Symmetric
- Areflexia

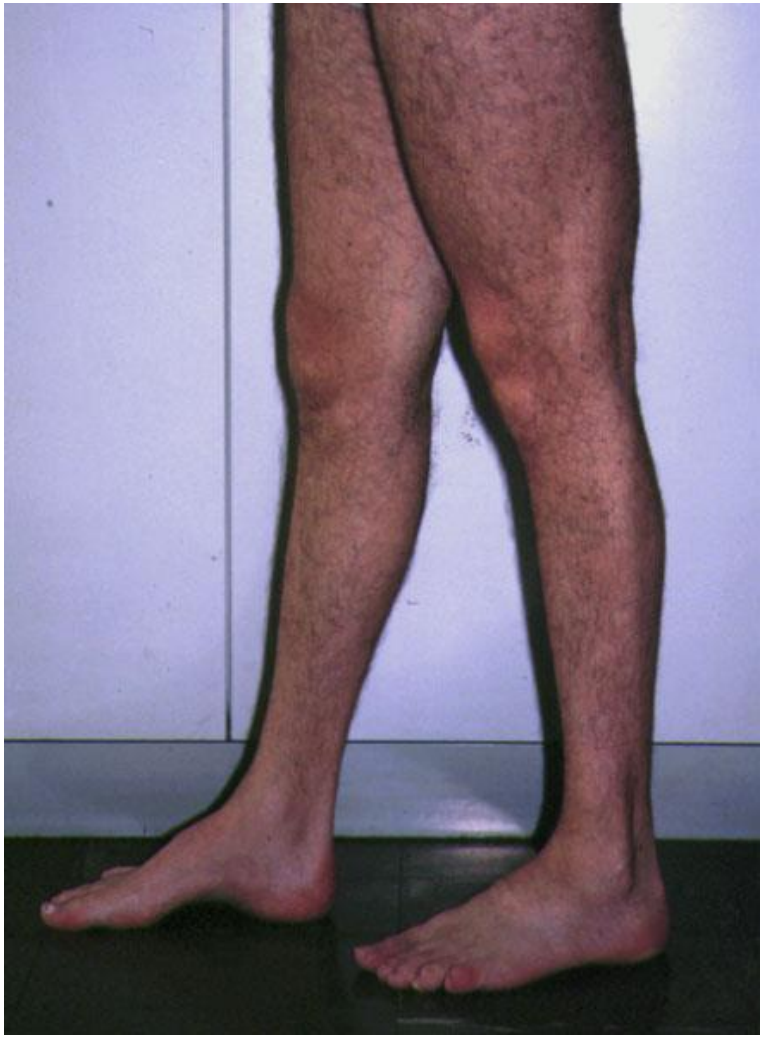
CMT



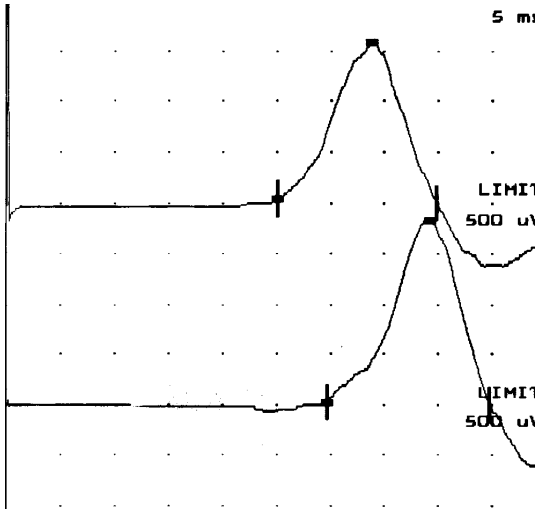


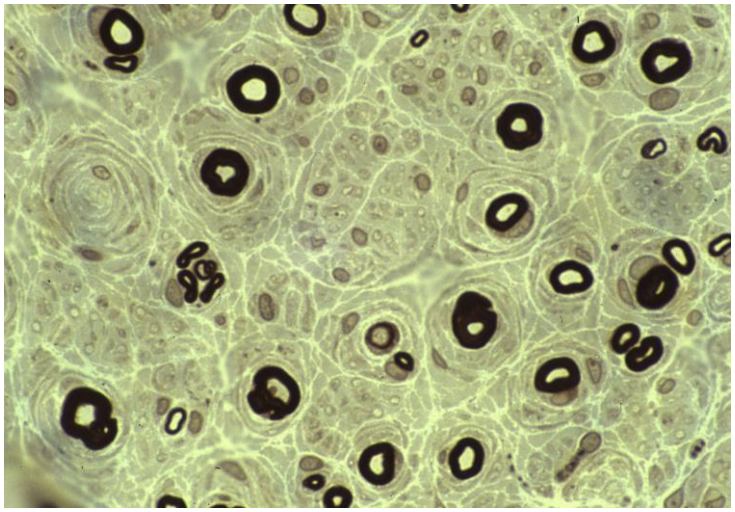


CMT1a



CMT





PMP22, P0, CX 32, MFN

Sensory signs + Foot-drop

Familial:

- Pes Cavus, Symmetric ➒CMT ➒PMP22 and others
- AD +multifocal nerve compression ➒HNNP ➒PMP22

Acquired:

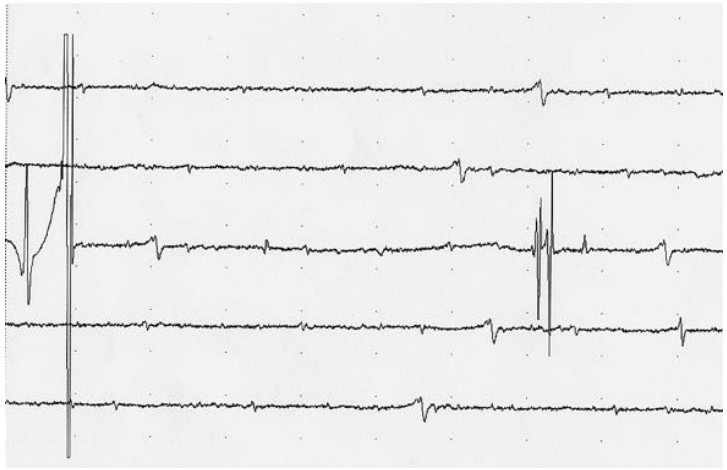
- Fibular nerve entrapment
- Proximo-distal +Symmetric+ areflexia ➒CIDP ➒NCS +CSF
- Distal + Multifocal + Asymmetric+ without atrophy ➒MMN ➒ NCS+GM1
- Lewis and Sumner
- Pain +Asymmetric +multifocal ➒vasculits➒NCS + Nerve biopsy

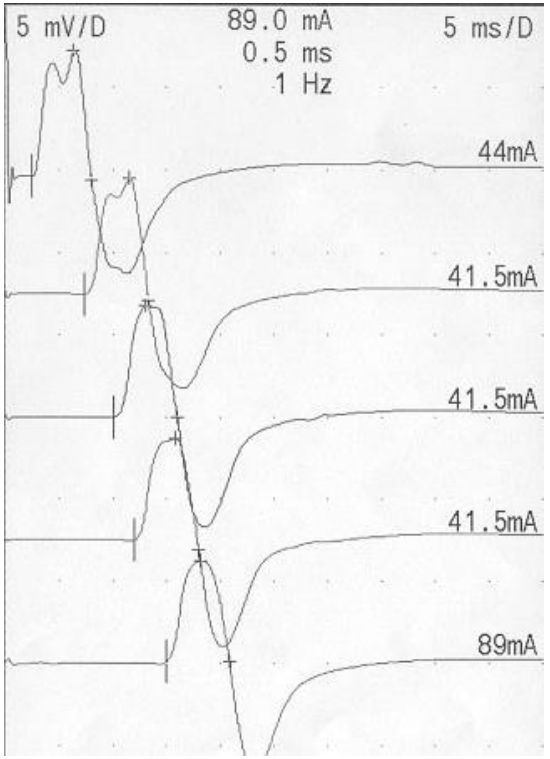
Anterior horn

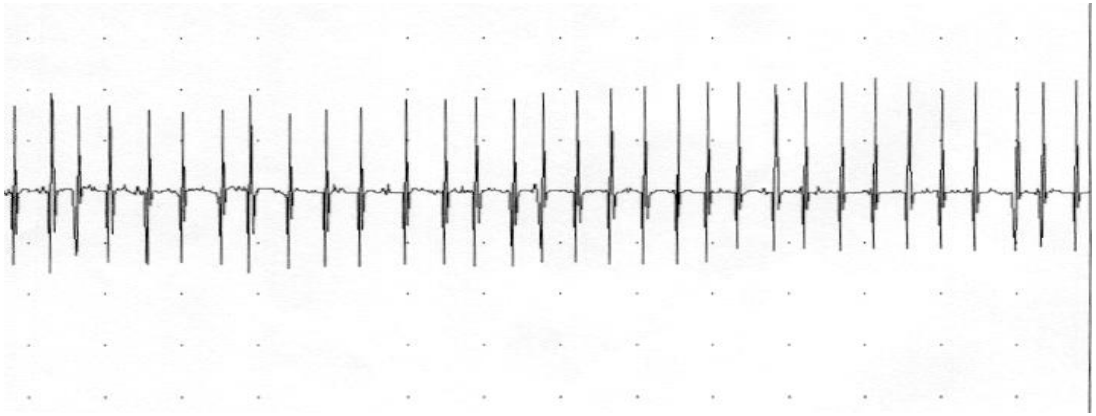
- ALS
- Post polio
- SMA: rare
  - SMA with SMN deletion
  - Stark kaeser sd
  - CMT spinal

ALS

- Family history ?
- Pure motor weakness
- Rapid progression
- Cramp & fasciculation
- Muscle Atrophy ++
- UMN signs
- Bulbar and respiratory involvement




5 mV/D
89.0 mA
0.5 ms
1 Hz
5 ms/D
44mA
41.5mA
41.5mA
41.5mA
89mA

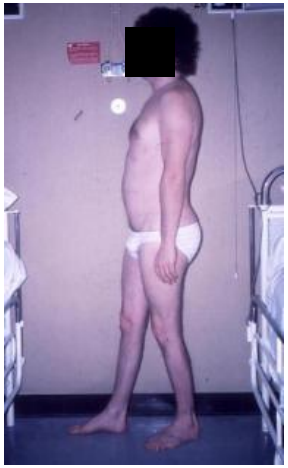


Scapulo-Peroneal peripheral neuropathy

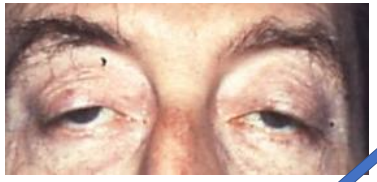
- Autosomal Dominant
- Weakness: Scapuloperoneal; Foot-drop , hand muscles
- Vocal Cord
  - Altered ; Hoarseness
  - Stridor
- Electrophysiology: Neurogenic EMG
- Mutation of ***TRPV4***

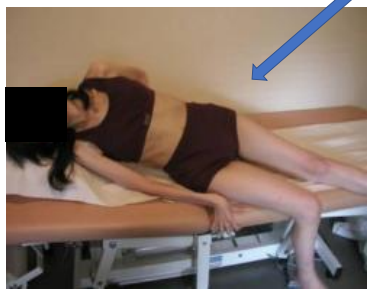
Démarche diagnostique I : Penser à une affection musculaire

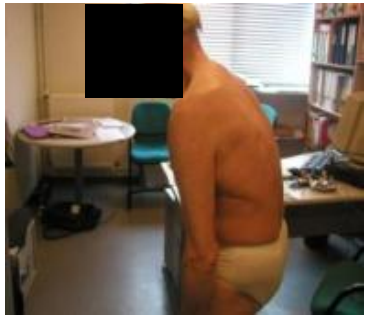
Moins aisé



Prédominance distale de l'atteinte (neurogène ?)







Hypotonie néonatale

Déficit axial au 1er plan

Atteinte respiratoire au 1er plan

Ophtalmoplégie (ptosis)

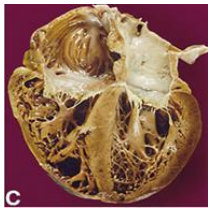
Absence de déficit

Symptomatologie transitoire

Fatigabilité / douleurs effort

Atteinte cardiaque au 1er plan

Atteinte systémique au Ier plan (SNC, neuropathie)



MÉDECINE SORBONNE UNIVERSITÉ

Faculté de Médecine Aix*Marseille Université

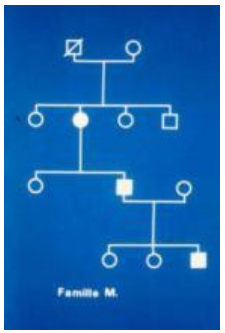
Diplôme Inter-Universitaire de Myologie

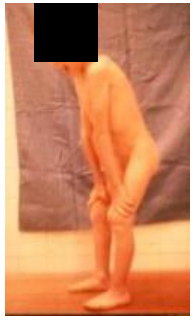
Année Universitaire 2019-2020

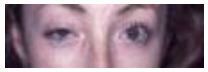
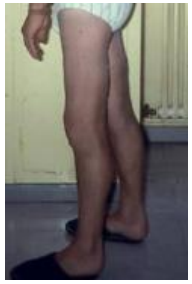
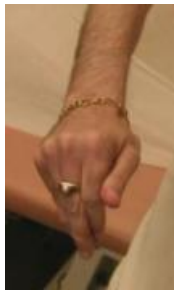
Responsables de l'Enseignement : *Dr Guillaume Bassez*
Mme Gisèle Bonne, Pr Shahram Attarian

Démarche diagnostique II :
Réunir les éléments cliniques et paracliniques

- hérédité
- âge de début / profil évolutif
- topographie du déficit :
  symétrie, sélectivité, atteinte de la face
- atrophie / hypertrophie
- rétractions (topographie)
- myotonie
- atteinte oculomotrice et/ou ptosis
- atteinte vélopharyngée
- intolérance à l'effort / rhabdomyolyse
- atteinte cardiaque et respiratoire
- autres organes (SNC, SNP, œil, thyroïde...)
- contexte iatrogène et général





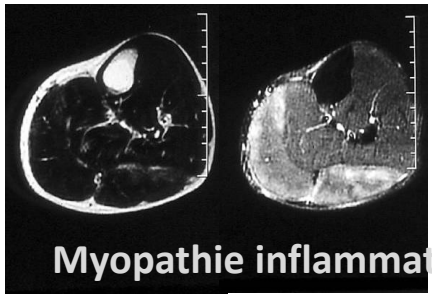




Affections musculaires : Examens complémentaires

- Confirmation du processus musculaire
  - CPK, électromyographie,
  - Imagerie : scanner et IRM musculaire
- Étiologie
  - Biopsie musculaire, biologie (immuno, dosages métaboliques…), génétique
- Dépister les complications
  - Bilan respiratoire : EFR, gaz du sang
  - Bilan cardiaque : ECG, Holter, échographie

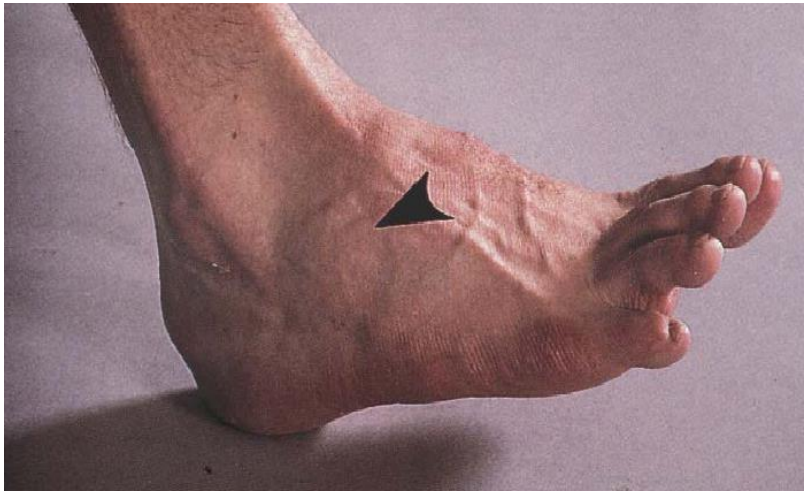

Calpaïnopathie


Bethlem (Collagène 6)

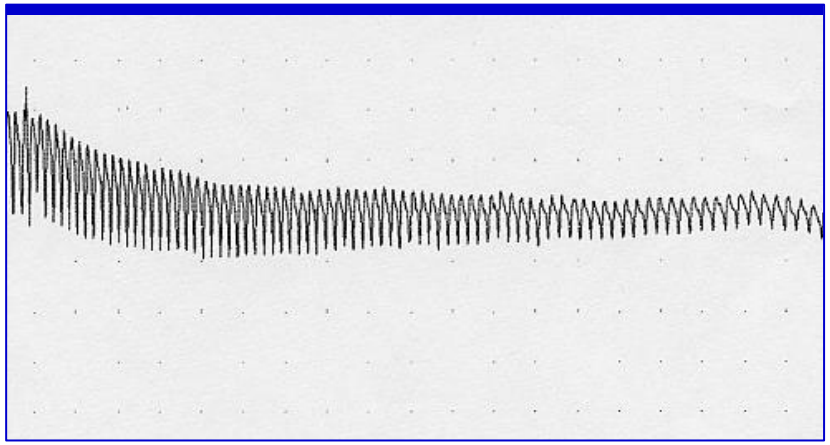

Myopathie inflammatoire

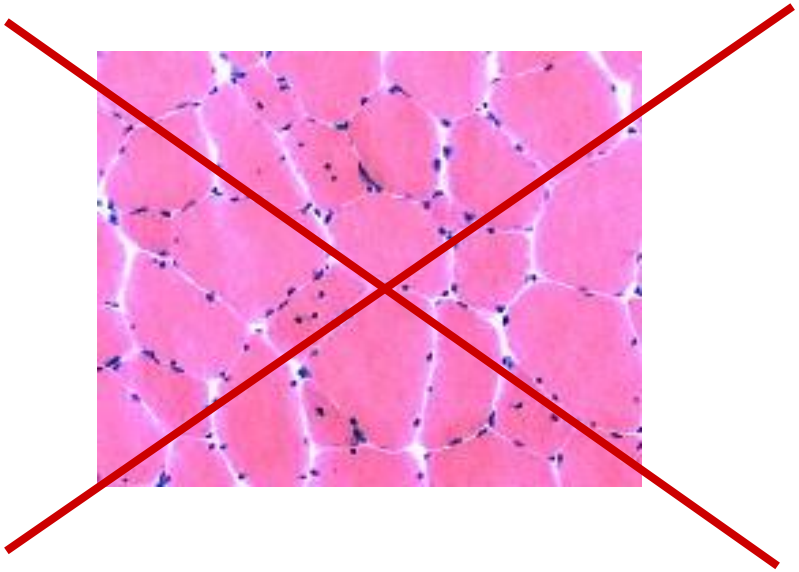
Myopathies

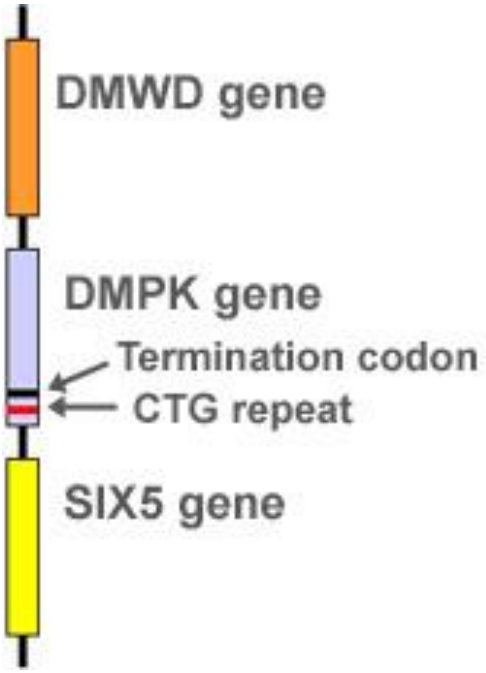
- Myotonic Dystrophy
- FSHD
- IBM
- Distal myopathies
- Scapuloperoneal myopathies
- Others: very rare



MD1







FSHD

Facial muscles

Shoulder-girdle muscles

Foot extensors

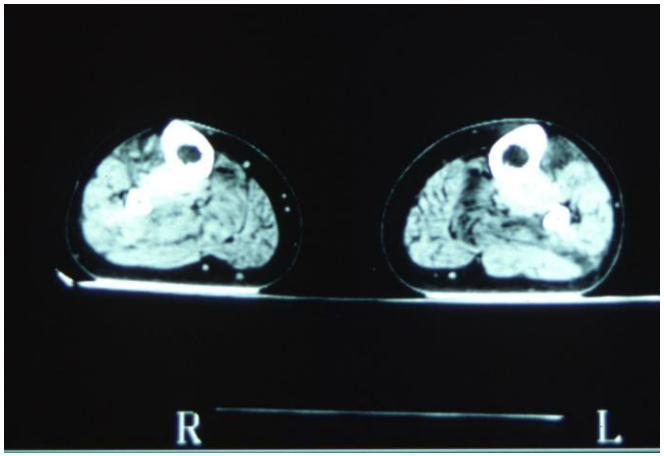
Pelvic girdle muscles

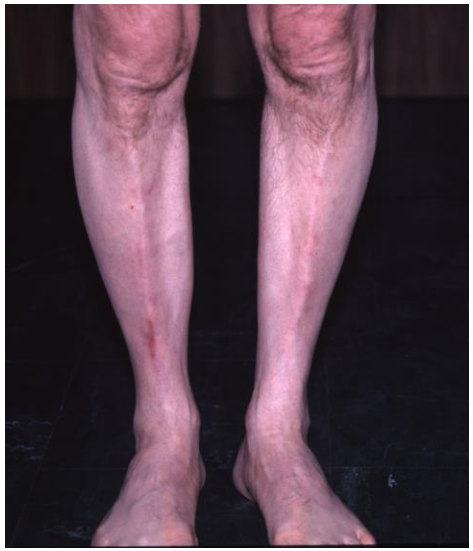
Respect of SCM, Deltoid

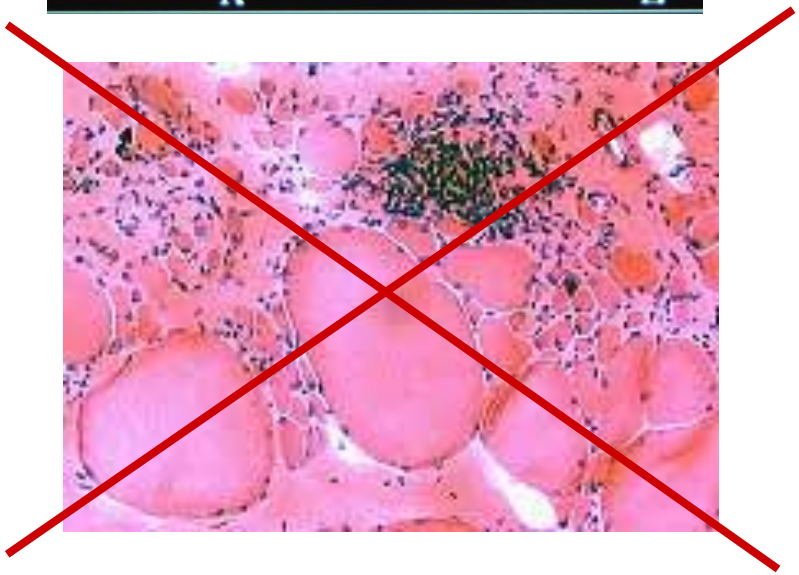
Asymmetry

Selectivity

FSHD: SP form

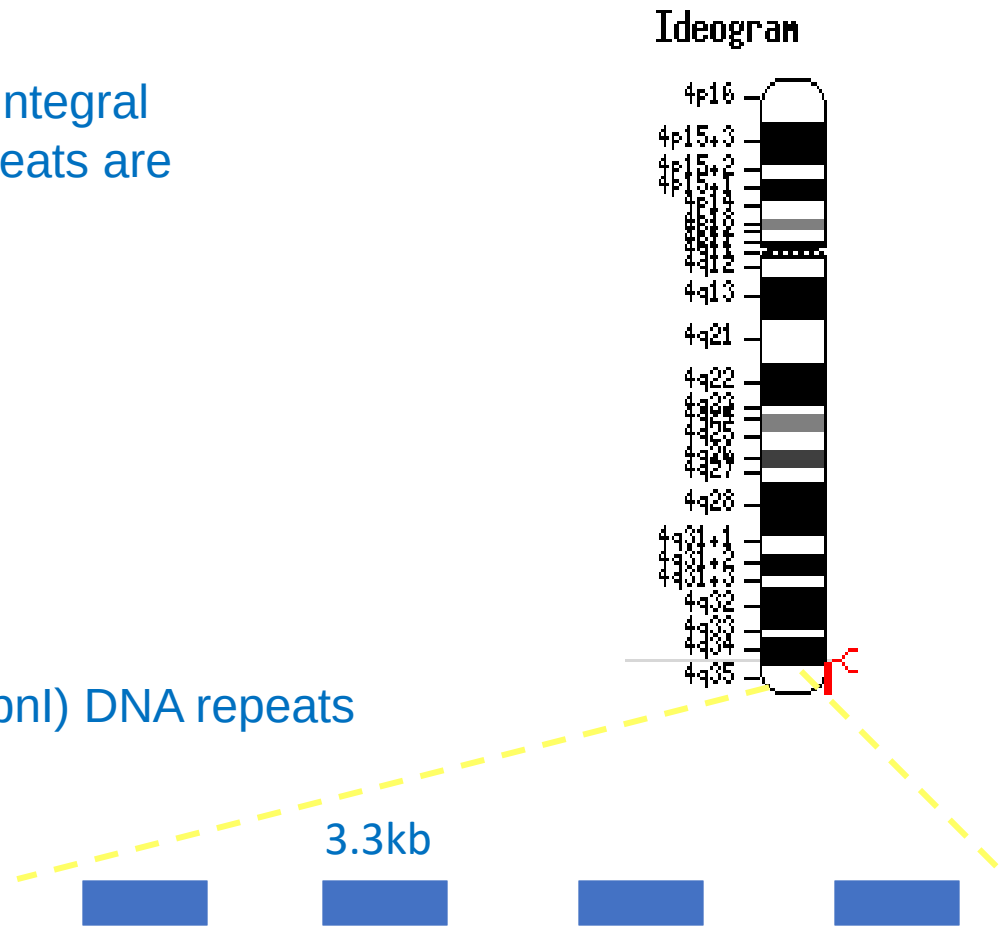






Molecular Genetics

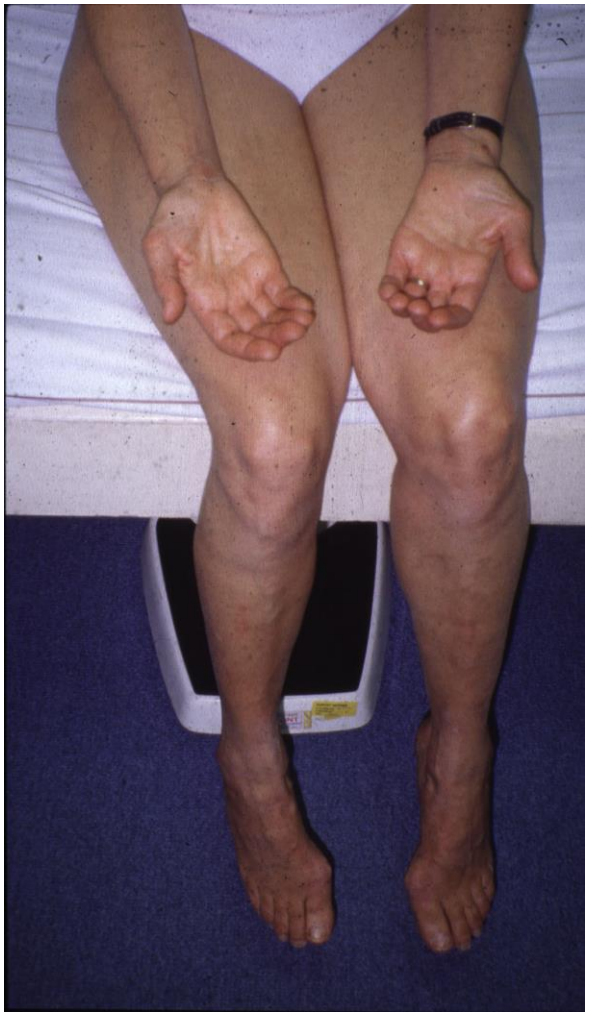
In patients an integral number of repeats are deleted

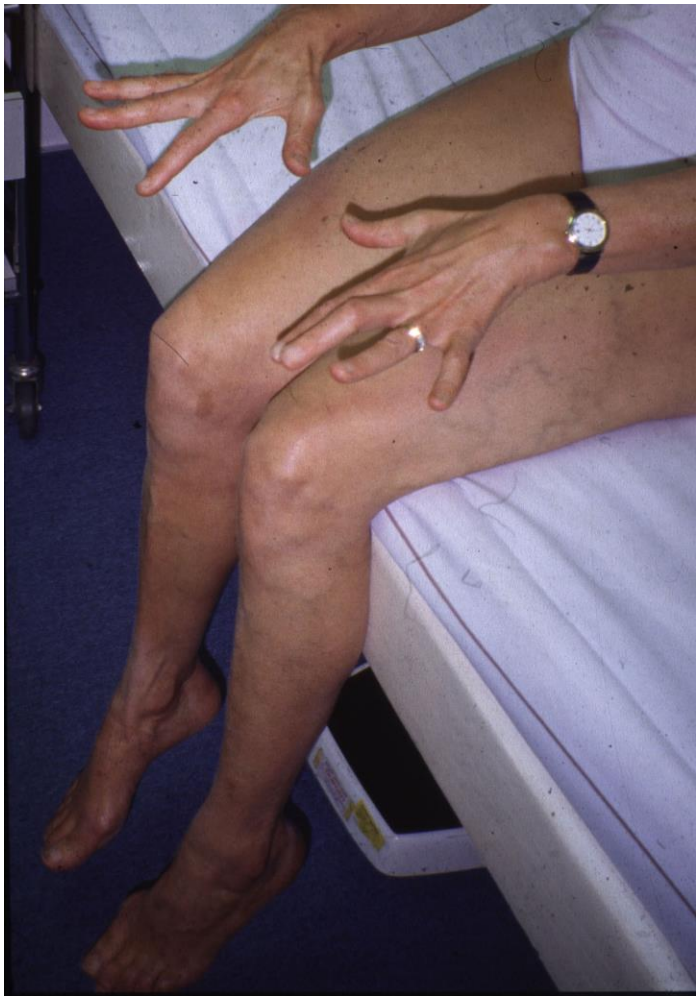


IBM

- Family history ?
- Onset age = late, 50-60
- Proximal and distal muscle weakness
- Hand Flexors +++
- Asymmetric
- Slow progression

IBM





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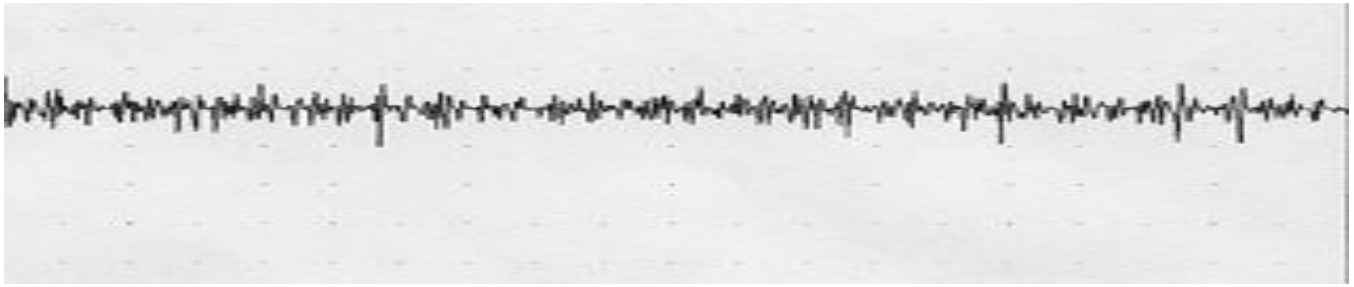

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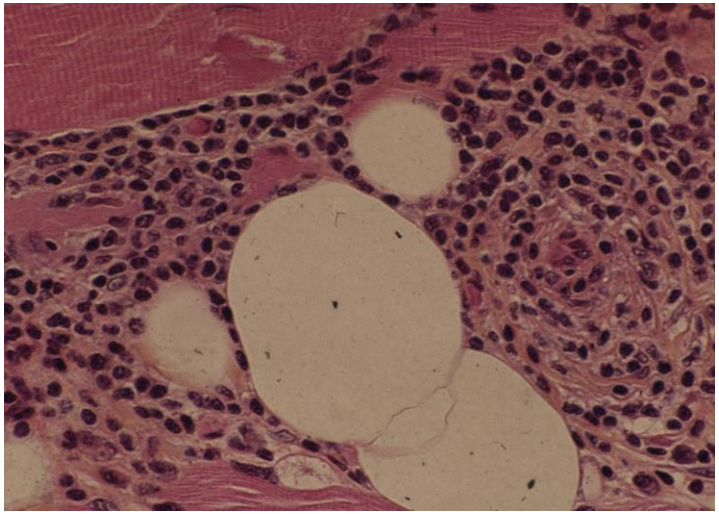
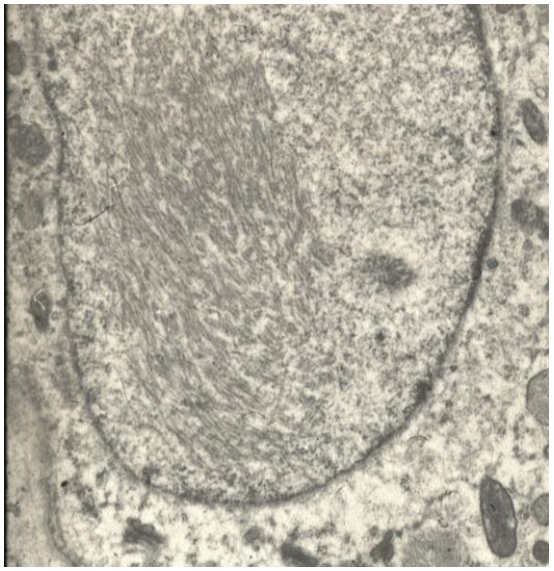
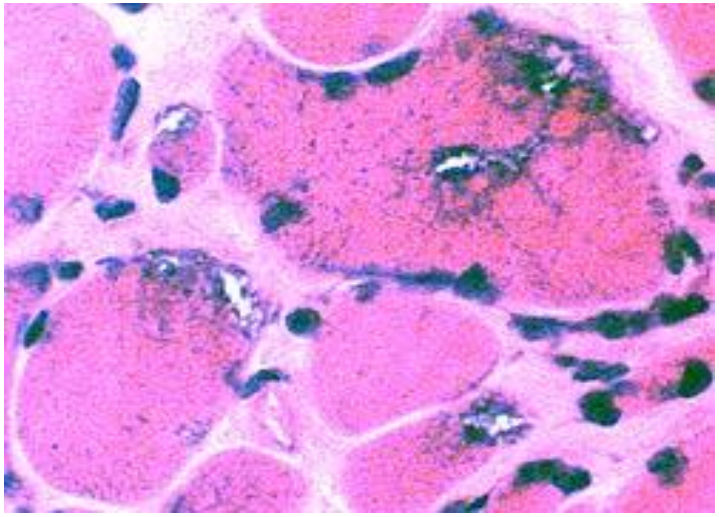
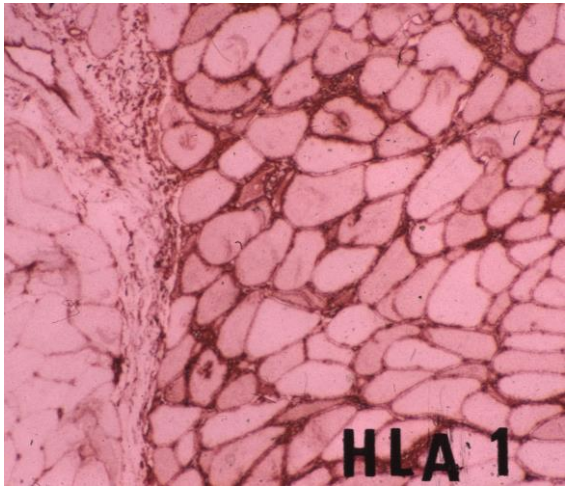








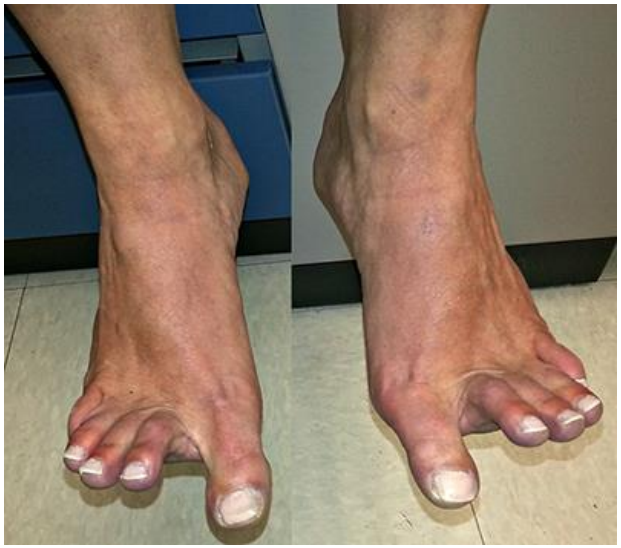
Myopathies

- SP muscular dystrophy, Type 1 (Hyaline body myopathy): FHL1
- SP muscular dystrophy, adult onset, Type 2 (Hyaline body myopathy): MYH7
- SP muscular dystrophy, Type 3 (Hyaline body myopathy; myosin storage): DES
- SP muscular dystrophy with mental retardation & lethal cardiomyopathy: LAMP-2
- Kaeser syndrome: Desmin
- Association of an ACTA1 Mutation with a dominant progressive SP Myopathy in an Extended Family (Zukosky et al, JAMA Neurol 2015)

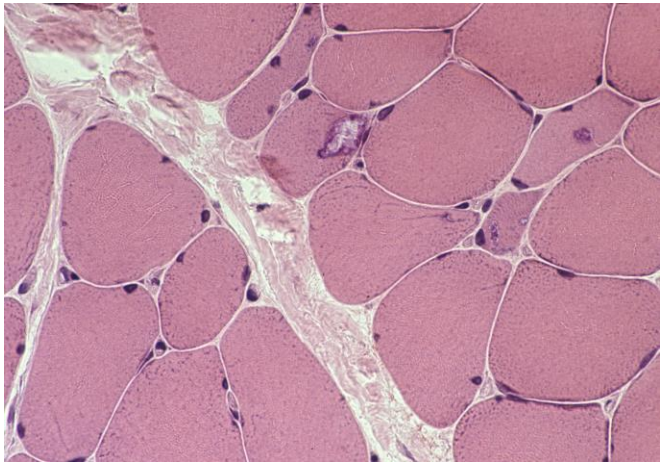
Distal Myopathies

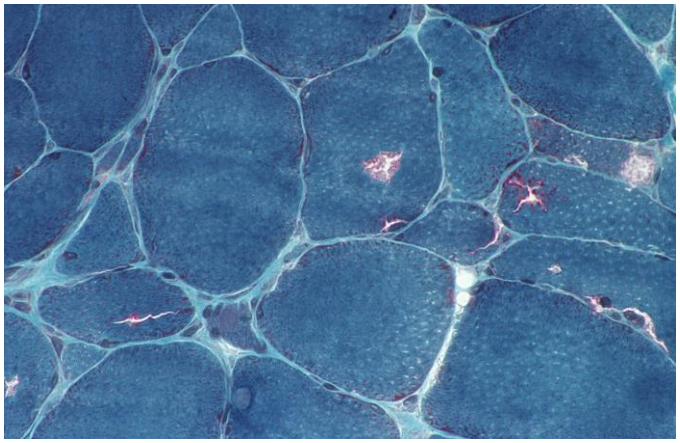
Anterior prominent

- MYH7
- FHL1
- NEB
- GNE
- Titin
- MATR3
- VCP
- + Lateral
  - DES
  - CRYAB

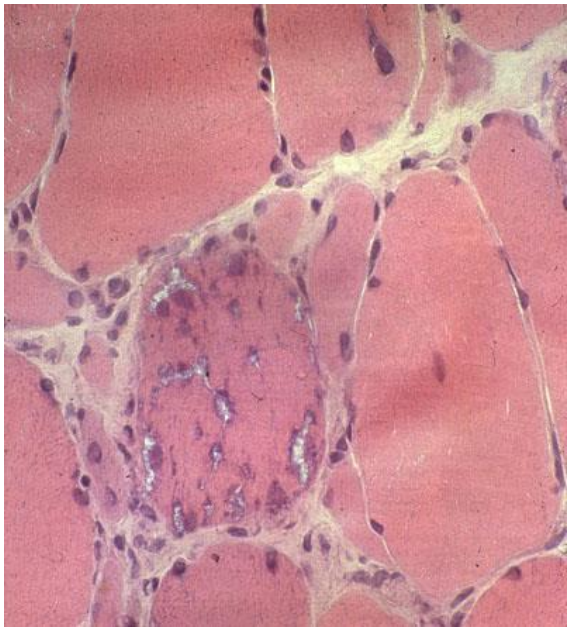


hIBM2; Nonaka

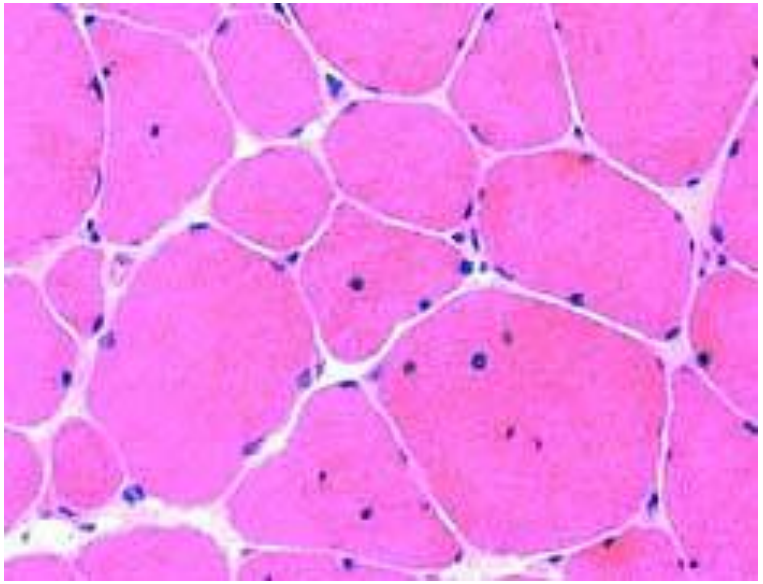


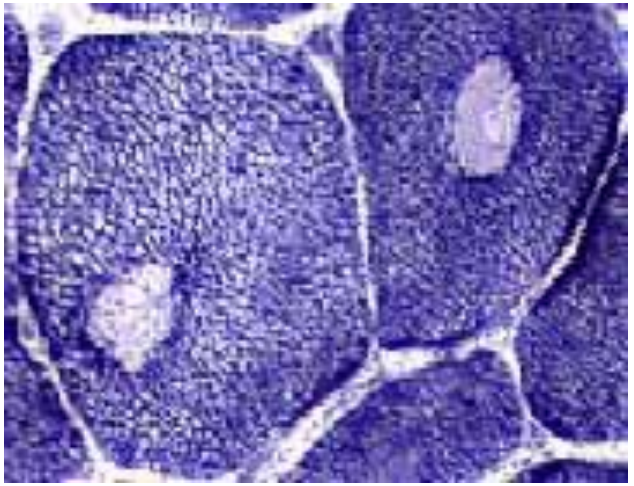


IBM2; Nonaka



Central core





34 Y

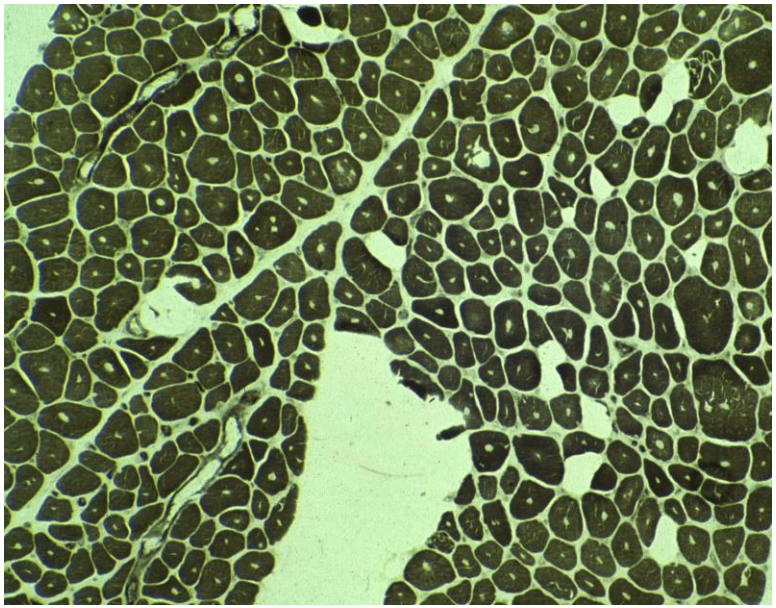
Scapuloperoneal weakness

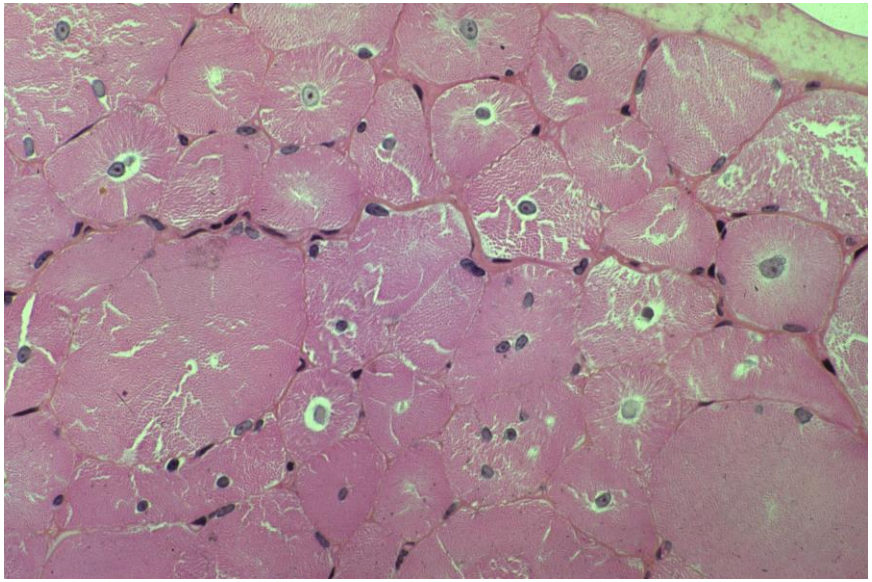
Facial weakness

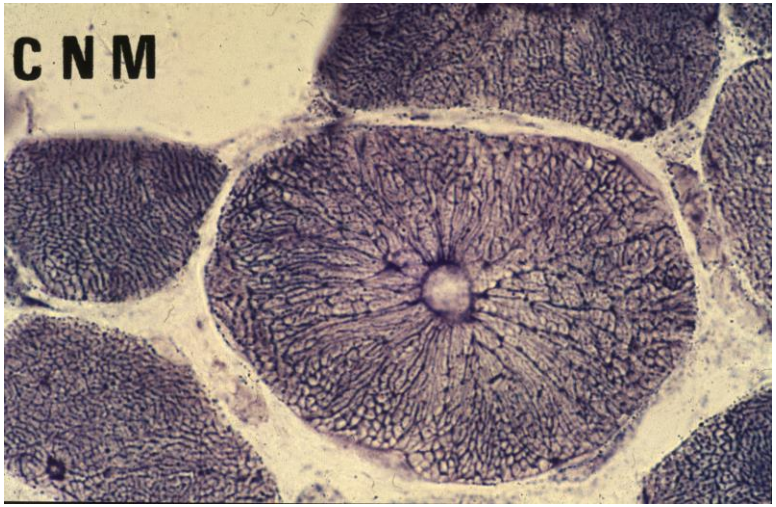
Onset in childhood

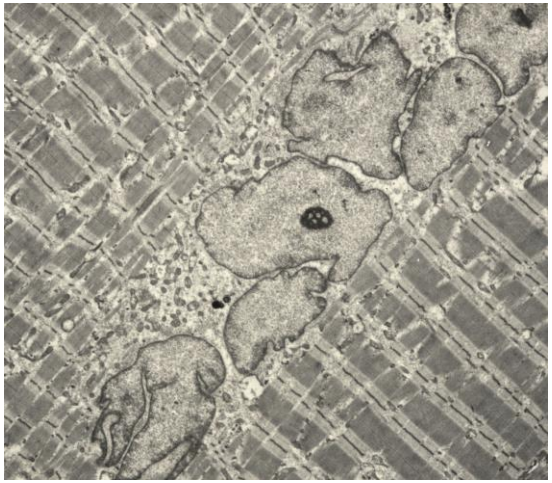
Mother had myopathy

Centronuclear









Myopathic Foot-drop

- MD
- FSHD
- IBM
- Distal myopthies
  - Titinopathy
  - hIBM
  - Scapuloperoneal

Sensory signs + Foot-drop

Familial:

- CMT
- HNNP

Acquired:

- CIDP
- MMN, Lewis and Sumner
- vasculits