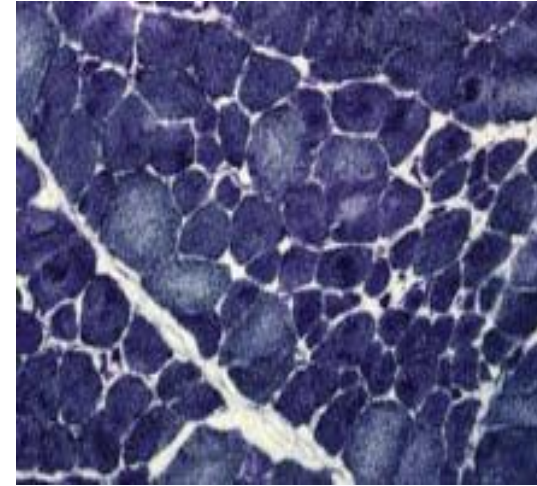




CONTRIBUCIÓN DE LA RESONANCIA MAGNÈTICA AL DIAGNÓSTICO DE LAS ENFERMEDADES MUSCULARES

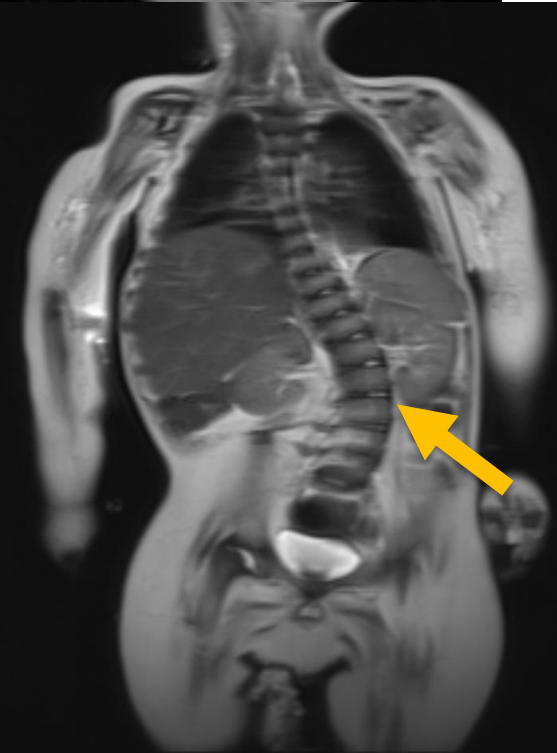
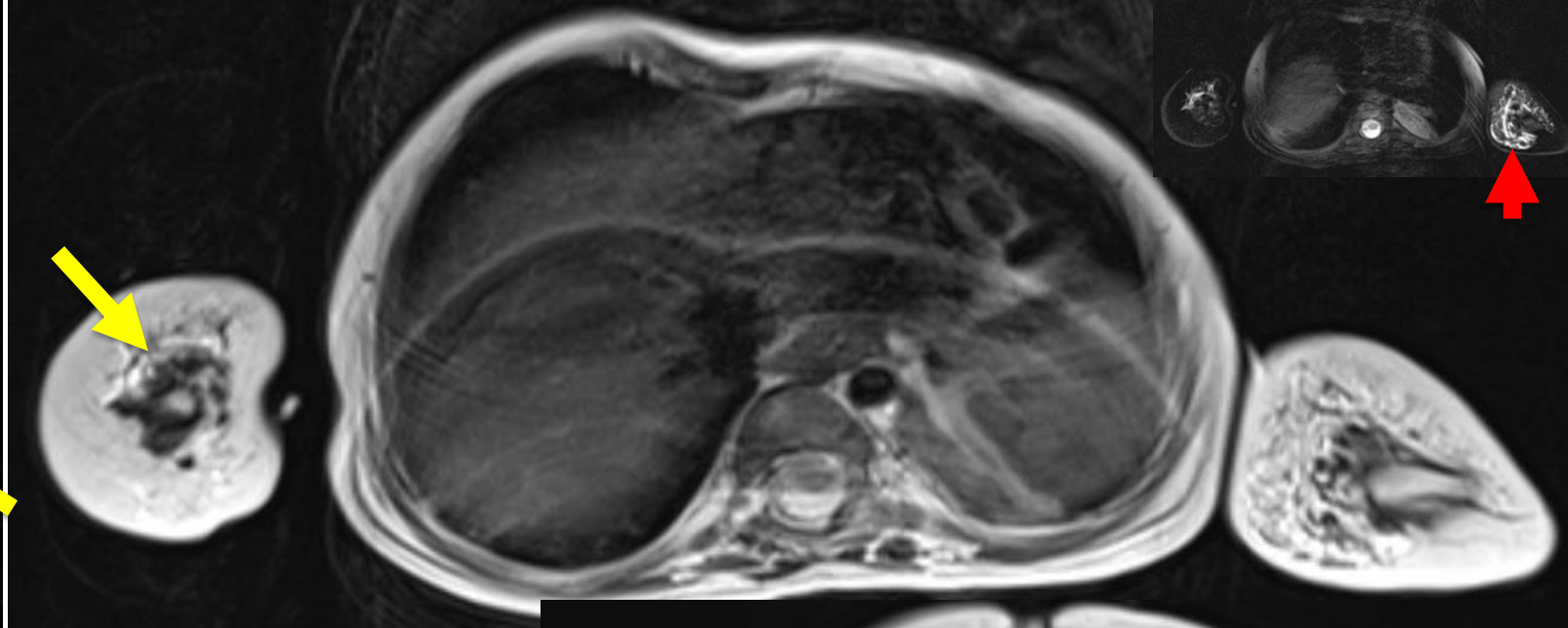
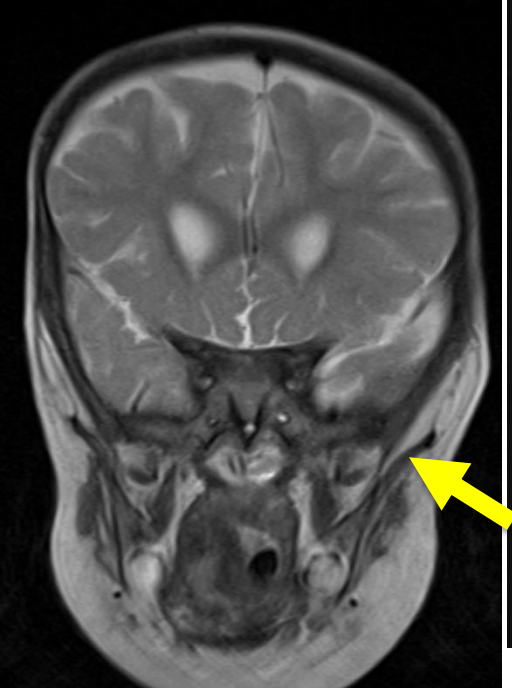
Francina Munell MD, PhD
Neurologia Pediàtrica
Hospital Universitario Vall d'Hebron
Barcelona





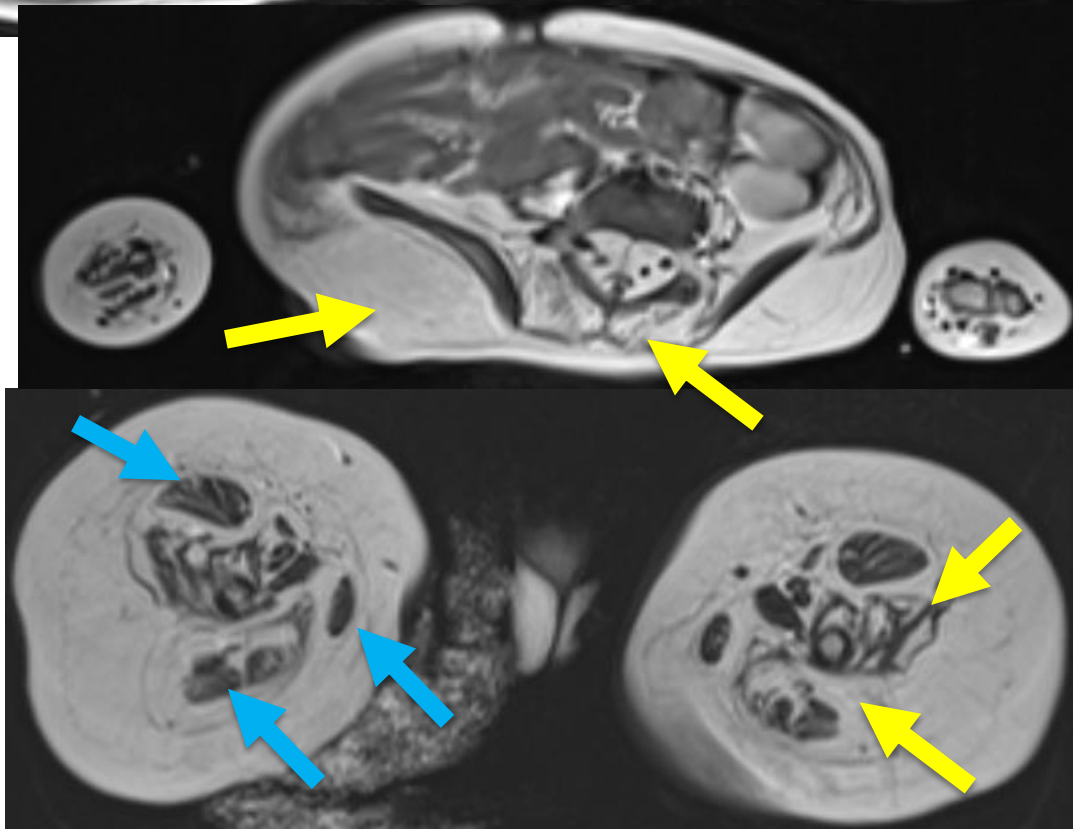
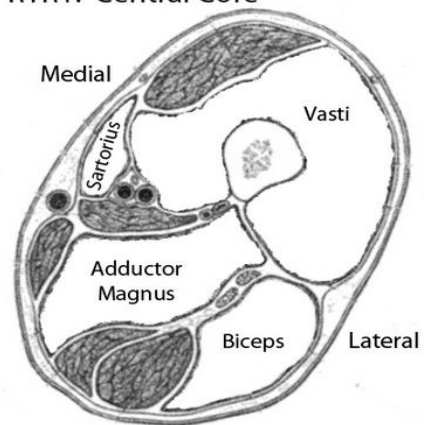
Muscle biopsy: large areas of myofibrillar disorganization

20 mo boy with hypotonia and
severe oromotor disorder



NFO 2012

RYR1: Central Core

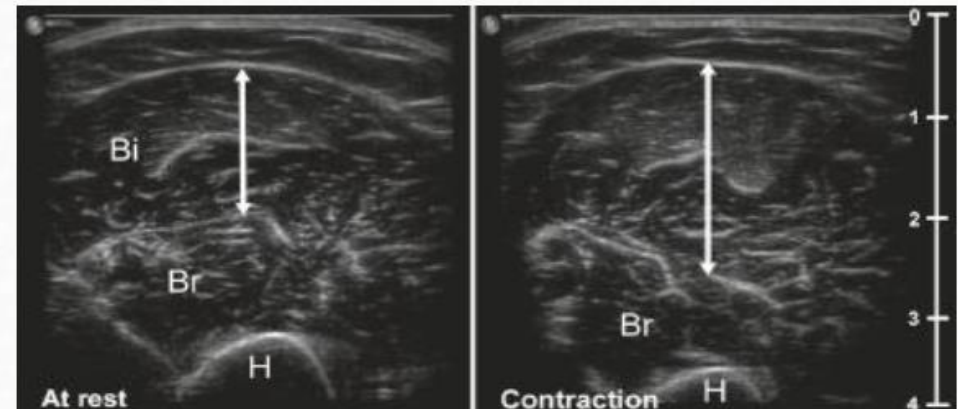
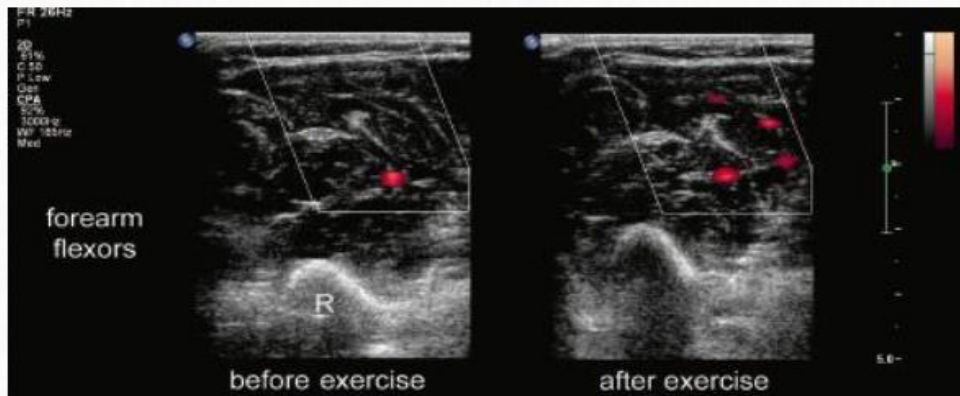
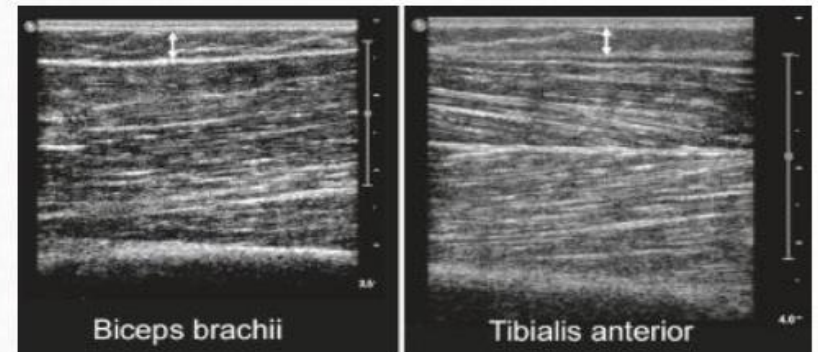


1. Imaging Modalities

1. US
2. CT
3. MRI

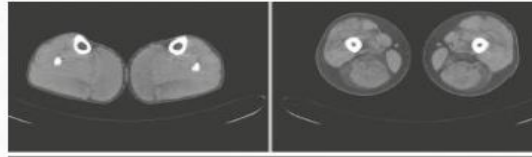
US

- Superficial Muscles
- No sedation
- Easy access, bedside
- Dynamic, real-time imaging
- Doppler
- Myopathies: hyperechogenic (fat and fibrosis)



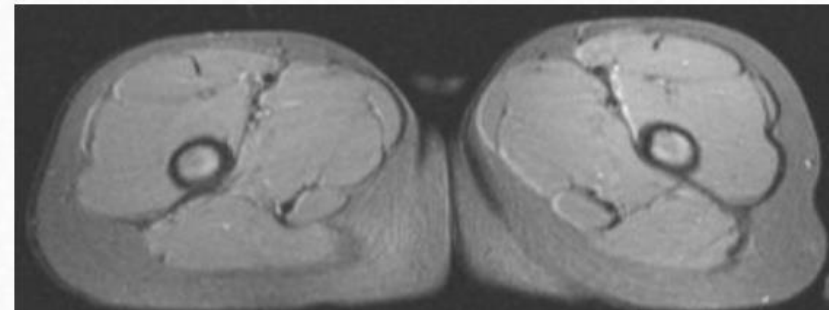
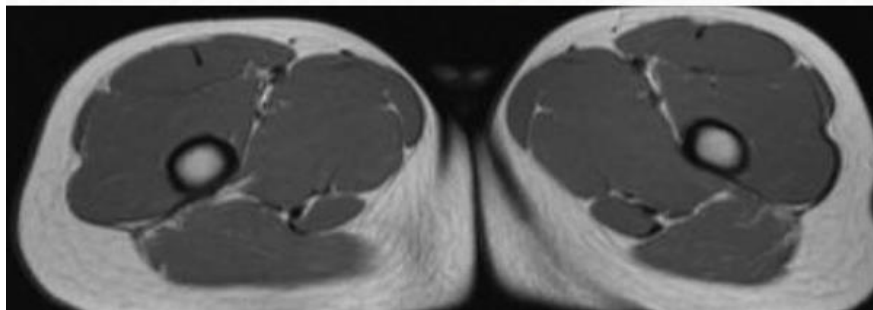
CT

- Replaced by MRI (exc.: pacemaker, claustrophobia...) / **RADIATION**
- Discrete inflammatory changes and muscle edema can be easily missed



MRI

- The most important imaging modality in the diagnosis of muscle pathology
- Whole-Body MRI: from head to toes (around 45')
- T1WI and fat-suppressed T2WI are the workhorses in the routine clinical setting
- Assessment of muscular pattern of involvement
- Visual rating scales for fatty replacement and muscle edema
- Muscle edema and represent the first stage of muscle degeneration
- Helpful in directing the muscle biopsy

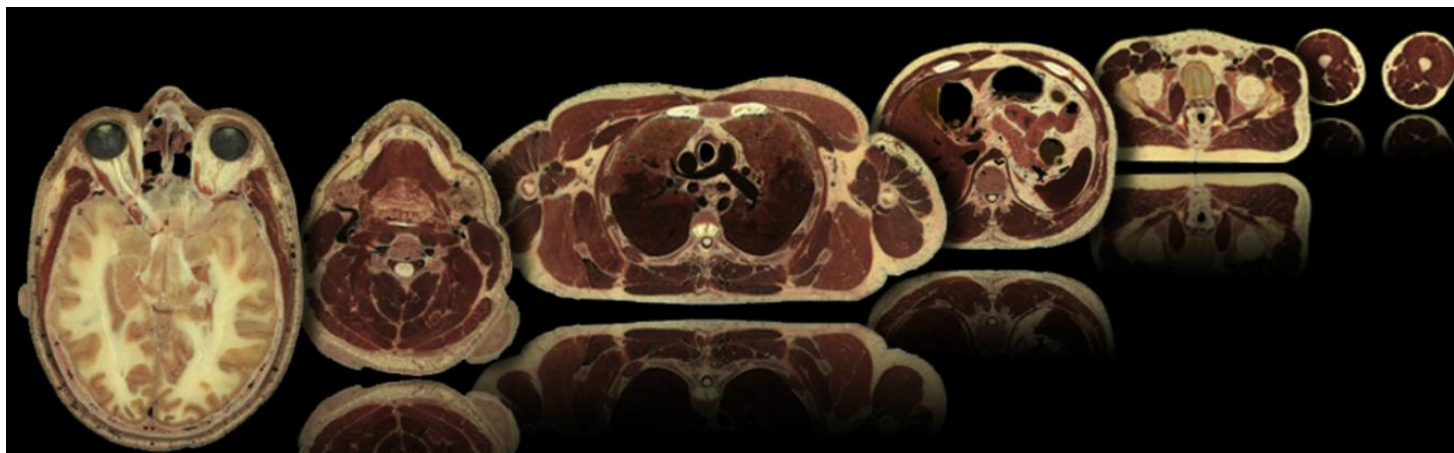
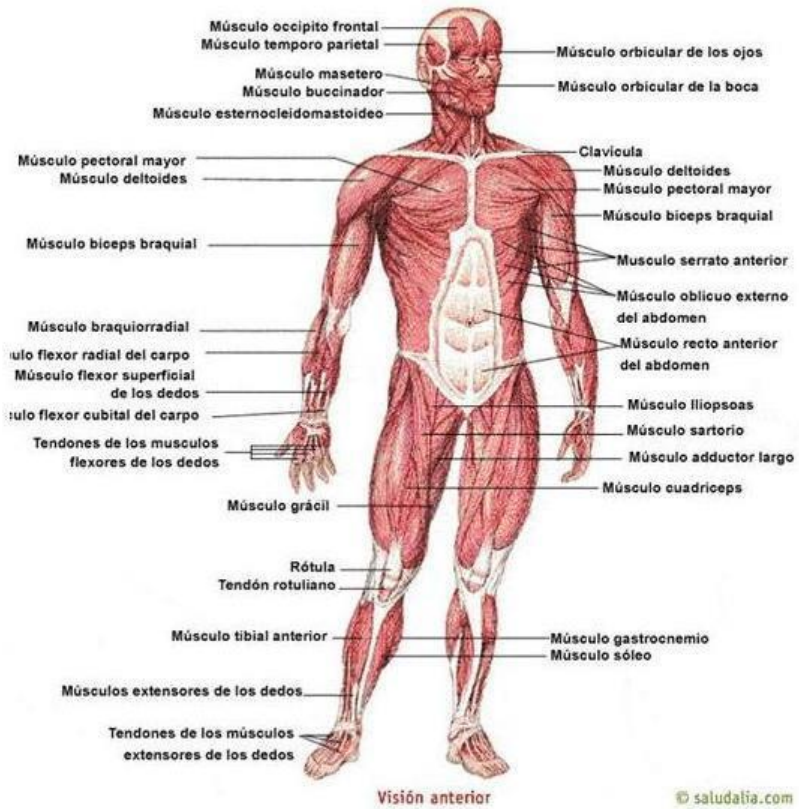
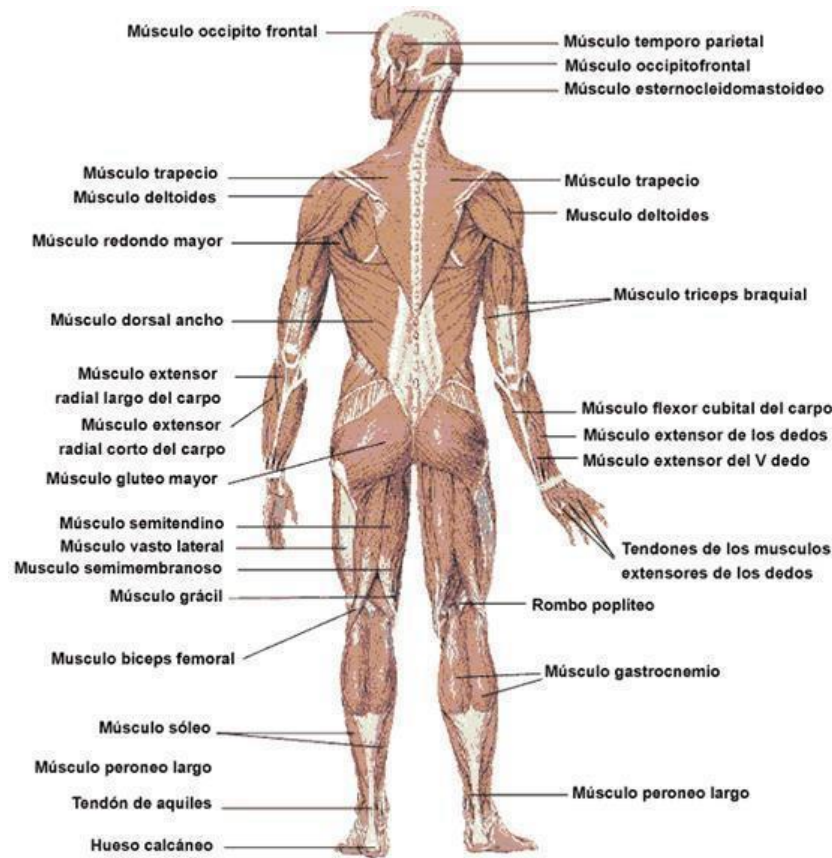




IDI
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Siemens Magnetom Trio (3,0 T)
Siemens Magnetom Avanto (1,5 T)

2009
2011



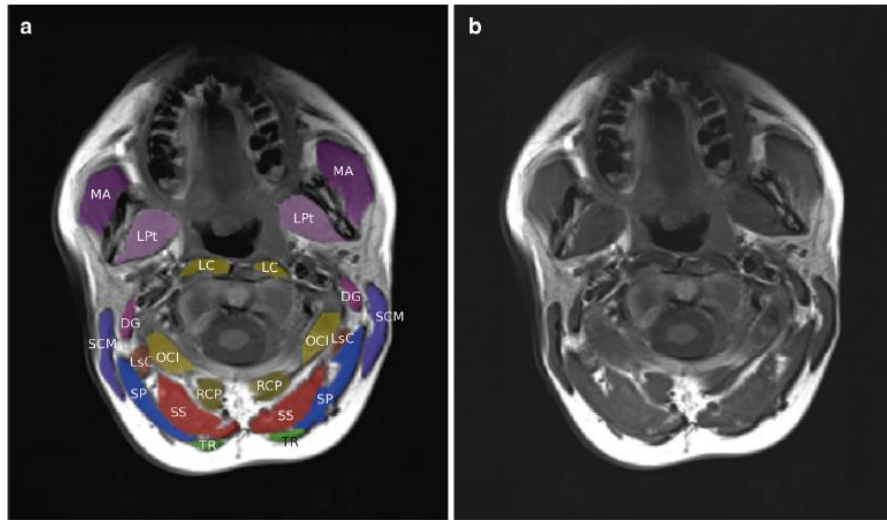


Fig. 10.2 Neck muscles at the level of C1/C2. *DG* digastric muscle, *LC* longus colli muscle, *LsC* longissimus colli, *LPt* lateral pterygoid muscle, *MA* masticator muscle, *OCI* obliquus capitis inferior muscle, *RCP* rectus capitis

posterior muscle, *SCM* sternocleidomastoid muscle, *SP* splenius capitis, *SS* semispinalis capitis and cervicis, *TR* trapezius muscle

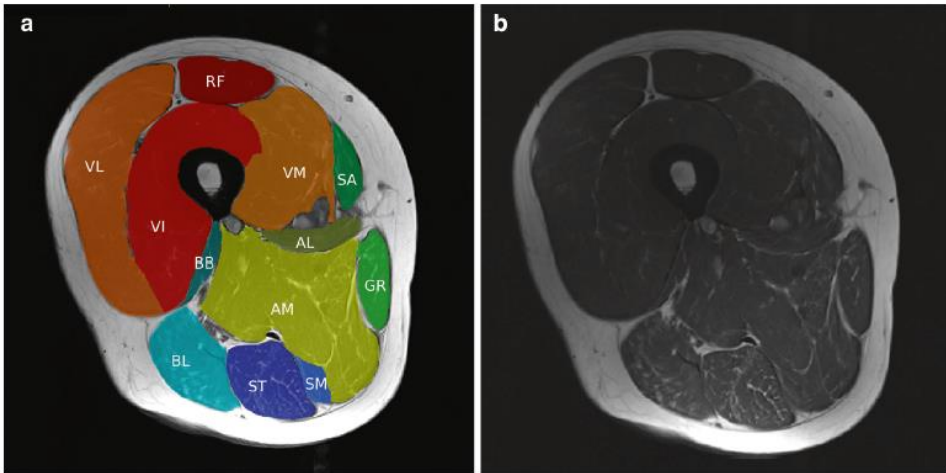


Fig. 10.24 Muscles of the thigh. *AL* adductor longus muscle, *AM* adductor magnus muscle, *BB* biceps femoris muscle short head, *BL* biceps femoris muscle long head, *GR* gracilis muscle, *RF* rectus femoris muscle, *SA* sartorius muscle, *SM* semimembranosus muscle, *ST* semitendi-

nus muscle, *VI* vastus intermedius muscle, *VL* vastus lateralis muscle, *VM* vastus medialis muscle. Quadriceps muscle in shades of *red*, hamstrings in *blue*, adductors in *yellow*

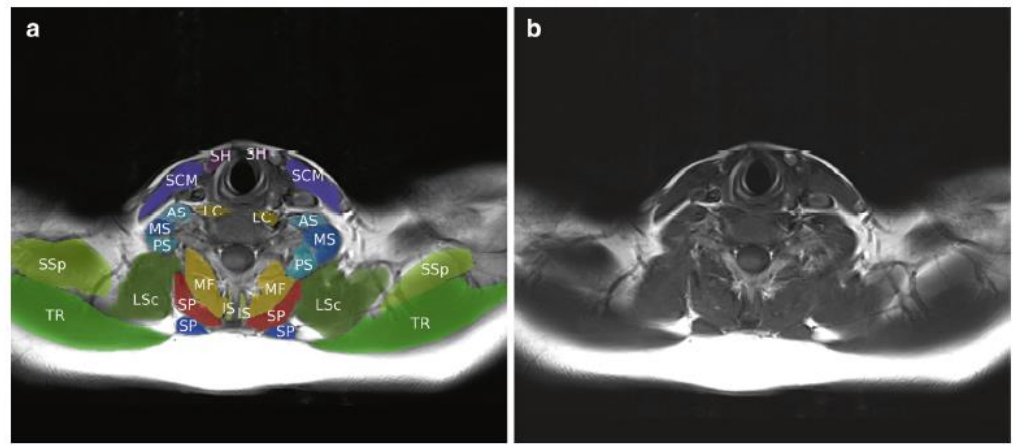


Fig. 10.4 Neck muscles at the level of C6/C7. *AS* anterior scalenus muscle, *IS* interspinosus muscle, *LC* longus colli muscle, *LSc* levator scapulae muscle, *MF* multifidus muscle

scalenus muscle, *SCM* sternocleidomastoid muscle, *SH* sternohyoid muscle, *SP* splenius capitis/cervicis muscle, *SSp* supraspinatus muscle, *TR* trapezius muscle

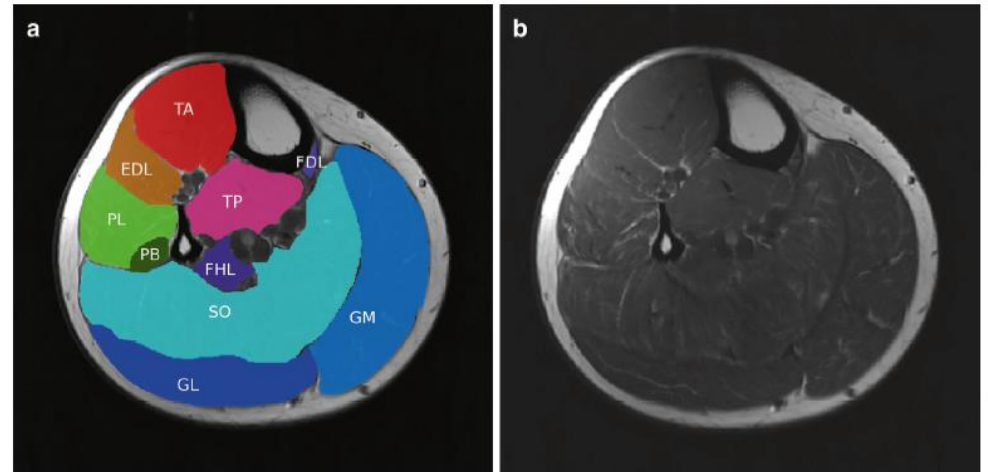
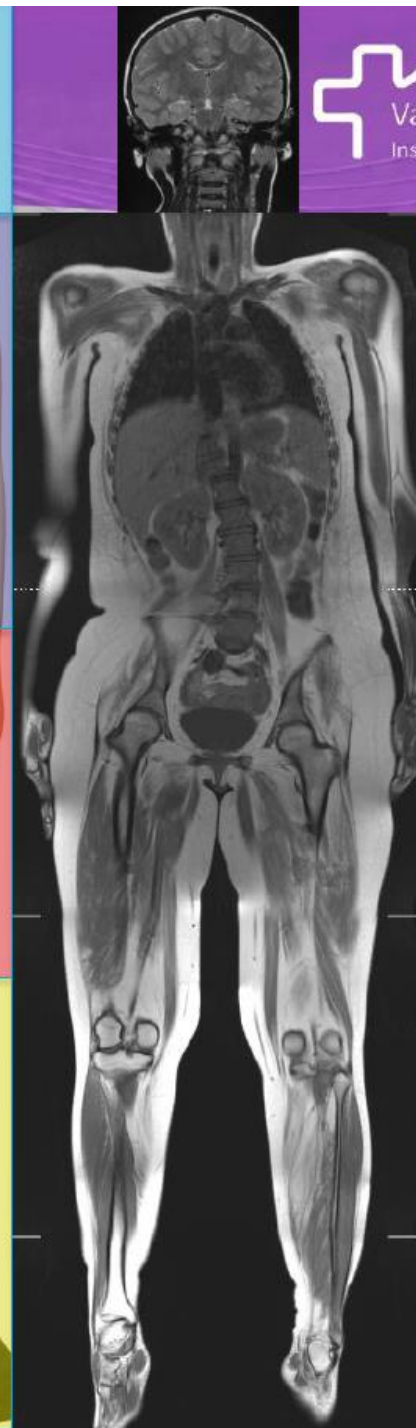
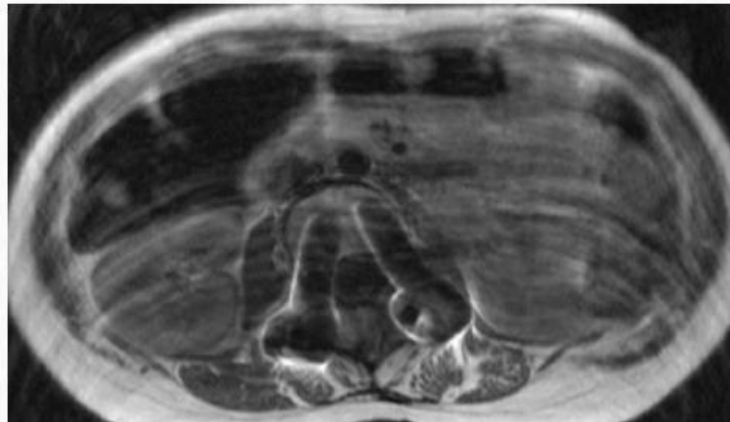
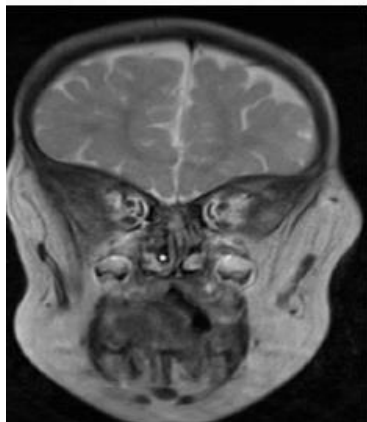


Fig. 10.29 Muscles of the calf. *EDL* extensor digitorum longus, *FDL* flexor digitorum longus muscle, *FHL* flexor hallucis longus muscle, *GM* medial gastrocnemius muscle, *GL* lateral gastrocnemius muscle, *PB* peroneus brevis

muscle, *PL* peroneus longus muscle, *TA* tibialis anterior muscle, *TP* tibialis posterior muscle. Triceps surae in shades of *blue*

3. Clinical Applications – WB MRI

- Muscular fatty replacement pattern
- Genetic diagnostic approach
- Muscular edema → treatment?
- Helpful in directing the muscle biopsy
- Follow-up
- Brain MRI
- Postsurgical evaluation (scoliosis...)
- Differential diagnosis



Workshop report

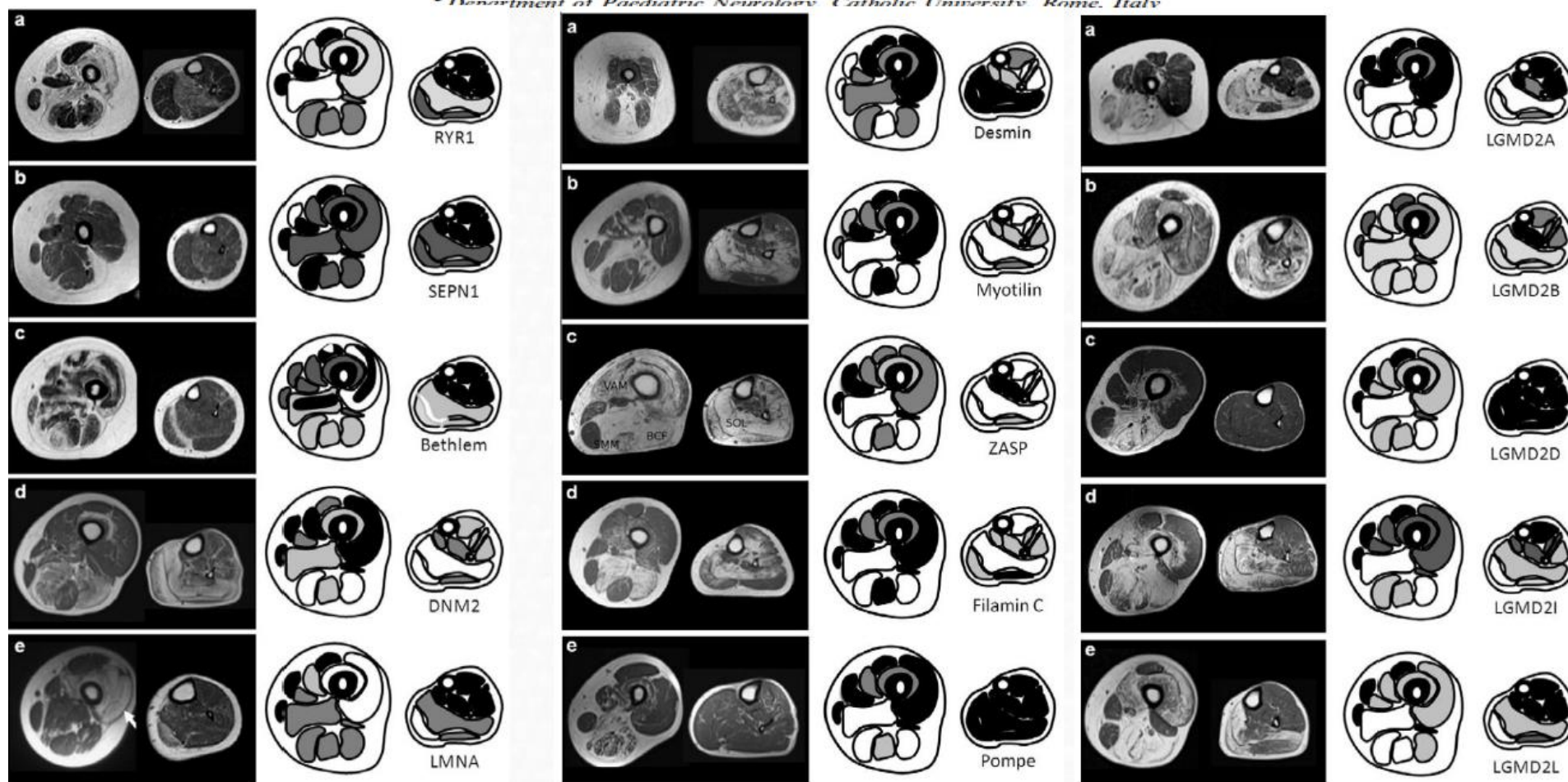
TREAT-NMD workshop: Pattern recognition in genetic muscle diseases using muscle MRI 25–26 February 2011, Rome, Italy

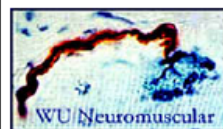
Volker Straub ^{a,*}, Pierre G. Carlier ^{b,1}, Eugenio Mercuri ^{c,1}

^a Institute of Genetic Medicine, Newcastle University, Newcastle upon Tyne, UK

^b Institut de Myologie, Groupe Hospitalier Pitié-Salpêtrière, 75651 Paris Cedex 13, France

^c Department of Paediatric Neurology, Catholic University, Rome, Italy

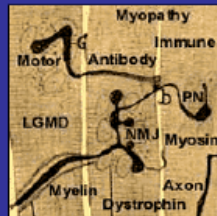




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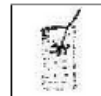
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[CMT-DIE: GN84; 3q26](#)
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Web Site Author: [Alan Pestrenk, MD](#)

Comments: pestrenka@neuro.wustl.edu

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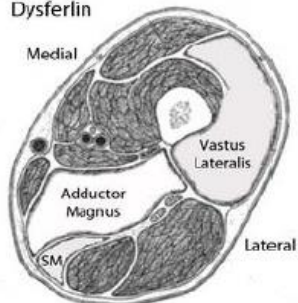
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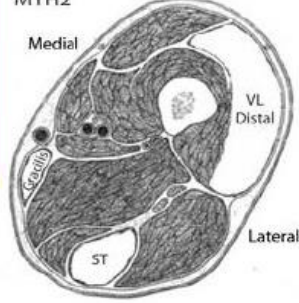
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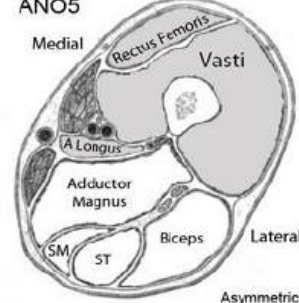
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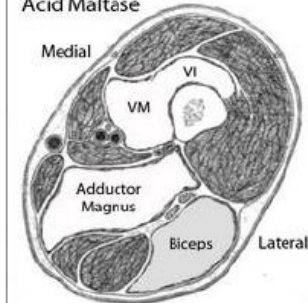
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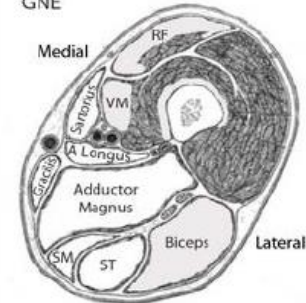
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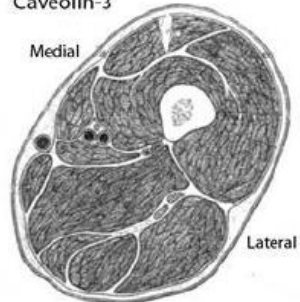
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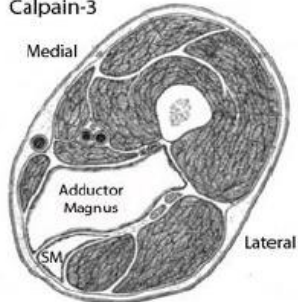
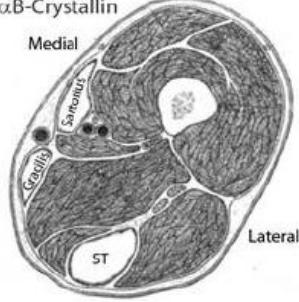
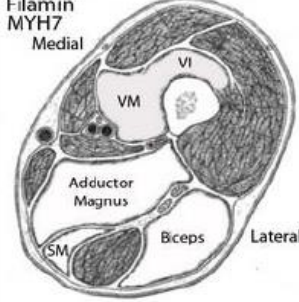
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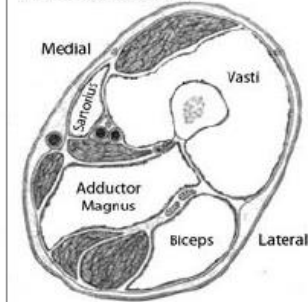
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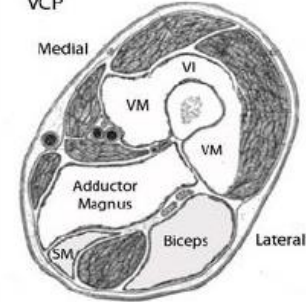
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 α B-CrystallinMyotilin
Filamin
MYH7

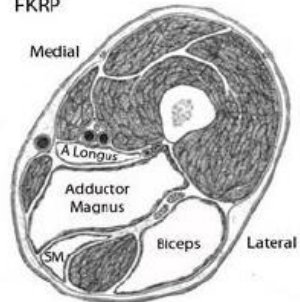
RYR1: Central Core



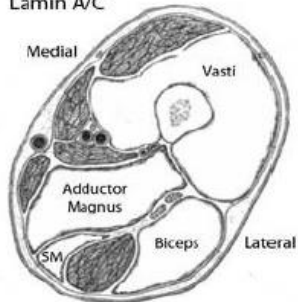
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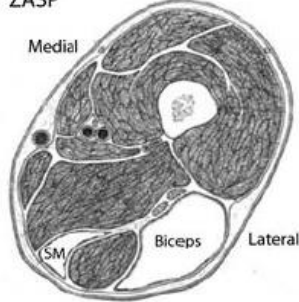
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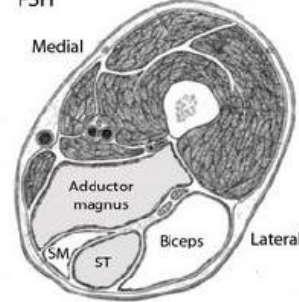
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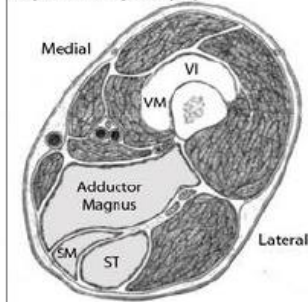
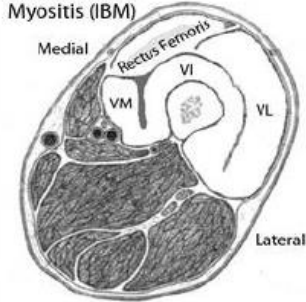
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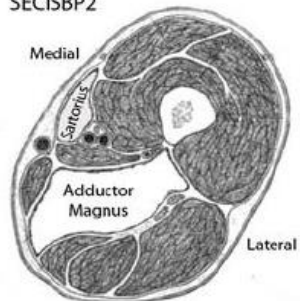
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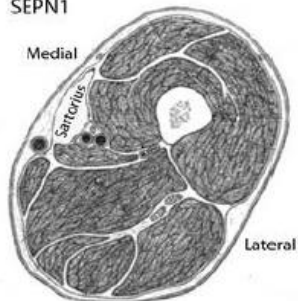
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Inclusion Body
Myositis (IBM)

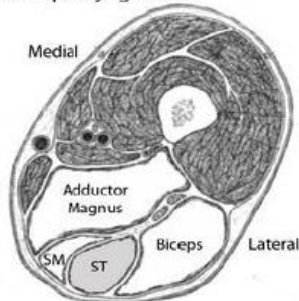
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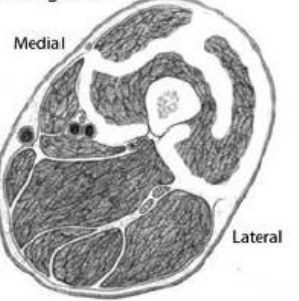
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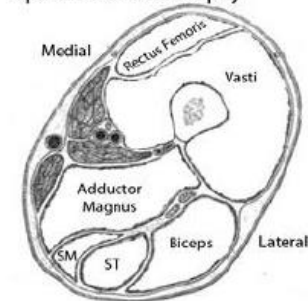
Oculopharyngeal MD



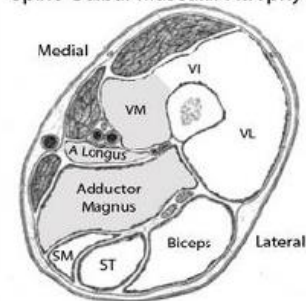
Collagen VI



Spinal Muscular Atrophy



Spino-Bulbar Muscular Atrophy



Gene (Disease/s)	Pattern	Series	Head	Neck	Shoulder girdle	Arm	Forearm	Trunk	Pelvic girdle	Thigh	Leg
COL6 (Ulrich-Bethlem Myopathy)	<i>Tigroid</i> pattern (bands)	13	0/1 * Tongue possible (mild) in severe	3	2 * Deltoid (radial bands) * SC (perpendicular)	2 Post > ant * Triceps (horizontal; single band)	2	3 * Lumbar PV > dorsal (radial bands) * IC possible in severe	3 * Glu M > Glu med, Glu m (longitudinal bands in frontal view)	3 Ant > post * RF (vertical shadow/notch) * VL (rolled cube band) SPARED: AL, sartorius, gracilis	2 Hypertrophy rim between soleus and MG-LG SPARED: TA
	No bands in early stages Gluteal & thigh: mild involvement	1	0	0	0	0	0	1 * Lumbar PV	1 * Glu M	1 * VL SPARED: RF, AL, sartorius, gracilis	0
SEPN1 (CMD; multicore myopathy)	SCM, axial & gluteal involvement Classic lower limb features ¹	4	0/1	2/3 * SCM (hypotrophy)	1	1	0/1 Post	2/3 * PV > IC, abdominal * Psoas variable	3 * Glu M	3 * Sartorius > GA, SM, BF, possible VL SPARED: RF, AL, gracilis	1/2 Post
LMNA (EDMD, LGMD1B)	Axial involvement Classic lower limb features ^{2,3}	2	0	0/1 * Longus colli	1 * Trapezius, SC, serratus	0	0	1/2 * PV lumbar > dorsal SPARED: psoas, abdominal	1 * Glutei, SPARED: pelvic	2 * Vastii, GA > RF, hamstrings SPARED: gracilis, sartorius	2 Post * MG > LG, soleus, peroneal SPARED: TA, TP
LMNA (L-CMD)	Diffuse "negative" pattern, sparing forearm & head ¹⁷	2	0	4	4 SPARED: SC	4	0/1	3 SPARED: psoas, abdominal	3 * Glu med, m > M SPARED: pelvic	4 SPARED: RF, Sartorius, gracilis	4 SPARED: TP, extensor digiti, peroneal
RYR1 (Congenital myopathy: central core; centro nuclear)	Diffuse: head, arm and girdles Classic lower limb features ⁹	3	2/3	2/3	2 * Latissimus dorsi * SC, serratus (variable)	2 Ant > post * Biceps	1	2/3 * PV lumbar > dorsal, IC, psoas, abdominal	3 * Glu M, med, m SPARED: Pelvic	3 * VL, VM, GA, SM, (ST) sartorius (variable) SPARED: RF, AL, gracilis	4 * Soleus, TP, flex digit. SPARED: TA, peroneal, MG LG
	Atypical pattern	1	0	1 * SCM (hypotrophy)	1 * Trapezius, deltoid	0	2 Ant	3 * Abdominal, PV	1	1/2 * Sartorius, ST	2/3 * Peroneus > TA TP, Soleus > LG MG
TPM2 (Congenital myopathy: Distal arthrogryposy)	Temporal & distal leg involvement	6	2/3 * Temporal > lat pteryg. masseter	0	0	0	0	0	0/1	1/2 * RF, other variable	2/3 * Ext digit, LG, soleus, peroneus
DNM2 (Congenital myopathy: centro-nuclear)	Head, axial & distal involvement (forearm ¹⁸ and leg ^{7,19})	2	3 * Lat pteryg temporalis SPARED: Med pteryg	2 * Neck extensors * SCM (hypotrophy)	1 * Trapezius SPARED Deltoid, SC	1 Ant > post * Biceps	2 * Deep forearm	2/3 * PV lumbar > dorsal	2/3 * Glu m > Glu M	2/3 * SM > ST * VI > VL SPARED: sartorius, gracilis	4 * Soleus, TA, peroneus * MG > LG SPARED: TP
GAA (Pompe's disease)	Tongue & subscapular involvement Variable in lower limb ²⁰	1	2 * Tongue	2 * Neck extensors	2 * SC, serratus	0	0	2 * IC, PV	2 * Glu med-min	2 Variable GA, AL, BF, SM	0
SMN1 (type 3 SMA)	'Striated' pattern Enlarged adductor longus muscle ²⁷	2	0	1 * Neck extensors, longus colli	1 * SS, SC, serratus	0	0	2/3 * PV lumbar > dorsal, *psoas, abdominal, belt, IC,	2 * Glu med-m > GluM	2 * GA, quadriceps SPARED: gracilis, ST Sartorius, *AL (Hypertrophy)	2 * Soleus, flex/ext digit, peroneal SPARED: MG, LG, TA

A relative degree of severity is given in each region for each entity, following the scoring proposed by Mercuri et al. [2], from 0 (normal) to 4 (severe fibroadipose infiltration). To facilitate reading of the table, a grading of colours from white to black, (including different shades of grey) is used in each body region. The darker, the more severe involvement of the muscles affected, compared to the other body regions.

AL: adductor longus; Ant: anterior; Arthrogryposy: arthrogryposis; BF: biceps femoris; CMD: congenital muscular dystrophy; Digit: digitorum; Ext: extensor; Flx: flexor; GA: great adductor; Glu M: gluteus maximus; Glu med: gluteus medius; Glu m: gluteus minimus; IC: intercostal; Lat: lateral; LG: lateral gastrocnemius; M: muscle; Med: medial; MG: medial gastrocnemius; Perpend: perpendicular; Post: posterior; Pteryg: pterygoid; PV: paravertebral; RF: rectus femoris; SC: subscapular; SCM: sternocleidomastoid; SM: semimembranous; SMA: spinal muscular atrophy; SS: supraspinatus; ST: semitendinosus; TA: tibialis anterior; TP: tibialis posterior; VI: vastus intermedius; VM: vastus medialis; VL: vastus lateralis.



Whole body muscle MRI protocol: Pattern recognition in early onset NM disorders

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Table 4.1 Overview of well-established rating scales on MRI concerning the visual rating of dystrophic changes (fatty degeneration) of striated muscle tissue

Grade	Mercuri et al. 2002	Kornblum et al. 2006	Fischer et al. 2008
0		Normal appearance	Normal appearance
1	Normal appearance	Discrete moth-eaten appearance with sporadic T1 hyperintense areas	Mild: traces of increased signal intensity on the T1-weighted MRI sequences
2	Mild involvement: Early moth-eaten appearance with scattered small areas of increased signal or with numerous discrete areas of increased signal with beginning confluence, comprising < 30 % of the volume of the individual muscle	(a) Moderate moth-eaten appearance with numerous scattered T1 hyperintense areas (b) Late moth-eaten appearance with numerous confluent T1 hyperintense areas	Moderate: increased T1-weighted signal intensity with beginning confluence in < 50 % of the muscle
3	Moderate involvement: Late moth-eaten appearance with numerous discrete areas of increased signal with beginning confluence, comprising 30–60 % of the volume of the individual muscle	Complete fatty degeneration, replacement of muscle by connective tissue and fat	Severe: increased T1-weighted signal intensity with beginning confluence in > 50 % of the muscle
4	Severe involvement: Washed-out appearance, fuzzy appearance due to confluent areas of increased signal, or an end-stage appearance, with muscle replaced by increased-density connective tissue and fat, and only a rim of fascia and neurovascular structures distinguishable		End-stage appearance, entire muscle replaced by increased-density connective tissue and fat

Reprinted from Wattjes MP, et al. (2010) Neuromuscular imaging in inherited muscle diseases. Eur Radiol 20: 2447–2460 with permission from Springer

3y-old boy, hypotonia

FH: No consanguinity. Father: transient neonatal hypotonia.

PF: Normal pregnancy, Normal fetal movements.

Breech presentation. Delivery 41wk GA.

At birth: **hypotonia, respiratory and swallowing problems.**

PhEx: Normal mental status. **Ptosis.** Microretrognathia.

Facial weakness. High arched palate.

Axial and peripheral hypotonia and weakness.

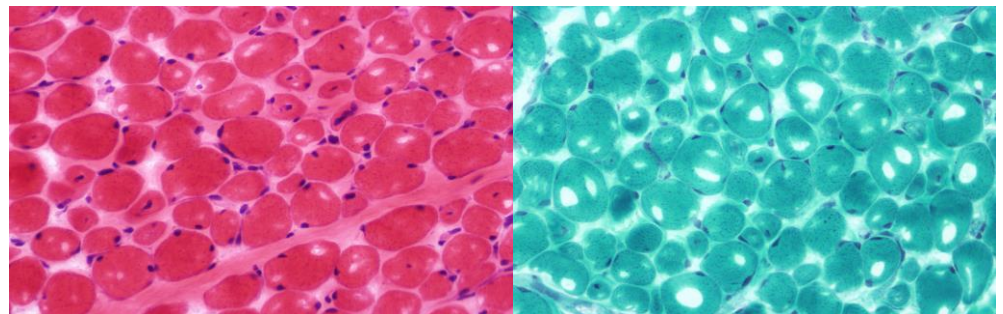
Distal hyperlaxity. Hyporreflexia.

CK: normal

EMG myogenic pattern

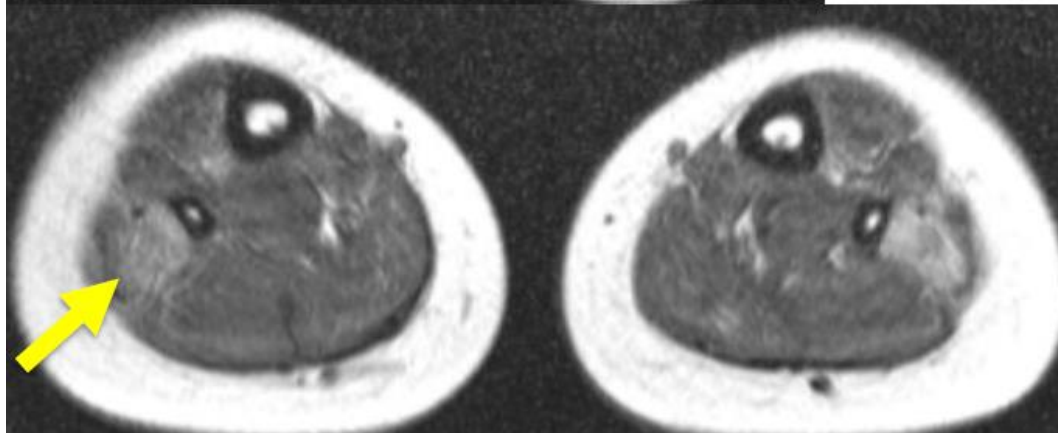
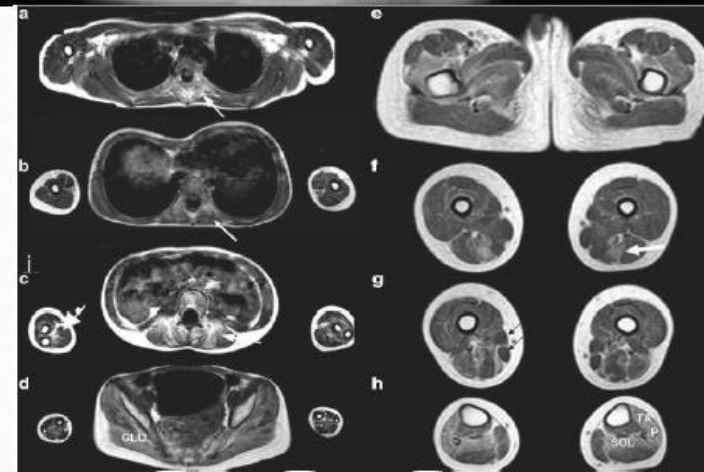
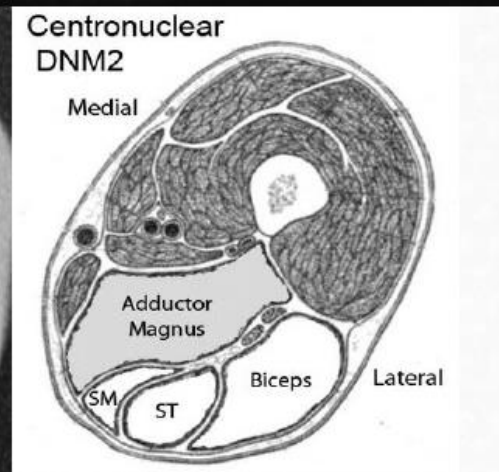
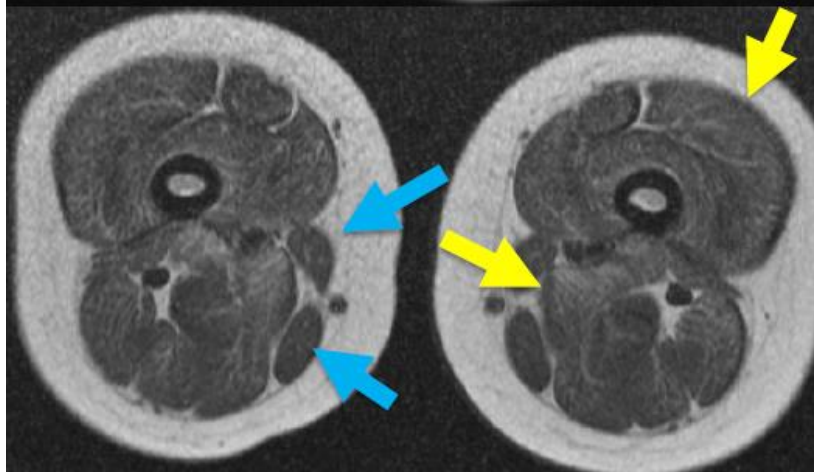
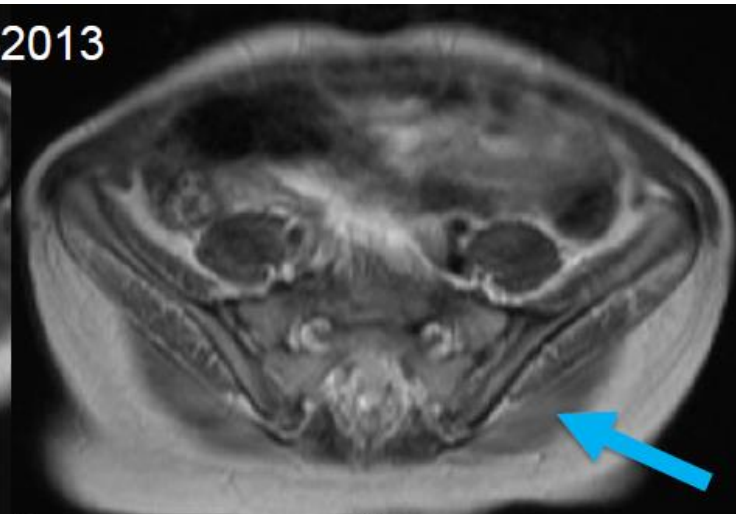
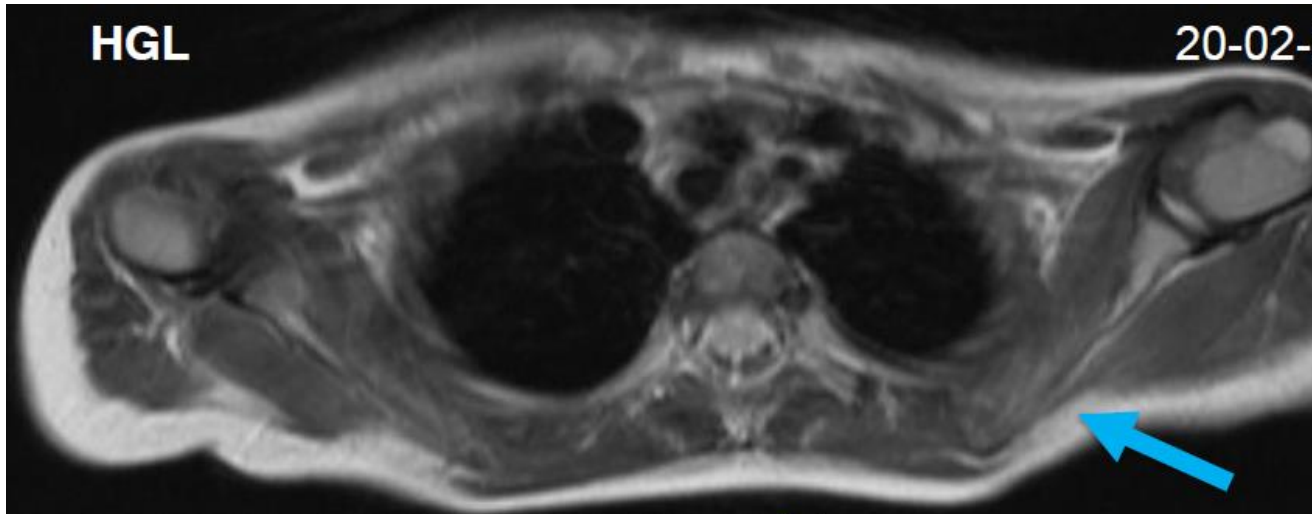
mMRI (11m) ?

MB: Central nuclei and
type I fiber atrophy



HGL

20-02-2013



10 y girl

Hypotonic newborn with poor suck and swallow and respiratory insufficiency

FH

Healthy and non consanguineous parents, no NMD family history

PH

- First, normal pregnancy, delivery at 40s
- newborn: **hypotonia, high arched palate and swallowing problems.**
- **Motor developmental delay (independent walk 20m)**

Ph Ex

Normal mental status. facial weakness, nasal voice.

Axial **hypotonia, distal hyperlaxity, proximal and distal limb weakness, areflexia**

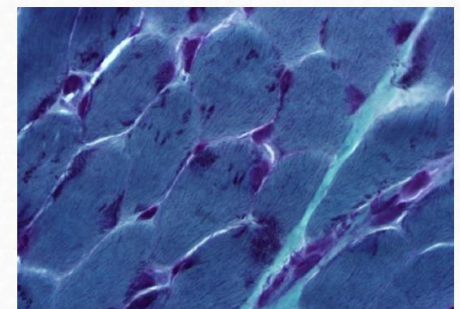
Non-progressive. Respiratory and swallowing improvement.

CK normal

EMG: (2y) myogenic pattern

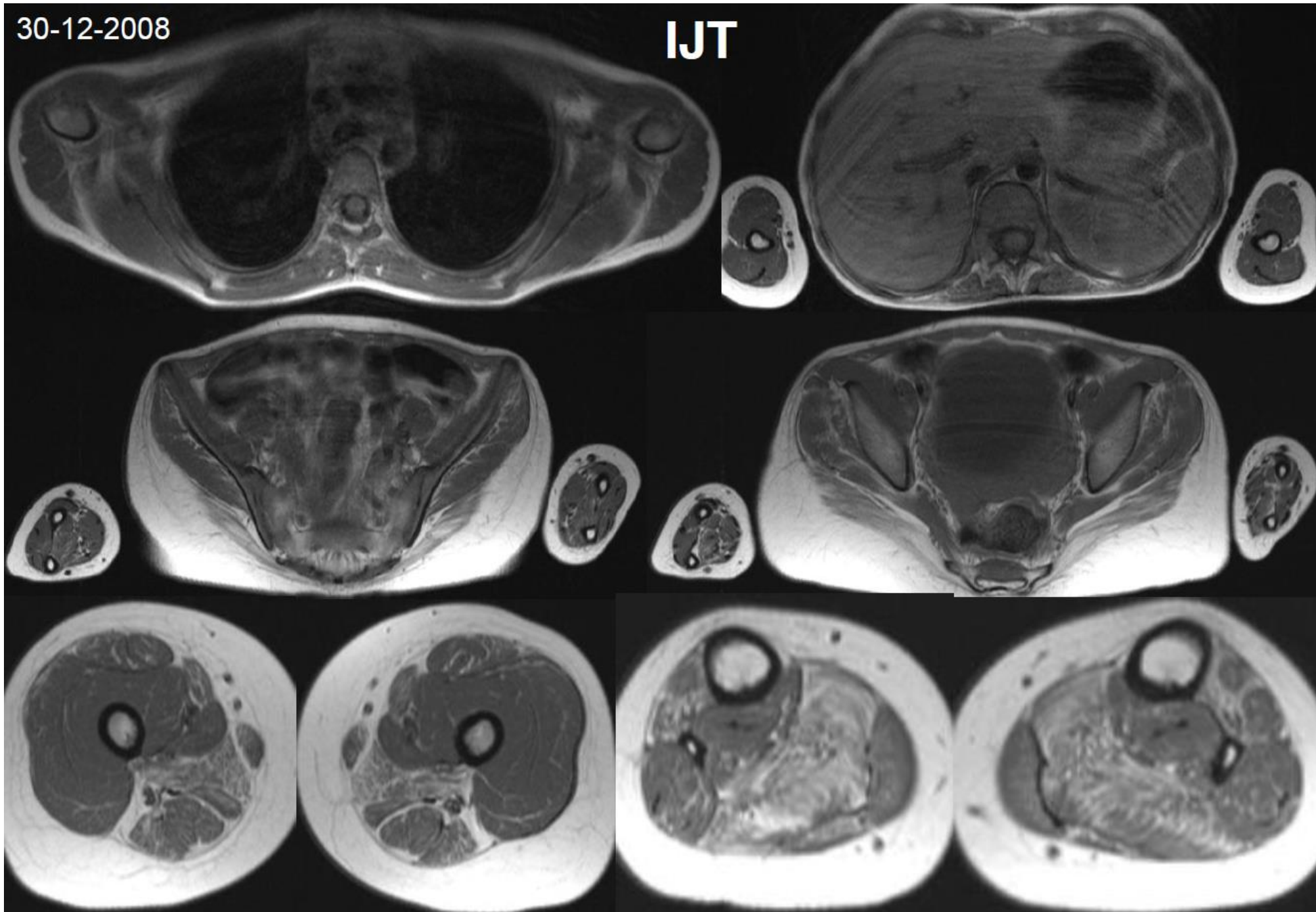
mMRI: Loss of volume and fatty substitution predominantly in gluteal, posterior compartment of thigh and soleus muscles. Upper limb girdle relatively spared.

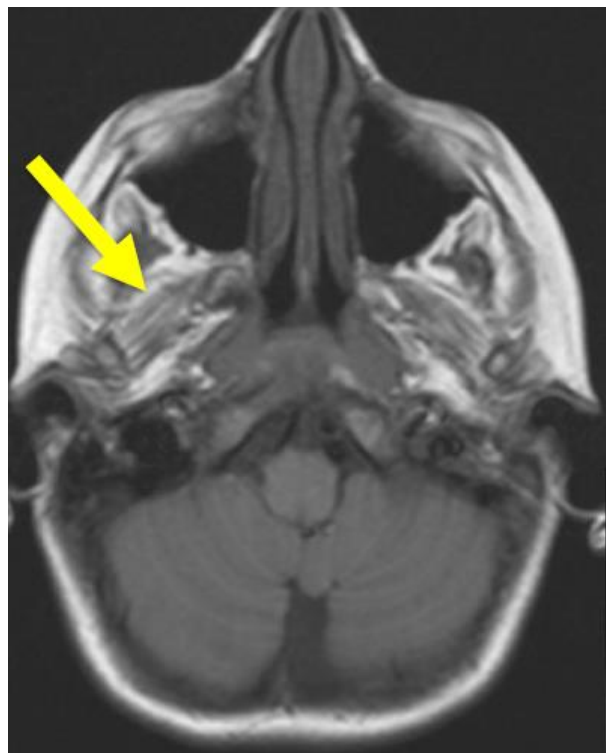
MB: (LM) Abundant subsarcoplasmic and also sarcolemmal nemaline bodies



30-12-2008

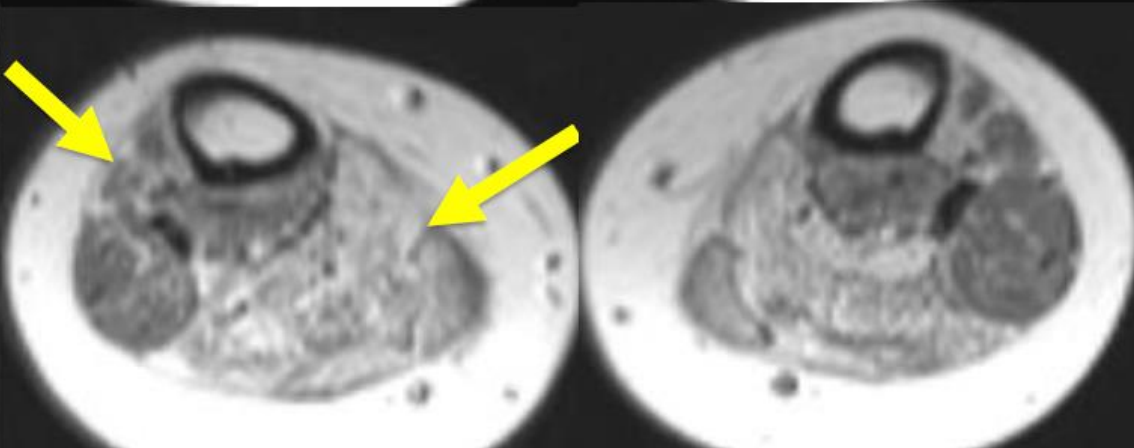
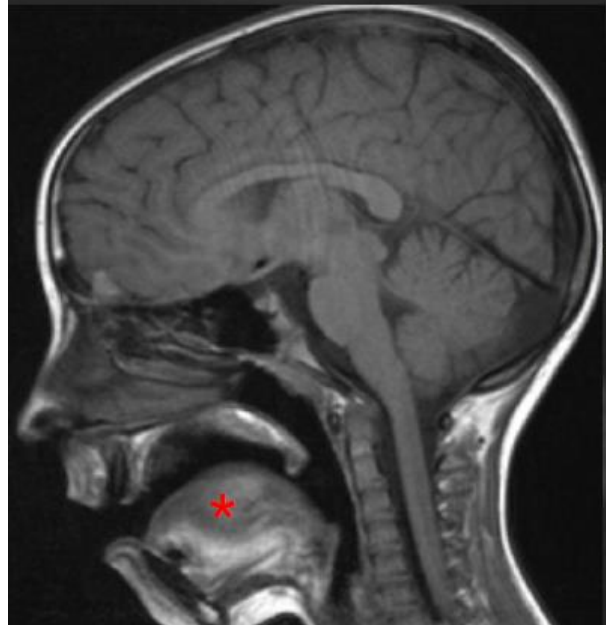
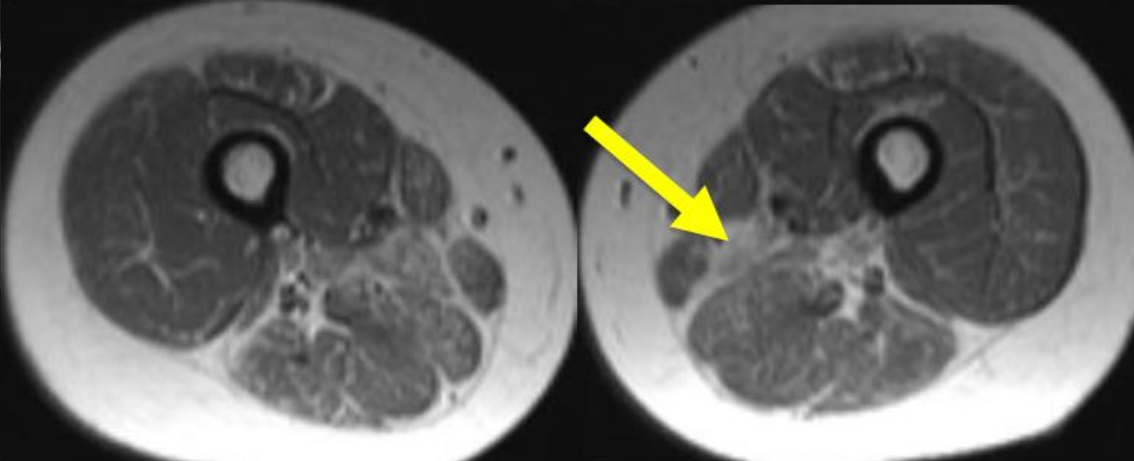
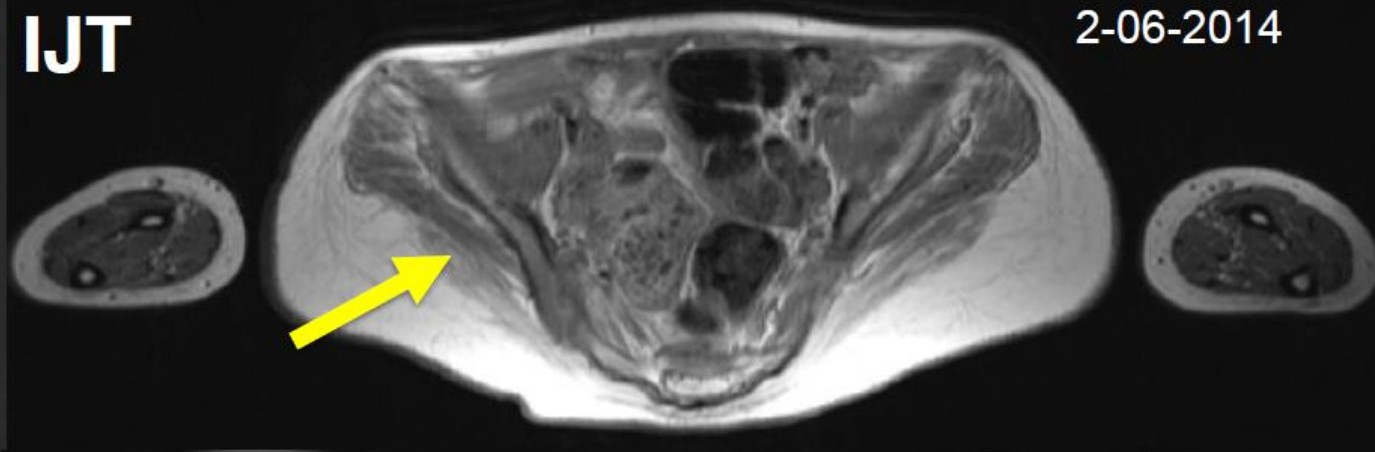
IJT





IJT

2-06-2014



2y-old girl

Hypotonia, weakness and respiratory insufficiency at birth

FH

Consanguineous parents, no NMD history

PH

- First normal pregnancy and delivery
- Respiratory distress at birth. Recurrent respiratory infections. **Diaphragm paralysis**. Swallowing problems.
- Mild hypotonia and weakness.

Ph Ex

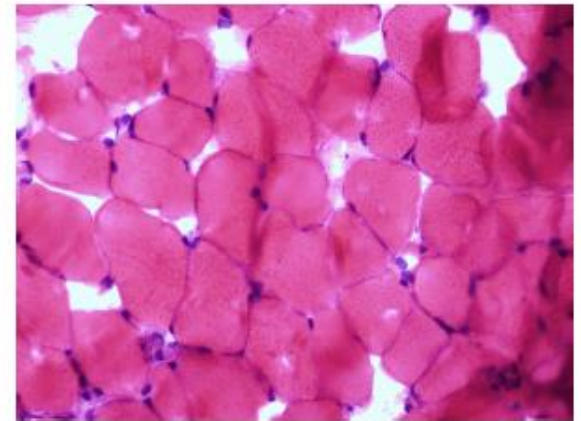
Normal mental status. Mild facial weakness. Hypotonia. No ocular involvement.
Axial weakness, head drop, scoliosis, hyporeflexia.

CK: normal.

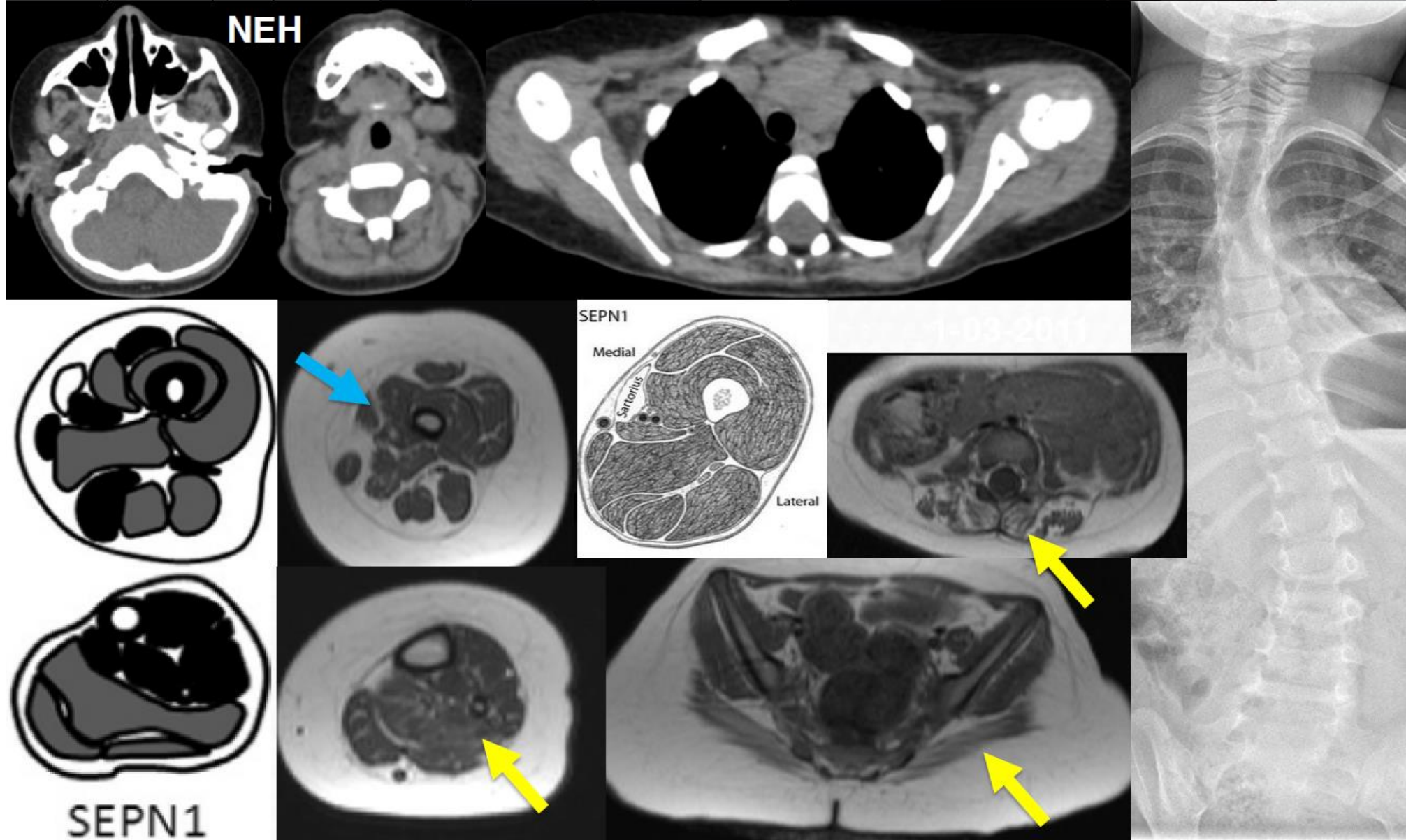
mMRI: loss of volume of paraspinal muscles and glutei.

EMG: myogenic pattern

MB: Fiber type disproportion.



SEPNI (CMD; multicore myopathy)	SCM, axial & gluteal involvement Classic lower limb features ³	4	0/1	2/3 *SCM (hypo- trophy)	1	1	0/1 <u>Post</u>	2/3 *PV > IC, abdominal *Psoas variable	3 * Glu M	3 *Sartorius> GA, SM, BF, possible VL SPARED: RF, AL, gracilis	1/2 <u>Post</u>
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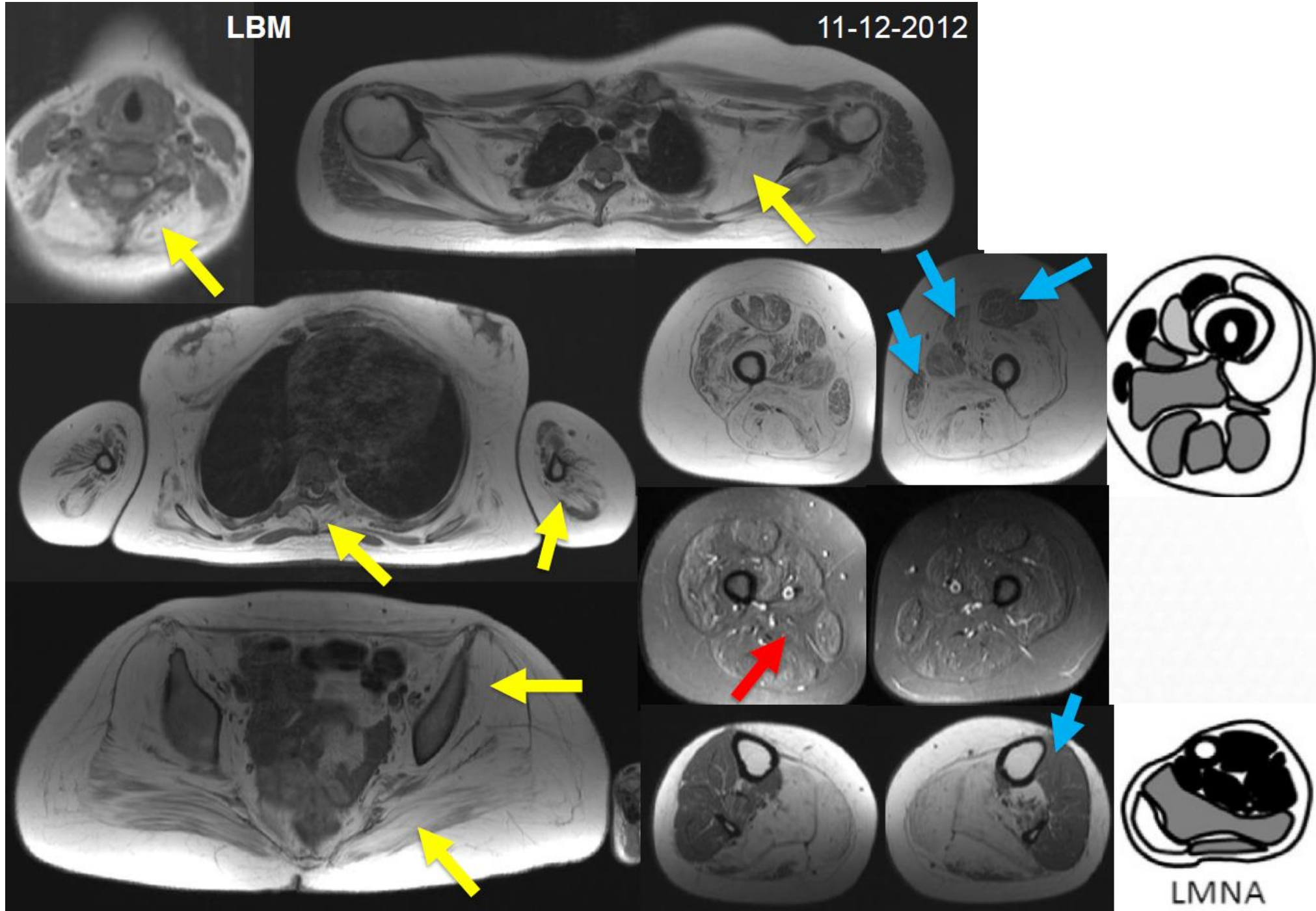


Patient LBM

- 12 yo
- Unrelated parents.
- 7 y of age: lower limb weakness.
- PhE: proximal lower limb weakness, scapula alata,
- myopathic gait.
- CK 300.
- EMG myogenic pattern
- BM fiber size variation.

LBM

11-12-2012



Patient IEC

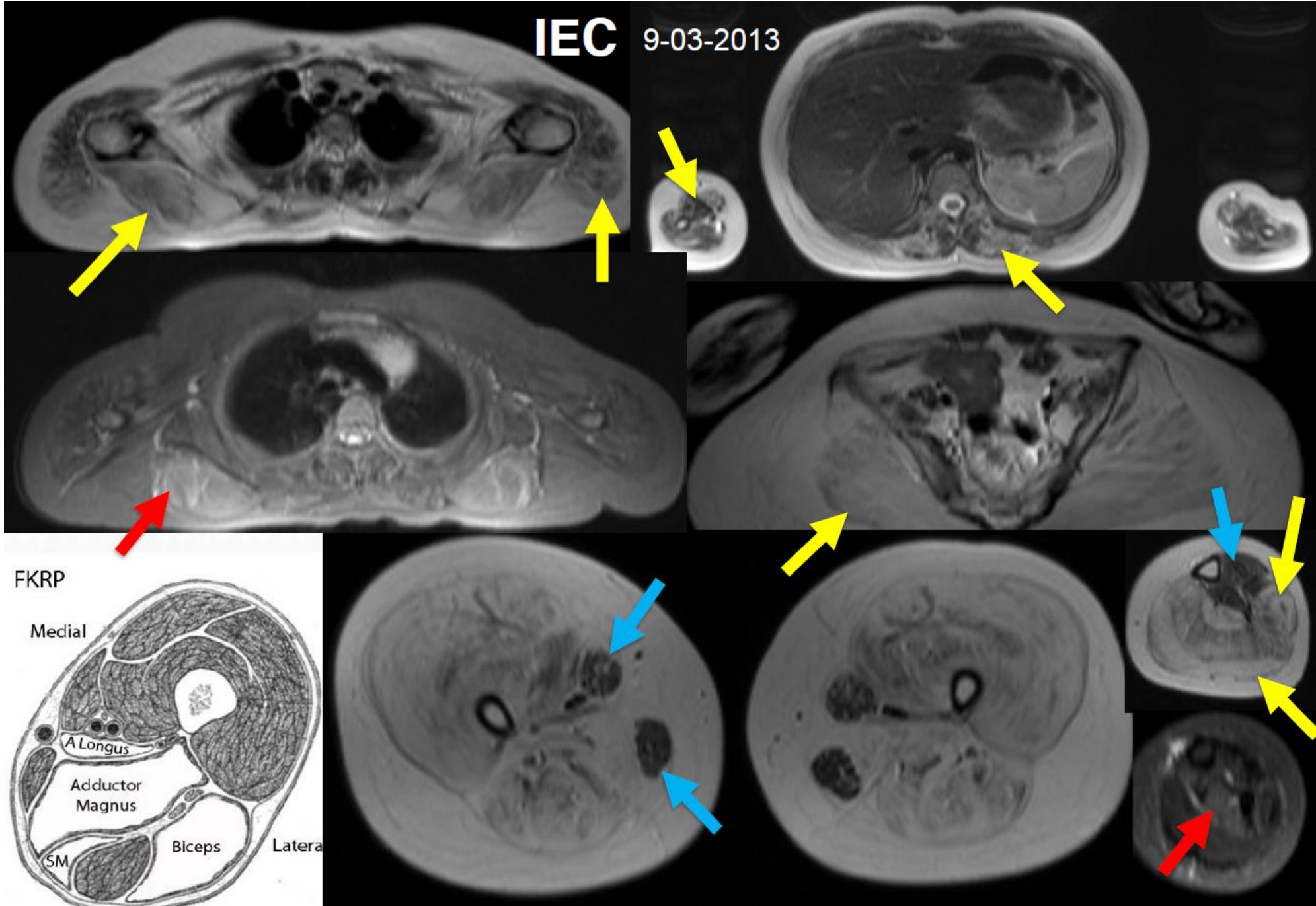
- 9 yo
- Consanguineous parents.
- Phenotype : CMD
- CK (10,000).
- Normal mental status.
- Loss of ambulation 7 years

PhE:

- Horizontal Ophthalmoplegia
 - Facial weakness
 - Limb girdle weakness
 - Proximal contractures in arms and legs
-
- Muscle Biopsy: dystrophy signs, connective and adipous tissue.

IEC

9-03-2013



Patient JHC

FH: Healthy parents and one healthy brother. Non consanguinity

Normal Pregnancy and delivery

Proximal lower limb weakness from 8 years of age, progressive

Distal hyperlaxity, keratosis pilaris

EMG miogenic pattern

CK: 3753 UI/L

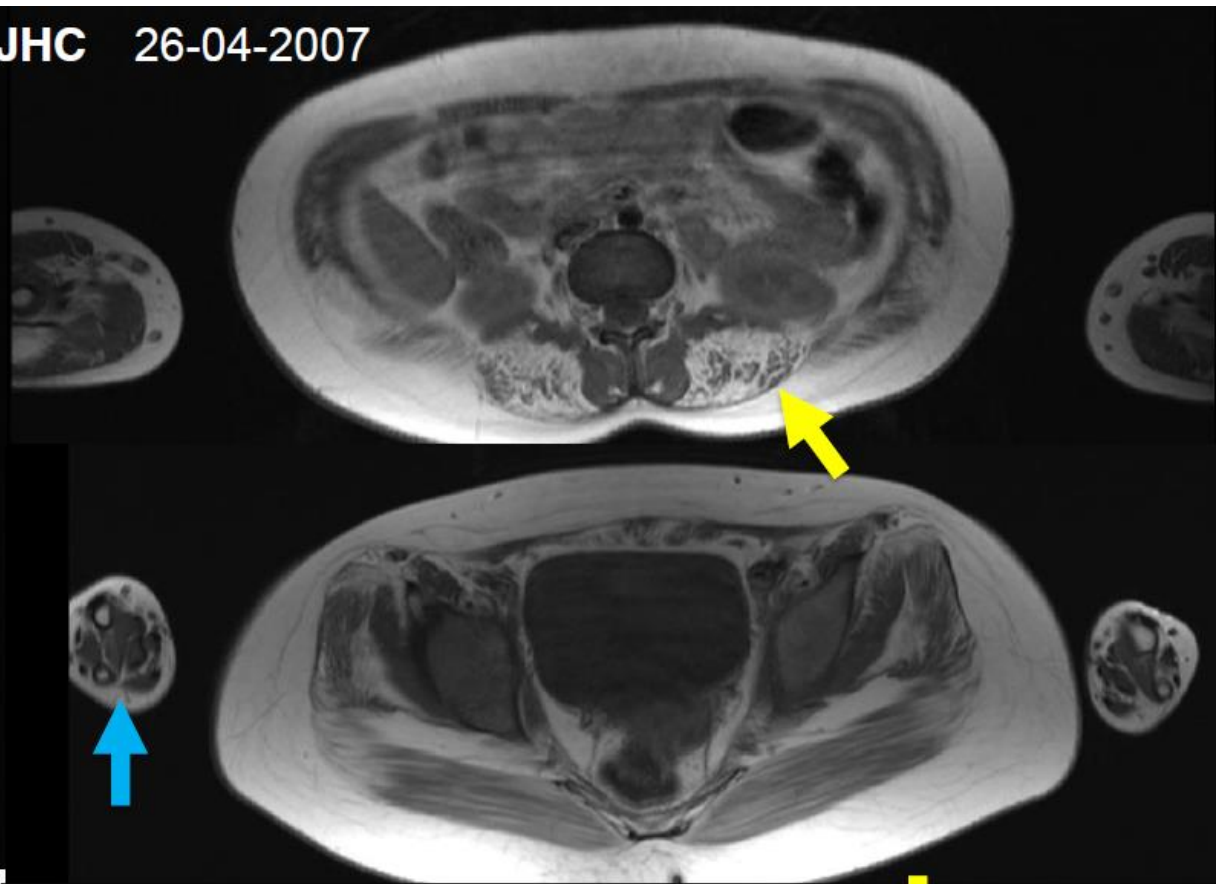
PhE: Normal metal status, no ocular involvement

Proximal limb weakness, progressive, Gower sign

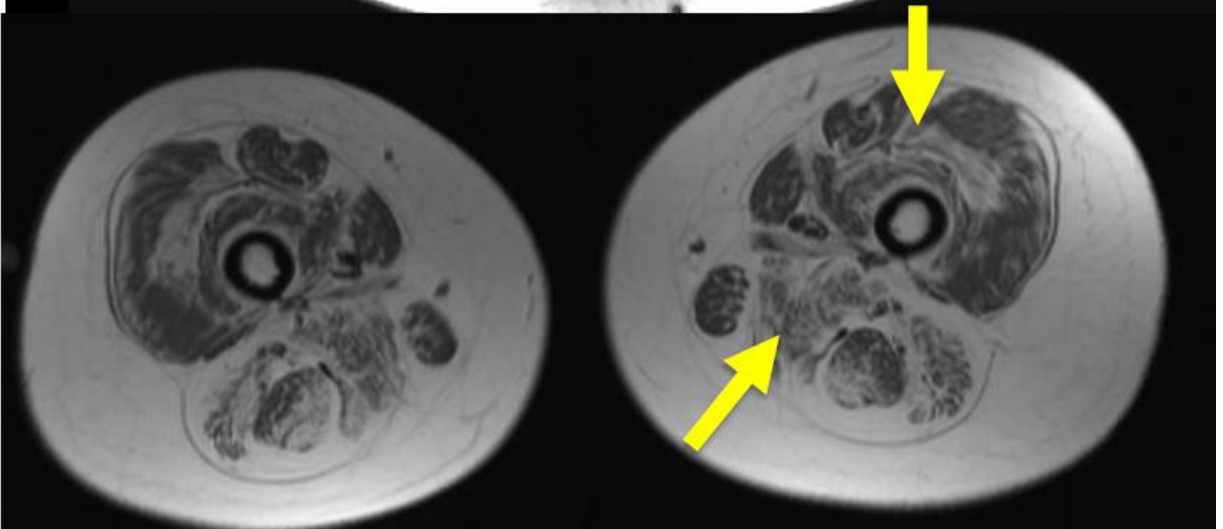
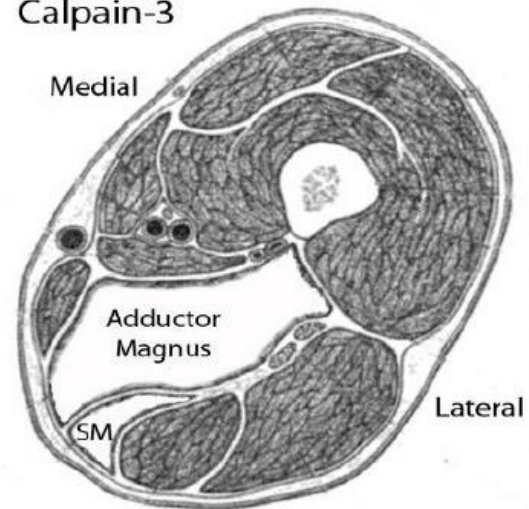
Col6??



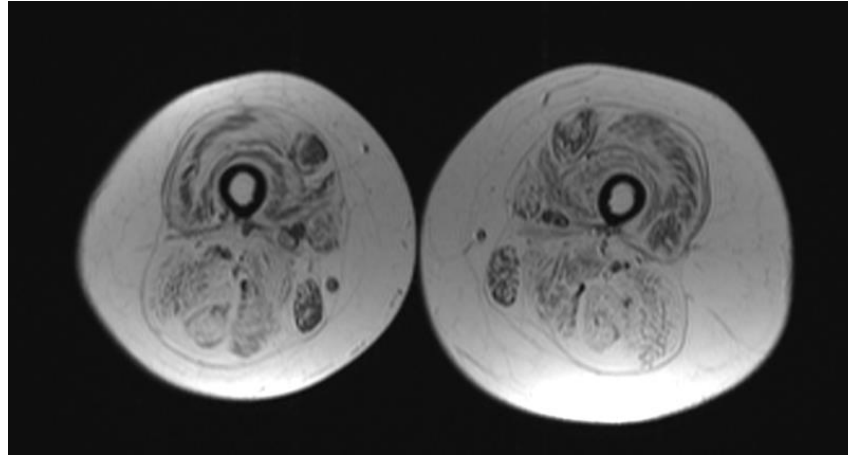
JHC 26-04-2007



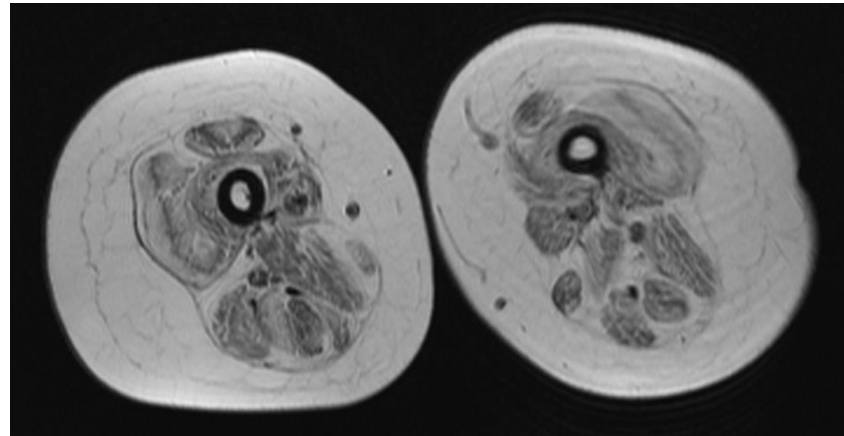
Calpain-3



ROLLER CAKE



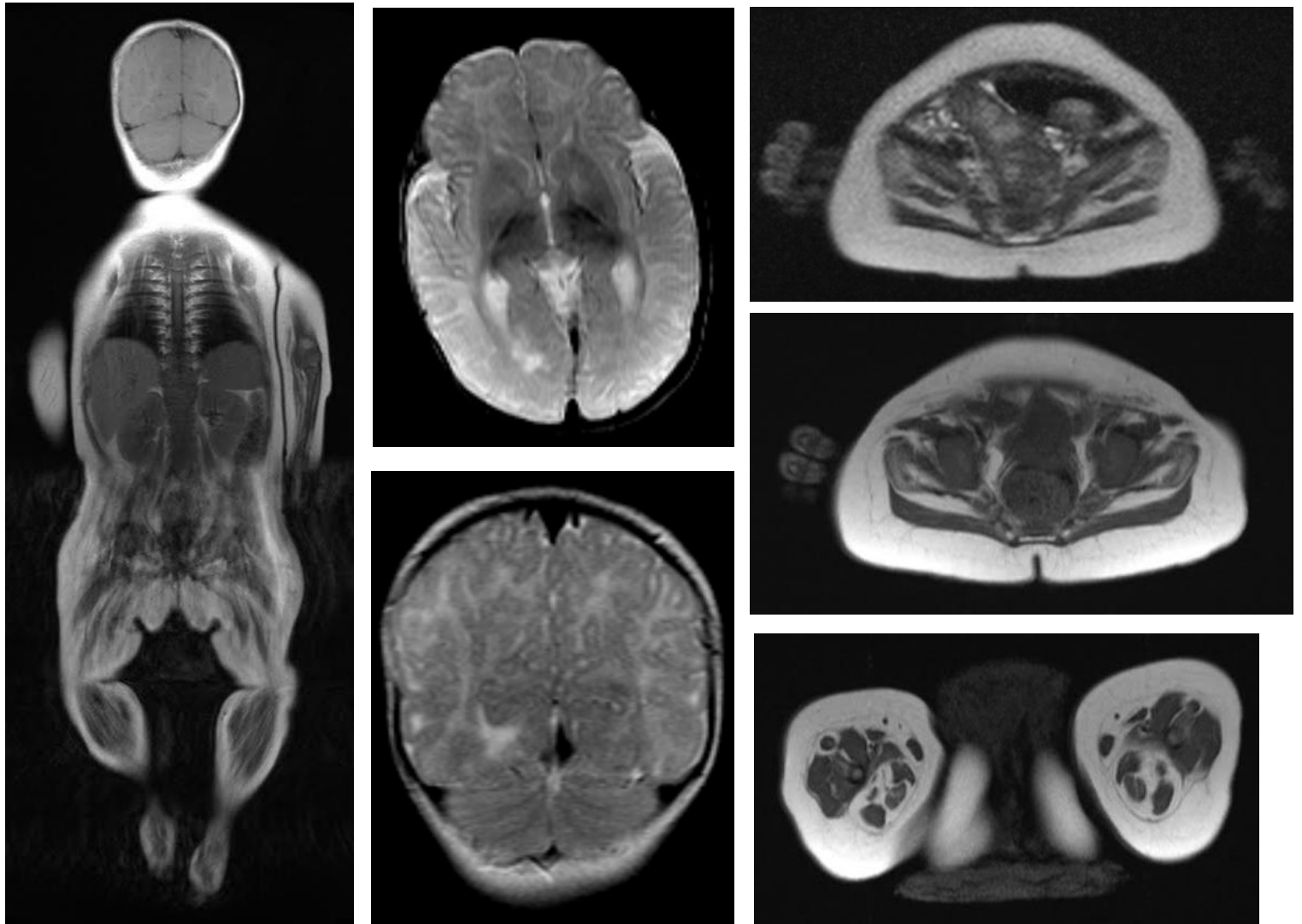
CAPN3



COL6A2

MDC1A, Merosin-Deficient Congenital Muscular Dystrophy Type 1A , LAMA2 DEFICIENCY

- 9 m. old girl
- **Severe hipotonia**
- **Face not involved**, alert
- **Pointed arch, weakness**
- CK **600-1000** UI/L
- EMG: miogenic
- MRI
- Clinical exome
LAMA2
c.1467+1G>C
c.3038-7G>A



Brain: white matter hyperintensity

WB: Gluteus medius, rectus femoris, adductor magnus, semitendinosus, peroneus and soleus. Muscle edema in deltoides and triceps.

CONGENITAL MYOPATHIES

Predominant involvement ?

Predominant lower leg involvement?

Predominant muscle ?

Mutation in ?

SEPNI ?

RYR1 ?

MTM1 ?

ACTA1 ?

MYH7 ?

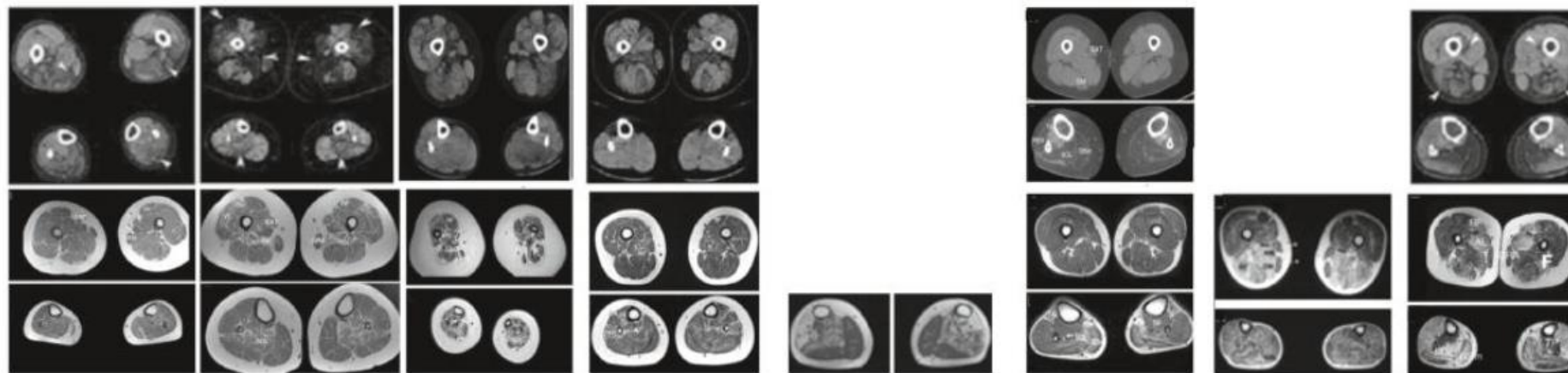
NEB1 ?

TPM2 ?

DNM2 ?

Muscle CT

Muscle MRI



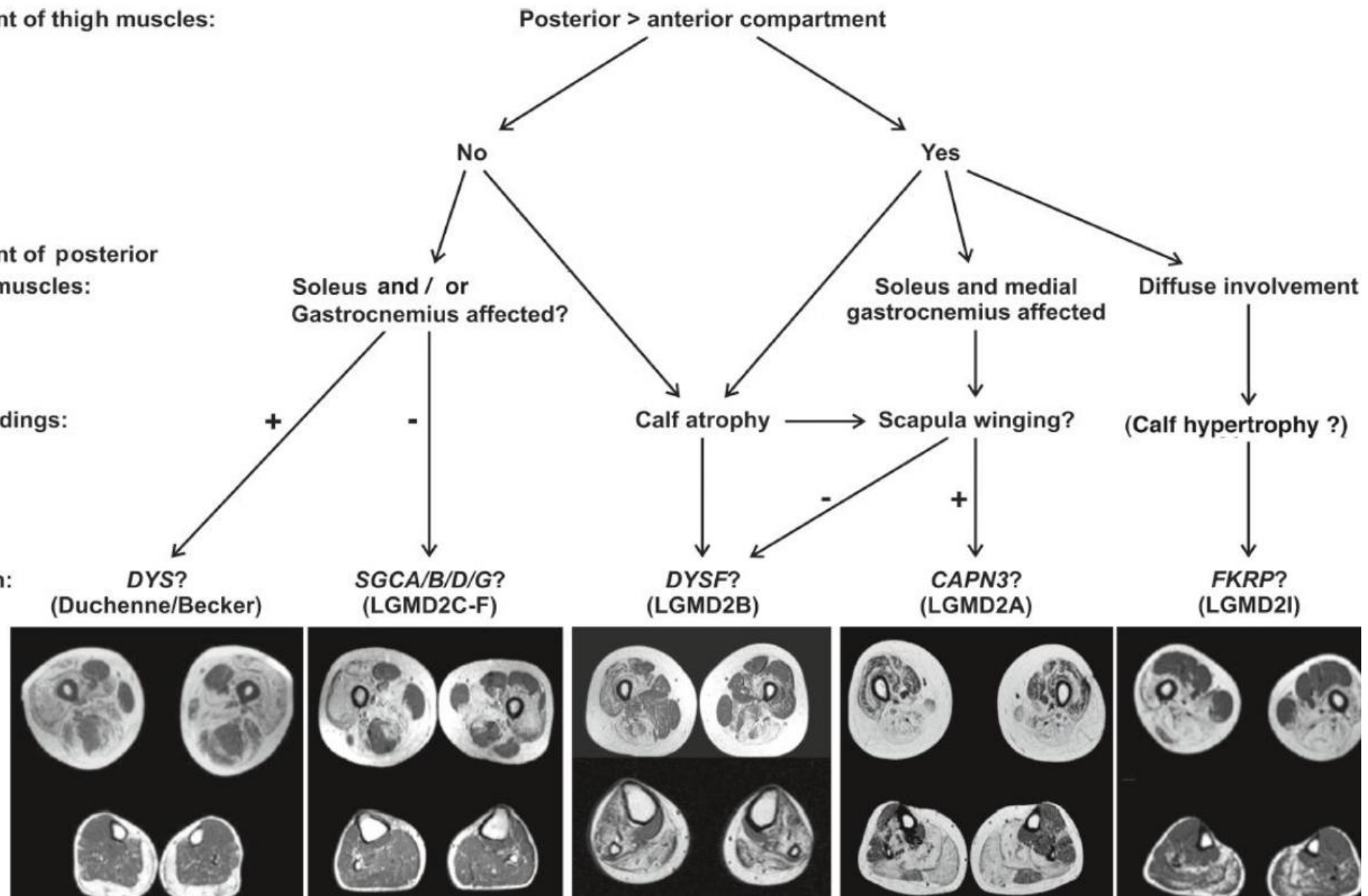
DYSTROPHINOPATHIES and LGMD

Involvement of thigh muscles:

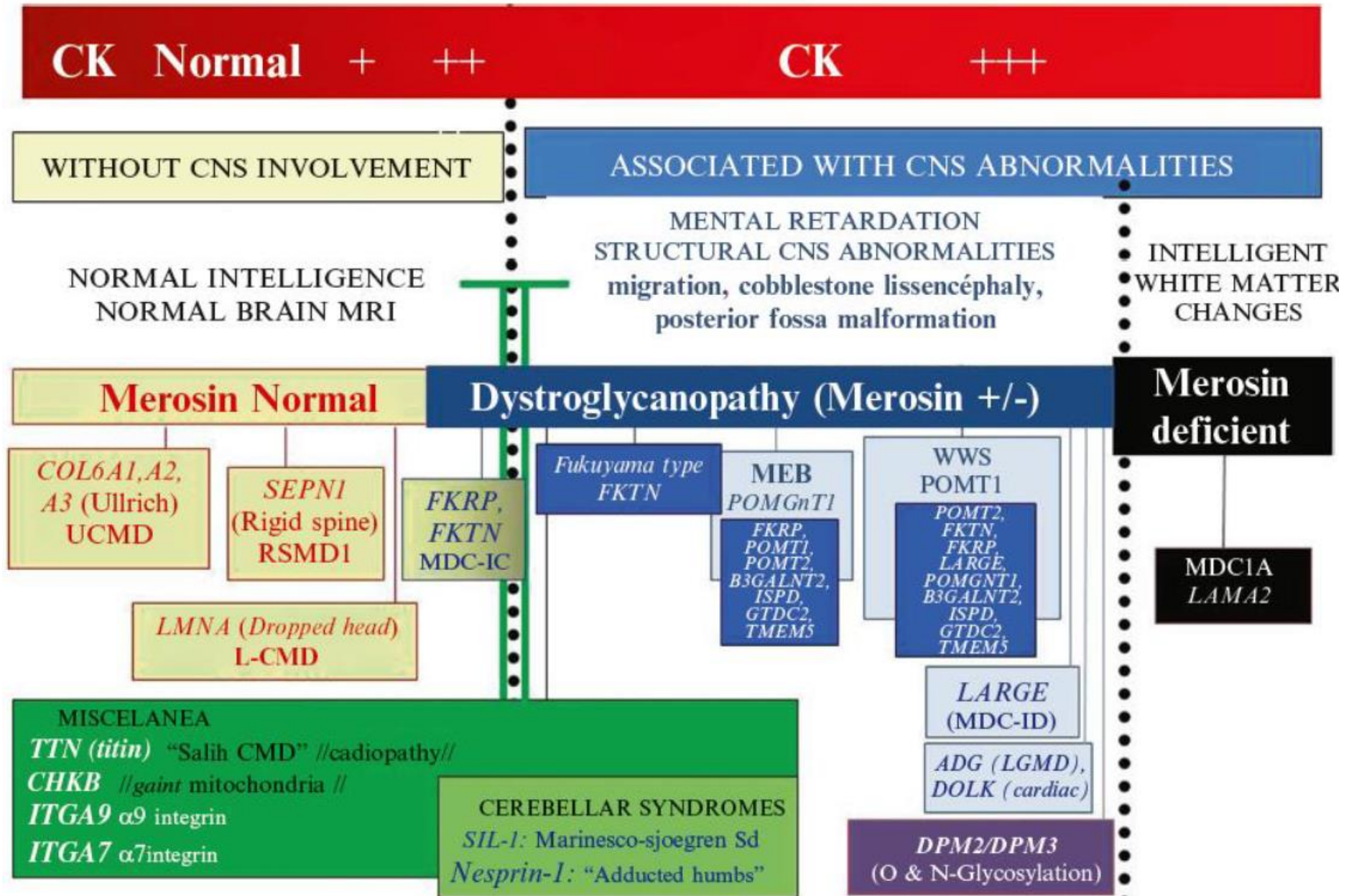
Involvement of posterior lower leg muscles:

Clinical findings:

Mutation in:

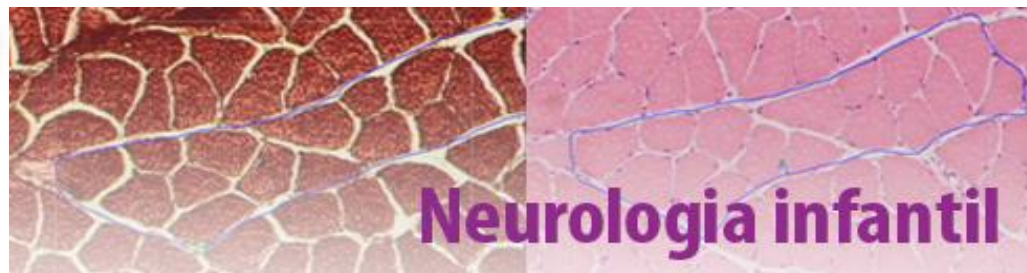


CONGENITAL MUSCULAR DYSTROPHIES



CONCLUSIONS

- The pattern of muscular involvement identified by WB MRI is a useful tool guiding the genetic diagnosis of congenital myopathies and congenital muscular dystrophies and in their follow-up
- The more difficult the pathology is, more people involved is needed (clinicians , neuropathologists , neurophysiologists , neuroradiologists, surgeons, basic researchers, bioinformaticians...)



Gràcies!!