# Case 14503

Eurorad

## Charcot-Marie-Tooth, MRI

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**ISSN:** 1563-4086
**Section:** Musculoskeletal system
**Area of Interest:** Musculoskeletal soft tissue Soft tissues / Skin
**Procedure:** Diagnostic procedure
**Imaging Technique:** MR
**Special Focus:** Genetic defects Inflammation Case Type: Clinical Cases
**Authors:** Coronel Tarancon Luis, Bello Baez Adan, Nieto Morales Maria Luisa, Benitez Rivero Sonia,Cavada Laza Arsenio.
**Patient:** 14 years, male

**Clinical History:**

A 14-year-old male patient presented with a history of life-long weakness, slight instability while walking with no associated falls, less physical aptitudes than his classmates and an uneven shoe wear.
Electrophysiological study shows motor and sensory polyneuropathy, mostly demyelinating, with severe and chronic axonal degeneration in upper and lower limbs.
**Imaging Findings:**

Severe atrophy of extensor digitorum brevis muscles (EDB), dorsal and plantar interosseus and quadratus plantae (QP).
No flexor muscle disorders.
There was no evidence of muscle atrophy, fatty infiltration or oedema at any other muscle.
**Discussion:**

Charcot–Marie–Tooth disease (CMTD), also known as hereditary motor and sensory neuropathy (HMSN), is the most frequent form of inherited polyneuropathy with a prevalence ratio of 17-40 cases per 100, 000 inhabitants. CMTD is caused by different gene mutations that produce modified proteins that affect peripheral axon or myelin. That correlates with changes affecting the normal neuron function or structure. It can be transmitted autosomal dominant, autosomal recessive or X-linked.
There are many different types of CMTD. The most frequent form of CMTD is CMT1A (around 70%), that is an autosomal dominant disease caused by duplication of PMP22 [1, 2] and it arise as a disorder of peripheral myelination.
Symptoms are present during the first decade of life in over two thirds of cases. Foot deformity, basically forefoot pes cavus and considerably less frequently pes planus, is a cardinal manifestation of any form of CMT subtype, although significantly more common in CMT1 than in CMT2 patients. Sensory changes are present but usually to a lesser degree. [3]
Diagnosis process begins with the anamnesis of personal and family background and neurological examination. If CMTD is suspected, then electrophysiological study may be requested. Characteristically, motor conduction velocity in upper-limb nerves is <38 m/s in CMT1. [3]
During the last 8 - 10 years MRI is showing it usefulness for differential diagnosis: besides intrinsic foot muscle involvement, variable and distally accentuated fatty infiltration of the lateral, anterior, and superficial posterior leg

muscle compartments and, to a lesser degree, of the deep posterior compartment [4]. Clinical MRI patterns of lower limb muscle atrophy vary with evolution of semiology. Selective involvement of intrinsic foot muscles is the characteristic pattern of CMT1A cases with minimal disease signs. Afterwards, this pattern usually combines variable and distally accentuated involvement of lower-leg muscles, especially those of the antero-lateral compartments.
The observed early selective fatty atrophy of intrinsic foot musculature, particularly of the lumbricals, in patients with mild phenotype but already showing pes cavus, supports its pathogenetic role in the initiation of forefoot pes cavus in CMT1A [5].
Positive diagnosis was traditionally made by peripheral nerve biopsy. Nowadays, it is made by genetic analysis. Genetic analysis can diagnose the existence of CMTD as well as it type.
There is no cure for CMTD, but physical therapy, orthotic and rehabilitation treatment or even orthopaedic surgery may be needed to help these patients to deal with the symptoms or disability caused by this disease.
**Differential Diagnosis List:** Charcot-Marie-Tooth type 1A., Dejerine-Sottas disease, Refsum dissease, Chronic inflammatory demyelinative neuropathy

**Final Diagnosis:** Charcot-Marie-Tooth type 1A.

**References:**

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Chung KW, Suh BC, Shy ME, Cho SY, Yoo JH, Park SW, Moon H, Park KD, Choi KG, Kim S, Kim SB, Shim DS, Kim SM, Sunwoo IN, Choi BO (2008) Different clinical and magnetic resonance imaging features between Charcot–Marie–Tooth dis- ease type 1A and 2A. Neuromuscul Disord 18:610–618 (PMID:18602827)
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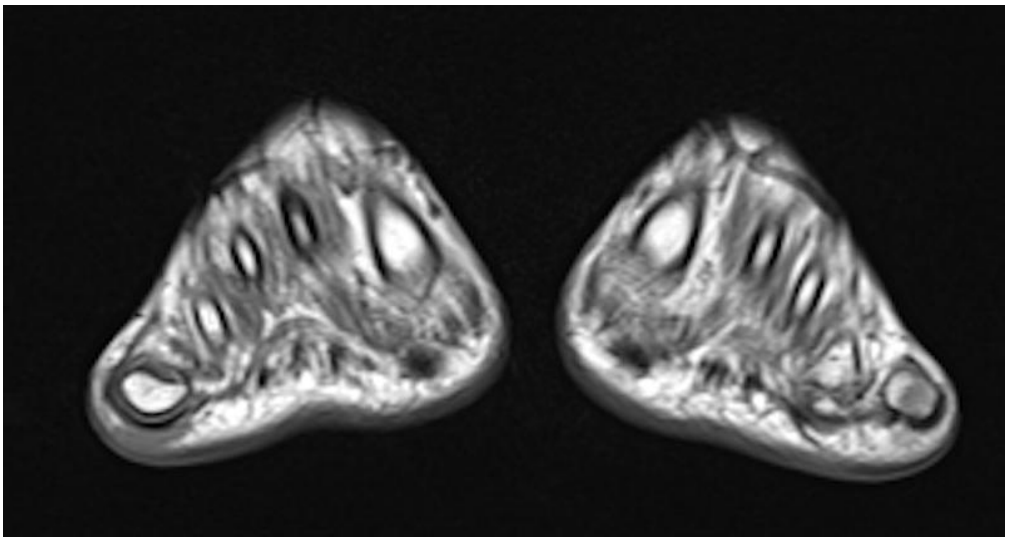
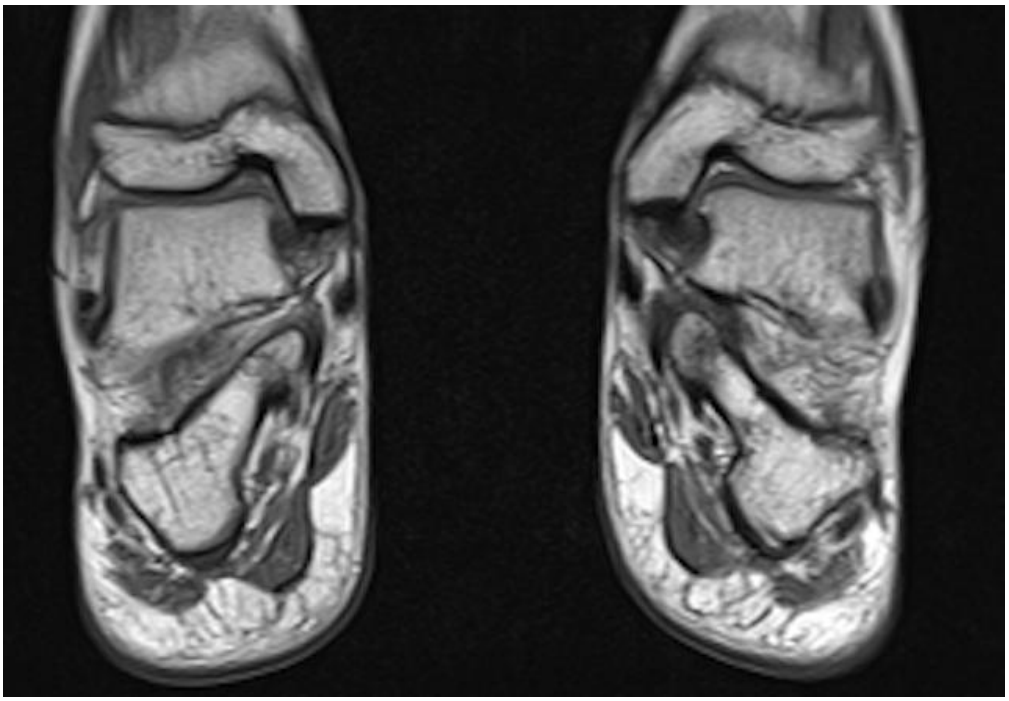
## Figure 1

**a**



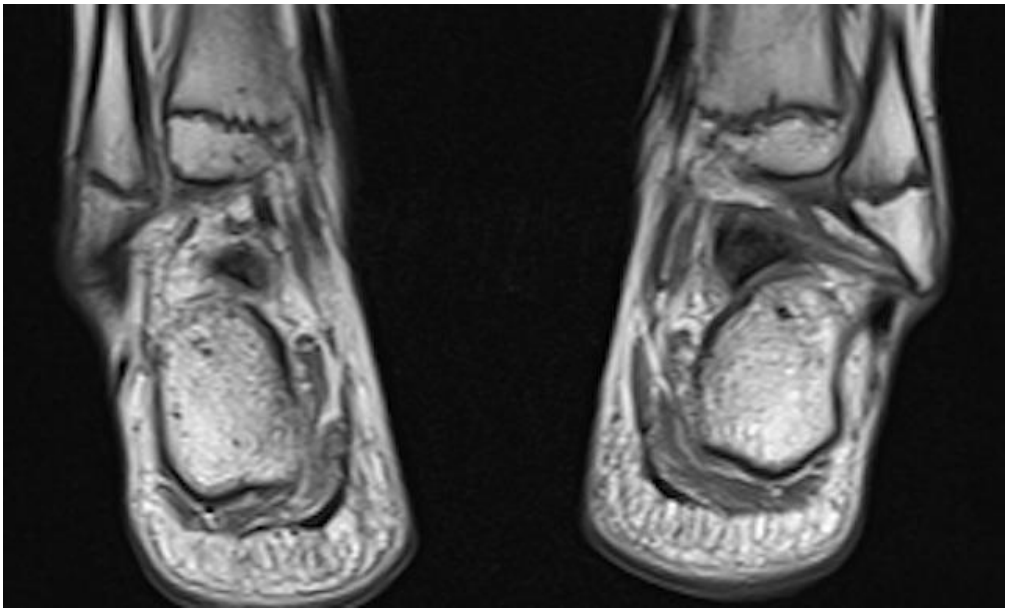
**Description:** Fatty infiltration in intrinsic foot muscles. Plantar interossei muscles show hyperintensity on T2FSE sequences because of severe atrophy. **Origin:** Bello Adán. Department of Radiology, HUNSC, Tenerife, Spain.

**b**



**Description:** Severe atrophy (fatty infiltration) of both quadratus plantae muscles. **Origin:** Bello Adán. Department of Radiology, HUNSC, Tenerife, Spain.

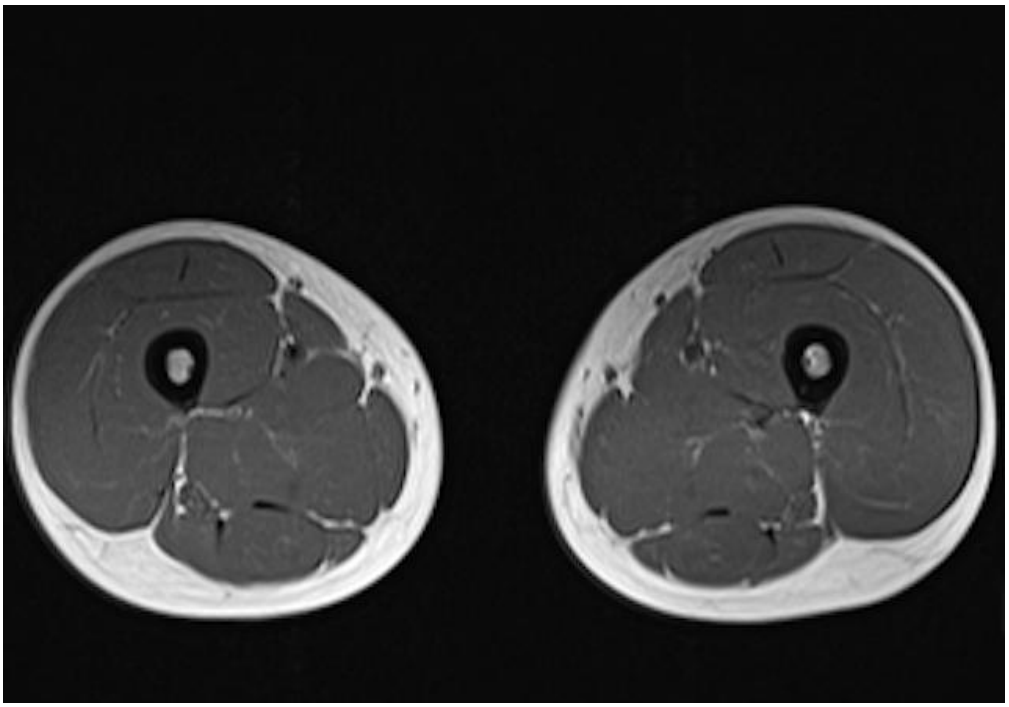
**c**



**Description:** Severe atrophy (fatty infiltration) of both quadratus plantae muscles. **Origin:** Bello Adán. Department of Radiology, HUNSC, Tenerife, Spain.

## Figure 2

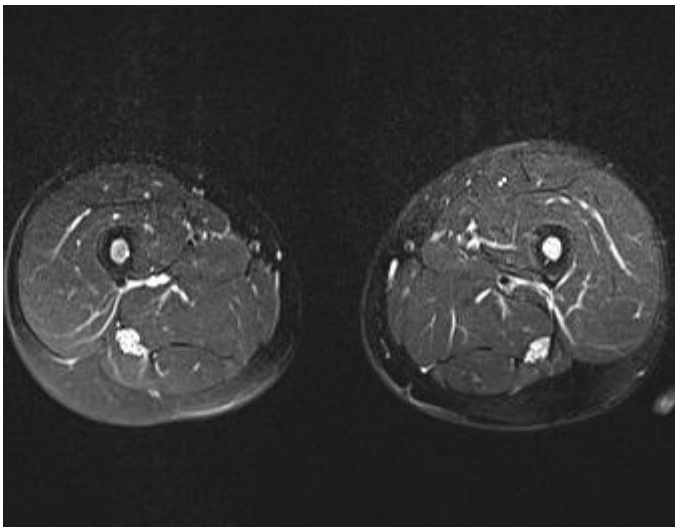
**a**



**Description:** Axial T1 FSE, midportion thigh level. Note that the selective involvement of intrinsic foot muscles without leg muscle involvement is a characteristic pattern of CMT1A and CMT2A with minimal disease. **Origin:** Adan Bello, Department of Radiology, HUNSC, Tenerife, Spain.

## Figure 3

**a**



**Description:** Axial T2 FSE fat sat. Midportion thigh level. Note that the selective involvement of intrinsic foot muscle without leg muscle involment is a characteristic pattern of CMT1A and CMT2A with minimal disease. **Origin:** Bello Báez Adán, Department of Radiology, HUNSC, Ternife, Spain.