Case 14967

Eurorad

Limb-girdle muscular dystrophy (LGMD2A)

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Section: Musculoskeletal system
Area of Interest: Musculoskeletal soft tissue
Procedure: Diagnostic procedure
Imaging Technique: MR
Special Focus: Genetic defects Case Type: Clinical Cases
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Patient: 33 years, female

Clinical History:

The authors present a 33-year-old woman with a gradual onset of lower limb proximal muscle weakness since she was 3 years old. The diagnostic work up detected elevated creatinine kinase levels and dystrophic changes on muscle biopsy. There was no family history. Now she presents a myopathic walk with reduced lower limb mobility.

Imaging Findings:

We performed a MR scan on the patient. There was a symmetric generalised muscle atrophy with fatty replacement seen in the muscles of the thigh (Fig. 1, 4). The vastus lateralis muscles were particularly affected (Fig. 2).
There was relative sparing of the quadratus femoris, adductors, rectus femoris and gracilis muscles (Fig. 1, 2, 3).
Axial gadolinium enhanced images showed diffuse enhancement of all the thigh muscles relatively spared to the fatty infiltration (Fig. 5, 6).
Hip joints were unremarkable and the underlying femur was normal.

Discussion:

A. Background

Limb-girdle muscular dystrophy (LGMD) is a group of progressive muscle weakness genetically determined diseases [1]. There are autosomal recessive (90%) and autosomal dominant (10%) forms. LGMD2A (autosomal recessive) is the most common variant, accounting for 8-26% of all LGMDs and is caused by a mutation in the gene encoding the proteolytic enzyme calpain-3 [2].

B. Clinical Perspective

LGMD have a variable age of onset with autossomal recessive forms presenting typically childhood onset and autosomal dominant forms with typical adult onset (as late as 40 years of age). The clinical picture consists of mostly symmetrical muscular atrophy with rare muscle hypertrophy or contractures. Slowly progressive muscular weakness with predominantly involvement of shoulder or pelvic-girdle muscles with variable rates of progression is

characteristic [3], a common feature with congenital dystrophinopathies such as Duchenne and Becker muscular dystrophies.

C. Imaging Perspective

Ultrasound allows evaluation of muscle echogenicity (fatty infiltration) and thickness. MR imaging is the modality of choice due to its superior soft tissue contrast allowing evaluation of atrophy and pseudohypertrophy [4]. T1W images are useful for evaluation of relative fatty infiltration, T2W for evaluation of oedema-like changes. MR also presents a potential tool for evaluation of therapy response and biopsy planning (by showing the areas of muscular oedema). Some studies demonstrate dystrophic muscles to show increased gadolinium-enhancement and advocate potential clinical use of contrast-enhanced MRI [7].
LGMD2A presents a common pattern of muscle atrophy with early atrophy of the posterior thigh muscles, the adductors and semimembranosus. The muscles relatively spared commonly are vastus lateralis and intermedius, sartorius and gracilis [5]. In this patient the atrophic vastus lateralis is unusual, probably due to the late disease state. The final diagnosis is obtained by muscle biopsy with immunohistochemical staining.

D. Outcome

There is a variable rate of disease progression with severe disability usually occurring within 20-30 years. Cardiac arrhythmias are a major cause of morbidity and mortality [6].
No specific treatment is available for any of the LGMD syndromes, though aggressive supportive care is essential. Low-impact aerobic exercise may improve cardiovascular and musculoskeletal function.

E. Take Home Message

Diagnosis and evaluation of muscular dystrophy depend on clinical, pathologic, and biochemical parameters. MR imaging has a role in the work up and management of muscular dystrophies. The variation in pattern of muscle involvement and relative sparing may allow differentiation of subtypes of muscular dystrophies. In LGMD2A patients usually have early atrophy of the adductors and semimembranosus.

Differential Diagnosis List: Limb-girdle muscular dystrophy, LGMD2A autosomal recessive., • Limb-girdle muscular dystrophy;, • Congenital muscular dystrophy;, • Dystrophinopathies (such as Duchenne muscular dystrophy), • Disuse bilateral muscular atrophy;

Final Diagnosis: Limb-girdle muscular dystrophy, LGMD2A autosomal recessive.

References:

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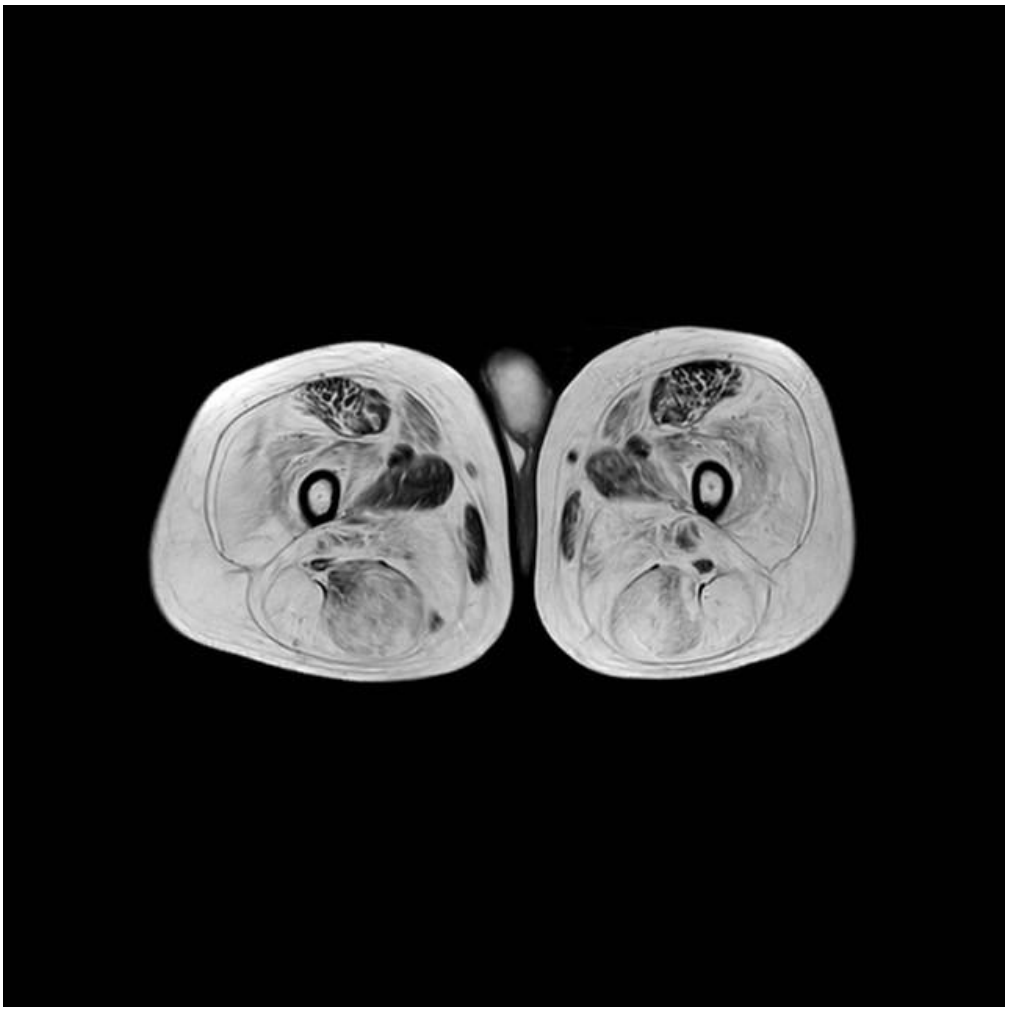
Figure 1

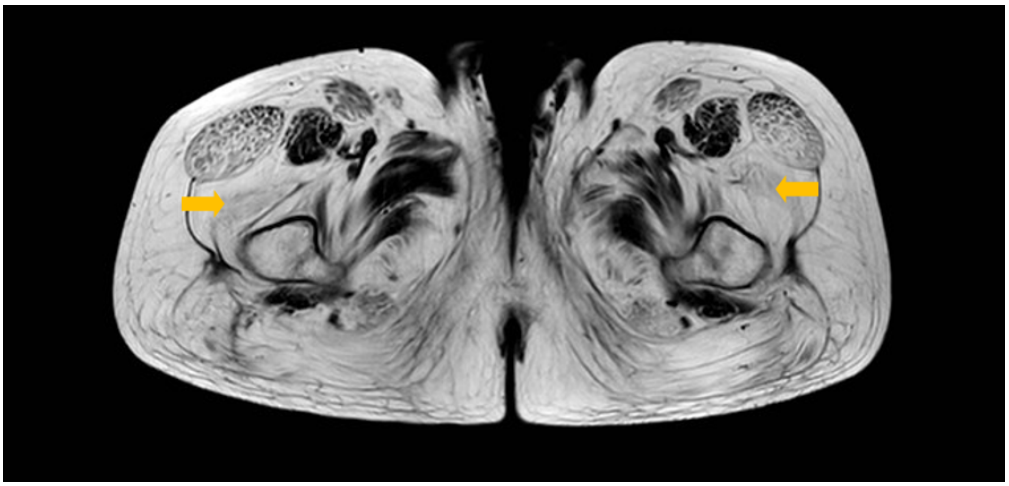
a



Description: Axial T2W MR image showing extensive bilateral and symmetric thigh muscle atrophy with fatty infiltration. There is a relatively sparing of the rectus femoris, adductors and gracilis muscles.
Origin: Prada R, Department of Radiology, POVISA Hospital, Vigo, Spain.

Figure 2

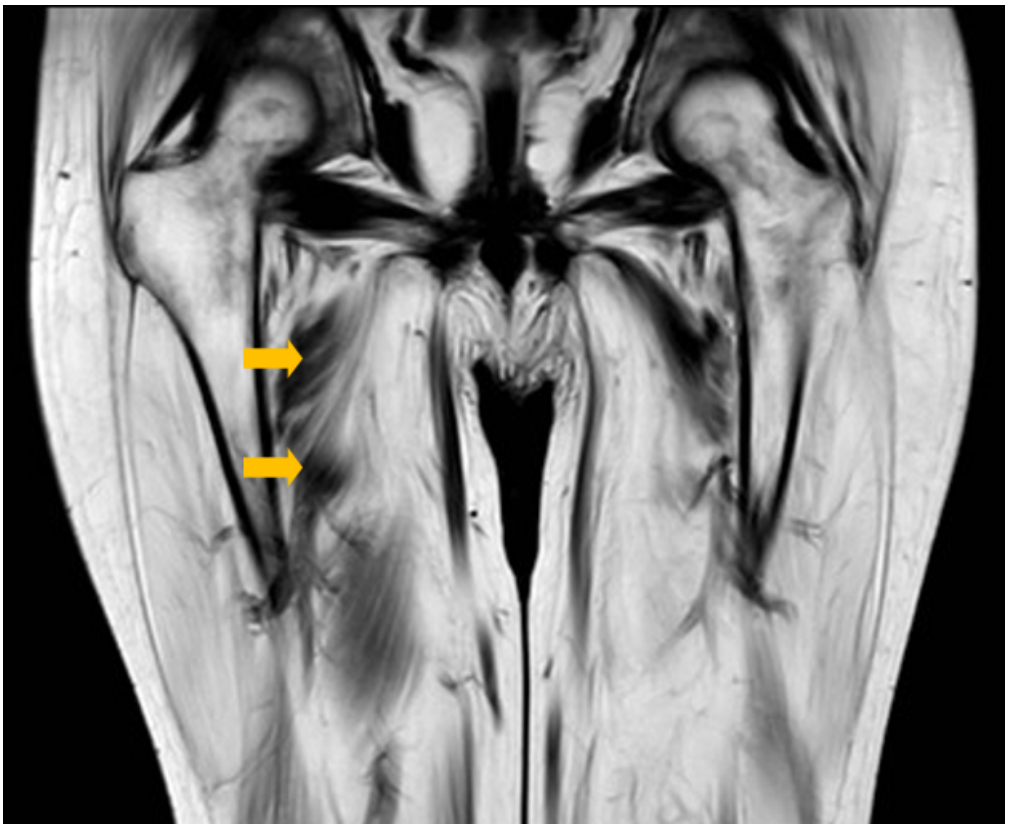
a



Description: Axial T2W MR image at a different level highlights the marked atrophy and fatty infiltration of the vastus lateralis muscles (arrows). **Origin:** Axial T2W MR image at a different level highlights the marked atrophy and fatty infiltration of the lateral vastus lateralis muscles (arrows).

Figure 3

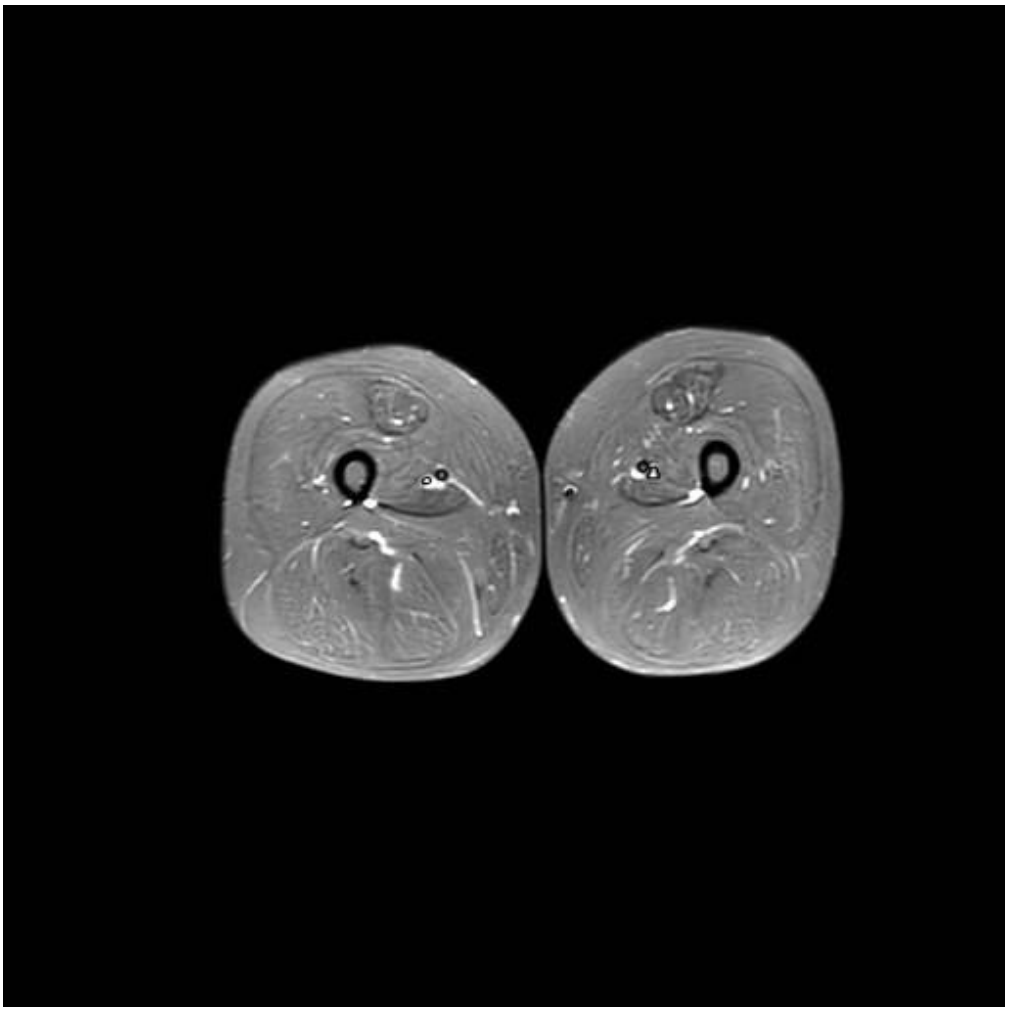
a



Description: Coronal T1 MR image highlighting fatty sparing of the adductors muscles (arrows). **Origin:** Prada R, Department of Radiology, POVISA Hospital, Vigo, Spain.

Figure 4

a



Description: Axial STIR MR image depicting diffuse muscular low signal intensity due to fatty infiltration. There is no oedema. **Origin:** Prada R, Department of Radiology, POVISA Hospital, Vigo, Spain.

Figure 5

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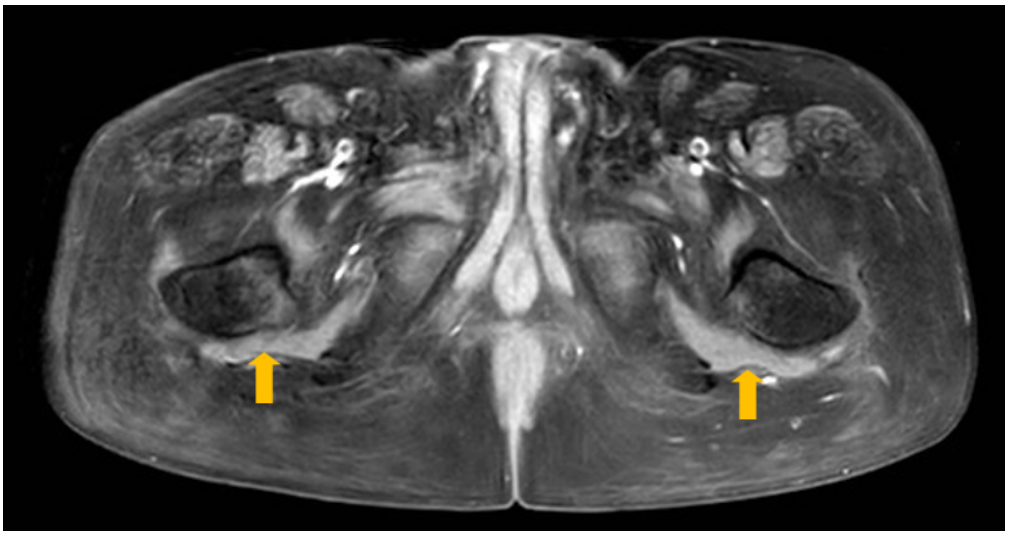

Description: Axial post-gadolinium MR image showing slight contrast enhancement of the adductors muscles and intense enhancement of the quadratus femoris muscles (arrows). **Origin:** Prada R, Department of Radiology, POVISA Hospital, Vigo, Spain.

Figure 6

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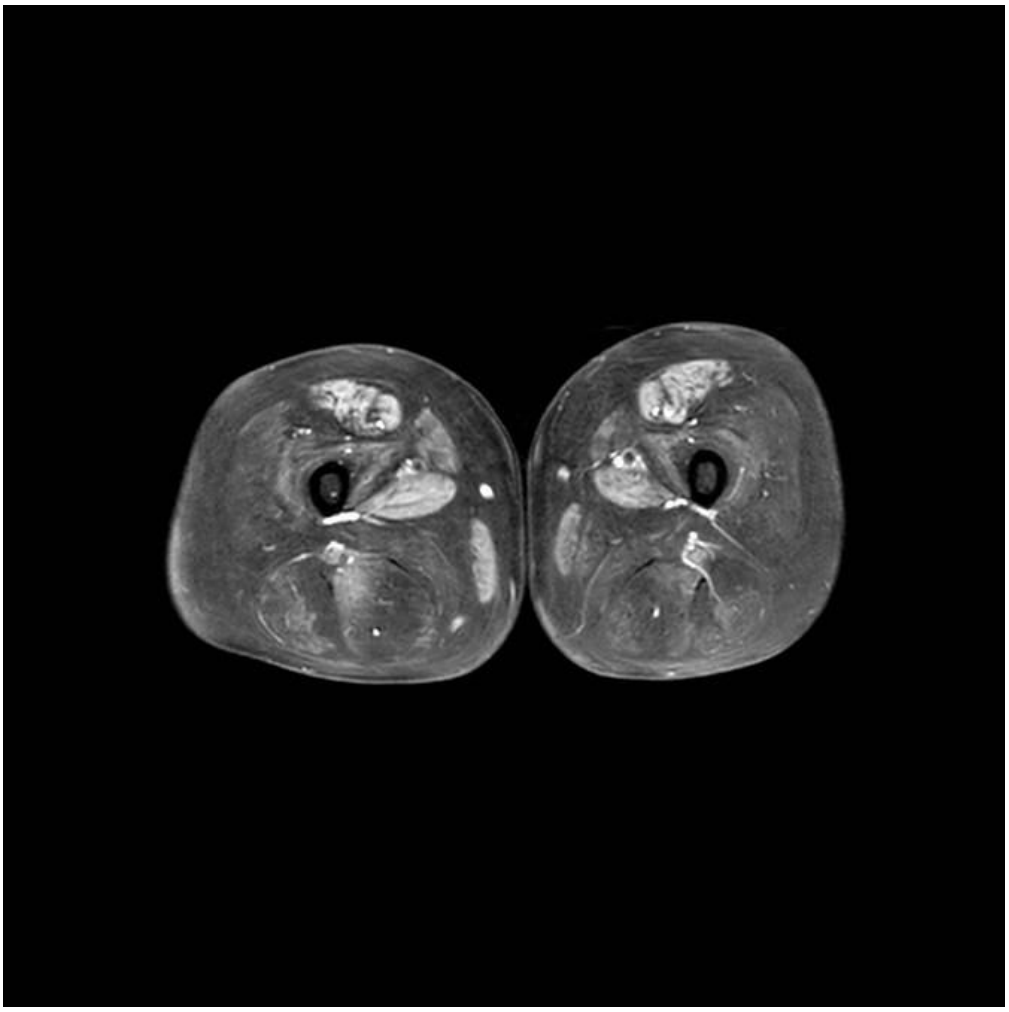

Description: Axial post-gadolinium MR image at a lower level than figure 5 showing diffuse muscle contrast enhancement. **Origin:** Prada R, Department of Radiology, POVISA Hospital, Vigo, Spain.