

Macro dystrophia lipomatosa - a rare congenital anomaly

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Keywords: Macro dystrophia lipomatosa; congenital

Introduction

Macro dystrophia lipomatosa is a rare non-hereditary developmental anomaly of localized or generalized gigantism of limbs or digits, predominantly consisting of fibroadipose tissue. It occurs at any age, predominantly in the distal lower limbs. We report a boy presenting with unilateral lower limb swelling compatible with the diagnosis of Macro dystrophia lipomatosa, which was subsequently confirmed by imaging and histopathology.

Case Report

A 14 year old, previously healthy boy presented with a unilateral painless, but progressive right-sided calf swelling which was more prominent on the ventral aspect of the for a duration of 4 years (Figure 1). He has no family history of musculoskeletal diseases. There were no dermatological or neurological abnormalities noted during further examination. X-rays of the lower limb revealed an enlargement of the soft tissue of the right calf region. The computerized tomography of the lower limbs revealed atrophy of the lateral head of the gastrocnemius muscle with fatty infiltration (Figure 2).



Figure 1. Hypertrophy of the left lower limb calf muscle, visibly more prominent on the ventral aspect.

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DOI: <http://10.4038/sljs.v34i3.8287>

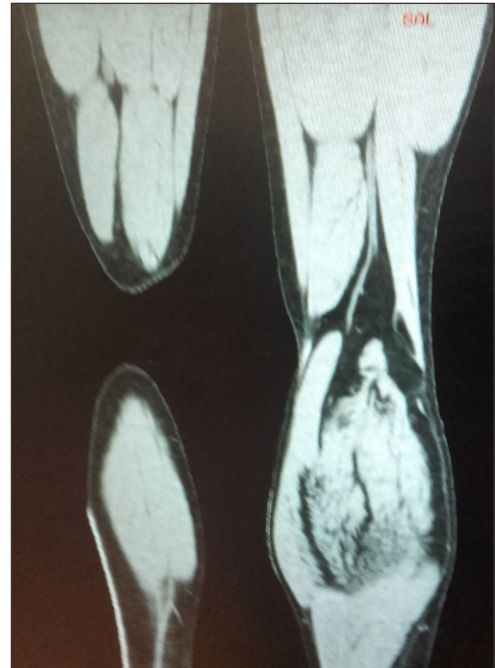


Figure 2. The CT of the lower limb revealed atrophy of the lateral head of the gastrocnemius muscle and fatty infiltration.

The biopsy of the calf muscle revealed fibroadipose tissue including a nerve with a thick fibrotic peri-neural sheath (Figure 3).

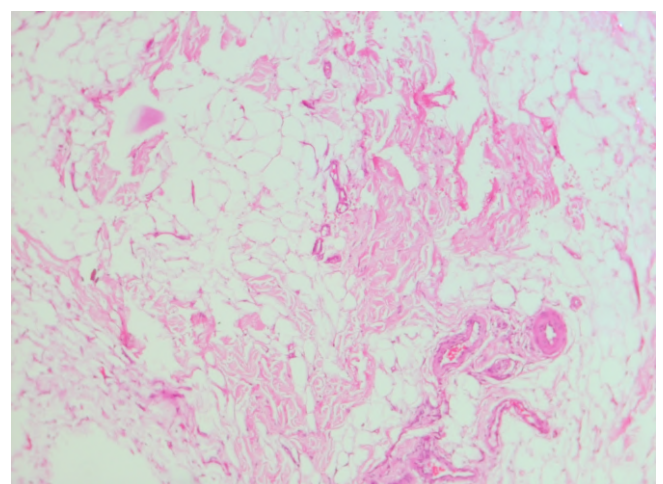


Figure 3. The histopathology of the muscle biopsy revealed fibrofatty infiltration into the gastrocnemius muscle.

His morphological, radiological and histopathological findings favoured a diagnosis of Macro dystrophia lipomatosa. He was referred to the plastic surgery team for consideration of reconstructive surgery.



Discussion

Macrodystrophia lipomatosa is a rare congenital localized or generalized gigantism of the limbs or digits [1]. It is characterized by an abnormal proliferation of all mesenchymal elements; mainly the fibroadipose tissue in digits or limbs. It occurs commonly in unilateral digits or limbs and is predominantly in males, involving the lower limbs [2-4]. The affected region continues to grow from childhood to the pubertal period and then reaches a plateau. We reported Macrodystrophia lipomatosa in a male patient involving the lower limb, diagnosed as a teenager.

The incidence and aetiology of Macrodystrophia lipomatosa is unknown. The lipomatosa degeneration, foetal circulation abnormality, damage of extremity bud, errors in the segmentation in intrauterine life and hypertrophy of the concerned nerve have been postulated to be involved in the aetiology of Macrodystrophia lipomatosa in literature[5].

There are two types which have been described; static and progressive [6]. The growth rate is normal in static form and is faster in progressive form [6]. The first case of Macrodystrophia lipomatosa was described by Feriz in 1925. Barsky described the static and progressive form of Macrodystrophia lipomatosa in 1967 [7].

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MRI scan revealed abundant fatty tissue and fibrous strands of low signal intensity, and was useful to exclude other differential diagnoses of Macrodystrophia lipomatosa such as neurofibromatosis, fibrolipomatous hamartoma and hemangiomas [8]. The histopathology showed the presence of abundant fibrofatty infiltration into the gastrocnemius muscle. The imaging and histopathological

study of this patient confirmed the diagnosis Macrodystrophia lipomatosa.

Patients are usually asymptomatic. However, conservative debulking surgery can be performed to improve cosmetic appearance. The recurrence rate after reconstruction surgery however is high [9].

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Key Points:

- The different imaging modalities and histopathological studies play an important role in the diagnosis of Macrodystrophia lipomatosa.
- Debulking surgery is an option to improve the cosmesis factor for patients.