Macrodystrophia lipomatosa: A rare entity with characteristic radiological findings

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Received - 19 November 2018 Initial Review - 27 November 2018 Accepted - 08 January 2019

Macrodystrophia lipomatosa is a rare congenital entity characterized by the proliferation of all the mesenchymal elements of a digit or digits with a disproportionate increase in the fibroadipose tissue [1]. In this condition, a progressive form of macrodactyly occurs with cessation of abnormal growth at puberty. Usually, one or more digits of a limb are affected. It frequently occurs along the median nerve distribution in the upper extremity and along the plantar nerve in the lower extremity.

Conventional radiographs have previously revealed hypertrophy of the soft tissues and osseous structures. Soft tissue lucency on radiographs has been found to result from hypertrophic adipose tissue, while the phalanges are typically elongated and widened. Magnetic resonance imaging (MRI) scans of macrodystrophia lipomatosa cases have shown the presence of abundant adipose tissue which exhibits same signal intensity as subcutaneous fat. Linear fibrous strands of low signal intensity, bone hypertrophy, and cortical thickening can also be detected on MRI [2].

Common differentials include neurofibromatosis, hemangiomatosis, and lymphangiomatosis which can usually be distinguished on imaging [3]. Neurofibromatosis is the most difficult condition to distinguish from macrodystrophia lipomatosa; however, only the median and plantar nerve affections are characteristic in macrodystrophia lipomatosa. In neurofibromatosis patients, other clinical features such as axillary freckles, cafe-au-lait spots, and Lisch nodules may be evident. The treatment of macrodystrophia lipomatosa is surgery, consisting of debulking and partial amputation of the affected limb [4]. Surgery should be delayed until the patient's growth period is complete if the deformity is not serious and no nervous system symptoms are present. Recurrent operations may be required for complete treatment.

A 15-year-female patient presented with progressive painless enlargement of the left forefoot since birth. Physical examination revealed unusually large 2nd, 3rd, and 4th toes (Fig. 1). No overlying cutaneous changes, pitting edema, or neurological deficit were found. A plain radiograph revealed enlargement of the soft tissues of 2nd, 3rd, and 4th toes and plantar aspect of the forefoot with a lengthening of the phalanges and corresponding metatarsals (Fig. 2). No evidence of any bone erosion was found.

For further characterization, MRI was carried out on a 1.5 Tesla scanner. MRI demonstrated enlargement and marked proliferation of adipose tissue characterized by high T1/T2 signal (Figs. 3a and 4a) with signal suppression on fat-saturated sequences (Figs. 3b and 4b). These imaging features were characteristic of macrodystrophia lipomatosa. The patient underwent a debulking procedure and partial amputation of 2nd, 3rd, and 4th toes. No post-operative complications were encountered. Post-operative histopathologic findings confirmed

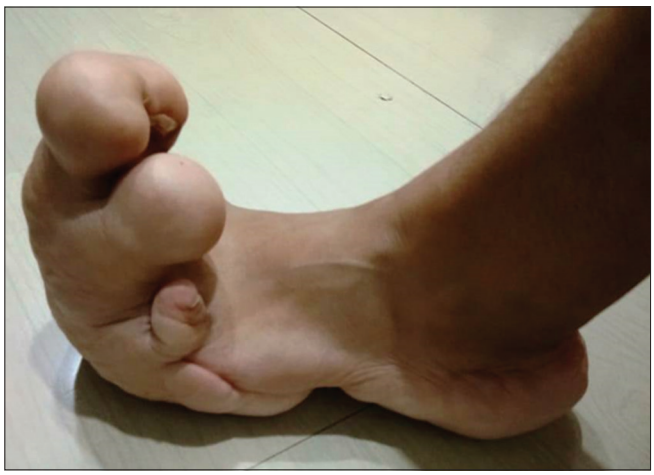

Figure 1: Photograph of the patient's left foot showing unusually large 2nd, 3rd, and 4th toes

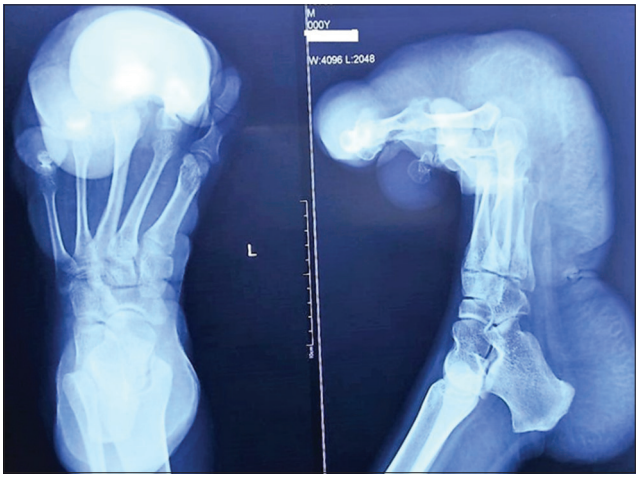

Figure 2: Radiograph showing enlargement of the soft tissues of 2nd, 3rd, and 4th toes and plantar aspect of forefoot with lengthening of the phalanges and corresponding metatarsals

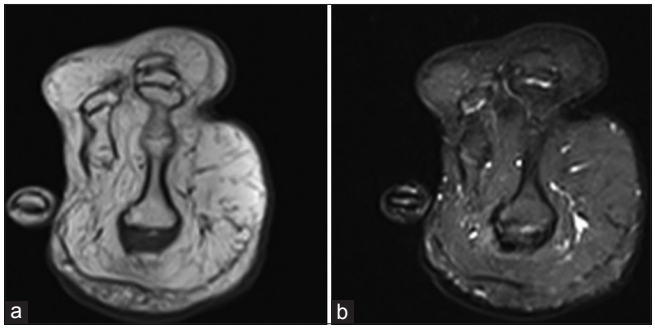

Figure 3: Axial magnetic resonance images demonstrating enlargement and marked proliferation of adipose tissue characterized by high signal on T1-weighted image (a) and signal suppression on fat-saturated T2-weighted image (b)

the diagnosis. At present, the patient is on follow-up to look for any recurrence.

This case highlights the typical imaging features of macrodystrophia lipomatosa. Awareness of characteristic symptoms, imaging features, and differential diagnosis of this rare entity is important for diagnosis and appropriate treatment.

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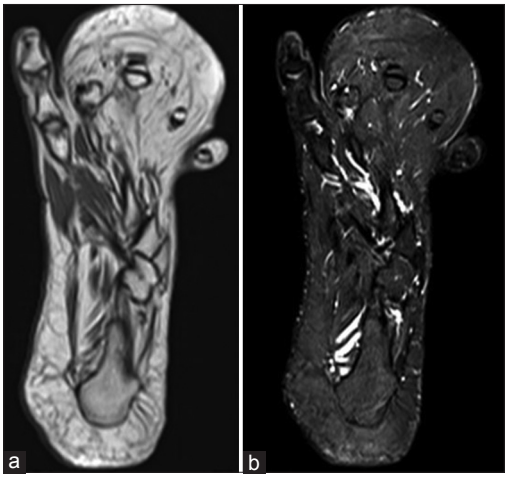

Figure 4: Coronal magnetic resonance images demonstrating enlargement and marked proliferation of adipose tissue characterized by high signal on T1-weighted image (a) and signal suppression on fat-saturated T2-weighted image (b)

Funding: None; Conflict of Interest: None Stated.

How to cite this article: Ray B, Vijayendra A. Macrodystrophia lipomatosa: A rare entity with characteristic radiological findings. Indian J Case Reports. 2019;5(1):90-91.

Doi: 10.32677/IJCR.2019.v05.i01.031