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Macrodystrophia Lipomatosa: A Rare Case of Ulnar Nerve Territory Involvement

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Abstract

Macrodystrophia lipomatosa (MDL) is a rare congenital, nonhereditary anomaly characterized by overgrowth of all the mesenchymal elements, predominantly the fibroadipose tissue in a sclerotomal distribution commonly involving the median nerve territory in the upper extremity and plantar nerve territory in the lower extremity. It can be either static or progressive, with the former being the more common. MDL is usually present since birth and the affected digit/region increases in length and girth, and growth ceases after puberty. We discuss a rare case of ulnar nerve territory involvement that progressed to grow even after puberty.

Keywords

- localized gigantism
- macrodystrophia lipomatosa
- ulnar territory macrodystrophia lipomatosa

Case Presentation

A 23-year-old male patient came to our hospital with complaints of diffuse swelling of the ulnar aspect of his left hand including the fourth and fifth digits for the past 17 years, which gradually increased to its present size. There was no associated pain. However, he experienced discomfort in movements of his fourth and fifth digits.

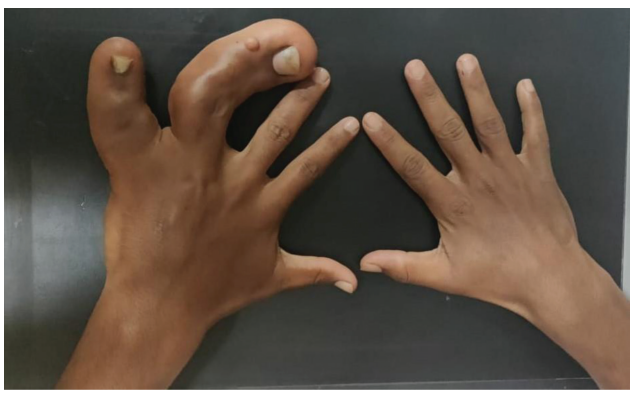

Fig. 1 Clinical image of both hands showing gross enlargement of the left fourth and fifth digits and medial aspect of the left hand with diffuse hyperpigmentation of the skin of the dorsal aspect of his left hand.

On Examination

There was diffuse swelling of the ulnar aspect of his left hand, including the fourth and fifth digits, limiting the movements at the metacarpophalangeal, proximal, and distal interphalangeal joints. There was diffuse hyperpigmentation of the skin of the dorsal aspect of his left hand compared to his right hand (►**Fig. 1**). No skin edema, pigmented nodules, or cutaneous angioma were seen. Neurovascular examination was unremarkable.

Radiographic Findings

Computed tomography (CT) revealed diffuse increase in fat tissue in the volar aspect of the left hand, dorsal and volar aspects of the fourth and fifth digits, and diffuse enlargement

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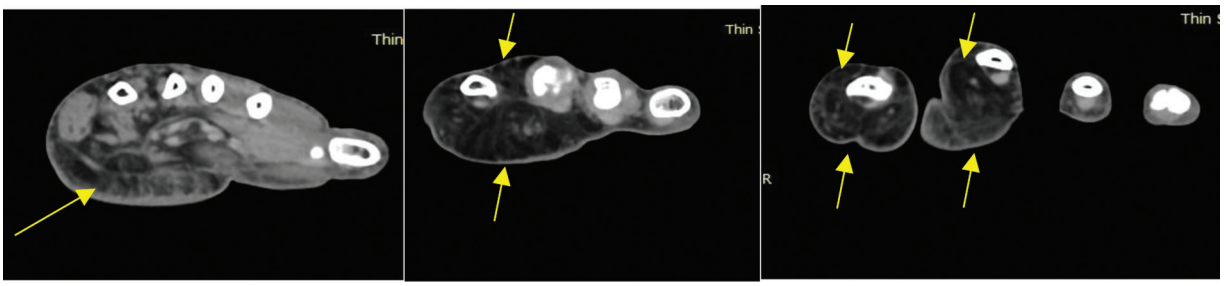

Fig. 2 Axial sections of plain computed tomography (CT) of the left hand in soft-tissue window showing diffuse increase in fat tissue in the volar aspect of the hand, and dorsal and volar aspects of the fourth and fifth digits (*yellow arrows*).

of the proximal, middle, and distal phalanges of the fourth and fifth digits with osteoarthritic changes in the lateral aspect of the distal interphalangeal joints of the fourth and fifth digits (►**Figs. 2**–**5**).

On magnetic resonance imaging (MRI), T1-weighted imaging (T1WI) and T2WI of the hand revealed diffuse hyperintensity in the fourth and fifth digits, which was suppressed on proton density (PD) fat-saturated image, and on time-of-flight (TOF) magnetic resonance angiography (MRA), the prominent palmar arteries of the fourth and fifth digits were noted. However, there was no arteriovenous (AV) malformation (►**Figs. 6** and **7**).

Discussion

Focal enlargement of a part of a body is known as localized gigantism and is always pathological. It can be either static or progressive, with the former being the more common.[1] In the static type, the affected part grows in proportion with the rest of the body, whereas in the progressive type, the affected part grows faster than the rest of the body.[2] Localized gigantism can be seen in various conditions such as neurofibromatosis type 1, Klippel–Trenaunay–Weber syndrome, lymphangiomatosis, hemangiomatosis, macrodystrophia lipomatosa (MDL), fibrolipomatous hamartoma of the nerve, and proteus syndrome.[2]

MDL is a rare congenital nonhereditary anomaly characterized by overgrowth of all the mesenchymal elements, predominantly the fibroadipose tissue. In 1925, Feriz first used the term "macrodystrophia lipomatosa" to describe localized gigantism involving the lower limb.[3] Goldman and Kaye suggested this term was also applicable to upper limb involvement.[4] The exact pathogenesis of macrodystrophia remains unknown. However, studies have shown an association with *PIK3CA* gene mutation.[5] Pathologically, there is predominant infiltration and hypertrophy of adipose tissue within subcutaneous tissue, nerve sheaths, and periosteum in a sclerotomal distribution.[4,5] MDL is usually

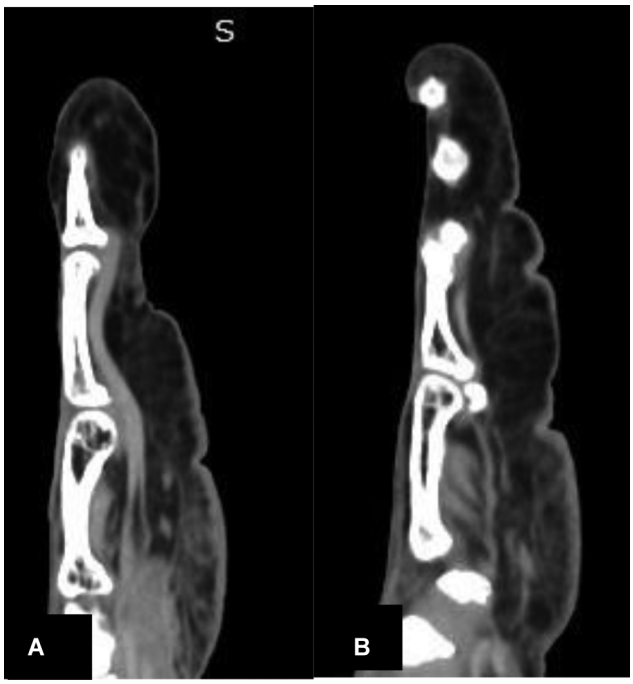

Fig. 3 Sagittal sections of plain computed tomography (CT) of the left hand in soft-tissue window showing a diffuse increase in fat tissue in the volar aspect of the (**A**) fourth and (**B**) fifth digits.

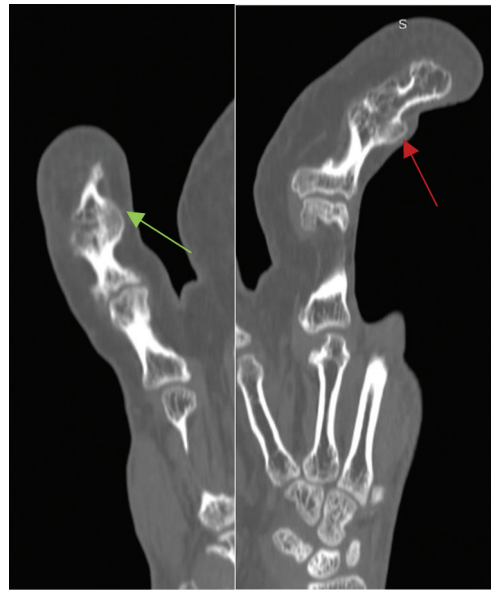

Fig. 4 Coronal section of computed tomography (CT) of the left hand in the bone window showing osteoarthritic changes in the lateral aspect of the distal interphalangeal joints of the fourth (*red arrow*) and fifth digits (*green arrow*).

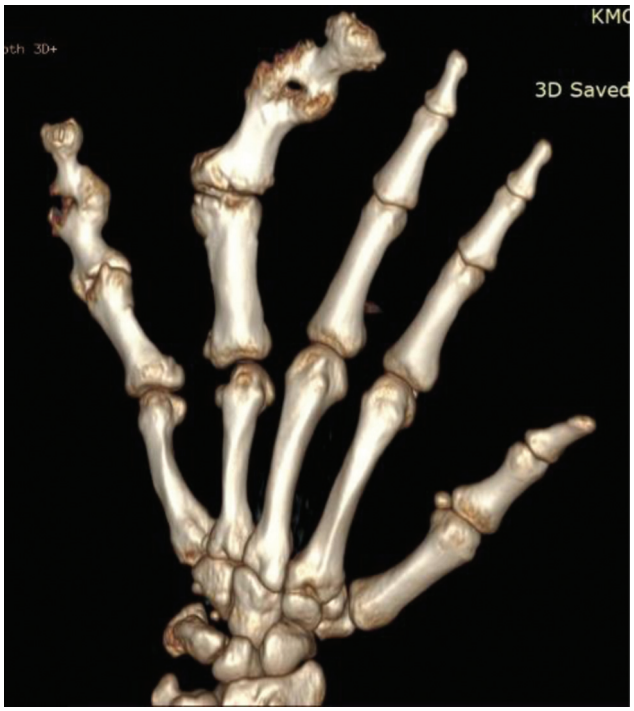

Fig. 5 Three-dimensional (3D) reconstruction of computed tomography (CT) of the left hand showing diffuse enlargement of the proximal, middle, and distal phalanges of the fourth and fifth digits.

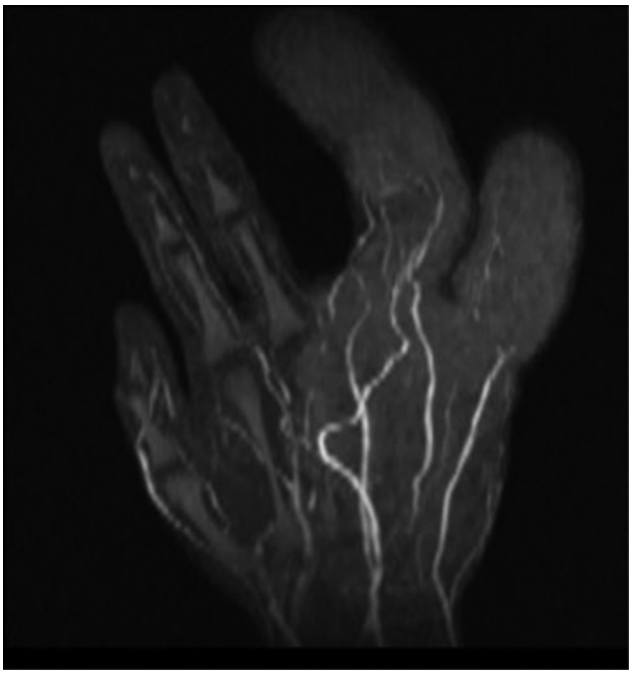

Fig. 7 Time-of-flight (TOF) magnetic resonance angiography (MRA) of the left hand showing prominent palmar arteries of the fourth and fifth digits.

present since birth and the affected digit/region increases in length and girth, and growth ceases after puberty.[6] In our case, progressive growth continued after puberty.

MDL has no sex predilection although a slight male preponderance is seen,[7] affects both the upper and lower extremities, and usually involves more than one adjacent digit. Most cases involve the index and middle fingers corresponding to the median nerve territory, medial aspect of the foot corresponding to plantar nerve territory.[8] So far, multiple cases involving the median nerve territory,[9,10] medial aspect of the foot,[7,11–13] lateral aspect of the foot,[5] entire limb,[14–16] abdominal wall,[17] and only one case involving the ulnar nerve territory[5] has been reported. To the best of our knowledge and with extensive literature review, this is the second case of MDL involving the fourth and fifth digits, corresponding to the ulnar nerve territory.

Conclusion

MDL is painless localized gigantism with sclerotomal involvement commonly involving the median nerve territory in the upper extremity and plantar nerve territory in the lower extremity, which ceases to grow after puberty. However, rarely, it can also involve the ulnar nerve territory and can progress to grow even after puberty as in our case. A plain radiograph coupled with ultrasound is sufficient for diagnosis. CT and MR

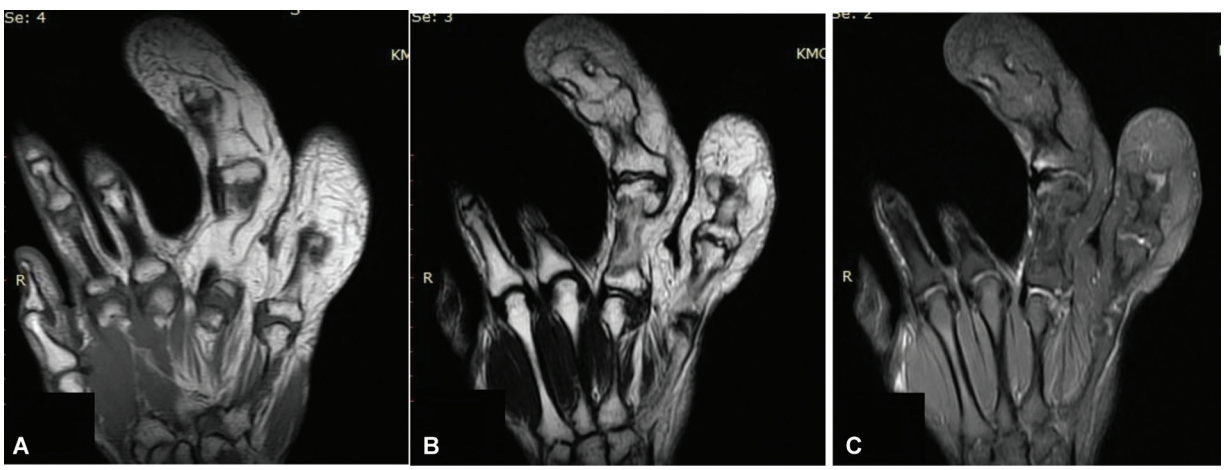

Fig. 6 (**A**) T1-weighted imaging (T1WI) and (**B**) T2WI of the coronal section of the hand showing a diffuse increase in fat tissue in the fourth and fifth digits, which is suppressed on (**C**) proton density (PD) fat-saturated image.

can help in differentiating MDL from other similar pathologies, assessing associated vascular malformation, and for operative planning by evaluating the full extent of the lesion.

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Conflict of Interest
None declared.

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