Macrodystrophia Lipomatosa with Median Nerve Involvement

Introduction:

Macrodystrophia Lipomatosa (MDL) is a rare cause of congenital macrodactyly, characterized by progressive proliferation of all mesenchymal elements, with a disproportionate increase in fibro-adipose tissue. This developmental anomaly is reportedly more common in the foot than in the hand, with a predilection for the plantar and median nerve distribution.

Clinical History:

A 25 year old female presented with history of enlargement of middle and ring finger since childhood. She had undergone surgery for the same in childhood. History of painful swelling over volar part of the wrist was noted past 4 to 5 months, because of which she was referred for MRI.

Imaging findings:

- Diffuse enlargement and hypertrophy of the median nerve with fatty proliferation, measuring 1.2 cm in maximum thickness. (cause of swelling)
- Splaying of third and fourth digits at their distal ends.
- Fibro-fatty proliferation of deep subcutaneous fat around the phalanges of third and fourth digits, showing fat signal intensity on all sequences.
- Expansion of medullary cavity of the phalanges of third digit.
- The terminal phalanges of third and fourth digits are not visualized (history of surgery noted).

Discussion:

MDL is commonly associated with fibro-lipomatous infiltration of peripheral nerves, with the median and plantar nerves being the most frequently affected. Although fibro-lipomatosis of the nerve can occur in isolation, i.e., without associated localized gigantism, in up to two thirds of the cases there is associated macrodactyly. Although overgrowth of all mesodermal elements of the digits is the dominant finding in MDL, associated clinical findings such as polydactyly can occur.

In most reported cases to date, the lesions are present at birth or develop within the first weeks of life. The lesions are unilateral and associated with uneven and progressive overgrowth that is more rapid compared to the rest of the limb.

Radiographs reveal soft-tissue and osseous overgrowth often with elongated, broadened, and splayed phalanges. The imaging appearance, particularly with sonography and MR imaging of advanced lipomatosis of a nerve is usually distinctively characteristic and reflects the underlying disease.

Sonography reveals alternating hyperechoic (fat) and hypoechoic (nerve fascicles) bands in a diffusely enlarged nerve, thus creating a cable-like appearance.

MR images show longitudinally oriented cylindrical areas of low to intermediate signal intensity (nerve fascicles) surrounded by adipose tissue in a diffusely thickened nerve. On MR images, increased fat content of the digits is also apparent in patients with macrodactyly.
The pathology of MDL is quite characteristic. The normal perineurium surrounding the nerve fascicles creates a barrier separating the nerve axons within the fascicle from the surrounding adipocytes. Because MDL usually regresses to a static lesion on completion of a child's growth, if surgery is to be performed it is usually delayed until growth has stopped. However, there have been cases of continuous enlargement into adulthood.

Fig 1: T2Wi Coronal.
Hypertrophy of subcutaneous fat around middle & index finger showing fat signal.

Fig 2: T2Wi Coronal.
Expansion of medullary cavity of phalanges of middle finger.

Fig 3 & 4:
T2Wi Axial, T1Wi Sag.
Diffuse thickening and hypertrophy of median nerve (arrows) with fatty proliferation.
Differential Diagnoses:

The differential diagnoses of MDL and macrodactyly include Neurofibromatosis, Hemangiomatosis, Lymphangiomatosis, Proteus syndrome, and Fibro-Lipomatous hamartomas.

Conclusion:

MR imaging is considered pathognomonic of MDL and, therefore, is advisable in the diagnostic approach of localized gigantism with questionable clinical features.

Radiological investigations, especially MRI, help to make a definitive diagnosis noninvasively and to differentiate it from other causes of macrodactyly.

Regards,

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N.B: This case is authentic and from the archives of Radiance Diagnostics. For any queries / suggestions/feedback write to us at radiance@radiancediagnostics.in