Macrodystrophia lipomatos (MDL) is a congenital, non-hereditary form of focal gigantism characterised by progressive overgrowth of all the mesenchymal elements (fat, skeletal muscle, nerve sheath and bones) of the digits. The overgrowth appears to develop along specific sclerotomes and is more common in the lower limbs along the distribution of the plantar nerves. Volar and distal portion of the digits are more commonly affected. The proliferative fatty growth is unencapsulated and can be appreciated as globular lucencies within the soft tissue on radiographs and confirmed on MRI. Biopsy revealed fragments of adipose tissue without any other spindle cell elements or malignant cells. Correlating the clinicoradiological picture and the biopsy findings, a diagnosis of macrodystrophia lipomatosa (MDL) was made. The patient has now been posted for corrective surgery.

Since childhood the first two toes of my right foot were enlarged. I used to hide it by wearing socks as I was afraid that my peers would tease me. I had difficulty in wearing shoes and my father used to get a customised shoe made for me. We consulted a doctor who conducted multiple tests and told that I had macrodystrophia lipomatosa. I was told that surgery could reduce the size of the deformity but the chances of recurrence were high. I was apprehensive and hence I refused the surgery. Finally when I was 20 years old, the swelling started to increase in size and it became a more significant cosmetic problem as well as a matter of discomfort while walking. I consulted the surgeon again. A biopsy was taken and I have consented for the surgery. (Translated perspective.)
MRI. The bones show broadening and elongation along with proliferative (hyperostosis) as well as degenerative changes due to altered biomechanics in the weight-bearing regions. MDL characteristically does not have systemic involvement or family history and occurs sporadically. Treatment is surgical debulking, but it is associated with high recurrence rates.4

A close differential on the radiograph is neural fibrolipomatous hamartoma since it also shows localised gigantism with fat lucencies along the distribution of a particular nerve. However, unlike MDL, there is no bony hypertrophy and on MRI, the fat is seen enclosed within the expanded nerve sheath and the nerve fibres are interspersed giving a ‘co-axial cable appearance’ on the transaxial images. The other differential diagnoses include neurofibromatosis type 1 (plexiform neurofibroma), vascular malformations (sometimes in association with Klippel-Trenaunay-Weber syndrome) and Proteus syndrome.5

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Learning points
► Whenever encountered with a case of focal gigantism with no systemic involvement, macrodystrophia lipomatosa should be in the list of differentials.
► On radiographs, the area of gigantism shows globular lucencies of fat as well as proliferative outgrowths and broadening of the underlying bones.
► The presence of bony hypertrophy of the affected digit on the radiograph differentiates it from fibrolipomatous hamartoma which is a close differential.