TOE MACRODACTYLY, MACRODYSTROPHIA LIPOMATOSA, FIBROLIPOMATOUS HAMARTOMA & LIPOMATOSIS OF NERVE. ARE THEY SIMILAR?

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Introduction: Macrodystrophia Lipomatosa (MDL) of toe is a rare, congenital, disproportionate overgrowth involving one or more digits in the lower limb. Despite being a benign condition, it may cause physical impairment and interfere with daily activities. This form of localized gigantism is the result of excessive proliferation of fibroadipose tissue within the nerve along with associated macrodactyly. The clinical, radiographic findings, and histopathological findings are presented. The difference between MDL, fibrolipomatous hamartoma (FLH) and lipomatosis of the nerve (LON) are also discussed.

Discussion: We report a case of a 15 years old boy, who presented with enlarged first and second toes which were present since birth. The enlarged toes were painless and slow growing. Radiograph revealed enlarged phalanges of the second toe, with the middle and distal phalanges deviated towards the third toe. Magnetic resonance imaging (MRI) showed thickened subcutaneous tissue surrounding the enlarged bony structures. He underwent Ray amputation of the 2nd toe and the specimen was examined. Histological examination showed infiltration of the epineurium and perineurium by adipose and fibrous tissue which dissected between and separated individual nerve bundles associated with concentric perineural fibrosis and pseudo-onion bulb formation. These findings were consistent with the diagnosis of lipomatosis of nerve. His review in clinic 6 months after the surgery showed no recurrence of the growth.

Conclusion: MDL is an extremely rare condition that can present with enlargement of toes or digits. It may present with or without lipomatosis of the nerve. However, in the existing literature, MDL, FLH & LON are used interchangeably and leads to confusion. The term FLH of nerve or LON can be used in cases when there is nerve enlargement without subcutaneous, inter-muscular and intra-muscular fibrofatty hypertrophy. Non-invasive investigations such as MRI may help establish a diagnosis and exclude other pathologies, but histological examination remains the gold standard.