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A rare case of macrodystrophia lipomatosa progressiva

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Image in medicine

A non-heritable congenital expansion of a digit is known as macrodactyly. It happens as a result of a mutation in the PIK3CA pathway, which regulates healthy cell proliferation. It is usually associated with gigantism, neurofibromatosis, lipofibromatosis, digital hyperostosis and hemihypertrophy. It is not always evident at birth, but at about age 2, the affected fingers begin to grow significantly faster than the normal fingers. Ninety percent (90%) of it is unilateral, and 70% of it involves more than one-digit most often the index. The expansion might only affect the digit or it might spread to the palm, forearm, or arm. The digit may exhibit angulation deformity. The distal

interphalangeal joint occasionally exhibits motion restriction. The patient presented to us with a loss of two-point discrimination and sensory deficit. In this condition, the motor deficit is rarely seen. Our patient reported a tingling sensation and the possibility of carpal tunnel syndrome was ruled out as an enlarged median nerve might result in carpal tunnel syndrome. Inspection and palpation revealed thick, fibrofatty tissue with restricted movements. The patient's symptoms were relieved by NSAIDs and Pregabalin 75mg, symptomatic relief was present, and the patient was advised debulking surgery as the treatment modality.

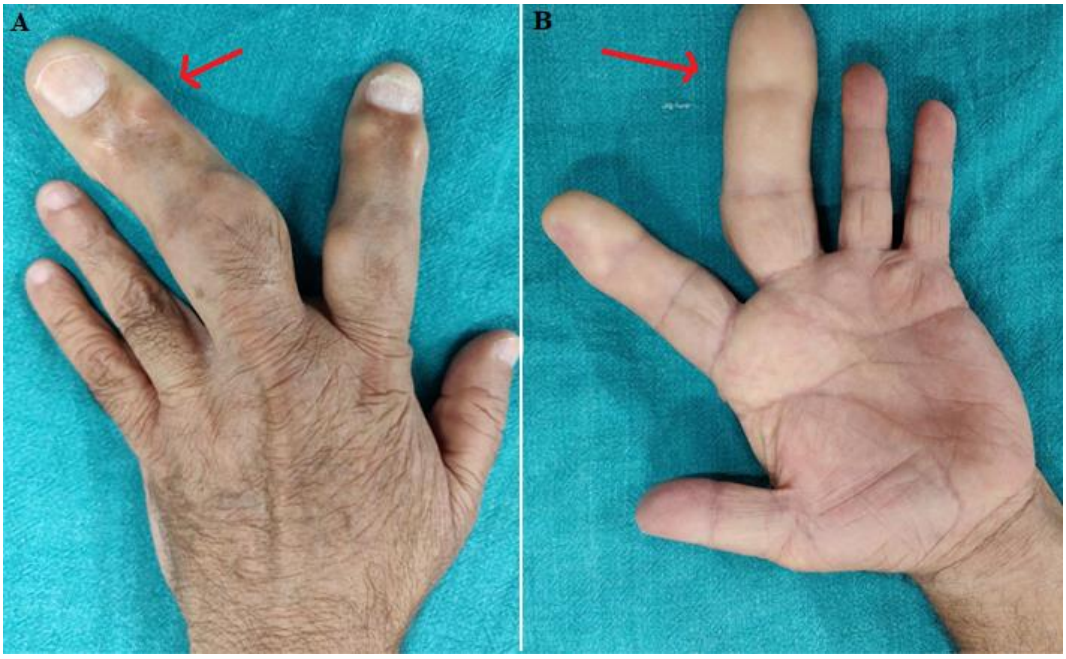

Figure 1: dorsal and volar aspect of the hand (A, B) showing macrodactyly of the index finger