Macrodystrophia lipomatosa is a congenital progressive gigantism of the hand and foot, which may be local or may involve the entire limb. Macrodystrophia lipomatosa comes to clinical attention mostly because of cosmetic reasons, and rarely due to development of neurovascular compression or mechanical problems like secondary degenerative joint disease. Magnetic resonance imaging (MRI) is considered pathognomonic of MDL and helps to differentiate it from other causes of localized gigantism (1). Although MDL of hand is more common along median nerve distribution; we report a rare case of MDL in a 20 years old female involving ring and little fingers of right hand with predominant ulnar nerve distribution.

Case report

A twenty-year-old girl presented to outpatient clinic of orthopaedics department with long standing swelling of the medial aspect of right hand. According to the history, she was born with disproportionately large medial two fingers with progressive growth in relation to the remaining digits, more marked in the ring finger. She had no pain or neurovascular symptoms. On examination, the patient had hypertrophy of the ring and the little finger (Fig. 1A). The other hand was normal. Swelling was firm on palpation without any inductions or tenderness. Assessment of the other systems showed no abnormality. Patient was subjected to routine lab examination along with radiological investigations, namely Radiograph & MRI. Radiograph of the right hand revealed enlarged phalanges of the ring and the little finger with prominent overlying soft tissue (Fig. 1B). MRI showed gross thickening and hypertrophy of osseous and soft tissue component. The soft tissue appeared hyperintense on both T1 and T2 weighted sequences (Fig. 2A, B) with evidence of suppression of bright signal on fat-suppressed STIR sequence (Fig. 3A, B) consistent with fatty proliferation of the soft tissues. Small tissue was biopsied for histological analysis, which revealed proliferation of normal-appearing adipose tissue. Patient was diagnosed as a case of macrodystrophia lipomatosa. Patient was advised cosmetic and reconstructive surgery.

Discussion

Macrodystrophia lipomatosa is a congenital progressive gigantism of the hand and foot that may be localized or may involve the entire limb. It is characterized by proliferation of all mesenchymal components, particularly fibroadipose tissue, most frequently in the distribution of the median nerve in the upper extremity and in the distribution of the plantar nerves in the lower extremity. Radiographs reveal soft-tissue and osseous overgrowth often with elongated, broadened, and splayed phalanges.
References


(2). The imaging appearance, particularly with sonography and MRI 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 (3, 4). MR images are similar, with longitudinally oriented cylindrical areas of low to intermediate signal intensity (nerve fascicles) surrounded by high signal intensity thickened adipose tissue (5). MDL should be differentiated from other forms of congenital localized gigantism such as Klippel-Trenaunay syndrome (KTS), Proteus syndrome and neurofibromatosis. KTS is differentiated from MDL by presence of varicose veins and anomalous lymph system. Proteus syndrome, also known as Wiedemann’s syndrome is characterized by mosaic overgrowth of skin, bones, muscles, fatty tissues, blood and lymphatic vessels, as well as by visceromegaly, lung cysts, and predisposition to benign and malignant tumors (6). Finally, neurofibromatosis cutaneous manifestations of the disease (cafe au lait macules, neurofibromas, freckling in the axillary and inguinal regions) and positive family history are typically present.

As the patient seeks medical advice usually for cosmetic reasons, surgical correction is the management of choice. The surgery is usually planned after puberty when the growth of affected parts stop. Multiple debulking procedures, osteotomies and epiphysiodesis may be required for satisfactory outcome (7).

Fig. 2. — Coronal (A) and sagittal (B) T1-weighted MR image of the right hand reveals enlargement of phalanges of 4th and 5th digit with gross thickening and hypertrophy of overlying soft tissue component that shows hyperintense signal intensity.

Fig. 3. — Coronal (A) and axial (B) STIR sequence showed suppression of bright signal intensity of soft tissue suggesting fatty proliferation consistent with the diagnosis of macrodystrophia lipomatosoa.