CASE REPORT

Imaging features of macrodystrophia lipomatosa: an unusual cause of a brawny arm

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SUMMARY

Macrodystrophia lipomatosa (MDL), a rare non-hereditary congenital disorder of localised gigantism, is characterised by progressive proliferation of all mesenchymal elements, with a disproportionate increase in fibroadipose tissue. We report a case of a 19-year-old man who presented with a history of painless enlargement of the left upper limb since childhood, which was gradually increasing in size and predominantly involving the radial aspect of the upper limb with relative sparing of the ulnar aspect. The patient was imaged with X-ray and MRI. Imaging and clinical features were classical of MDL. The patient underwent stage 1 reduction plasty of the left forearm; preoperative and histopathological findings confirmed the preoperative diagnosis.

BACKGROUND

In patients presenting with localised gigantism of the extremity, a differential of macrodystrophia lipomatosa (MDL) should be kept in mind apart from neurofibromatosis, haemangiomatosis, lymphangiomatosis, Proteus syndrome, fibrolipomatous haematoma and Klippel-Trenaunay-Weber syndrome, since MDL is treatable mainly for cosmetic and psychological reasons, depending on the patient's age and health status. Investigations such as plain radiograph, ultrasonography and MRI can help to determine the cause and aid in better management of this condition.

CASE PRESENTATION

A 19-year-old man presented with a history of painless enlargement of the left upper limb

(figure 1) since childhood. There was a gradual increase in size predominantly involving the radial aspect of the upper limb with relative sparing of the ulnar aspect. Six years earlier (2009) a surgery was performed for similar symptoms. The skin over the limb was thick and non-tender. There were no associated nodules, café-au-lait spots or pitting oedema, and no audible bruits or thrills. The patient's range of movement was reduced in the affected limb, and he was stressed and depressed owing to his inability to engage in normal social activities and interactions. No other significant history was present.

INVESTIGATIONS

X-ray findings

Asymmetrical soft tissue prominence on the lateral aspect of the arm (black star in figure 2A, B), forearm (black star in figure 2C) and hand, including first and second digits (white arrow in figure 3A) were seen on X-ray. Also seen was a smooth enlargement of the cortex of the midshaft of humerus towards the lateral aspect (black arrow in figure 2B). Mild radial bowing of the radius and ulna was noted (white arrow in figure 2C), with prominent subcutaneous veins (black arrow in figure 2C). The first and second metacarpals and the phalanges of the first and second digits were diffusely enlarged and showed irregular margins with degenerative changes in first and second carpometacarpal (black arrow in figure 3B) and first metacarpal-phalangeal joints (black arrow in figure 3A) and interphalangeal joint. No soft tissue calcifications were seen.

Figure 1 Clinical images showing localised soft tissue swelling involving the radial aspect of the left upper limb, with sparing of third, fourth and fifth fingers.

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Figure 2 (A) Chest radiograph demonstrates diffuse soft tissue opacity involving the left shoulder (black star). (B) Radiograph of the left arm and elbow joint shows soft tissue prominence on the lateral aspect of the arm and forearm (black star), which are lucent, suggestive of fat; and smooth enlargement of the cortex of the mid part of humerus (black arrow). (C) Prominent subcutaneous veins (black arrow), soft tissue and skin folds (black star) are also seen.

MRI findings

Shoulder and arm

Asymmetrical increase in fatty elements (which appear hyperintense on T1-weighted and T2-weighted images and show complete suppression on short τ inversion recovery and T1 fat suppressed images) without a definite capsule was noted in the subcutaneous plane in the lateral aspect of the shoulder joint and arm (black arrow in figure 4A, B). Fatty infiltration was also noted within the deltoid (black arrow in figure 5A), supraspinatus (black arrow in figure 5A) and infraspinatus (white arrow in figure 5B) muscles. The subcutaneous veins were prominent. No focal mass lesions were seen.

Cortical thickening of the midshaft of humerus towards the lateral aspect was noted (white arrow in figure 5A). The bone marrow did not show any abnormal signal intensity.

Hand and distal forearm

Asymmetrical increase in fatty elements was seen in the subcutaneous tissue and muscles in the radial aspect of forearm, hand, thumb and index finger (stars in figure 6A, B). Prominence of subcutaneous veins was also noted. Enlargement of the first and second metacarpal, trapezium and trapezoid bones, and phalanges of first and second digits were seen with degenerative changes involving the carpometacarpal (black arrow in figure 6A) and interphalangeal joints (white arrow in figure 6A). Third, fourth and fifth digits were normal. No abnormal signal intensity was noted within bone marrow.

Figure 3 (A) Radiograph of the left hand showing soft tissue swelling causing enlargement of first and second fingers with splaying (white arrows). First, second metacarpals and phalanges are diffusely enlarged and show irregular margins with degenerative changes predominantly involving first (B, black arrow) and second carpometacarpal joint, and first metacarpal-phalangeal (A, black arrow) and interphalangeal joints. The third, fourth and fifth digits appear normal.

DIFFERENTIAL DIAGNOSIS

Based on findings of an asymmetrical increase in fatty elements involving the subcutaneous tissue and muscles, MDL was the primary diagnosis after the differentials of neurofibromatosis, haemangiomas, lymphangiomas, Proteus syndrome, fibrolipomatous hamartoma and Klippel-Trenaunay-Weber syndrome were also considered.¹

Type-1 neurofibromatosis (plexiform type) is often bilateral, and is associated with a positive family history; clinical cutaneous lesions (café-au-lait spots, skin nodules) often exist, which when seen on MRI T2-weighted sequence show hyperintense neurofibromas close to nerves; all these were absent in our patient.

Haemangiomas are diagnosed clinically with a palpable bruit, which on MRI—T2-weighted sequence shows increased signal from serpiginous vascular channels within the haemangiomas. Lymphangiomas clinically presents with a diffuse swelling and pitting oedema. On MRI it appears hyperintense compared to muscle in T1-weighted sequences, and hyperintense compared to fat on T2-weighted sequences. Neither of these images showed osseous growth in the varied MRI findings, hence these differential diagnoses were ruled out.^{1 2}

In Proteus syndrome calvarial changes, pulmonary cysts, pigmented naevi and intra-abdominal lipoma often coexists. Fibrolipomatous hamartoma is a tumour where the nerve sheath, usually an isolated nerve, is infiltrated with fat causing enlargement of the nerve, which is significant on MRI, showing a speckled appearance, which was absent in our patient.

Klippel-Trenaunay-Weber syndrome clinically presents with co-existing limb hypertrophy, cutaneous haemangiomas, varicose veins and arteriovenous fistulas. On MRI T2-weighted sequence low signal areas are seen which represent haemosiderin deposit or areas of calcification.³ These were not noted in our patient.

TREATMENT

The patient underwent surgery; stage 1—reduction plasty of the left forearm was performed. Histopathological examination of the specimen confirmed the diagnosis of MDL.

OUTCOME AND FOLLOW-UP

The postsurgical phase was normal, the patient's stress level reduced to a certain extent and he was psychologically relieved. He was discharged and advised follow-up imaging after 1 year or if the swelling reappear. There was no further follow-up with the patient.

DISCUSSION

- In 1925, this condition was first described by Feriz, who used the term MDL to refer to localised gigantism of a lower

Figure 4 (A and B) MRI T1-weighted and T2-weighted axial sections of the left shoulder joint show prominent subcutaneous fatty tissue (black star) in the lateral aspect, without a definite capsule. The deltoid muscle (black arrow) is enlarged with fatty infiltration.

Figure 5 MRI T2-weighted coronal sections of the left shoulder joint show prominent fatty soft tissue on lateral aspect with fatty infiltration within the deltoid (A, black arrow), supraspinatus (B, black arrow) and infraspinatus (B, white arrow) muscles. Cortical thickening (A, white arrow) of the midshaft of humerus towards the lateral aspect is seen. The marrow of the bones does not show any abnormal signal intensity.

Figure 6 (A and B) MRI T1 weighted, T1-weighted fat-saturated coronal sections of the left wrist joint show enlarged index finger (black star) with degenerative changes involving the carpometacarpal (black arrow) and interphalangeal (white arrow) joints.

limb. Golding extended the term, in 1960, to include involvement of an upper limb.⁴

- MDL has been given various names such as macrodactyly, megalodactyly, digital gigantism, macromelia, partial acromegaly, macrosomy, limited gigantism, fibrolipoma of nerve,

fibrolipomatous hamartoma of nerve and lipomatosis of nerve, often described as having a 'coaxial cable-like' appearance on axial scans.^{5 6}

- MDL is not a hereditary disorder. The aetiology remains obscure; proposed mechanisms include lipomatous

degeneration, disturbed fetal circulation, an error in segmentation, the trophic influence of a tumified nerve and in utero disturbance of a growth-limiting factor.⁷

Patient's perspective

The patient is a 19-year-old adolescent male who developed painless enlargement of his left upper limb since childhood. The disproportionate increase of the upper limb began to cause him worry. Local remedies failed to cure his ailment and the worry gradually turned to depression. The patient admitted that he did not see the point of taking care of himself anymore and at times felt suicidal. He found it more convenient to eat at home and felt uncomfortable to dine at restaurants. He frequently missed the numerous medications that he was prescribed. A sense of disability engulfed him and his future appeared bleak.

Learning points

- ▶ When a patient presents with a localised gigantism of an extremity since childhood, one of the differentials should be macrodystrophia lipomatosa (MDL).
- ▶ Features such as asymmetrical soft tissue prominence, smooth enlargement of the cortex with irregular margins and degenerative changes on plain X-rays should be taken note of.
- ▶ MRI showing characteristic features of MDL, such as an asymmetrical increase in fatty elements without a definite capsule involving the subcutaneous plane and muscles, with prominent subcutaneous veins and normal bone marrow will clinch the diagnosis.
- ▶ Complications such as hamartomas and underlying bone involvement may also be assessed.
- ▶ Imaging provides vital clues to an accurate diagnosis, which is confirmed by histopathology. Surgery can be offered to patients after puberty for cosmetic as well as functional reasons, mainly to reduce the rate of recurrent fatty tissue proliferation.
- ▶ A localised recurrence rate of 33–60% makes the management of MDL demanding.¹¹

- ▶ Two subtypes of MDL exist. One is static, where the growth of the enlarged digit is at the same rate as the other digits, the other is progressive, where the growth of the enlarged digit is more rapid than the rest of the extremity; the latter is less common. Our patient was of the progressive type.
- ▶ The abnormality corresponds to the zone of innervation by sclerotome. It is mostly unilateral, bilateral involvement is rare.⁸ Involvement of a lower limb is more frequent than an upper limb, and the most common sites are the second and third digits.⁹
- ▶ Demonstration of a hypertrophic nerve is described, but may not always be identified within the subcutaneous tissue due to fatty infiltration of the nerve. Electroneurography and nerve conduction velocity tests performed in these patients may reveal slowed distal motor and sensory conduction, local (segmental) conduction block or slowing of the peripheral nerves at the entrapment sites.¹⁰

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