

## Case Report

# MR imaging of macrodystrophia lipomatosa

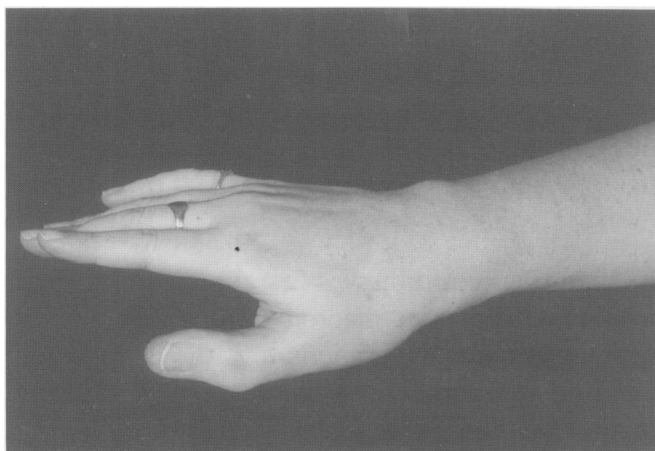
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Macrodystrophia lipomatosa is a rare nonhereditary congenital form of localised gigantism usually involving the 2nd or 3rd digit of the hand or foot. Pathologically an increase in adipose tissue involving subcutaneous tissue periosteum and bone marrow is present. Typical clinical and radiological appearances are described in this case report.

**Case Report** A 36-year-old female presented with enlargement of her right thumb and first metacarpo-phalangeal (MCP) joint since birth. Over the last four years the joints of the thumb had become more painful and had reduced movement. This was impacting on her work as holding a pen and writing was now difficult.

On examination bony enlargement and tenderness were present around the interphalangeal (IP) and MCP joints of the thumb, second MCP joint and the distal radius. Deviation at the IP joint was also present (*Fig 1*).



*Fig 1.* Enlargement of the right thumb and adjacent soft tissues with ulnar deviation of IP joint is demonstrated.



*Fig 2.* X-ray of both hands showing soft tissue swelling in the thenar eminence of the right thumb, compared to the left. Juxta articular osteophytes and degenerative joint space narrowing of first CMC, MCP and IP joints of the right thumb are demonstrated.

No cutaneous skin lesions, oedema or bruits were present. X-Ray of both hands (*Fig. 2*) and Magnetic Resonance Imaging (MRI) of the right thumb were performed (*Fig 3-5*).

A diagnosis of macrodystrophia lipomatosa was made from the imaging findings.

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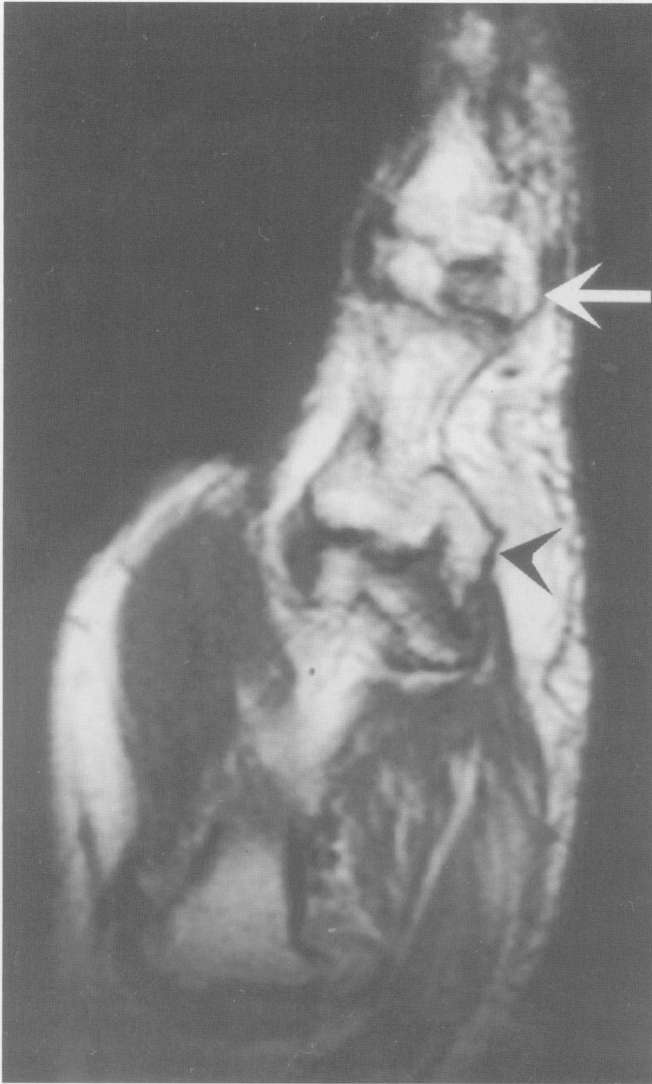
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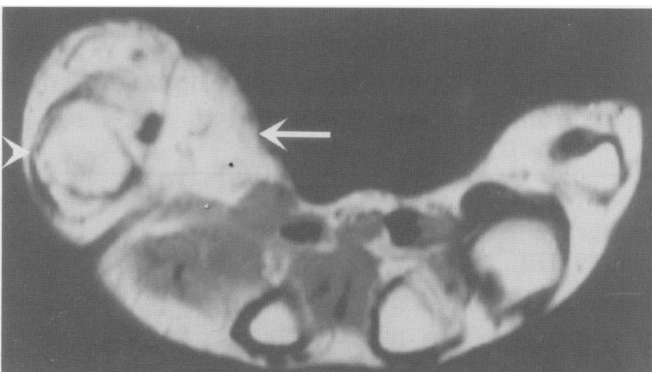
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*Fig 3.* Coronal T1 weighted MRI of right thumb demonstrating the high signal, excess fatty tissue and prominent hook shaped osteophytes at the IP joint (white arrow) and first MCP joint (black arrowhead). Note the low signal fibrous stands within the fatty tissue.



*Fig 4.* Axial T1 weighted MRI of right thumb again showing the surrounding excess high signal fatty tissue (white arrow). The first metacarpal is labelled (white arrowhead).



*Fig 5.* Coronal STIR MRI of right thumb demonstrating suppression of signal of the tissue surrounding the right thumb (white arrow) confirming that this excess tissue is fatty in nature.

## DISCUSSION

Macrodystrophia lipomatosa is a non hereditary, congenital and progressive overgrowth of all of the mesenchymal elements of a digit.<sup>3</sup> There is disproportionate increase in the amount of fibroadipose tissue.<sup>1</sup> Usually the lateral aspect of the upper limb and the medial aspect of the lower limb are affected.

In 1925 this condition was first described by Feriz, who used the term *macrodystrophia lipomatosa* to refer to localised gigantism of the lower limb.<sup>4</sup> Golding extended the term, in 1960, to include involvement of the upper limb.<sup>4</sup>

Clinically, findings are present at birth. An equal incidence is present in males and females.<sup>2</sup> Involvement of an extremity is always unilateral

and adjacent digits of the extremity can be involved. The lower extremity is more often involved than the upper extremity. The 2nd and 3rd digits are the usual sites (in the distribution of the median and plantar nerves).<sup>2</sup>

Involvement usually causes cosmetic disfigurement and mechanical problems are encountered in adolescence due to secondary degenerative joint disease causing reduced function, as in this case. Osteophyte overgrowth may also cause compression of adjacent nerves and vessels. Syndactyly, polydactyly and clinodactyly can occur.<sup>3</sup>

The affected digit increases in length and girth until puberty, when growth ceases.

Aetiology is unknown and several theories exist, including lipomatosis degeneration, disturbed foetal circulation and disturbance of growth factor in utero. Pathologically there is an increase in a fine mesh of fibrous tissue that involves the bone marrow, periosteum, muscles, nerve sheaths and subcutaneous tissues.<sup>4</sup> Phalanges are enlarged due both to endosteal and periosteal deposition of bone.

Two subtypes of congenital macrodystrophia lipomatosa exist.<sup>1</sup> These are static and progressive cases. In static cases the growth of the enlarged digit(s) is at the same rate as the other digits. In progressive cases the growth of the enlarged digit(s) is more rapid than the rest of the extremity. The progressive form is the less common. Involvement of the metacarpal and metatarsal bones is more likely in the progressive group; however in both groups the changes are most pronounced at the distal end of the digit (s).

#### RADIOGRAPHIC FINDINGS

On conventional X-Ray images macrodactyly and soft tissue overgrowth are visible and are most marked along the volar aspect of the digit and at its distal end. This overgrowth can produce dorsal deviation of affected parts,<sup>3</sup> whilst in this case ulnar deviation is present (*Fig 2*). Soft tissue radiolucency, representing overgrowth of fatty tissue, is occasionally seen.<sup>2</sup> The phalanges are elongated, broad, and the distal ends are splayed and can have a "mushroom" shape.<sup>4</sup> Slanting of the articular surfaces can occur and this leads to secondary degenerative joint disease,

manifesting in subchondral cyst and osteophyte formation.<sup>5</sup> The aetiology of the development of the secondary degenerative change, which occurs in adults, is unclear. We postulate however that it may be due to abnormal stresses across the joints due to the deformity.

MRI demonstrates an excess of fibro-fatty tissue around the affected digits.<sup>1, 8</sup> This will therefore have the same signal characteristics as fat on MRI; i.e. high signal on T1 and T2 weighted sequences and low signal on fat suppression sequences (STIR). Fibrous strands within the fatty tissue will be demonstrated as low signal linear strands on T1 weighted sequences. The fatty tissue may also be seen to infiltrate the adjacent muscles. Any bony abnormalities such as cortical thickening and secondary degenerative changes are also identified with MRI. Fibrous thickening of a nerve may also be seen.<sup>6</sup>

In our case the digit enlargement was not as gross clinically as some textbook examples of this condition; however it had been enlarged since birth. This clinical history, together with the radiographic findings of excess soft tissue swelling and advanced degenerative changes, out of keeping in a patient of this age, suggested the diagnosis of macrodystrophia lipomatosa (*Figs 1, 2*). Therefore MRI was performed to identify if the excess soft tissue was fibro-fatty in nature. This was demonstrated as the signal from this tissue suppressed on the fat suppression (STIR) sequence (*Fig 5*). Occasionally a thickened nerve can be demonstrated in the region of the soft tissue overgrowth on MR imaging. This is not visualised in all patients with macrodystrophia lipomatosa, as in our case, probably because fatty infiltration into the nerve sheath can make its detection difficult within the subcutaneous tissue.<sup>6</sup>

In the clinical scenario of a patient with congenital digit enlargement, MRI is a useful imaging modality to aid diagnosis. The differential diagnosis of congenital macrodactyly includes neurofibromatosis, Klippel-Trenaunay-Weber syndrome, lymphangiomatosis, haemangiomatosis and fibrolipomatosis of the nerve.<sup>1</sup> MR imaging, by characterising the type of soft tissue proliferation can be used to differentiate between most of these diagnoses.

In neurofibromatosis (NF) T2 weighted MR images show high signal hyperintense neurofibromas, which

will be situated close to the nerve.<sup>8</sup> In addition cutaneous manifestations of NF and a family history will be present.

In Klippel-Trenaunay-Weber syndrome limb hypertrophy, haemangiomas and arteriovenous fistulae are present. Plain x-ray can show soft tissue and bone enlargement together with phieboliths within the vascular anomalies. MRI is a non invasive method of identifying the presence and the extent of the vascular anomalies. These tend to be of high signal on T2 weighted images, although areas of low signal can be seen and represent haemosiderin deposition or areas of calcification.<sup>2</sup>

In haemangiomas T2 weighted MR imaging shows increased signal from the serpiginous vascular channels within the haemangiomas.<sup>8</sup> Bruits may be detected on clinical examination.

In lymphangiomas, the lymphangiomas are hyperintense to muscle on T1 weighted images and hyperintense to fat on T2 weighted images.<sup>1</sup> Clinically, diffuse limb swelling and pitting oedema are found.

Fibrolipomatosis of the median nerve can be seen with macrodactyly. In this condition MRI will identify fat deposits within the nerve sheath causing the marked enlargement of the nerve.<sup>7</sup> In comparison in cases of macrodystrophia lipomatosa the fat deposits can be within the nerve sheath, subcutaneous tissues, bone marrow, periosteum and muscles.<sup>7</sup>

Our patient is currently awaiting an athrodesis of the interphalangeal joint to relieve the pain caused by the secondary degenerative joint disease.

## CONCLUSION

MRI is an extremely useful imaging modality in a patient who presents with congenital digit enlargement. An excess of fibro-fatty tissue, together with proportional enlargement of other mesenchymal tissues is characteristic of macrodystrophia lipomatosa. Demonstration of a hypertrophic nerve is described, but may not always be identified within the subcutaneous tissue due to fatty infiltration of the nerve.

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