Imaging of a rare disorder: macrodystrophia lipomatosa

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Introduction

Macrodystrophia lipomatosa (MDL) is described as a rare, non-hereditary, congenital condition presenting with localised macrodactyly and a proliferation of mesenchymal elements. There is in particular a marked increase in fibroadipose tissue.[1]

We describe 2 young patients presenting to our department in a 6-month period, with a history of disproportionately large limbs since birth. While our first case demonstrated all the typical features of MDL, our second case failed to demonstrate osseous gigantism, but all other features of MDL were present. An extensive search of the literature failed to yield any cases described without osseus gigantism; but at the same time, the other radiological features failed to fit in with any other causes of focal gigantism, and the most appropriate diagnosis appears to be MDL. We therefore concluded that this may be a case of a rare, atypical MDL that was arrested or frustrated and so failed to demonstrate full expression of the syndrome.

Case 1

A 33-month-old boy was referred by a paediatric surgeon for radiographs of the left arm. According to the history, he was born with the left arm disproportionately larger than the rest of the body. Clinically, there was marked increase in the soft-tissue bulk of the entire left arm with focal gigantism involving the thumb and second finger. Good functionality of the arm was maintained.

Radiographs revealed lucent soft-tissue thickening of the affected arm, suggesting the presence of fat (Fig. 1). Focal gigantism involving the metacarpals and phalanges of the thumb and second finger were noted. The rest of the bony elements were normal.

Ultrasound (US) examination demonstrated an extensive increase in subcutaneous tissue with poor visualisation of muscles and nerves. Normal vascularity was noted on Doppler US. US of the abdomen was normal.

MRI studies showed diffuse proliferation of fatty tissue throughout the arm, with fat infiltration of the muscles (Figs 2 - 4). The nerves

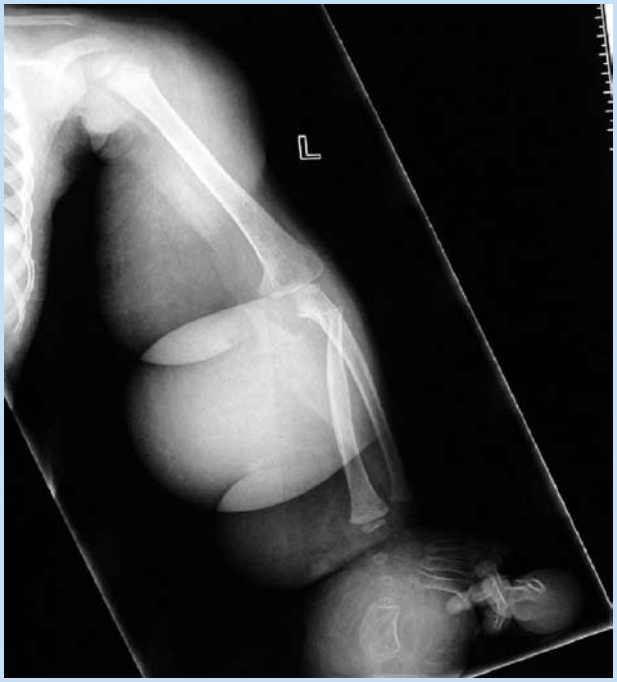

Fig. 1. Radiograph of the left upper extremity demonstrates diffuse soft-tissue swelling involving the entire upper limb. Soft tissues are lucent, suggestive of fat. Multiple skin folds are evident. Marked osseous gigantism of the 1st and 2nd fingers noted. Humerus, radius ulnar and bony elements of the 3rd, 4th and 5th fingers are within normal limits.

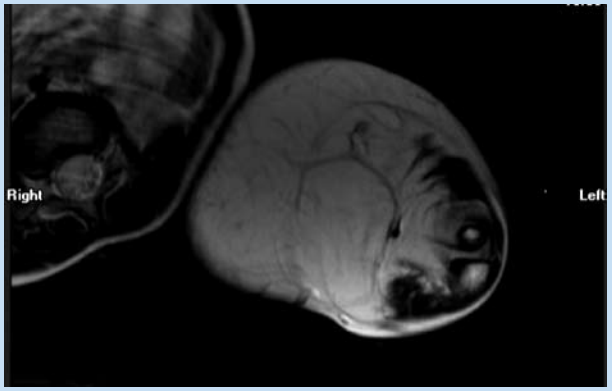

Fig. 2. Axial T1WI through the forearm. There is proliferation of subcutaneous fat that is not encapsulated. The muscles are markedly infiltrated by fat, and the nerve cannot be identified.

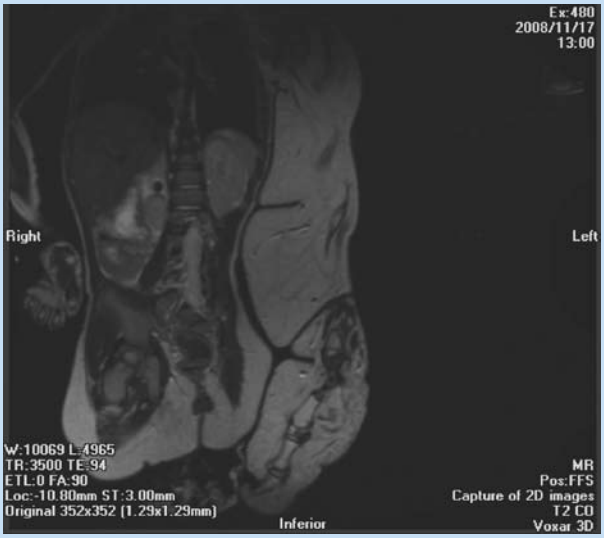

Fig. 3. Coronal proton density image of the left upper limb showing osseous gigantism of the 1st finger and diffuse increase of subcutaneous fat with very little muscle bulk.

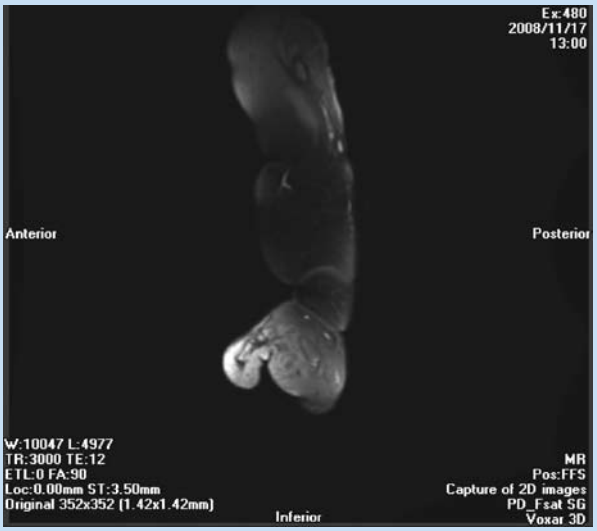

Fig. 4. Fat-suppressed images of the left upper limb.There is marked suppression of the signal from the subcutaneous fat, confirming the diagnosis of MDL.

could not be identified on MRI, probably owing to infiltration by fat. Histological analysis of the tissue revealed adipose tissue only, which was in keeping with a diagnosis of MDL.

Case 2

An 11-month-old girl was referred by a plastic surgeon for MRI to exclude lymphoedema of the right lower limb. According to the history, she was born with the right lower limb disproportionately bigger than the rest of the body. Clinically, there was marked increase in soft tissue of the entire right lower limb. Good function of the limb was maintained, and the child was otherwise normal.

Radiographs revealed increased soft tissue throughout the limb, and the underlying bones were normal (Fig. 5). US was not performed.

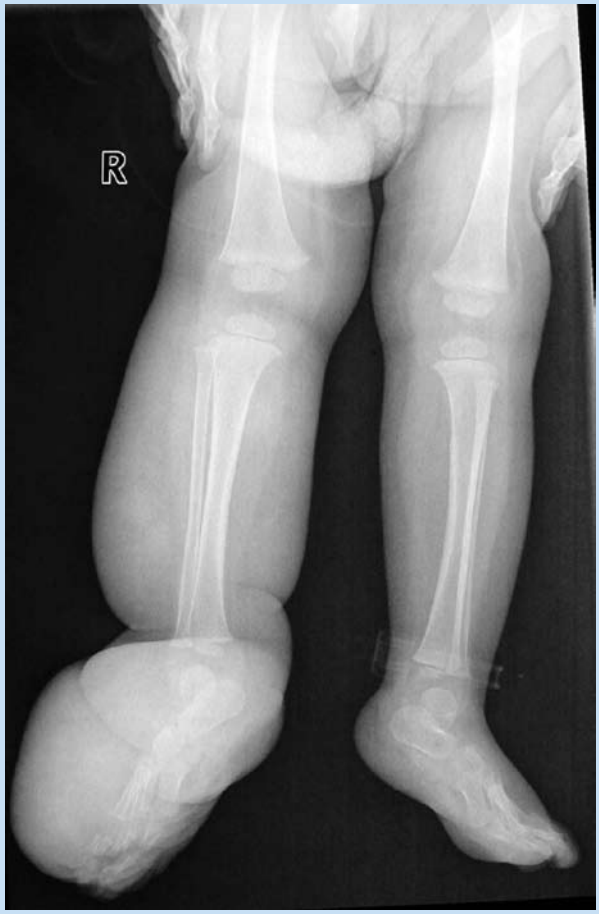

Fig. 5. Frontal radiograph of the lower limbs. There is diffuse soft-tissue swelling involving the subcutaneous fat of the right lower extremity more marked in the distal lower leg and foot. Soft-tissue skin folds are evident. There is no discrepancy in the bony elements in comparison with the left side.

MRI showed diffuse increase of fat in the soft tissues, as well as scattered cystic lesions of varying signal intensity (Figs 6 - 8). This feature has not been described in the literature reports of other cases of MDL, and was thought to be due to either fat necrosis or old haematomas secondary to trauma. There was no fat infiltration of the muscles, and the neurovascular bundles were intact.

Once again, histological analysis revealed proliferation of adipose tissue in keeping with a diagnosis of MDL.

Literature review

MDL is a rare congenital disorder of focal gigantism affecting the extremities – more commonly, the second and third digits of the foot, but it may also affect the hand.[2,3] It is characterised by the proliferation of all mesenchymal elements but especially of fibroadipose tissue.[2,4,5] It is typically described in a particular nerve distribution; usually the medial plantar nerve or median nerve.[3]

Radiological investigations include plain film radiographs, US and MRI, where findings may be typical. Plain films show lucent soft-tissue overgrowth as well as hypertrophy of osseous structures in the distribution of the median and plantar nerves.[1] In our cases, only one patient demonstrated the osseous overgrowth. In the second case, the bony ele-

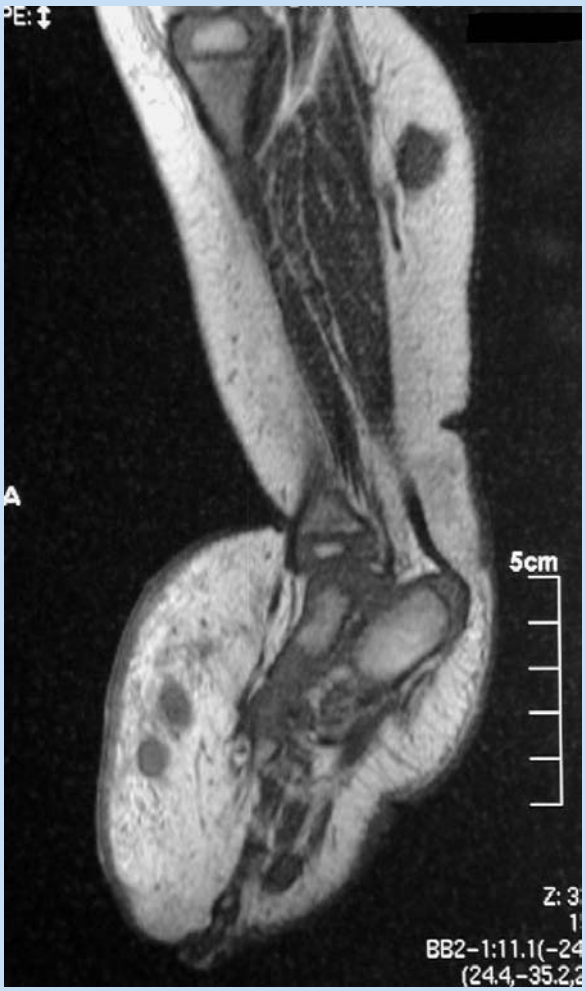

Fig. 6. T1WI of the right lower limb shows marked thickening of subcutaneous fat with associated well-circumscribed lesions of low signal intensity.

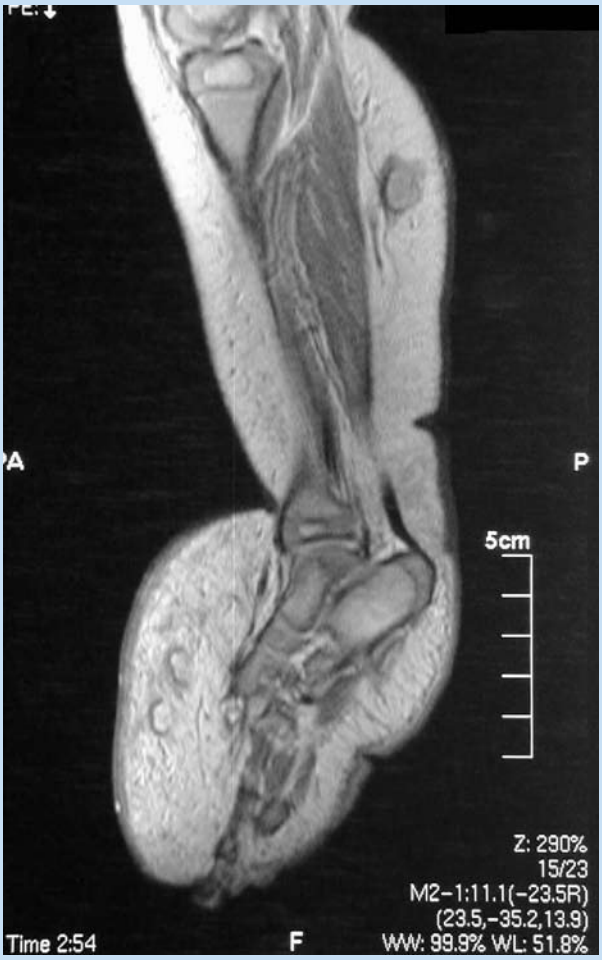

Fig. 7. T2WI of the right lower limb confirms fat signal in the thickened soft tissue. The well-circumscribed lesions demonstrate varying signal intensities.

ments were within normal limits. In older patients, secondary osteoarthritis changes may also be seen.

US reveals large amounts of subcutaneous tissue, infiltration of the muscle and thickening of the affected nerves. Doppler studies reveal an absence of any increased vascularity.

MRI findings are most useful in confining the differential diagnosis.[4] MRI investigations reveal overgrowth of unencapsulated fatty tissue that demonstrates high signal on T1WI and T2WI, with suppression of the signal on short inversion time inversion recovery (STIR) sequences, as demonstrated by our patients. Thickened nerves may, or may not, be demonstrated.

Lack of flow voids and calcifications helps to differentiate the condition from vascular malformations of Klippel-Trenauney-Weber syndrome. Lymphangiomas and neurofibromas may be hyperintense to fat on T2WI,[4] distinguishing MDL from these conditions. Proteus syndrome may be similar but has other associated features such as skull anomalies, lung cysts and pigmented naevi.[1] Some authors suggest that MDL is a localised form of Proteus syndrome.1 Fibrolipomatous harmatoma of the nerve displays features similar to MDL;[1] however, fat deposition is within the nerve, giving a speckled appearance on MRI.

Two subtypes of the condition are described in the literature: the static and progressive types. In the static type, the enlarged digits grow at the same rate as the other digits. In the more rare progressive type, the growth of the enlarged digits is more rapid. Our patients were thought to have the static type clinically.

Conclusion

Numerous aetiologies of focal gigantism exist. While clinically the distinction may be difficult, radiological investigation is very useful in confining the differential diagnosis. MRI in particular shows characteristic findings of MDL, and radiologists therefore play an important role in the diagnosis of these conditions.

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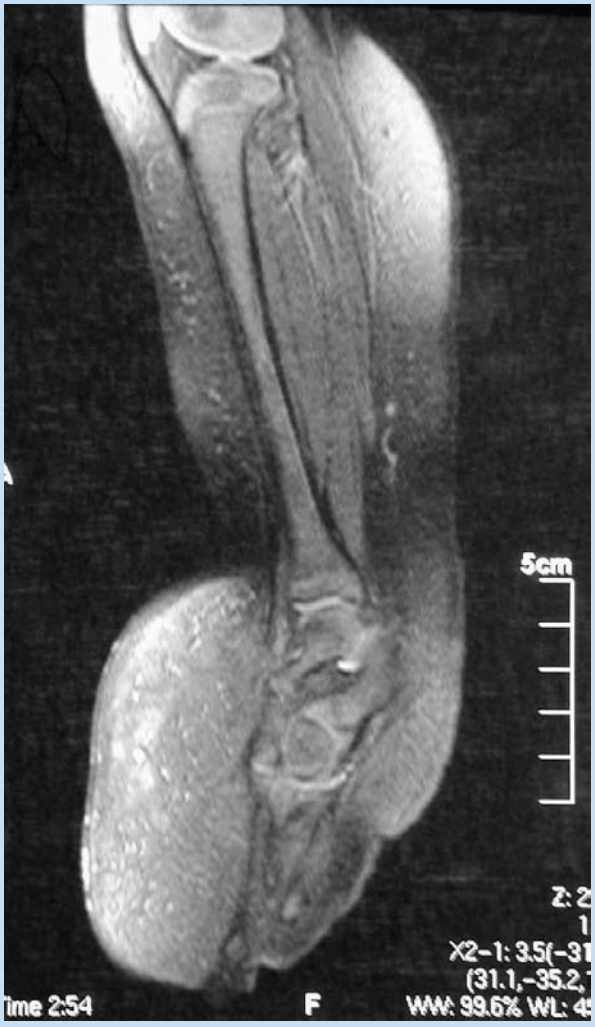

Fig. 8. Fat-suppressed image of the right lower limb. There is loss of signal in the thickened soft tissue. The muscles and osseous elements appear normal in all sequences.

Straight talking on CT scanning

Bradley M Tipler, *MD*

Dr Tipler is a private-practice radiologist in Waynesboro, VA, USA. We are indebted to him and to the website diagnosticimaging.com for this viewpoint.

For the past 10 years I have been lecturing on America's radiation phobia. Obviously, I am not doing a good job, because it is growing. What I find particularly distressing is the problem's growth among radiologists. Recently, phobic radiologists have been publishing and lecturing like rabbits on Viagra.

I have always endorsed and applied ALARA. I heartily endorse the Image Lightly campaign. If there is any risk from medical radiation, it is to children. But I raise cattle on my farm, and I occasionally step in the stuff being printed and propagated by a lot of radiologists now.

While reports on biological effects of ionising radiation (BEIR) and the alphabet soup that formulates radiation standards for the USA are monumental works of statistics, they are not facts. Historically, they were developed to help us to formulate national and international policy on radiation safety and work out radiation regulations. Still today, they are based in large part on data gathered from survivors of Hiroshima and Nagasaki.

The American public's exposure to radiation from medical imaging has nothing in common with that of Japanese atom bomb survivors. The figures are great for developing radiation regulation guidelines, but they are not appropriate for determining the risk/benefit ratio for an acutely ill patient in an emergency room. What US radiologist in his or her right mind uses Japanese statistics for gastric cancer when reading an upper GI?

Every one of the data now being headlined by the lay press is based on the linear no-threshold (LNT) theory. Is there a radiologist who actually believes that the effects of low-dose radiation over a lifetime are the same as those of one massive dose? The LNT made sense in the early development of radiation safety guidelines. Rule-makers need to err on the side of safety.

We all know CT is being overused. And we all know there is one primary reason: money. Clinicians with their own scanners want to produce income for themselves, and good docs who don't own a machine want to avoid generating income for plaintiffs' lawyers.

Radiation regulations are to clinical decision-making what highway construction regulations are to NASCAR. You don't see speed limit signs on NASCAR tracks because that would be dumb. It is just as dumb to put theoretical numbers on the risk of a CT scan for an individual patient. If the patient needs the test, the theoretical radiation risk is immaterial. If the patient doesn't need the test – duh, don't do the test. If you are not sure, the American Trial Lawyers Association would love to review your decision later.

As I say in my lectures, this is the classic American approach to risks. We love scary movies, but we want mother hen to eliminate all real risk from our lives. We can save thousands of lives by lowering the speed limits, but we want to drive fast. When we wreck, we hire a lawyer and sue anyone who didn't make our fast driving safer.

We need to emphasize .DAM (dot DAM = don't order the test if it doesn't alter management) and make sure we use the lowest possible dose for a given exam. Recent literature has shown we are lousy at the latter. We do not need to complicate the ordering process, especially with biased statistics that were never intended to be applied on an individual basis.

I think putting a note on a CT order form about the theoretical risk of cancer from a single CT means the radiologist is clueless about the conflicts facing the ordering doctor. The radiologist looks like, and probably is, a fool.

Americans like to compare CT doses to our background level of 3 mSv per year. Background radiation on the coast of Brazil exceeds 150 mSv a year. Have you seen those poor, over-irradiated Brazilian beach babes? And they live longer than us.