CASE REPORT

Lipofibromatosis – a case report

L Naidoo, *MB ChB, FCRad (D)*
Helen Joseph and Coronation Hospitals, Johannesburg

Introduction

Lipofibromatosis is a rare paediatric tumour described by Fetsch *et al.*[1] in 2000. They documented 45 cases of this soft-tissue entity. Since then there have been a few reports of this tumour and its imaging characteristics.[2] Lipofibromatosis is a poorly demarcated mass involving the subcutis or deep soft tissues.[1] The histology is that of abundant mature adipose tissue with a spindled fibroblastic element involving the septa of fat tissue.[1] It forms a slowly growing painless mass arising in children and in some cases like the one described below – congenitally.[1] Fetsch *et al.*[1] documented ages ranging from 11 days to 12 years (median age 1 year) at the time of initial biopsy and resection as well as a 2:1 male predominance. This case report serves to describe the clinicoradiological features of this condition as well as the radiological differential diagnosis.

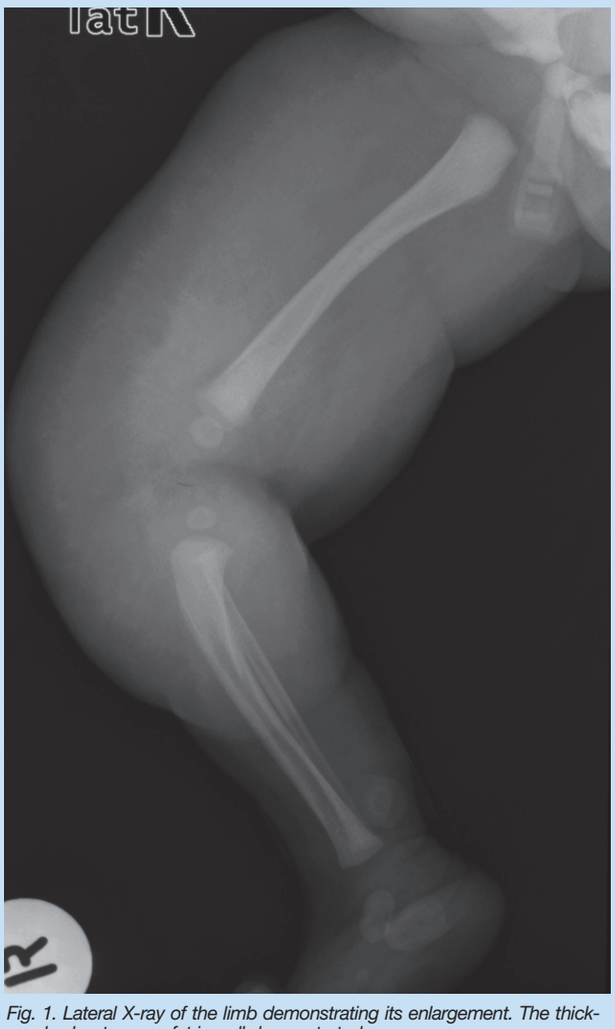

Fig. 1. Lateral X-ray of the limb demonstrating its enlargement. The thickened subcutaneous fat is well demonstrated.

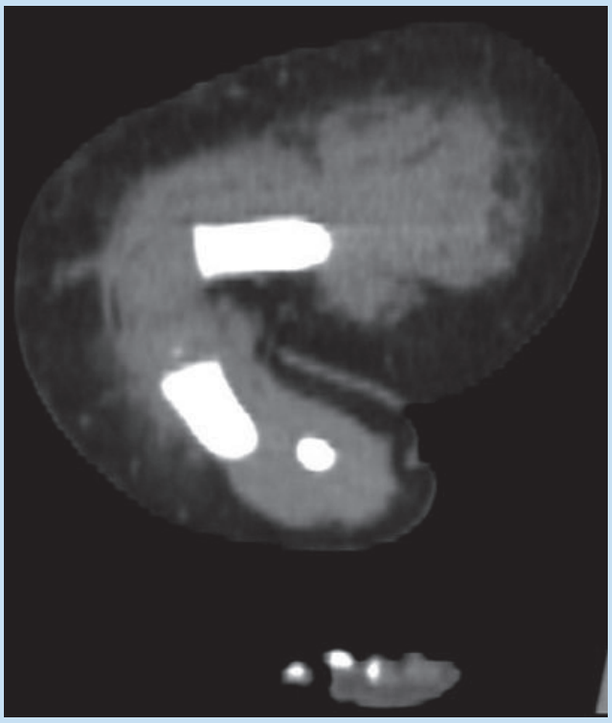

Fig. 2. Sagittal axial CT showing the fatty interdigitation into poorly marginated bulky musculature.

Case report

A male infant presented at birth with diffuse swelling of his right thigh and upper leg. Antenatal and family history proved unremarkable. The patient was born via normal vaginal delivery with good birth Apgar scores. Examination revealed enlargement of the right upper leg extending from the thigh to the mid lower leg spanning a distance of 24 cm. There was overlying skin hypertrichosis. The leg was firm with no pulsatility or bruit. There was no obvious tenderness and no other lesions could be detected.

On plain radiography (Fig. 1) there was marked increase of limb

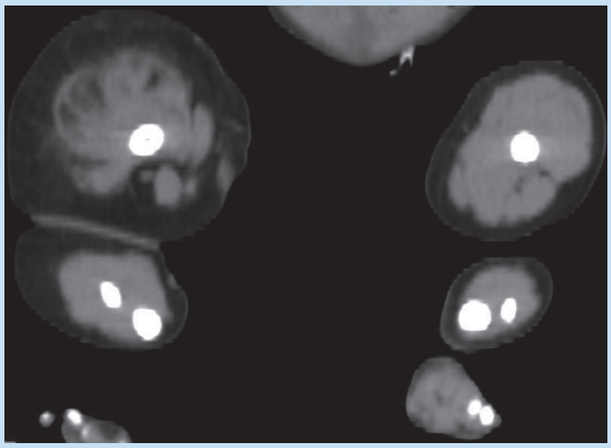

Fig. 3. Coronal CT with fatty infiltration of musculature.

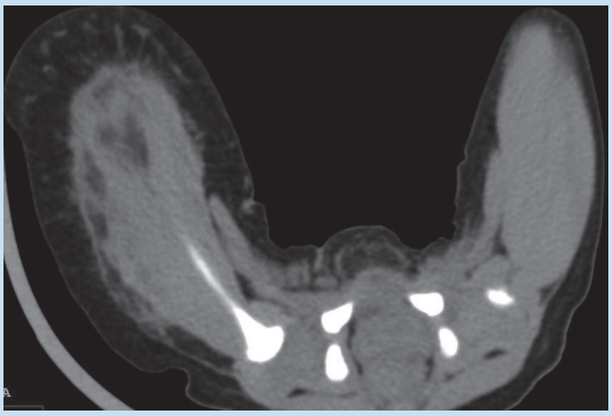

Fig. 4. Axial CT demonstrating fibrous stranding within the thickened subcutaneous fatty tissue.

diameter with diffuse thickening of the fatty subcutaneous tissue. The underlying muscular compartment was bulky with radiolucencies – that appeared isodense to adjacent subcutaneous fat – suggestive of a fatty infiltration. No calcifications could be detected. Bony outlines appeared intact.

Computed tomography (CT) scans (Figs 2 - 4) demonstrated a soft-tissue mass with diffuse thickening of the subcutaneous adipose tissue and fibrous stranding. There was interdigitation of the fatty tissue into the musculature. Fatty tumour components were interspersed in part through musculature that appeared infiltrated and poorly defined. There was no vascular component, nor nerve infiltration. No bony overgrowth was demonstrated.

Although infiltrating, the adipose tissue overgrowth was clinically slow growing and the radiological differential was for that of superficial adipocytic peripheral benign tumours of infancy.

Wedge biopsies from the upper thigh and knee were submitted for histology. The findings were those of a tumour composed of alternating streaks of mature adipose tissue as well as a fibrous spindle-cell component consistent with lipofibromatosis.

Radiological differential diagnosis

The differential diagnoses for lipofibromatosis that were considered included lipofibromatosis hamartoma of the nerves, Proteus syndrome and lipoblastoma(tosis).

Lipofibromatosis hamartoma of the nerves is a slow growing mass of fibrofatty tissue surrounding and infiltrating major nerves.[3] When this condition is associated with macrodactyly and bone overgrowth it is described by the term 'macrodystrophia lipomatosa'.[3] Owing to absence of nerve infiltration in the described case, this possibilty was considered unlikely.

Proteus syndrome is a disorder characterised by progressive overgrowth of multiple tissues with a propensity to develop particular neoplasms.[4] The clinical criteria include the presence of a connective tissue nevus, ovarian cystadenomas or parotid adenomas as well as disproportionate overgrowth of specific tissue, vascular malformations and specific facial phenotypes.[4] This differential was a consideration on the basis of the adipose tissue overgrowth as well as the prescence of hypertrichosis.[3] Although other criteria could not be met, one has to consider this as the abnormalities described above tend to increase with age.[4]

Lipoblastomas are benign mesenchymal tumours of embryonal white fat.[5] Histologically the well-circumscribed and encapsulated features of lipoblastoma differentiate it from the multicentric and infiltrative nature of lipoblastomatosis.[5] This tumour has a wide anatomical distribution but chiefly affects the extremities.[5] Imaging differentiation from lipofibromatosis is often impossible because of the predominance of fatty tissue component in both tumours. Biopsy forms the mainstay of diagnosis and displays mainly immature fat cells (unlike lipofibromatosis).[5]

Discussion

Lipofibromatosis has previously been interpreted as a form of infantile fibromatosis.[1] In 2000, Fetsch *et al.*[1] proposed that although the tumour is likely to be a part of the infantile fibromatosis spectra, it should be considered a distinctive entity because of its histology. Fibromatosis exhibits a more sheet-like growth and does not contain fat as an integral component.[1,2] Although biopsy offers a definitive diagnosis, imaging modalities are useful adjuncts for diagnosis as well as management. Imaging is especially useful in determining the degree and depth of anatomical involvement which is helpful for evaluation prior to complete removal of the tumour, reconstruction or excision of recurrent diffuse lesions.

Imaging generally reveals fat that appears exaggerated, adipose tissue that is more disorganised than normal with poorly demarcated lobules, infiltration and entrapment as well as displacement of muscle with fibroblastic elements within the fat septa.[5]

Ultrasound usually demonstrates poor musculature planes with hyperechoic content.

CT is useful in outlining the tumour and demonstrating a low-density non-enhancing mass measuring fat in Hounsfield units. Magnetic resonance imaging (MRI), though not available in the case described above, plays an important role in tissue characterisation with increased T1 and T2 signals that are consistent with fat. Intralesional areas of signal change that are increased on T1 and become fat saturated on T2 are also reflective of fatty content.[2,5] MRI can also be useful in differentiating between lipofibromatosis and lipoblastoma(tosis) on the basis of the immature cell content of the latter. This finding sometimes results in the isointense T1 signal of lipoblastoma(tosis).[5]

Lipofibromatosis is best managed by complete removal because of its predeliction for recurrence in incompletely excised lesions.[2] However there are some cases where long-term follow-up has shown no progress.[2] Metastases have not been reported.

Conclusion

Although biopsies form the mainstay of diagnosis in peripheral fatty tumours of infancy, CT and MRI form valuable adjuncts in differentiating fatty tumours from other soft-tissue tumours. These imaging modalities also serve to delineate lesions for surgical excision and reconstruction procedures.

1. Fetsch JF, Miettinen M, Laskin WB, Michal M, Enzinger FM. A clinicopathologic study of 45 pediatric soft tissue tumours with an admixture of adipose tissue and fibroblastic elements, and a proposal for classification as lipofibromatosis. *Am J Surg Pathol* 2000; 24: 1491-1500.
2. Sasaki D, Hatori M, Hosaka M, Watanabe M, Kokubun S. Lipofibromatosis arising in a pediatric forearm – a case report. *Upsala J Med Sci* 2005; 11: 81-106.
3. Kameh D, Perez-Berenguer J, Pearl G. Lipofibromatous hamartoma and related peripheral nerve lesions. *South Med J* 2000; 93: 800-802.
4. Barclay L, Vogin G, Vega C. Cutaneous manifestations of Proteus syndrome reflect general severity. *Arch Dermatol* 2004; 140: 947-953,1001-1002.
5. Chien A, Song D, Stein S. Two young girls with lipoblastoma and a review of the literature *Pediatr Dermatol* 2006; 23: 152-156.