



Lipoblastoma – an unfamiliar but important diagnosis A case series and literature review

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Lipoblastoma and its infiltrative variant lipoblastomatosis are rare adipose tissue tumours seen in infants and children. Many surgeons are unfamiliar with these uncommon lesions and hence they are suboptimally treated. We report a case series of six patients in our tertiary paediatric hospital. Cases were reviewed retrospectively with reference to demographics, investigations, diagnosis and their management. Lipoblastomas are easily misdiagnosed and excision before proper investigations may result in incomplete resection, recurrence and further potentially mutilating surgery.

Keywords : lipoblastoma ; lipoblastomatosis.

INTRODUCTION

Lipoblastoma and its infiltrative form lipoblastomatosis are rare adipose tissue tumours seen in infants and children (5). They are benign tumours of embryonic mesenchymal cells, composed of lipoblasts that continue to proliferate during the postnatal period. Many surgeons are unaware of this lesion, owing to its rarity. Due to clinical similarity it is not uncommon for these lesions to be wrongly diagnosed as simple lipomas, leading to excision without further investigation. It is important to recognise the diagnosis preoperatively because incomplete excision can result in local recurrence, which makes further treatment very difficult. Orthopaedic surgeons should be aware of this lipomatous tumour because they may be encoun-

tered in orthopaedic practice. We present this case series to emphasize the importance of proper diagnosis and management of this uncommon paediatric tumour.

MATERIALS AND METHODS

We retrospectively reviewed six cases of lipoblastoma / lipoblastomatosis which presented in our tertiary referral paediatric hospital between 2002 and 2006. Case notes were used to gather data regarding age, gender, ethnicity, side, location, presentation and clinical features. Preoperative / postoperative diagnosis, radiology results and pathology reports were all reviewed. Minimum and maximum follow-up was two and four years respectively. The data is summarised in table I.

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Table I. — Demographics of the six patients

Case	Sex	Age	Site	Side	Follow-up	Complications	Recurrence
1	M	4 months	Forearm	Left	2 years	Nil	Nil
2	F	5 years	Ankle	Right	2 years	Nil	Nil
3	M	3 years	Thigh	Left	3 years	Nil	Nil
4	M	6 months	Foot	Right	4 years	Nil	Nil
5	M	18 months	Back	Left	4 years	Nil	Yes
6	M	3 years	Back	Right	3 years	Nil	Nil

Case 1

A four-month-old boy was referred from a general paediatric clinic for excision of a lipoma from his left forearm. The swelling was soft and non tender. It moved under the skin with slippery margins and measured approximately 4×2 cm. There were no neurocutaneous stigmata, pressure symptoms, skin puckering or any history of rapid increase in size. Ultrasonography showed a well defined oval lesion within the subcutaneous tissues approximately 3.9×1.8 cm, with the same echogenicity as the surrounding tissues, solid in nature with no colour flow. The final report read “*The findings are non specific but would be consistent with a lipoma*”. An excision biopsy of the swelling was performed as a day case and the specimen was sent for histopathology. The post-operative recovery was uneventful and the wound healed normally. The histopathology result came back as lipoblastoma. There was recurrence of the swelling two months later. A wide excision with rotational flap cover was undertaken. There was no further recurrence at three year follow-up.

Case 2

A five-year-old girl was referred to the orthopaedic department for a large soft tissue swelling over the lateral aspect of her right ankle. The swelling had been present since birth with a gradual increase in its size. Clinical examination was consistent with lipoma. Active and passive movements in the subtalar joint were painful and limited. There were no neurocutaneous stigmata, pressure symptoms or skin puckering. Radiographs showed a soft tissue swelling but no bony abnormality. Ultrasound revealed a subcutaneous lipoma with deep extension. An MRI scan raised the suspicion of lipoblastoma (fig 1). She was referred to a specialist centre and surgical excision was done which confirmed the diagnosis. There is no evidence of any recurrence at the latest follow-up done at 2 years.



Fig. 1. — MRI with arrow indicating a lipoblastoma at the lateral aspect of the right ankle (Ref : case 2).

Case 3

A three-year-old boy presented with a swelling on the lateral aspect of his left thigh. The parents had noticed the swelling for the past four months but denied any rapid increase in size. On examination there was a firm, partially tethered subcutaneous lump. There were no associated sinister features or lymphadenopathy. An ultrasound scan suggested that the lesion was superficial to the quadriceps muscle, although there was a degree of tethering

clinically. The provisional diagnosis was lipoma. An excision biopsy was done. Histopathological examination revealed the diagnosis of lipoblastoma. There was no evidence of recurrence at three years follow-up.

Case 4

A six-month-old baby was referred to the orthopaedic department for a soft tissue swelling on the medial side of his right foot. According to the parents, the swelling was there for at least two months. Examination showed a non tender fullness of his medial longitudinal arch area. Radiographs and ultrasonography did not reveal any obvious pathology. The swelling was excised and sent for histopathological examination. The report came back as consistent with a mature lipoblastoma. There was no evidence of recurrence after follow-up of four years.

Case 5

An eighteen-month-old baby was referred to the paediatric surgery department with a swelling over his back. Earlier an ultrasonography had reported this swelling as an encapsulated superficial lipoma. The swelling was excised as a day case. There was no evidence of any recurrence at two year follow-up.

Case 6

A three-year-old girl was seen in the paediatric surgery department for a swelling over her back. Examination showed a smooth, soft, well defined swelling to the right of her thoracic spine. Ultrasound was done and was reported as possible lipoma without any deeper extension or vascular component. The excision biopsy report came back as lipoblastoma. No recurrence has been noted to date.

DISCUSSION

Adipose tumours are relatively rare in children. They comprise about 6% of soft tissue tumours seen in the first two decades of life. Up to 30% of them are lipoblastomas (3). Lipoblastomas are characterised by their benign nature, early presentation (4) (90% < 3 years), male predominance (15) and rapid growth (9). They have no metastatic potential, but a single case of cervico-axillary lipoblastoma – recurrence in the right atrium

6 months after macroscopic complete excision has been reported by Abel *et al* (1).

Two clinicopathological types of lipoblastomas have been described (4). The commonest is the circumscribed and encapsulated type, which is generally located in the superficial soft tissue, simulating a lipoma. The second is the diffuse type – lipoblastomatosis which has an infiltrative pattern and extends into the deeper soft tissue (4).

Histologically lipoblastomas are neoplasms composed of adipocytes in different stages of maturation. Microscopy shows lobules separated with well defined septa. The stroma is often a myxoid and rich delicate plexiform vascular network. No atypical mitotic figures and no nuclear atypia are seen. An infiltrative pattern is suggestive of lipoblastomatosis rather than lipoblastoma. The most important differential diagnosis is myxoid liposarcoma from which it is difficult to distinguish. Both lipoblastoma and myxoid liposarcoma have similar histological and radiological features, but myxoid liposarcoma is exceedingly rare in a paediatric age group (12). The average age of presentation for lipoblastoma is 3 years whereas for myxoid liposarcoma it is 30 years (2). One way of differentiating them is by karyotyping. A break point in the long arm of chromosome 8 is the commonest abnormality seen in lipoblastoma (7) whereas karyotyping shows a characteristic clonal chromosomal anomaly t(12;16)(q13;p11) in myxoid liposarcoma (10,13).

Magnetic resonance imaging (MRI) is the investigation of choice (7,14). Plain radiographs, ultrasound, colour doppler and computed tomography (CT) all have a complementary role in diagnosis of lipoblastomas (1). Plain radiographs help by detecting bone overgrowth (3), which suggests the diagnosis of neurofibrolipoma or proteus syndrome. Plain radiographs also show soft tissue swelling. Ultrasonography (USG) usually shows a homogenous hyperechoic (3) mass. Cystic areas can also be seen. Fine lobulations may also be demonstrated by USG. Colour doppler helps to rule out angiomatous variants of a lipomatous tumour. Computed tomography allows conclusive identification of the fatty components (3), as well as intratumoral stranding. CT will show a low attenuation (equivalent to fat) and a relatively well-delineated tumour interspersed

by numerous thin septa. CT also shows other soft tissue density components. MRI helps in localising, staging and characterising these tumours. MRI also demonstrates anatomical details (1), which is essential for successful tumour excision. MRI gives important information regarding tumour components including cells, fibrous septa and capsular infiltration. The presence of poorly defined margins indicates infiltration into nearby structures including muscle and subcutaneous fat. MRI will also describe the lobulations and the fibrous framework. Fat suppression sequences (9) using short T₁ inversion recovery (STIR) help to identify the fat content and help to differentiate lipoblastomas.

In infants and young children, a heterogeneous mass with significant fatty components on radiological investigation should raise the possibility of lipoblastoma (6). Complete excision with negative margins should be attempted rather than biopsy (9) when ever possible. There is no role for radiation or chemotherapy in the treatment of these tumours. Complete resection is the only known definitive treatment. Recurrence is common after incomplete excision as demonstrated by Case 1, with an incidence as high as 14% to 25% in various studies (9) and hence a follow-up of at least 5 years is ideal.

Although maturation may eventually occur in some lipoblastomas (8). regardless of the site, surgical resection is the treatment of choice (9). However, a more conservative observational approach may be justified in infants with large invasive lesions requiring mutilating resection. In 2000 Mognato *et al* described the first and only case in the literature of spontaneous and complete resolution of a diffuse lipoblastoma (11).

In infancy and early childhood, the identification of a tumour composed mostly of fat should suggest the diagnosis of lipoblastoma rather than lipoma (12). In one of the largest series by Mc Vay *et al* (9) the pre-operative diagnosis was accurate only in 3 out of 16 cases.

In conclusion our own experience in a tertiary referral paediatric centre suggests that many of these lesions are excised before appropriate investigations have been made. We therefore believe that it is important to raise the profile of this potential diag-

nosis. Awareness and recognition of this rare tumour is important because lipoblastomas are easily misdiagnosed and excision before proper investigation may result in incomplete resection, recurrence and further potentially mutilating surgery.

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