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Myxoid Lipoblastoma

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A rapidly growing soft tissue mass in the axilla of an infant raises the suspicion of a lipoblastoma or a liposarcoma. Excisional/incisional biopsy is vital in confirming the diagnosis and hence avoiding aggressive extirpation. This case report highlights the role of histopathology and immunohistochemistry as the gold standard in differentiating a lipoblastoma from a liposarcoma. In some cases where the histopathology is inconclusive, genetic rearrangement of the PLAG1 (pleomorphic adenoma gene 1) oncogene on chromosome 8q12 helps in confirming the diagnosis of lipoblastoma.

Key words: *Axillary mass, Lipoblastoma, Infant.*

The presence of an axillary mass in infancy entertains the diagnosis of a cystic hygroma, hamartoma or a soft tissue neoplasm. Lipomatous tumors, namely lipoblastomas are rare, benign tumors of infancy and early childhood. They arise from the embryonal fat cells which persist and continue to proliferate into postnatal life. They are characterized by their rate of rapid growth, local invasion and increased incidence of local recurrence of 14-25% [1]. The diagnosis of a liposarcoma in infancy should be made with caution, owing to its rarity and the aggressive treatment involved. The role of excisional biopsy, histopathology with immunohistochemistry and cytogenetics in the establishment of an accurate diagnosis, is highlighted in the following case report.

CASE REPORT

An 8-month-old female child presented to us with a swelling in the right anterior chest wall, extending through the axilla to the back (***Fig.* 1**). It was noticed since the age of 5 months, with rapid increase in size over the last one month, to the present size. The mass was non tender, soft to firm in consistency, bosselated surface, with no skin changes. The differential diagnoses included a vascular hamartoma, cystic hygroma, a soft tissue tumor such as lipoblastoma or a liposarcoma, or a matted lymph node mass.

The ultrasound examination showed an 8×7.5×4.5cm iso- to hyperechoic, lobulated solid mass, with posterior border extending beneath the lower margin of the scapula. MRI chest confirmed an 8×5×4cm right shoulder girdle,

well encapsulated soft tissue mass probably arising from the subscapularis muscle and probably of neoplastic etiology. There was no intrathoracic or bony involvement. In view of the well encapsulated nature of the tumor, an excisional biopsy was planned. The rapid growth in the tumor size warranted immediate excision to prevent mass effect and rule out malignancy which is rare, but not unknown.

Surgical exploration revealed an 8×5×6 cm vascular fleshy mass arising from the subcutaneous tissue in the axilla, closely adherent to the muscles of the chest wall and the axillary neurovascular bundle. The use of a nerve stimulator and bipolar diathermy facilitated fine dissection. The mass was completely excised. On histopathology, gross examination showed an encapsulated lobulated mass with pale cut surface with myxoid changes. On microscopy, lobular architecture with interspersed myxoid and mature adipose tissue was seen. Each lobule contained vacuolated adipocytes in various stages of maturation. Maturation of the adipocytes was more in the centre than in the periphery. There was absence of invasion in the surrounding skeletal muscle. The wide CD34, focal S100 positivity, Mib negativity and morphology rendered the diagnosis of a myxoid lipoblastoma. A PLAG1 gene analysis for determining the aggressiveness of the tumor was recommended, but was unavailable. No recurrence has been noted 2 years postoperatively.

DISCUSSION

Lipoblastoma is a tumor of infancy, with 90% before 3 years of age and 40% in the first year of life [3] A male preponderance of 3:1 has been noted [1]. Though most commonly found in the extremities -70% [4], it can also be seen in the head and neck area, trunk, mediastinum, retroperitoneum, and various organs like lung, heart and parotid gland [5]. It is recognized as a benign neoplasm with tendency of local recurrence of 14% to 25 % [1]. They can be locally invasive making complete surgical excision difficult and hence increasing the risk of local recurrence.

Histopathology with immunohistochemistry in conjunction with the morphology remains the gold standard in differentiating a lipoblastoma from a myxoid liposarcoma. Myxoid morphology in lipoblastoma is not very common. Characterstic features and lipoblastoma have been previously described [1,3,6].

In cases where these features are inconsistent, cytogenetic advancement has led to the confirmation of LPB, where consistent rearrangements in the PLAG1 oncogene on chromosome 8q12 have been noted [7].

FIG. 1 *An 8×5 cm swelling in right axilla extending to the back.*

It has been found that 70% of lipoblastoma have *PLAG1* gene rearrangement on chromosome 8q12 and up to 18% are associated with polysomy for chromosome 8 [7]. *PLAG-1* is involved in mitogenesis, proliferation, apoptosis and IGF-2 up-regulation. In humans it is expressed mainly in fetal tissues and in low levels postnatally [7, 8].

Surgical resection is the treatment of choice except in those infiltrating tumors requiring mutilating excision [1]. The aim of surgery is complete gross excision without sacrificing the surrounding vital structures or extirpation of tissue that could lead to major deformity. Incomplete gross excision, infiltrating LPB, is notorious for its recurrence. Hence, sequential close postoperative follow up with MRI is essential. A follow up of at least 2 years postoperatively is recommended [9].

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Rhizomelic Chondrodysplasia Punctata With Maternal Systemic Lupus Erythromatosus

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We report Rhizomelic Chondrodysplasia Punctata (RDCP), a rare, autosomal recessive disorder with rhizomelic shortening of limbs, congenital cataracts and seizures but without any biochemical abnormality. The mother of the baby developed Systemic Lupus Erythromatosus (SLE) with Ro/SSA antibodies 11 months after delivery. Ro/SSA antibodies may generate calreticulin antibodies causing characteristic skeletal changes.

Key words: *Anti Ro/SSA, Punctate epiphyseal calcification.*

The classic form of rhizomelic chondrodysplasia punctata (RCDP) a rare, autosomal recessive peroxisomal disorder is characterized by proximal shortening of the limbs, cataracts, distinct facial appearance, growth failure, psychomotor retardation and seizures[1]. Common radiological features are punctate epiphyseal calcifications, metaphyseal abnormalities, coronal clefts in vertebral bodies [1]. RCDP is usually lethal with 60% deaths occurring by age 1 year. [2] The characteristic biochemical profile has been previously described [3]. Recently, patients with RCDP phenotype but without abnormal peroxisomal function have been reported usually secondary to teratogen exposure or maternal diseases [4]. We report a neonate with features of RCDP without biochemical abnormality but whose mother was diagnosed having SLE 2 months prior to delivery.

CASE REPORT

This male baby was the first child of healthy unrelated Indian Hindu parents born at term by spontaneous vaginal delivery. His mother and father were 25 and 29 years old, respectively. There was no history of spontaneous abortions or antenatal teratogen exposure. His birthweight was 2459 g (10-25th percentile), length was 42.5 cm (<10th percentile), and head circumference was 33 cm (50th percentile). His upper segment to lower segment ratio was 1.8:1. He was a disproportionately

FIG. 1 *Skiagram showing punctate epiphyseal calcification of shoulder, elbow, hip and knee joints with metaphyseal flaring of humerus.*