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Lipofibromatosis: Report of An Extremely Rare Paediatric Soft Tissue Tumour Presenting As A Huge Abdominal Mass

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Abstract: *Lipofibromatosis is a rare paediatric soft tissue neoplasm usually presenting within 1 year of age and often confused with other diagnosis. In this case, 3 year old girl presented to us with a huge abdominal mass which was slowing growing without any other associated symptoms. Lipofibromatosis usually have predilection for male gender and in majority of the cases involves the extremities unlike this case. On further work up tumour was thought to be lipoblastoma until the specimen was sent for histopathology which showed abundance of mature adipose tissue with proliferative fibroblasts in adipose septa and the diagnosis of lipofibromatosis was also supported by immunohistochemisty. The tumour was completely excised and the patient was followed up for 1 year with no signs of recurrence.*

Keywords*: Abdominal mass, Lipoblastoma, Lipofibromatosis, Paediatic neoplasm, Recurrence*

I. Introduction

Lipofibromatosis is a rare paediatric soft tissue tumour with predilection for extremities and also occurs in wide variety of other sites like trunk, head and neck region,[1] slightly more common in males and histologically showing predominance of mature adipose tissues with septal fibroblastic component. These lesions are seen exclusively in children from birth to the early second decade of life. [3] It was previously interpreted as type of infantile or juvenile fibromatosis, fibrous hamartoma or fibrosing lipoblastoma and 1st described in 2000 by Fetsch et al as lipofibromatosis by reporting 45 cases [1] and since then only a few additional cases has been reported. [2] Although benign, it is known for very high recurrence after surgical excision. Although the original clinical and pathological descriptions are comprehensive, documentation of imaging findings in this entity is sparse.[4] Here we are presenting a case of lipofibromatosis in a 3 year old girl who presented to us as a large abdominal mass.

II. Case Report

A 3 year old female child presented with a firm huge swelling on the left side of the abdomen which was first noticed at 7 months of age and was progressively increasing in size. Apart from this, there were no significant symptoms associated with the swelling. The child was delivered by normal vaginal delivery. There were no significant pre or post natal history. The child was otherwise normal with normal developmental milestones for her age. On local examination, the swelling was on the left lateral abdominal wall measuring approximately 12cm x 10cmx 8cm which was globular, well circumscribed, firm, non tender with dilated veins over it. It extended superiorly upto 9th rib, inferiorly beyond the pelvic brim and posteriorly upto 2 cm lateral to midline [fig: 1(A, B, C)]. There were no other swellings and the rest of the physical examinations were within normal limits. No other associated congenital anomalies were present.

Radiography showed soft tissue thickening and fat type radiolucency. On CECT abdomen there was a well circumscribed hypoattenuating(-35 to -60HU) lesion with intervening soft tissue attenuation(20-40HU) involving the left lateral abdominal wall with retroperitoneal extension showing no significant enhancement on post contrast study[fig:2(a,b,c)]. All other abdominal organs were within normal limit. The tumour was excised and sent for histopathological examination. The excised specimen weighed 586gm and measured 14cm x13cm x10cm [fig: 3 (I, II, III)]. On gross examination, external surface was bosselated and cut section showed lobules of fatty tissue separated by broad bands of collagen [fig: 3-III & 4-i]. On microscopy the specimen showed a tumour composed of benign spindle cells arranged in loose singles, in broad fascicles and in fish stream like pattern. Lobules of mature adipocytes were seen separated by broad bundles of fibrocollagenous stroma. Peripheral areas displayed the spindle cells intersecting through bundles of skeletal muscle. There was no nuclear atypia, mitotic activity or area of necrosis. No lipoblast and immature mesenchymal tissue is seen [fig: 4 (ii,iii,iv)]. Thus overall features were consistent with that of the lipofibromatosis. Spindled fibroblastic cells stained negative for Desmin on immunohistochemistry[fig: 4-v].

III. Discussion

Lipofibromatosis is a rare benign soft tissue tumour reported from birth to early second decade of life, previously designated as infantile fibromatosis of non-desmoid type but now delineated as distinct entity after study of 45 cases in 2000 by Fetsch et al, as it has distinct clinical and histological features. [1] These tumors had been variously diagnosed as a type of infantile fibromatosis, a variant of fibrous hamartoma of infancy and a fibrosing lipoblastoma. This tumour is more common in male, with male to female ratio of 2:1. [5] It is most commonly seen in extremities and is slightly less common in the thigh, trunk and head. [5, 7, 8] Aetiology of this tumour remains unknown till date. [6] Lipofibromatosis mostly presents as a painless slow progressively growing mass and usually measures 1 to 3 cm, rarely exceeding 5cm, [1] unlike our case where the size of the tumour is relatively large measuring about 14cms in largest dimension involving the trunk and probably is the first case to be reported in the literature till date.

Imaging generally reveals fat that appears as 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. Ultrasound usually demonstrates poor musculature planes with hyperechoic content. CECT is useful in outlining the tumour and demonstrating a low density non-enhancing mass measuring fat in Hounsfield units as also seen in the present case. As in this case, the diagnosis of lipoblastoma was made after CECT findings. Magnetic resonance imaging (MRI), though not available in the case described, plays an important role in tissue characterisation with increased T1 andT2 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. [9] The confirmatory diagnosis is always made after histopathological examination which consists of abundant mature adipose tissue separated by septa containing spindle fibroblast-like cells. [1] As this lesion has prominent fat component, various adipocytic tumors may be considered in the differential diagnosis including angiolipoma, atypical lipomatous tumor, lipomatosis, lipoblastoma as the diagnosis. Immunohistological studies are also an invaluable aid in doubtful cases. [1,6] Lipofibromatosis is focal staining of the spindle cells with CD34, CD99, SMA, BCL-2 and typically negative staining with desmin.[1,6,10] As in present case, spindle cells stained negative for desmin which is the only immunohistological studies done. There is a high predilection for recurrence in incompletely excised lesions hence complete surgical excision is the mainstay of treatment. [11] This case has not shown any sign of recurrence since 1 year following excision and the tumour was completely excised without causing any functional compromise.

IV. Figures

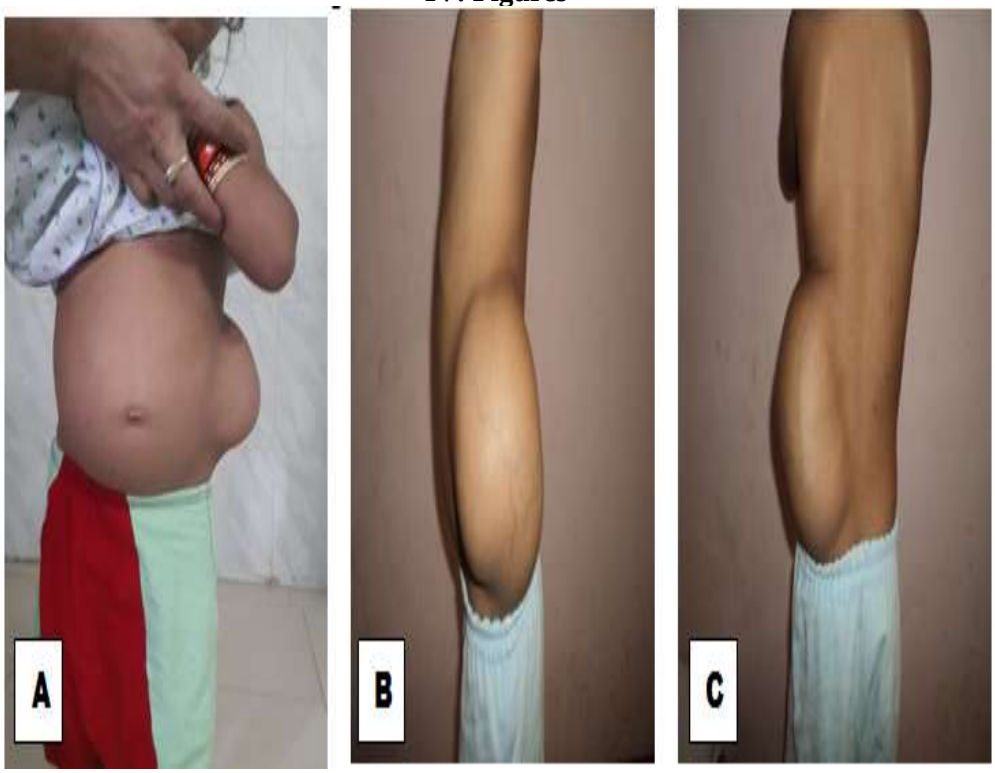

Figure 1: The patient presented with the painless slowly growing abdominal wall mass with dilated veins over it [A] Anterior view [B] left lateral view [C] posterior view

Figure 2: CECT abdomen showing a well circumscribed fat attenuating (-35 to -60 HU) lesion with intervening soft tissue attenuation (20-40 HU) involving the left lateral abdominal wall with no significant enhancement on post contract study. [a] Coronal section [b] left lateral view [c] Axial section

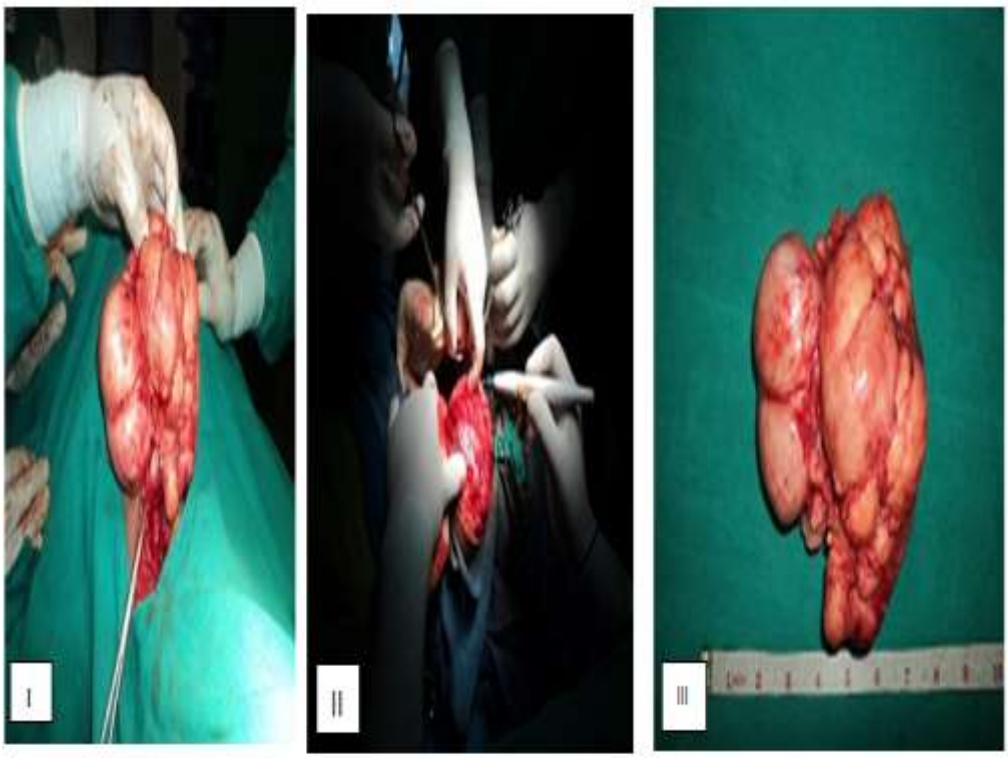

Figure 3: Intraoperative pictures of the tumour [I] Dissection following the skin incision in search of the correct tissue plane [II] Isolating the tumour from the surrounding tissues just before the complete excision [III] Excised specimen of the tumour consisting of a globular soft tissue mass with bosselated external surface measuring 14x13x10cms and weighing 586gms.

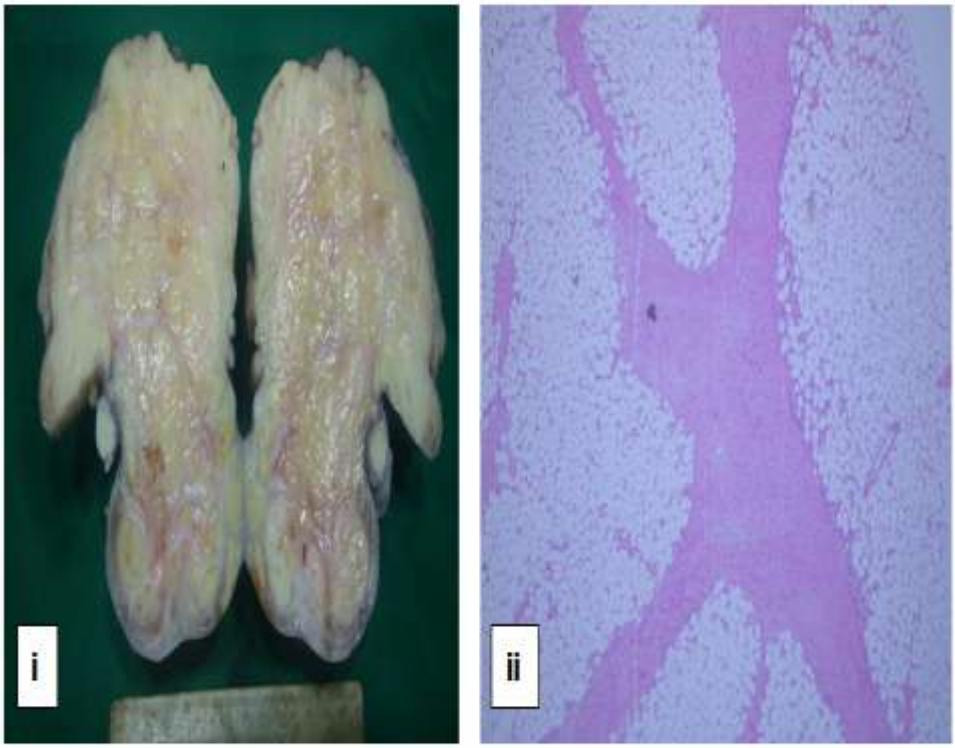

Figure 4: [i] Cut section of the tumour showing lobules of fatty tissue separated by broad bundles of collagen and peripheral areas shows bundles of skeletal muscles. [ii] Microscopic picture showing lobules of mature adipocytes separated by the broad bundles of fibrocollagenous stroma.

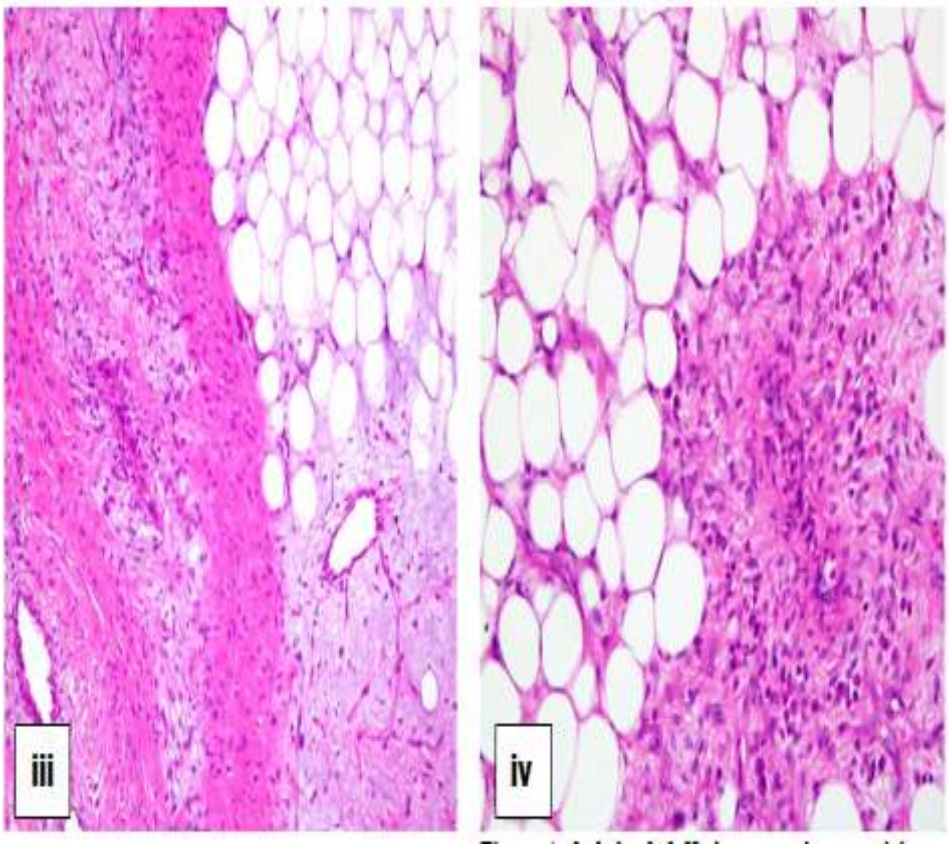

Figure 4: [iii] & [iv] Higher magnification of the microscopic pictures with benign spindle cells in a myxoid stromal background interspersed by numerous thin walled capillaries lined by plump endothelium. There is no

evidence of mitotic activity, nuclear atypia or areas of necrosis. No lipoblasts and immature mesenchymal tissue seen.

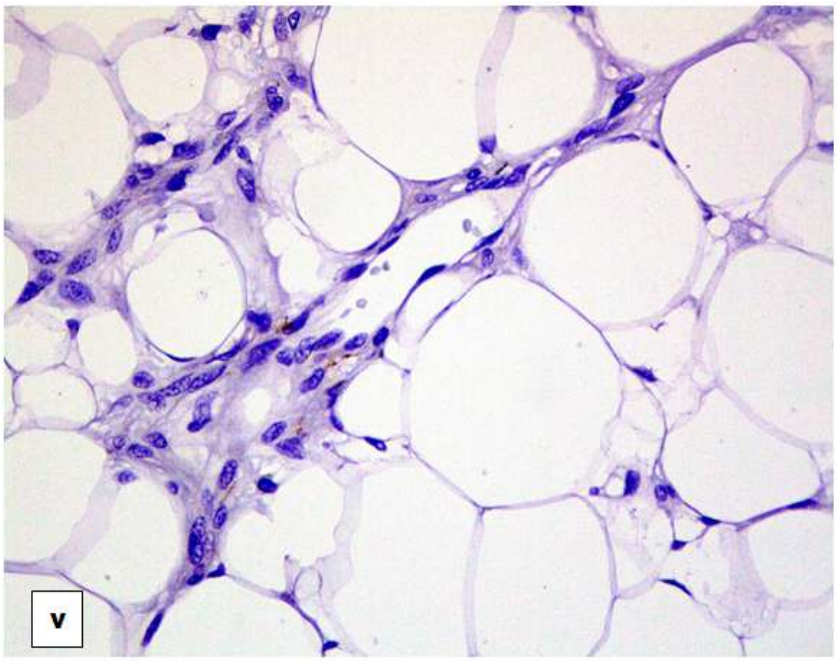

Figure 4 : [v] Immunohistochemistry marker, Desmin negative spindle cells

V. Conclusion

Putting together the clinical presentation, findings on examination and investigations, this case points towards the diagnosis of lipofibromatosis which is a very rare soft tissue tumour in the paediatic age group and lipofibromatosis should be taken into consideration as a differential diagnosis when came across child with abdominal wall mass.

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