PHILIPPINE JOURNAL OF OTOLARYNGOLOGY-HEAD AND NECK SURGERY VOL. 26 NO. 1 JANUARY – JUNE 2011

Lipofibromatosis: An Unusual Head and Neck Mass in the Paediatric Age Group

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ABSTRACT

Objective: To describe a rare case of lipofibromatosis presenting as a head and neck mass in a 6-year-old child.

Method:

Design: Case Report
Setting: Tertiary Public General Hospital
Patient: One

Result: A six-year-old male child admitted with a large right head and neck region mass underwent complete excision of a possible soft tissue neoplasm following investigations which included Fine Needle Aspiration Cytology, Ultrasonography and Computed Tomography. Histopathological examination yielded lipofibromatosis, a very rare lesion with a distinctive fibrofatty pattern. The patient was well with no recurrence after three months of follow up.

Conclusion: Although lipofibromatosis is a rare lesion in children and has a predilection for distal extremities, it may also present as a mass in the head and neck area. Complete surgical excision is feasible and is the only treatment option available for this rare lesion

Keywords*: lipofibromatosis*

Lipofibromatosis is a rare fibrofatty tumour of the paediatric age group. It has only recently been described as a clinicopathologic entity in 2000 by Fetch *et al.* and subsequently only few case reports have been published in the literature.[1] These lesions are seen exclusively in children from birth to the early second decade of life[2] with an over 2:1 male predominance and predilection for distal extremities, being less common in the trunk, head and neck.[3] A MEDLINE PubMed search using the terms "lipofibromatosis" and "neck mass" yielded only two cases of this lesion presenting as neck mass in the paediatric age group.[4,5] The case described in this report is another instance that presented as a head and neck mass and subsequently turned out to be lipofibromatosis on histopathological examination.

CASE REPORT

A six-year-old boy was admitted to the paediatric surgery department with a slowly growing painless mass on the right side of his neck and face for the last two years. Upon examination, the mass extended from the orbital floor to the lower border of the mandible, also occupying

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the submandibular triangle in the neck. It was firm in consistency and not fixed to skin and underlying structures *(Figure 1)*.There was no significant lymphadenopathy of the neck.

Ultrasonography of the mass revealed an isoechoic to hyperechoic lesion without any flow on Colour Doppler. Contrast-Enhanced Computed Tomography (CECT) scan showed a large non-enhancing soft tissue mass mainly of fat density with lots of fibrous septa displacing the carotid vessels posteriorly *(Figure 2)*. Fine Needle Aspiration Cytology (FNAC) showed spindle cells suggesting a mesenchymal lesion. The haematological investigations were within the normal range. The tumor was excised via a large semilunar cervicofacial skin flap that was resutured without any facial reconstruction. The completely-excised tumour was 8x4cms in size on gross examination *(Figure 3)*. Histopathological examination revealed lipofibromatosis *(Figure 4)*. The patient was discharged in good condition after ten days of hospitalization and he was well after three months of follow up.

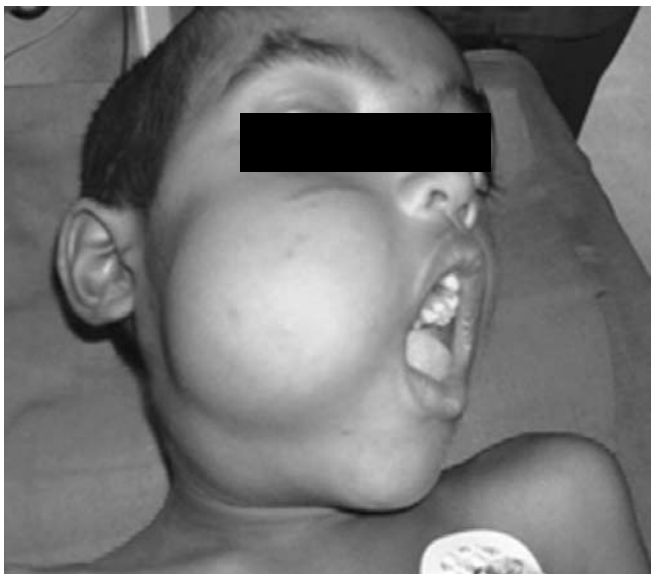

Figure 1. Clinical photograph shows a huge mass on the face and neck.

DISCUSSION

Lilpofibromatosis is a rare benign soft tissue neoplasm of childhood, previously designated as infantile fibromatosis of non-desmoid type.[5] In 2000, Fetch *et al*. proposed that although this tumour is likely to be a part of the infantile fibromatosis spectrum, it should be considered a distinctive entity because of its histological pattern and they coined the term lipofibromatosis for this rare lesion.[1] These tumors had been variously diagnosed as a type of infantile fibromatosis, a variant of fibrous hamartoma of infancy and a fibrosing lipoblastoma.[3] Lipofibromatosis has been described from birth to the early second decade and the median age for the first surgery is one year[2.] There is a male predominance with a male to female ratio of 2:1.[3] It is most commonly seen in hands and feet and is slightly less common in the thigh, trunk and head.[3] The etiology remains unknown.[4] The lesion usually measures 1 to 3 cm, with a median size of 2 cm, and it presents as a poorly circumscribed mass involving the subcutis and/or deep soft tissues. It is rare for lipofibromatosis to be over 5 cm in diameter[1] and the present lesion measuring 8 x 4 cm and extending from the neck and face to the orbital floor could probably be the first case of this size in the literature.

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. 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.[6]

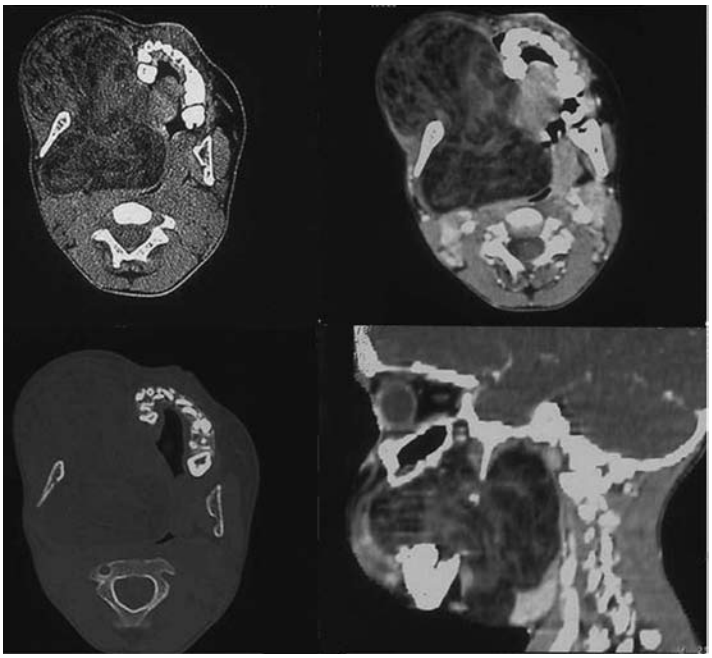

Figure 2. Axial and reformatted sagittal non-contrast and contrast-enhanced computed tomographic sections reveal a well-defined dumbbell-shaped lesion measuring 6 x 8 x 10cm between the maxilla and mandibular ramus on the right side, grossly displacing the adjacent bones.

As this lesion has prominent fat component, various adipocytic tumors may be considered in the differential diagnosis including angiolipoma, atypical lipomatous tumor, lipomatosis, lipoblastoma/

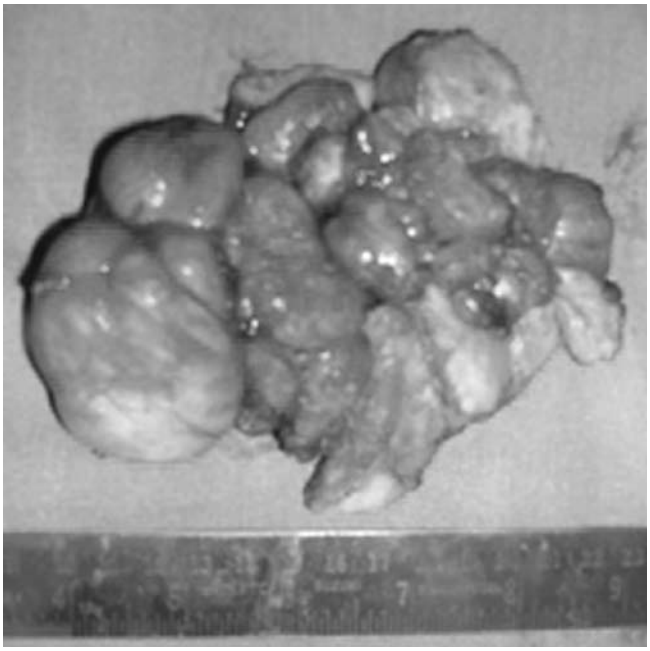

Figure 3. Photograph of gross specimen, excised tumour

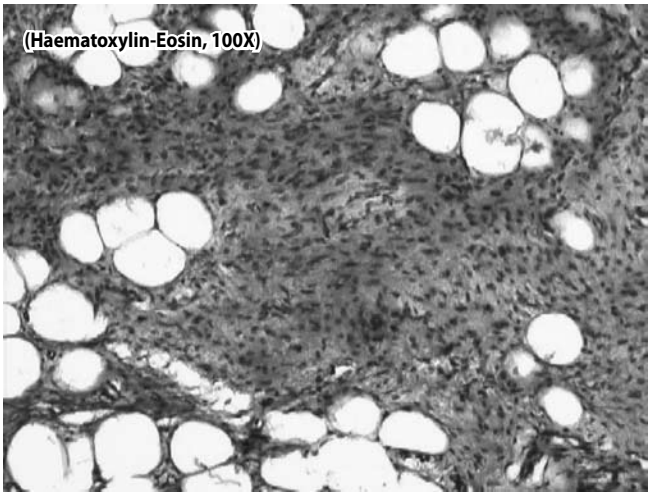

Figure 4. Haematoxylin and Eosin (H&E) Slide, high power magnification (100X) showing lipofibromatosis- primitive fibroblasts infiltrating between lipocytes.

lipoblastomatosis, fibrous hamartoma of infancy and fibrohisticytic lipoma.[7] However, the microscopic examination of this tumor revealing uniformly-sized mature adipocytes with lack of nuclear atypia, necrosis and mitotic activity exclude malignancy. Moreover, the simple morphological pattern with spindle cells and fatty tissue septa without primitive mesenchymal components exclude other non –malignant conditions like lipomatosis and fibrous hamartoma of infancy.[7] If facilities for immunohistochemistry are available in the hospital, immunohistochemical examination of this lesion may help in making the diagnosis. The most common immunoprofile of lipofibromatosis is focal staining of the spindle cells with CD99, SMA, BCL-2 and typically negative staining with desmin.[1]

Complete surgical resection is the mainstay of treatment because of high predilection for recurrence in incompletely excised lesions.[5] Since most lesions have infiltrating borders, complete removal of the tumor might cause functional compromise. In the present case, the tumour was removed completely without any significant functional compromise. However, there are some cases with longer follow-up who experienced no recurrence even though the lesion was incompletely excised.[1,5] In view of these circumstances and due to the paucity of literature that accurately predicts outcome, the conclusions regarding patient management as well as prognosis must be individualized based on the patient's condition.

In conclusion, although more documented cases of this entity have been published, insufficient clinical experience in treatment still remains. Despite the rarity of its presentation as a head and neck mass, lipofibromatosis should be taken into account as a differential diagnosis in the management of head and neck masses in the paediatric age group.

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