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CLINICAL REPORT

Maxillary Lipofibromatosis in a pediatric patient. Case report.

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ABSTRACT

This report discusses a rare case of a soft palate deformity in a young girl due to lipofibromatosis (LPF). This rare benign pediatric soft tissue tumour usually arises in the distal extremities. We believe this case represents the first report of lipofibromatosis involving only the maxillary bone.

KEY WORDS:

Lipofibromatosis; Childhood/paediatric; Fibroblastic/myofibroblastic tumour.

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INTRODUCTION

Lipofibromatosis (LPF) is an uncommon benign soft tissue tumor of childhood first described by Fetsch et al. in 2000 and was previously named infantile/juvenile fibromatosis[1,3]. In 2002 LF was included in the World Health Organization (WHO) under the fibroblastic/myofibroblastic tumors and afterward reclassified based on the biological behavior as an intermediate- locally aggressive lesion1. In the past, this tumor was misinterpreted and has recently been considered a distinct entity from other pediatric fibro-fatty lesions[2]. Lipofibromatosis is characteristic of its high recurrence rate, reported from 33% to 72% in the literature, and the diagnosis is mainly pathological since the radiological and clinical characteristics are non-specific[1]. Therefore, the primary treatment is complete surgical resection, and only in cases where the lesion is diffusely infiltrating could partial resection be considered to reduce the patient's postoperative morbidity[4]. Fortunately, there is no metastatic potential[3].

To our knowledge, two reports detail a case of exclusive intraoral LPF, both on the left side of the mandible. The first one, a single case of the original 45 examples study of LPF, was in the oral cavity[5], and the second one was described by Wollenberg et al. in 2016[6]. This review discusses the diagnosis and treatment of lipofibromatosis in this uncommon location.

CASE REPORT

A six-year-old girl presented with her mother to the Maxillofacial Surgery Department with a painless swelling of the left side of the soft palate after three months of evolution. The mass gradually increased and was not associated with aggravating or relieving factors. Her medical and family history was otherwise non-contributory.

Upon examination, a non-tender and firm mass was palpable on the soft palate measuring approximately 1.5 x 1.5 cm. It was not fixed to underlying structures, and the overlying mucosa was slightly erythematous. There was no significant neck lymphadenopathy and no other abnormality in physical examination (Figure 1)

Contrast Computed Tomography scan and Magnetic resonance imaging (MRI) showed a circumscribed soft tissue lobulated lesion in the left soft palate region with focal differences of density and fibrous tissue displacing without evidence of bony involvement (Figure 2).

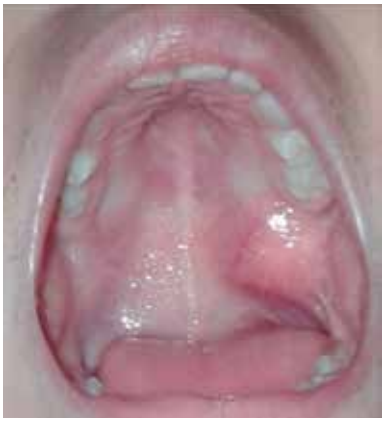

Figure 1. The image shows a soft palate mass, with clear borders. measuring approximately 1.5 x 1.5 cm.

Pre-surgical laboratory tests were requested: complete blood count, VHS, protein C, and prothrombin time to assess the patient's systemic condition were within the normal range.

The patient underwent surgical excision of the mass, considering wide safety margins to reduce recurrence risk. The incision was made between the maxillary tuberosity and the left palatoglossus muscle without midline compromise.

Gross examination showed a white-tan nodular fragment of 2.0 x 1.5 x 1.0 cm. Microscopically, a non-encapsulated lesion composed of fused fibroblastic proliferation with lax areas and myxoid foci, few mitoses, septa of mature adipose tissue, and a concentrate of bone tissue included was seen. There was no atypia. Immunohistochemically, the lesion showed focal positivity for vimentin, S-100, smooth muscle actin (SMA), and CD34. (Figure 3). Negative results were for desmin, pan-cytokeratin, and Ki-67 <5%. Based on these findings, a diagnosis of lipofibromatosis was

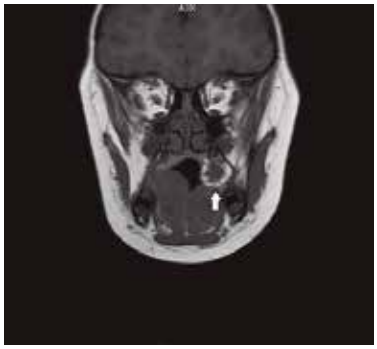

Figure 2. Magnetic resonance imaging (MRI) showed a circumscribed soft tissue lobulated lesion.

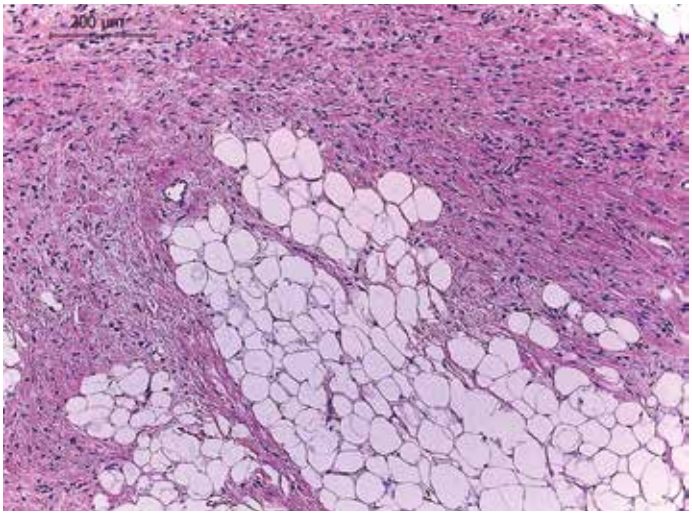

Figure 3. Photomicrograph showing a non-encapsulated lesion composed of fused fibroblastic proliferation with lax areas and myxoid foci, few mitoses, septa of mature adipose tissue with no atypia.

made. After hospitalization, the patient was discharged in good condition and continued well after a year of follow-up.

DISCUSSION

Lipofibromatosis is a rare benign soft tissue neoplasm of childhood, previously designed as an infantile/juvenile fibromatosis variant of non-desmoid type[7-8]. Patients have ranged in age from newborns to teenagers[1,7,9]. These tumors have a male-to-female predominance of a 2.7:1 ratio[3,4], but other studies have reported no gender predilection[9]. Even though numerous theories have been proposed, the etiology is still uncertain[7,10]. Approximately 20% of cases are congenital[7]. Clinical features described a slow-growing, solitary, painless, ill-defined, and firm mass located in an extremity's subcutis or deep soft tissue[1,7]and only about 17% of cases have affected the head and neck or a truncal location[7,9]. Tumors range in size from 1-3 cm; however, reported cases have exceeded 5 cm in diameter3. They can infiltrate adjacent neurovascular bundles and muscles but generally do not impair function[9].

Imaging studies have a low sensitivity in diagnosing LPF, and images are variable depending on the proportion of adipocytic and fibrous component[1,2]. Ultrasound is an initial functional screen to determine whether the lesion is of vascular origin[4] but is non-specific and has poor diagnostic utility[1]. Contrast Computed Tomography helps outline the tumor and demonstrate a low-density non-enhancing mass measuring fat in Hounsfield units[8]. In this case, initial imaging includes MRI, which is better suited to distinguish soft tissue, obtains a better cleavage plane image, and is considered the imaging modality of choice[2,4,9,11]. It helps evaluate the tumor extension and characterization with hyperintense signaling that isointense to fat on T1- and T2- weighted imaging[1,3,4,8]; even though this cannot definitively diagnose LPF, it can exclude other diagnoses in preoperative planning[4].

Another type of diagnosis complement is the fine needle aspiration cytology (FNAC) which may be helpful to suggest a probable preoperative diagnosis in these lesions[2,11] but was not performed in this case.

Macroscopy showed a yellowish or tan-white color and a rubbery, firm, or gritty consistency[5]. Histopathology contains abundant adipose tissue that usually accounts for >50% of the tumor, and the fat cells generally mature and lack atypia. A fibroblastic spindle element characteristically forms fascicles concentrated in septal regions and along the perimysial surface of skeletal muscle. The fibroblastic part has only mild atypia and a low mitotic rate[7]. This description was consistent with the appearance of the resected mass.

Immunohistochemistry is not required to establish the diagnosis as it is variable and non-specific[1,2,3,4,5]. The spindle cell element of the tumor may exhibit immunoreactivity for CD99, CD34, alfa smooth muscle actin (SMA), BCL-2, S-100, muscle-specific actin (MSA), and rarely epithelial membrane antigen (EMA)[1,3,5,8]. Stains for desmin and keratin are usually negative[1,5,8]. Adipocytes highlight S-1004,5 and fibroblasts typically express only vimentin.

The pathogenesis of LPF has been controversial and has recently been identified as involving ligands FN1-EGF genetic fusion as a genetic aberration in calcifying aponeurotic fibroma. The genetic overlap of these tumors raises the possibility that a subset of LPF may represent an early form of calcifying aponeurotic fibroma without calcification or matrix formation[1,12]. The fusions involved ligands (EGF, HBEGF, TGFA) to the epidermal growth factor receptor (EGFR) itself or other receptor tyrosine kinases (ROS1, RET, PDGFRB). These are all known to activate the PI3K-AKT-mTOR pathway; thus, deregulation has been suggested as a potential pathogenetic mechanism in developing lipofibromatosis[1,7].

LPF should be differentiated from other benign fibrous and adipocytic tumors of childhood, including calcifying aponeurotic fibroma (CAF), recently mentioned fibrous hamartoma of infancy, lipoblastoma, intramuscular lipoma, and lipofibromatosis neural tumor[1,2,4]. In addition, clinical features like the site of involvement and tumor appearance may provide a clue to differentiation among these diagnoses. It is important to highlight as a differential diagnosis with lipofibromatosis -like neural tumor, which also is a soft tissue lesion usually seen in the pediatric age that manifests as a slowly growing mass in the deep dermis or subcutis of extremities, head, neck, and trunk[1], but the positive staining for S-100 only in adipose tissue and negative in the fibrous component rule out this tumour. In addition, lipofibromatosis -like neural tumor requires positivity with CD34 in the spindle cell component, which is appreciated, but negative S-100 staining discard the hypothesis.

About the prognostic factors, LPF has no known metastatic potential[1,3,4,7,9,13]. Although it is a benign lesion, it may exhibit progressive, infiltrative local growth and recurrence owing to its infiltrative margins. It can become locally destructive and may be of cosmetic or functional significance[4]. Congenital lesions, male sex, acral location, mitotic activity in the fibroblastic component, and incomplete excision have been suggested as predisposing factors for recurrence[1,7].

Complete surgical excision with ample margins is the preferred treatment method for LPF[2]. Radio and medical therapy have been attempted as adjunct therapy but without known benefits[1,10]. In literature, spontaneous regression has not been reported[10]. Fortunately, in this six-year-old girl, the tumor was entirely removed without any significant physical alteration. However, in some circumstances where the lesion is diffusely infiltrative, partial resection is recommended to reduce the morbidity and functional compromise of the patient[1,4]. Some cases with longer follow-up experienced no recurrence even though the lesion was incompletely excised[5,8].

CONCLUSION

Given the rarity of this benign tumor and even more of this case, since the anatomical site and gender are not the most reported in the literature, the conclusions highlighted regarding diagnosis, management, and prognosis must be individualized based on the patient's condition. The maxillary bone is a strange site for developing lipofibromatosis. However, LPF is uncommon; we might suspect and not rule out its diagnosis when the clinical characteristics point to soft tissue lesions, especially at the pediatric age. Therefore, this entity should be considered in the differential diagnosis of soft tissue tumors in childhood.

CONFLICTS OF INTREREST

The authors have no conflicts of interest to declare.

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ETHICS APPROVAL

No ethical approval was required for this study.

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