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A Case report: Aggressive Fibromatosis in a One-year Old Child

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ABSTRACT

Aggressive fibromatosis, also defined as desmoid tumor, is an uncommon tumor. A review of literature detected less than 100 cases that have been reported of pediatrics aged 16 years or younger with aggressive fibromatosis in the head and neck. In this study we report a 1-year old girl who presented with an oral mass . The clinical , radiological, the histopathological features, and treatment are discussed .

Key words : aggressive fibromatosis, desmoids tumor, pediatric, and head and neck .

INTRODUCTION

Aggressive fibromatosis, previously referred to as desmoids tumor, is described as uncommon soft tissue tumor. It often rises from deep-seated musculoaponeurotic structures [1, 2] .The tumor margins sometimes interdigitate with surrounding tissues, and have a high potential for local invasion and recurrence. Head and neck fibromatosis comprise around 12–15% of all cases [3]. In 1839, Dupuytren first described the features of fibromatosis, but it was not fully published until 1954 by Stout .

The etiology of aggressive fibromatosis remains unknown. However, other various etiologies such as genetic predisposition, trauma, and association with Gardner's syndrome, etc, have been suggested in the literature [3] .In relation to a potential endocrine etiology, there have been cases reported in the literature that have shown progression or regression in response to either puberty or hormonal therapy. We report a 27 month old patient that has been diagnosed as a case of aggressive fibromatosis, along with literature review of similar cases reported in the literature.

CASE REPORT

A 27 months old female presented with an oral mass noticed by her mother since she was 3 months of age. This mass was stable in size until a biopsy was taken at the age of 3 months. Since then, the mass started growing slowly. The mother noticed decrease in oral intake. The family seeked medical advice. The patient was presented to the ORL clinic and a biopsy was taken and the preliminary diagnosis was suspicion for leiomyoma. On examination, there was 3X4 cm oropharyngeal mass projecting on the right side of the mouth. It was occupying most of the soft palate on the right and crossing the midline pushing the tonsil inferiorly and the uvula medially (Figure 1A) .

The mass had a smooth outer surface but firm to palpation. Flexible nasopharyngoscopy showed no intranasal involvement. There was no airway compromise with only a small bulge on the right lateral pharyngeal wall at the level of the palate. The patient had no palpable neck nodes and the rest of the head and neck examination was unremarkable. An MRI was performed and showed hypo-intense parapharyngeal tumor on T2 and Hyper-intense on T1 (Figure 2).

The patient was taken to the operating theatre for resection of the parapharyngeal tumor. The patient's guardian was consented for transoral and possible transcervical approaches. The tumor was resected transorally. Upon excision the tumor was found to extended posteriolaterally to the ptyregoid muscles. Biposy from ptyregoid muscles, was taken as a deep margin. The right tonsil was removed with reconstruction of the remaining soft palate primarily. The patient was discharged after recovery and was able to tolerate oral feeding (Figure 1B).

Histologically, sections from oropharengeal mass reveal multiple fragments of soft tissue some of it composed of mucus secreting glands in which there was spindle cell proliferation

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(Figure 3). The tumor cells were composed of elliptical – oval nuclei with inconspicuous nucleoli. The cytoplasm was abundant. Scattered mitosis was seen. The tumor cells were arranged in a storiform pattern. Occasional cartwheel pattern were also present. Immunostains showed that the tumor cells were positive with vimentin, Desmin, Actin HHM35, Caldesmin and B Catanin. Ki 67 was 20% nuclear positivity. All other stains including, CK-pan, S100, NFP, ER, PR and HHV8 were negative.

Clinical examination two years post treatment showed no evidence of recurrence (Figure 1 C). The child was planned for MRI in another year to check for recurrence.

DISCUSSION

Aggressive fibromatosis is a soft tissue tumor, arising from musculoaponeurotic structures. Many attempts have been done by Enzinger and Weiss to classify it by dividing it into two categories either superficial or deep [4] .

Spindle cell tumors in children comprise a clinically heterogeneous group of neoplasms with similar morphologies. The benign differential diagnosis included nodular fasciitis & fibromatoses. Other differential diagnosis of intermediate features of malignancy included inflammatory myofibroblastic tumor, infantile fibrosarcoma, dermatofibrosarcoma protuberans, low grade fibrosarcoma, low grade leiomyosarcoma, monophasic synovial sarcoma and malignant peripheral nerve sheath tumor. Immunostains was helpful to differentiate the most important morphological differential diagnosis. (See table I) > Based on the immunostains the most compatible diagnosis was fibromatosis. Electron microscopy (EM) was performed from the block that was formalin fixed paraffin embedded material . It showed spindle cells with large number of intermediate filaments, endoplasmic reticulum and absence of basal lamina and pinocytotic vesicles (Figure 4). Morphology, immunohistochemistry and EM are all supportive of the diagnosis of fibromatosis. Incidence of fibromatosis peaks in individuals from 6 to 15 years of age. Another incidence between puberty and 40 years of age in women.

Both infants and young children have a higher propensity for tumors in the head and neck accounting for 12–15% of all cases of Fibromatosis [5] .Typically fibromatosis present as a firm, painless mass that is hurriedly enlarging. The most important histologic differences between desmoid tumors in children and adults is that, children fibromatosis is more cellular with higher mitotic rates. The relation of fibromatosis and Gardner syndrome have been discussed in details in the literature. Patients with Gardner syndrome have number of extracolonic manifestations such as osteoma and desmoid tumor [6] .

In addition both Familial adenomatosis polypi (FAP) and Gardner syndrome are associated with germline mutation in the adenomatous polyposis coli (APC) gene. The APC protein acts to promote degradation of Beta-catenin, which is an intracellular protein that aids in the transduction of cell proliferation signals to the nucleus. Somatic mutations in APC and CTNNB1. The later is the gene encoding catenin, in sporadic desmoid tumor cases. Both mutations ultimately enhance -catenin activity. Presence of Beta- catenin nuclear stain by immunohistochemistry confirm the presence of mutation in CTNNB1 gene, that can be either acquired or inherited mutation, however conclusion cannot be drawn from this result [6] .

The treatment for aggressive fibromatosis is surgery with clear margins [3] .The clinical behavior of aggressive fibromatosis remains random and there are reports of spontaneous regression with minimal to no treatment [7] .Fibromatosis do not metastasize, on the other hand without adequate resection, they are locally aggressive and tend to recur [4] .The overall recurrence rates reported to be around 50% [8] .The recurrence rate for those who received primary surgery was around 19%. Examples of Adjuvant therapy, such as chemotherapy, radiation therapy, and hormonal therapy are occasionally given for persistent disease, recurrent disease, or unrespectable disease. Adjuvant therapy is hardly given as a primary therapy for destructive fibromatosis, and there is little evidence to support its use [3,9] .In only few cases , spontaneous regression without treatment was seen. Regression of fibromatosis has been defined at menarche, suggesting a hormonal influence. Tamoxifen, an anti- estrogen has been shown to have an inhibitory effect on aggressive fibromatosis. In the literature, two patients

received hormonal therapy, only 1 of which was certainly tested for estrogen receptors. Both patients responded well to Tamoxifen [10] . Nonsteroidal anti-inflammatory drugs have also been used in the treatment of aggressive fibromatosis. Sulindac has been used with some accomplishment in patients with aggressive fibromatosis who have familial adenomatous polyposis. In our review, we found no patients who had experienced of NSAID therapy

CONCLUSION

Head and neck fibromatosis, although rare, Yet it present a complex problem for the otolaryngologist as they are often aggressive, and invade local bone, nerve and muscles, and can leave patients with a cosmetic and / or functional deformity.

Head and neck fibromatosis remains until now very hard to treat. The standard treatment of this kind of disease is aggressive surgical resection with an attempt at negative margins. The chemotherapy and radiation therapy are reserved for those cases that are either not curable or for those patients that experience a recurrence during their disease process.

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FIGURES

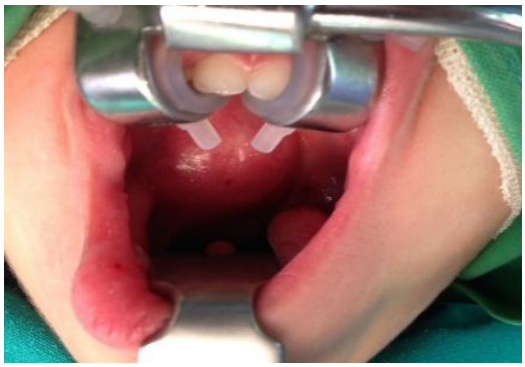

Figure 1. A. large oropharyngeal mass

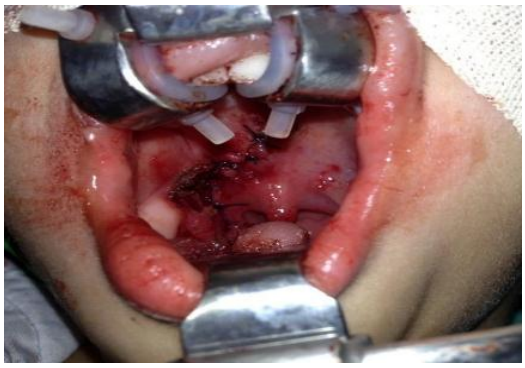

Figure 1. B. post transoral excision and repair of soft palate

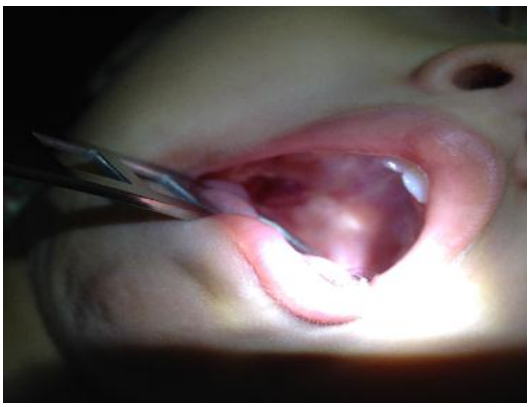

Figure 1. C. One year clinical follow up showing no evidence of recurrence

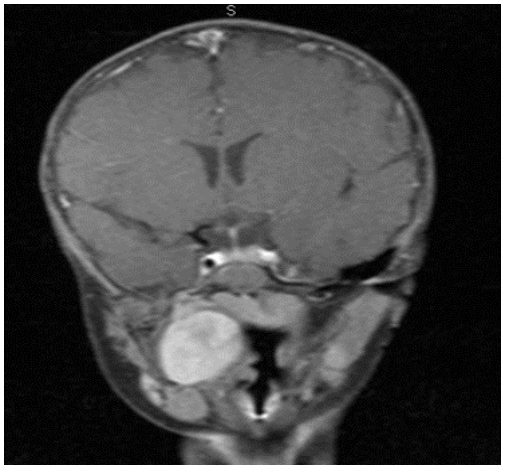

Figure 2: MRI coronal cuts showing parapharyngeal extension of the tumor which was hypo-intense on T2 and hyper-intense on T1.

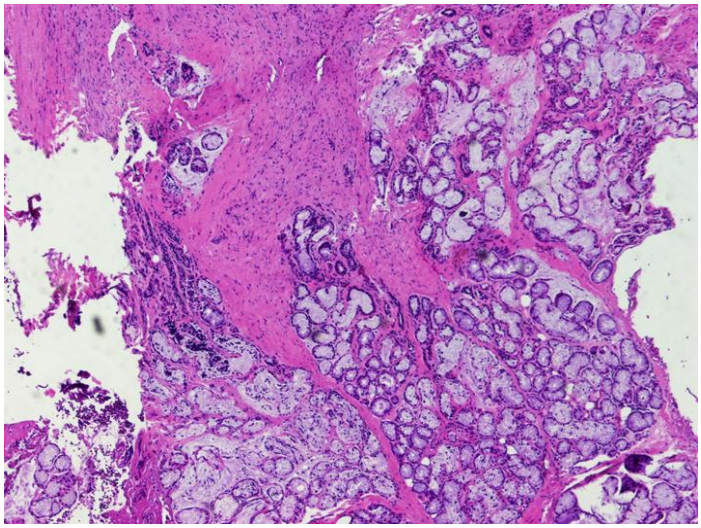

Figure 3: Spindle cell proliferation around mucus secreting glands of nasopharynx

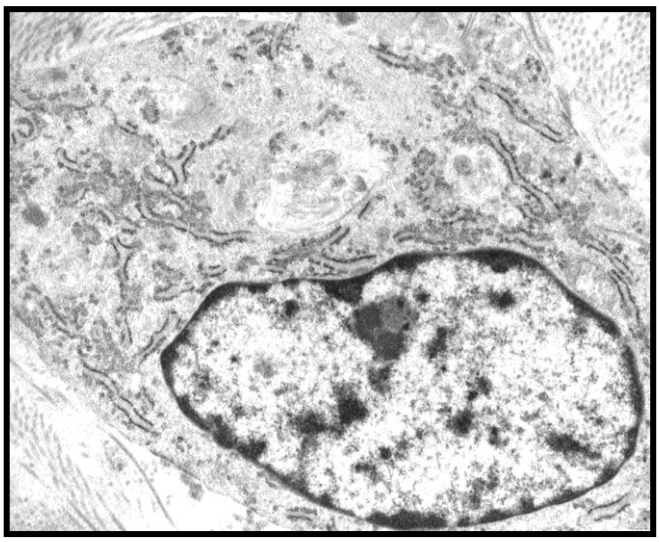

Figure 4 : EM Photograph to demonstrate the spindle cells with intermediate filaments in its cytoplasm and lack of basal lamina and prominent endoplasmic reticulum

Table 1 :
- To demonstrate the different immunohistochemical stains used to differentiate the spindle cell lesions in the differential diagnosis.
- Immunohistochemical stains that helped in the diagnosis in fibromatosis & to differentiate it from other spindle cell tumors.

	Our case	Fibromatosis	Myofibroma	Fibrosarcoma	Leiomyosarcoma
Vimentin	+	+	+	+	+
Desmin	+	-	+	-	+
Actin HHM35	+	+	+	+	+
CD34	F+	-	+	+	-
Caldesmin	+	+	-	_	+
B catenin	+	Nuclear ++	cytoplasmic++	cytoplasmic++	cytoplasmic++
CP-Pan	-	-	-	-	-
Myogen	-	-	+	-	+
S100	-	-	-	-	-
NFP	-	-	-	-	-
ER&PR	-	+	-	_	-
HHV8	-	-	-	-	-