

## Juvenile Myofibromatosis of Mandible- A Case Report

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### ABSTRACT

Myofibroma is a solitary benign tumor of myofibroblasts. Myofibromatosis describes multiple, simultaneous myofibromas at different sites in various organs. Myofibromatosis is a rare but well recognized entity which was originally thought to affect only neonates and infants. It is now apparent however that adults may also be affected. Solitary cases affecting the oral cavity appear to be rare. This case report documents a case of myofibromatosis in a 13-year-old boy. The elongated spindle cells were identified as myofibroblasts by immunohistochemistry. The lesions showed characteristic features which enabled them to be distinguished from other fibrous lesions and from benign or malignant smooth muscle tumors with which they have been often confused.

**Key Words:** Myofibroma, multicentric, solitary

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## Introduction

Myofibromatosis is a rare, but well recognized, entity which was first described by Stout as congenital generalized fibromatosis.<sup>1</sup> In 1965, Kauffman and Stout divided the condition into two categories: a multiple form limited to the skin, soft tissue, and bone, and a generalized form with visceral involvement.<sup>2</sup> Wiswell et al 1988 sub classified the condition into solitary and multiple lesions with further subdivision into the presence or absence of visceral involvement.<sup>2</sup> The solitary form is the most common mode of presentation, it particularly affects the skin, muscle, bone and subcutaneous tissue in the head, neck and trunk.<sup>3</sup> The solitary form tends to occur predominately in males and the reported incidence of solitary osseous myofibromatosis is rare.<sup>4</sup>

The present case report is probably the 40<sup>th</sup> documented case of solitary myofibroma of mandible in a 13-year-old male patient with detailed description of clinical, radiographic, histopathological and immunohistochemical findings.

## Case Report

A 13 year old male patient reported in the Department of Oral Pathology & Microbiology with complaint of slow growing, painless swelling in the right side

of the posterior region of the jaw since 4 months.

On extra oral examination mild facial asymmetry was revealed on right half of the face (Figure 1). Swelling was measured with the help of Verneir caliper and the dimensions were 3.7 x 2.1cms. Anteriorly swelling extended from mentalis to tragus area, superiorly 1cm away from angle of mouth and inferiorly upto lower border of mandible.

On palpation swelling was well circumscribed, soft in consistency and indurated. Intraorally swelling extended from 47 region to retromolar trigone (figure 2). No abnormality was detected on Orthopantomogram radiograph (OPG) (Figure 3). Computed tomography (CT) Face/ Orbit Plane (Figure 4), impression revealed a large lytic, expansile lesion in the right mandibular ramus measuring approx 3.7 (CC) x 3.3 (AP) x 2.1 (TR) cm. Thinning of medial cortex with a large exophytic component medially and anteriorly, abutting eth pterygoid muscle and posterior part of right upper alveolar arch was observed. There was a large area of breach in the cortex along its medial surface. Inferiorly the lesion was in close relation with the crown of impacted lower right 3<sup>rd</sup> molar tooth. Based on clinical and radiographic features differential diagnosis of ameloblastoma, dentigerous cyst,

odontogenic keratocyst, leiomyoma was considered. On excisional biopsy H & E stained section revealed well encapsulated lesion (figure 5) with interlacing bundles of collagen fibers along with numerous spindle & plump shaped fibroblasts (figure 6). Spindle shaped smooth muscle cells were observed. Lesional cells exhibited little pleomorphism and were accompanied by giant cells in focal areas with numerous endothelium lined capillaries which showed branching. At few places hyalinized areas were also seen (figure 7). In the immune histochemical staining the lesional tissue was strongly positive for smooth muscle actin (Figure 8). However vimentin & desmin were focally positive in tumor cells (Figure 9) and S-100 was negative. Overall clinical, radiological, histopathological and immunohistochemical findings were suggestive of myofibromatosis.

### Discussion

Myofibromatosis is a benign self-limiting lesion.<sup>5</sup> The literature dealing with myofibroma/myofibromatosis is confusing and lesions have been reported under different terms, such as congenital generalized fibromatosis, congenital mesenchymal hamartomas and infantile myofibromatosis. However, in the current



**Figure 1:** Extraoral photograph revealing mild swelling over the lower 1/3<sup>rd</sup> of the face.

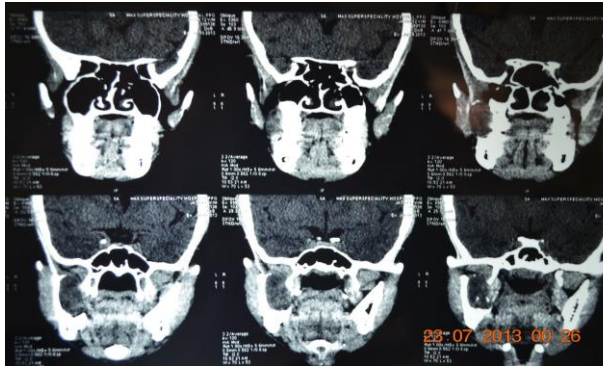


**Figure 2:** Intraoral photograph revealing



**Figure 3:** Panoramic view with impacted 18,28,38,48. No significant findings regarding the lesion.

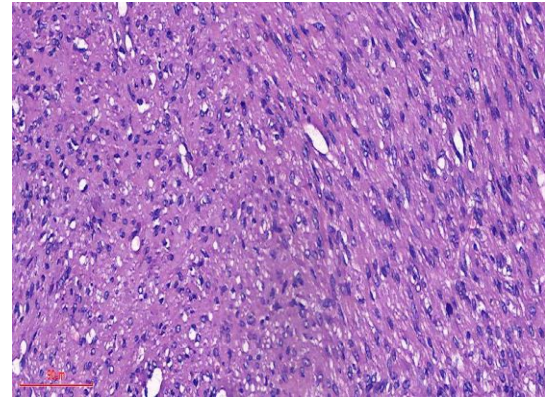




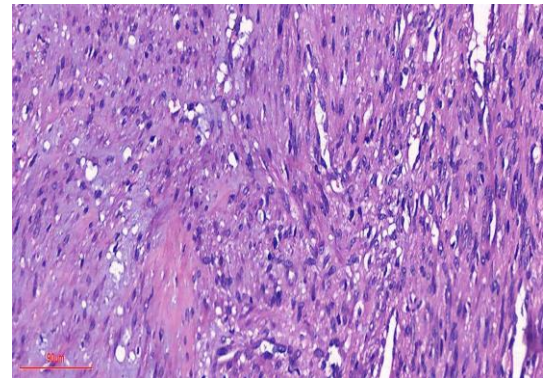
**Figure 4:** CT face/orbit showing large lytic, expansile lesion in the right mandibular ramus measuring approx 3.7 (CC) x 3.3 (AP) x 2.1 (TR) cm. Thinning of medial cortex with a large exophytic component medially and anteriorly, abutting the pterygoid muscle and posterior part of right upper alveolar arch and large area of breach in the cortex along its medial surface. Inferiorly the lesion is in close relation with the crown of impacted lower right 3<sup>rd</sup> molar tooth.



**Figure 5:** H & E stained section Low power view X 100 showing well encapsulated tumor with richly cellular areas.



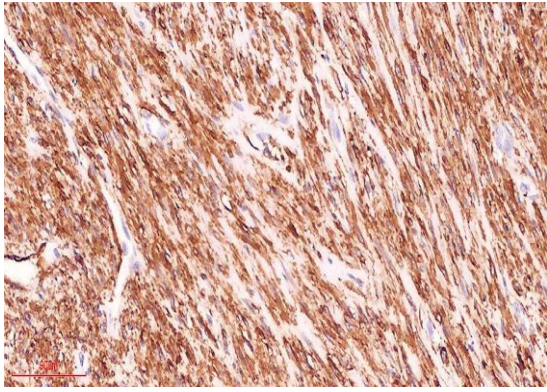
**Figure 6:** H & E stained section High power view X 400 showing plump spindle shaped cells and few areas showing round cells.



**Figure 7:** H & E stained section High power view X 400 showing hyalinized areas in focal areas and numerous spindle shaped cells.



**Figure 8:** Immunohistochemical staining showing tumor cells strongly positive for smooth muscle actin.



**Figure 9:** Immunohistochemical staining focally positive in tumor cells for vimentin.

WHO classification of soft tissue tumors the accepted terminology is myofibroma for solitary lesions and myofibromatosis for multicentric lesions.<sup>6</sup>

Allon et al., in 2007 reported four new cases of myofibroma of the mandible, in addition to the 19 other well-documented cases that had been reported until then.

The 12 cases mentioned in

AFIP review were excluded from their analysis due to lack of sufficient data.<sup>7</sup>

Since then, only four additional cases of myofibroma of mandible have been reported, making our case probably the 40<sup>th</sup> such report documented in medical literature.

The etiology of this disease is not well understood. One of the proposed hypotheses is the upregulation of the estrogen receptors on the fetal smooth muscles, which leaves them more sensitive to maternal estrogen and may induce their

proliferation.<sup>4</sup> Jennings et al (1984) suggested autosomal dominant inheritance with reduced penetration after careful study. On the other hand, Venencie et al in 1987 proposed the hypothesis that it is an autosomal recessive condition.<sup>8</sup> Studies with regard to the specific genetic aberration have been limited, with monosomy 9q, trisomy 16q and del (q12; q15) being the few cytogenetic abnormalities that have been reported.<sup>5</sup> Some authors proposed that it might be due to fetal stimulation by estrogenic hormone, due to proliferation of lesions similar to myofibromatosis in guinea pigs after administration of the hormone.<sup>9</sup> Clinically, lesions present as an asymptomatic jaw swelling and rarely as a soft tissue mass when there is cortical plate perforation.

Common findings of bone lesions on X-ray include well-defined lytic lesions with or without sclerotic borders.<sup>10</sup> In the present case OPG did not show any such findings.

Histologically myofibromatosis show a zoning phenomenon.<sup>10</sup> At low magnification, there is typically a nodular or multinodular growth pattern that appears biphasic owing to the alternation of light- and dark-staining areas. The light-staining areas consist of plump myoid spindle cells with eosinophilic cytoplasm

arranged in nodules, short fascicles, or whorls. The nuclei are elongated and tapering or cigar-shaped and lack nuclear atypia.<sup>11</sup> The dark-staining areas of the lesion, usually centrally located<sup>11</sup>, consists of more primitive appearing, round to polygonal cells with hyperchromatic nuclei and scanty cytoplasm, associated with thin-walled branching vessels, resembling hemangiopericytoma.<sup>12</sup>

Differentiation from fibrous lesions such as nodular fasciitis and fibrous histiocytoma may also be difficult. Nodular fasciitis has a predilection for the head and neck but is composed of plump myofibroblasts set in a loose, often myxoid stroma with little collagen producing the typical feathery pattern.<sup>13</sup> Fibrous histiocytomas have a characteristic storiform pattern, frequently contain giant cells and show an infiltrate of inflammatory cells and foamy histiocytes.<sup>13</sup>

Because of rapid proliferation and features of increased cellularity, rich vascularity and extensive necrosis, these tumors can easily be misdiagnosed as malignant neoplasms. Fibrosarcoma represents the most common misdiagnosis.<sup>14</sup>

Immunohistochemistry can be helpful for diagnosing infantile myofibromatosis (SMA+, CD34± and desmin-).<sup>14</sup> The myofibroblastic phenotype are reactive for

markers such as vimentin and SMA, whereas reactivity for desmin<sup>4</sup> and S-100<sup>1</sup> is variable.<sup>4</sup>

The prognosis of this tumor is mostly excellent in solitary cases.<sup>15</sup> Excision of solitary lesions is usually performed for diagnosis and is typically curative. The recurrence rate is low (10%) and usually successfully treated with re-excision.<sup>16</sup> In aggressive cases, there was limited experience of success with radiation therapy, different combination of chemotherapy, steroid injection and alpha interferon,<sup>2</sup> vincristine, actinomycin – D, methotrexate and 2 cholodeoxyadenosine ,<sup>17</sup> cyclophosphamine.<sup>5</sup>

### Conclusion

Myofibromatosis is usually seen in infants however in our case it was reported in a juvenile. Follow up in these cases is desirable as these lesions are aggressive and show high recurrence rate. Also care should be taken not to confuse these lesions as malignant tumors to which they often resemble.

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