

Immunohistochemistry: An indispensable aid in diagnosis and management of infantile myofibroblastoma

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Abstract

Myofibroma is a benign tumor of myofibroblasts occurring commonly in infants and children. It can occur as a solitary lesion or as multiple lesions in the soft tissues or intraosseously. The presence of the solitary lesion is common in the soft tissues of the head-and-neck region but rare in jawbones. Histologically, it has a biphasic pattern of presentation and mimics spindle cell tumors. Immunohistochemistry is essential for its confirmatory diagnosis. This article describes a case of this rare lesion presenting as a solitary lesion in the left body of the mandible in a 7-month-old boy baby. The diagnostic and therapeutic challenges have been discussed.

Keywords: Immunohistochemistry, infant, intraosseous, mandible, myofibroma, spindle cell

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INTRODUCTION

Myofibroma is a rare, benign nodular tumor of myofibroblastic origin presenting as solitary or multiple nodules. It occurs in newborns and infants and is hence termed as “infantile myofibromatosis.”^[1] Since the routine histopathology characterizes it as a spindle cell tumor, immunohistochemical evaluation for definitive diagnosis and treatment planning is necessary. This article reports one such rare case of infantile myofibroma in a 7-month-old boy baby.

CASE REPORT

A 7-month-old boy baby was brought with the complaint of diffuse swelling on the left lower third of the face that developed gradually over 1-month duration. On examination, it was firm, nontender,

measuring about 2 cm × 3 cm on the left body of the mandible [Figure 1]. The regional lymph nodes were palpable and nontender. Radiograph showed a well-defined unilocular radiolucency in the left body of the mandible [Figure 2]. The histopathology of the excised mass revealed an unencapsulated lesion with biphasic pattern or zoning phenomenon consisting of areas of spindle cells in interlacing bundles/fascicles and whorls [Figure 3a] and round-to-spindle cells arranged around hemangiopericytoma-like multiple slit-like vascular spaces [Figure 3b]. The supporting stroma was focally myxoid with interspersed areas of hyalinization. There was no cellular atypia, but few typical mitotic figures were seen. The spindle cells showed blunt-ended nuclei with prominent nucleoli [Figure 4]. Based on these findings, it was diagnosed as a locally invasive, benign neoplastic lesion with biphasic pattern and myofibroma, leiomyoma,

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neurofibroma, nodular fasciitis and solitary fibrous tumor were considered in the differential diagnosis. The specimen was subjected to immunohistochemistry (IHC) with a panel of markers – S100, desmin, CD34+, α -SMA and vimentin to arrive at confirmatory diagnosis. It was positive to α -SMA and vimentin and negative to S100, desmin and CD34+ [Figure 5]. Based on the immunohistochemical findings a conclusive histopathological diagnosis of myofibroma was given and the final diagnosis of infantile myofibroma of solitary type was arrived at by correlating with the clinical findings. The follow-up of the patient after the surgical curettage for 5 years has been uneventful.

DISCUSSION

Myofibromas can occur in soft tissue, bone or internal organs and affects all ages but common in infants. It has been known by various terminologies since its identification in 1951. However, finally, it was termed as “infantile myofibromatosis” to indicate its occurrence in infants and its myofibroblastic nature.^[1] These lesions are categorized and currently adopted by the WHO as myofibroma and myofibromatosis to describe the solitary and multiple lesions, respectively.^[2]



Figure 1: Clinical photograph showing diffuse swelling on the left lower third of the face, measuring about 2 cm × 3 cm

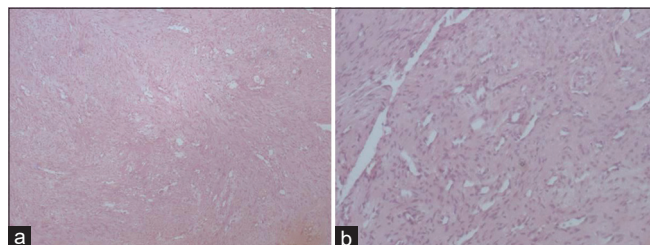


Figure 3: H&E-stained sections showing biphasic pattern or zoning phenomenon. Spindle cells in interlacing bundles or fascicles and whorls (a); round-to-spindle cells arranged around hemangiopericytoma-like multiple slit-like vascular spaces (b)

Most cases of myofibromas are reported as sporadic in occurrence with probable heredity and trauma as etiologic agents.^[3,4] Ninety percent of cases of myofibromas manifest before the age of 2 years, with most cases occurring in the first decade of life and few cases in adults.^[5] The oral lesions typically present as intraosseous lesions in the mandible or as soft-tissue lesions in the lips, cheek and tongue.^[6]

Clinically, lesions of the jaws present as a painless, firm, nodular swelling of varying duration causing cortical expansion and facial asymmetry. At times, the lesion perforates the bone and can be seen as a nodular mass in the mucosa overlying the involved bone.^[7] Radiologically, they present mostly as well-circumscribed unilocular radiolucent lesions with thick clear sclerotic margins without perforation of the cortices of bone.^[8]

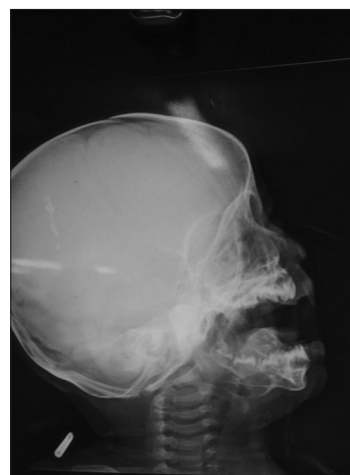


Figure 2: Radiograph showing well-defined unilocular radiolucency in the left body of the mandible

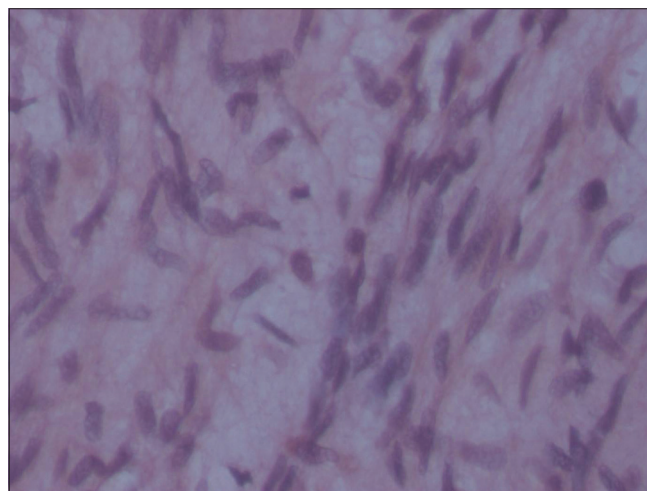


Figure 4: The spindle cells with blunt-ended nuclei and prominent nucleoli ($\times 40$ magnification)

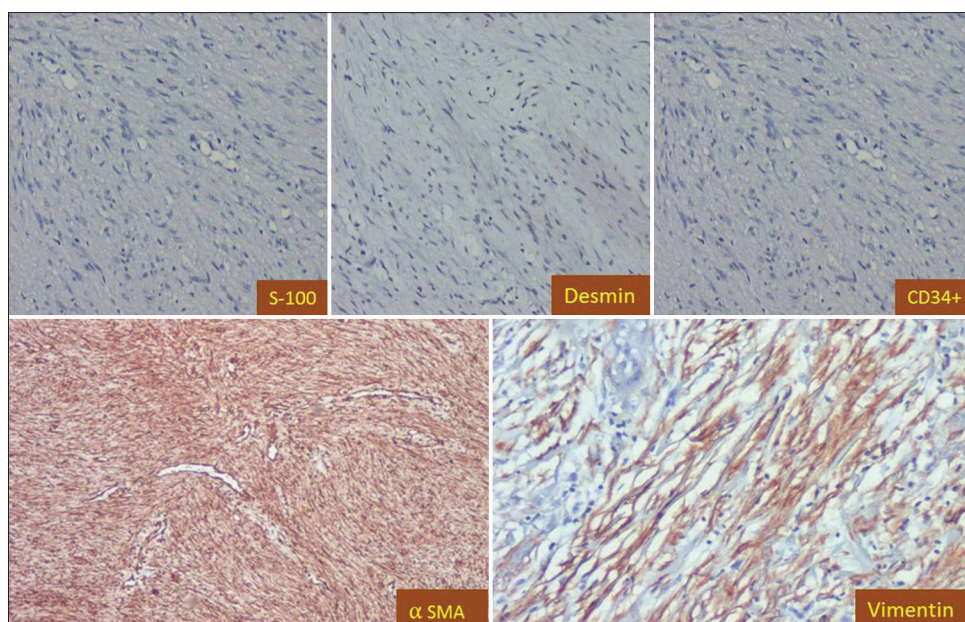


Figure 5: Results of immunohistochemistry with a panel of markers negative for S100, desmin and CD34+ and positive for α -SMA and vimentin ($\times 20$ magnification)

The present case occurred as a solitary growth with a well-defined unilocular radiolucency, in the left body of the mandible, in a 7-month-old boy baby. These findings correlated with the findings in the literature of a solitary lesion in the mandible occurring within 2 years of age and more commonly in males. No specific etiology could be attributed, and there was no perforation of the cortices in the present case.

Histopathologically, the distinct features such as biphasic pattern of arrangement of spindle and round cells and the zoning phenomenon as seen in myofibromas were evident in the present case.^[9] However, since other tumors, such as leiomyoma, neurofibroma, nodular fasciitis and solitary fibrous tumor, mimic the histopathologic findings of myofibroblastoma, they had to be considered in the differential diagnosis.

IHC becomes an invaluable tool in cases like this, to arrive at a final conclusive diagnosis. Analysis of a combination of markers (α -SMA, vimentin, desmin, S100 and CD34+) for positive and negative findings will aid in differentiating between these neoplasms. Very strong positivity to α -SMA in most areas and to vimentin in few areas is noted in myofibromas. Leiomyoma shows positive staining to desmin, neurofibroma to S100 and solitary fibrous tumor to CD34+. Faint positivity to α -SMA is also noted in nodular fasciitis.^[1,9] In our case, the neoplasm showed strong positivity to α -SMA. It was evident even in the walls of the blood vessels. Vimentin was also positive in few areas but was negative for desmin, S100 and CD34+. Based on the

IHC findings in the present case, leiomyoma, neurofibroma, nodular fasciitis and solitary fibrous tumor were ruled out and a definitive diagnosis of myofibroblastoma was made.

Since these lesions occur at young age and do not recur, the treatment of choice would be conservative surgical excision and use of potential antimyofibroblast pharmacological therapeutic agents. An aggressive surgical approach should be avoided as it would require additional reconstructive procedures that could be detrimental to the developmental, functional, esthetic and psychological aspects of the patient.^[10]

Conservative surgical excision of the lesion with a thorough follow-up of 5 years in the present case has been uneventful.

CONCLUSION

IHC is imperative to the definitive diagnosis of myofibroblastoma. The early lesions can be treated by conservative surgical methods, and since they do not have any tendency for recurrence, establishing a confirmatory early diagnosis needs no emphasis in their management. Thus, this article highlights the importance of considering myofibromas, although rare, in the differential diagnosis of central lesions of the mandible, especially in young patients so as to avert unnecessary management of such cases.

Declaration of patient consent

The authors certify that they have obtained all appropriate

patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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