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Solitary myofibroma in children: a report of two cases

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ABSTRACT

Myofibromas are rare benign tumors of myofibroblasts, seen more commonly in children. These tumors typically involve soft tissues with a predilection for the head and neck. Malignant neoplasia is often suspected for these rapidly growing tumors in early childhood. Clinical and radiological findings are not typical, and histopathological examination makes the definitive diagnosis. This pathology requires the intervention of a multidisciplinary team and regular follow-up. We report our experience with two children with myofibromas, one in the right arm of a 5-year-old girl and the other in the right axilla of a 9-year-old boy. Tumors were totally resected in both children, who are under follow-up with no adjuvant treatment free of disease after surgery. Myofibroma should be considered in the differential diagnosis of pediatric soft tissue tumors. Surgical resection is sufficient for treatment and patients should be followed regularly for possible recurrences.

Keywords: Solitary myofibroma, arm, axilla, immunohistochemistry, surgery

Myofibroma is a benign neoplastic proliferation of myofibroblastic cells affecting mainly the early pediatric age group [1]. The tumor may arise in a solitary or multicentric form, with similar histopathological findings, but varied clinical features and prognosis [2]. Myofibromas usually consist of benign nodules in the skin, muscle, or bone. Less often, they can occur in the lungs, heart, gastrointestinal tract, or orbit [3]. The rarity of this lesion may complicate the diagnosis for clinicians and pathologists. Differential diagnosis is important to accurately distinguish neurofibromas and other lesions like benign and locally aggressive vascular tumors and various malignant soft tissue tumors [3]. Here, two cases of solitary myofibroma are presented.

PRESENTATION OF CASES

CASE 1

In November 2020, a five-year-old girl presented with a 10-day history of a painless subcutaneous lump in the upper right arm. She had no history of trauma. Physical examination revealed a firm, slightly tender, immobile palpable mass under the skin proximally in the right arm medial to the humerus measuring approximately 2.5 × 1.5 cm in size with normal color of the overlying skin. Ultrasonography (US) showed a 2.5 × 1.5 × 1.2 cm subcutaneous soft tissue solid lesion with smooth lobulated contours, close to the middle third of the right humerus. Magnetic resonance imaging (MRI) displayed a non-specific fusiform solid

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mass in the middle third of the right arm, abutting the brachial artery, basilic vein, and median nerve; malignancy could not be ruled out (Fig. 1a and b). A trucut biopsy was performed and histopathological examination revealed myofibroma and surgery was planned based on the biopsy result. In the operation, a mass of approximately 3 × 3 cm was removed totally, preserving the vascular and neural structures. Histological examination revealed the diagnosis of myofibroma showing diffuse positivity with SMA (smooth muscle actin) in the tumor cells, which were negative for caldesmon and desmin (Fig. 2); surgical margins were clear. In the postoperative control examination, no palpable mass or limitation of arm movements was detected. Considering the benign features of the tumor, no adjuvant treatment was indicated. Regular follow-up was planned with physical examination and periodical MR imaging. At a follow-up of 14 months, there was no evidence of disease locally or elsewhere.

CASE 2

In June 2021, a 9-year-old boy presented to our hospital with the complaint of a painless nodule in the right axillary region which was noticed by her parents a few days earlier. He had café-au-lait spots spread over his body. The family history was positive for neurofibromatosis type 1 (NF-1). His brother, uncle, and grandmother had documented NF-1, with multiple neurofibromas and café-au-lait spots. The local US revealed a subcutaneous 0.6 × 0.8 cm, slightly echogenic, hypoechoic solid nodular lesion in the right axilla (Fig. 1c); myofibroma and neurofibroma were considered in the differential diagnosis. The patient was consulted by the plastic and reconstructive surgery department, whereupon the lesion was excised totally over the right posterior axillary line. The lesional cells were immunoreactive for SMA and desmin, but negative for S-100 protein, CD34 (Fig. 3). Thus, the pathologic diagnosis of myofibroma was rendered. During a 6-month follow-up, no recurrence was identified either locally or systemically. Parents

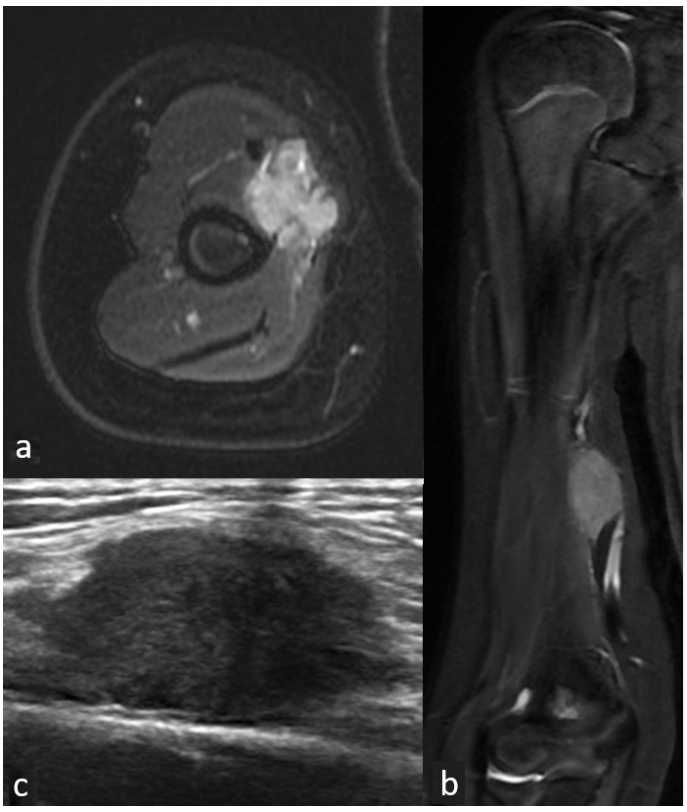

Fig. 1. Magnetic resonance images of case 1. (a) axial fat-saturated T2-weighted image and (b) coronal STIR sequences show an ovoid-shaped hyperintense lesion with lobulated margins in the middle part of the right arm. Ultrasonography of case 2 (c): a well-delineated hypoechoic nodule appeared in the subcutaneous tissue.

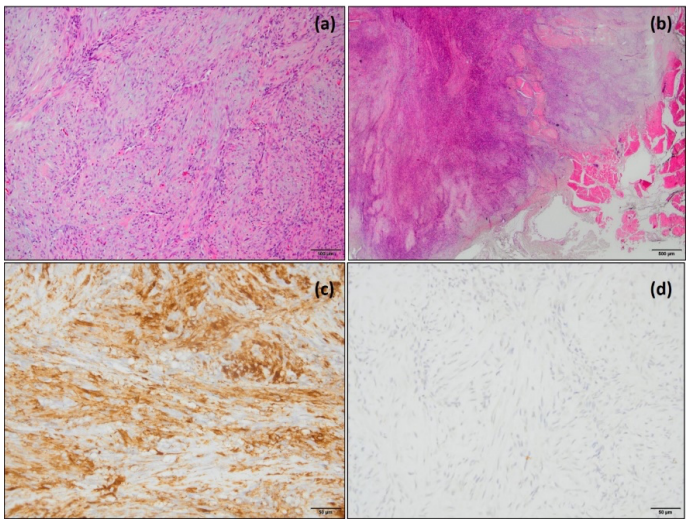

Fig. 2. Histopathological images of case 1. (a and b) well-circumscribed but unencapsulated mesenchymal tumor with intermingled hypocellular and hypercellular areas forming a vague plexiform pattern, benign spindle cell proliferation with hemangiopericytic pattern; (c) SMA showed strong positivity while (d) desmin was negative.

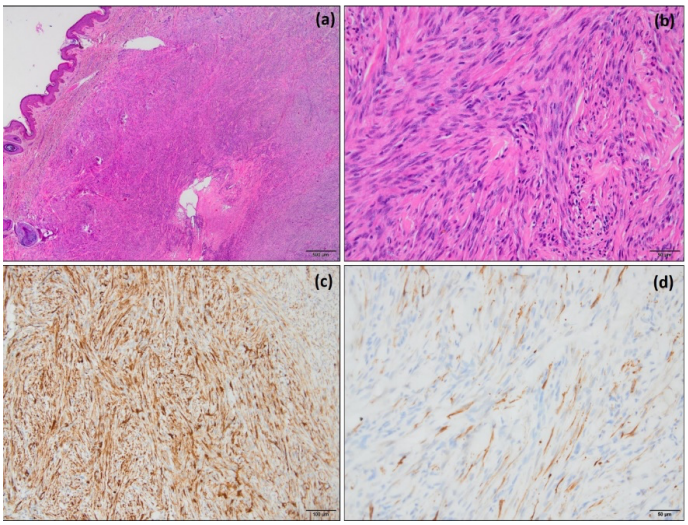

Fig. 3. Histopathological images of case 2. (a) dermal nodule with central degeneration, (b) neoplasm composed of intersecting fascicles of benign spindle mesenchymal cells; (c) SMA was positive in tram-tract (myofibroblastic) pattern, and (d) desmin showed focal positivity.

and patients were informed about the purpose of the case report, and informed consent was obtained from both families for this publication.

DISCUSSION

Myofibroma is a fibrous tumor of childhood and infancy that is characterized by the development of nodular lesions involving the skin, subcutaneous tissue, visceral organs, or bones. It was first described as myofibroblastic proliferation in a newborn in 1951 by Williams and Schrum [3] and was classified according to clinical signs and clinicopathological features by Chung and Enzinger in 1981 [3]. In the WHO 2020 classification of soft tissue tumors, myofibromatosis is classified under the category of pericytic (perivascular) tumors [4].

Myofibroma is classified into three clinical subgroups such as solitary, multiple without or with visceral involvement [5]. In our cases that did not show any evidence of internal organ involvement, the diagnosis was solitary myofibroma. Dhupar *et al*. [6] reported that solitary myofibroma accounts for approximately 70% of cases and Mahajan *et al*. [1] found that 38 (90%) of 42 patients had solitary lesions.

The age of patients ranges from the first few weeks of life to the end of adolescence. In the study by Mahajan *et al*. [1], the median age at diagnosis of 42 patients was 37 months (range; birth-17 years). A series of 114 cases including adults reported by Qudijk *et al*. [7] in 2012 found that two-thirds presented in the first 2 years of life and 91% before the age of 18. Our cases were 5 and 9 years old, respectively, both within the age range reported in the literature.

The etiology of myofibroma is not well known. Some investigators reported that patients with familial inheritance and inherited genes with autosomal or genetic heterogeneity chromosomes are more susceptible to myofibroma formation [6]. Trauma, injury, and the effects of estrogen are other proposed hypotheses [6]. In our cases, there was no family history of myofibroma or history of trauma.

Both benign and malignant tumors can develop in patients with NF-1, neurofibromas being the most common. Malignant peripheral nerve sheath tumors (MPNSTs) can develop within an associated plexiform neurofibroma and have an extremely poor prognosis with widespread recurrence and distant metastases [8]. The brother of our second case was diagnosed with NF-1 and MPNST had developed based on NF1. The myofibromas described in our cases were benign, and no association of myofibroma with NF1 was found in the literature.

Myofibroma typically develops in the soft tissues of the head, neck, and trunk. However, extremities or the skeleton may also be affected. In our cases, myofibroma localizations were upper extremity and axilla, both of which are rare localizations. In the literature, Qudijk *et al*. [7] reported that myofibromas were located in the arm in four patients and the axilla in a single patient. In the study of Weiliang Wu *et al*. [2], bone myofibromas in the upper extremities were reported in two cases. In addition, Mahajan *et al*. [1] identified 10 of 38 patients had solitary lesions in their extremities.

The presenting symptoms usually reflect the location of the tumor and mainly include swelling or an enlarging mass or nodüle; lesions are typically painless. Hemangioma-like discoloration, skin atrophy, and ulceration have also been described [3]. Similar to the literature, our cases presented to the clinic with painless masses. The imaging features of myofibromas are not specific and in the US masses may show a hypoechoic or anechoic center. In our cases, well-con-

toured, solid nodular lesions, hypoechoic in the US, were revealed [2].

Myofibroma shows a characteristic histopathological bi-phasic pattern composed of elongated spindle cells at the periphery and polygonal cells with hyperchromatic nuclei at the center6. Tumor cells have characteristic eosinophilic cytoplasm when stained with hematoxylin-eosin, with no atypical pleomorphic or malignant features [2]. Immunohistochemical staining may be helpful in diagnosis. Myofibromas are benign tumors with a characteristic histological and immunohistochemical pattern, displaying SMA- and, to a lesser extent, SMA-positivity, with a low rate of desmin expression7. SMA and desmin were positive at immunohistological staining in our cases (Figs. 2 and 3).

The differential diagnosis of myofibroma is extensive, including nodular fasciitis, neurofibroma, fibrous histiocytoma, desmoid tumor, lipofibromatosis, infantile myofibroelastic tumor, and congenital infantile fibrosarcoma. The histology of myofibroma is typical in most cases and immunohistochemistry along with clinical findings may help in diagnosis. Nodular fasciitis is rare in children and is an important differential diagnosis in the adult age group. The histology of nodular fasciitis shows a more prominent myxoid matrix, a tissue culture-like growth pattern, often scattered chronic inflammatory cells, and some erythrocytes. The hemangiopericytoma-like vascular pattern typical of myofibroma is not seen in nodular fasciitis. The peripheral areas of myofibroma may resemble neurofibroma, but the myofibroblastic cells lack S-100 protein and do not have the typical buckled nuclei of neuronal cells. A fibrous histiocytoma consists of cells arranged in a stratiform arrangement and may be SMA positive, such as myofibroma. Generally, fibrous histiocytoma cells express factor XIIIa and CD68. Inflammatory myofibroblastic tumors are characterized by a proliferation of myofibroblastic spindled cells and an inflammatory, predominantly lymphoplasmacytic infiltrate. Inflammatory myofibroblastic tumors can often show rearrangements involving the ALK gene in the 2p23 chromosomal region. Congenital infantile fibrosarcoma is included in the differential diagnosis and can be differentiated from myofibroma by defining the t (12,15) translocation. In general, the identification of myoid cells in the periphery of the lesion is a useful feature for myofibroma [7].

The recommended treatment for myofibroma is total surgical excision9, which was performed in our cases with negative margins resulting in local control. In cases with difficulty in total excision, conservative debulking and then close observation can be planned. In the study consisting of 38 cases with solitary lesions, 12 patients underwent complete and 26 had incomplete resections [1]. In myofibroma with visceral involvement, cytotoxic chemotherapy agents or radiotherapy can be used. However, there is no evidence in the literature of the overall success of these treatments [10]. Recurrences have also been reported, the recurrence rate being 7%-10% in the literature [3]. In a study published in 2017, the median follow-up was 50.5 months, and only 1 in 42 patients developed tumor progression1. In our cases, although follow-up is short, recurrence did not occur 6- and 15-months after surgery, respectively.

The most common site of involvement of myofibroma is the head and neck, followed by the trunk. On the other hand, myofibromas located in the upper extremity and axilla in our cases were unusual in terms of their localizations. Radiological imaging can define lesion boundaries. Histological analysis is required for a definitive diagnosis. The primary management modality is usually excision of the lesion. This pathological condition requires the intervention and regular follow-up of a multidisciplinary team.

CONCLUSION

In conclusion, although myofibroma is rare, it should be considered in the differential diagnosis of pediatric soft tissue tumors. Surgical resection is sufficient for the more common solitary type, no additional treatment is indicated in most cases. The overall prognosis is favorable even in tumors resected with positive margins. Patients should be followed regularly for possible recurrences.

Patients' Consent

Parents and patients were informed about the purpose of the case report, and informed consent was obtained from both families for this publication.

Authors' Contribution

Study Conception: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY; Study Design: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY; Supervision: KK, MA, IV, ÜA, HNÖ, AV, BY; Funding:N/A; Materials: N/A; Data Collection and/or Processing: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY; Statistical Analysis and/or Data Interpretation: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY; Literature Review: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY; Manuscript Preparation: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY and Critical Review: ÇC, KK, MA, IV, ÜA, HNÖ, AV, BY.

Conflict of interest

The authors disclosed no conflict of interest during the preparation or publication of this manuscript.

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