Solitary orbital myofibroma in a child: A rare case report with literature review

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Myofibroma is a rare benign mesenchymal tumor of uncertain histogenesis. A six-year-old boy presented with a unilateral lower eyelid mass of six weeks' duration. MRI revealed a circumscribed mass in the inferolateral orbit with bony erosion. A systemic examination was unremarkable. Excision with histopathology revealed a partially infiltrative spindle cell tumor with bland nuclear morphology expressing smooth muscle actin and muscle-specific actin, compatible with myofibroma. Solitary myofibroma is a rare childhood orbital tumor and may clinico-radiologically closely mimic a malignancy. Histopathology and immunohistochemistry can help reach a definitive diagnosis. Systemic evaluation and close follow up are crucial in such cases.

Key words: Benign, bone erosion, children, myofibromatosis, orbital myofibroma

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Myofibromas are benign soft tissue neoplasms, and were previously classified as fibroblastic/myofibroblastic in origin. WHO classification of tumors of the soft tissue in 2013, reclassified them as pericytic tumors.[1] They commonly occur in early infancy, and are clinically classified as solitary, multicentric without visceral involvement and multicentric with visceral involvement (generalized).[2,3] Though labeled as the commonest fibrous tissue tumor of infancy,[1,2] they are still quite rare. The term "myofibroma" is used to describe the solitary form and is the commonest form of presentation, whereas "myofibromatosis" denotes the multicentric form.[2] The most common site is the head and neck regions followed by limbs and trunk.[4]

Orbital myofibromas are extremely rare. Herein, we report a case of an orbital myofibroma presenting in a six-year-old child and a review of literature of pediatric orbital myofibromas.

Case Report

A six-year-old male child presented with a painless progressive swelling over the lateral left lower eyelid of about six weeks' duration. There was no other associated ocular or systemic complaint.

On examination, a well-defined, firm, nodular, non-tender soft tissue mass was palpated in the lower eyelid at the lateral edge of the inferior orbital rim [Fig. 1a and b]. The lower fornix and overlying skin were uninvolved. There was no globe displacement, proptosis or ocular motility restriction. Visual acuity in both eyes, and the ocular examination was normal. A detailed systemic clinical examination was unremarkable. A chest radiograph and ultrasonography of the abdomen and the pelvis were both within normal limits. On MRI of the orbits, the mass appeared hypointense on T1W images [Fig. 1c, red arrow] and lay adjacent to the left zygomatic bone with changes suggestive of underlying bony erosion. On T2W images, it was isointense, relatively homogeneous, and well circumscribed [Fig. 1d, yellow arrow].

An excision biopsy was performed. Intraoperatively, the mass appeared pinkish and firm. The underlying bony orbital rim was eroded [Fig. 2a, yellow arrow]. The mass was excised from its bony attachment with a ragged base [Fig. 2a]. Clear surgical margins were not obtained. The eroded bony base was curetted. Intra-operative squash and imprint cytology demonstrated bland appearing spindle cells suggestive of a benign spindle cell tumor.

Gross examination showed a partially circumscribed mass, with a pseudocapsule surrounding three-quarters of the tumor [Fig. 2b, red arrow]. One-quarter displayed irregular edges [Fig. 2b; yellow arrow]. The tumor comprised predominantly of variably sized spindle cells in fascicles, and whorls intersecting at places [Fig. 3a and b]. It displayed a rich vascularity [Fig. 3c] with thin-walled slit-like branching vessels imparting a staghorn appearance [Fig. 3c, black arrow]. Individual cells showed ill-defined cell membranes, abundant eosinophilic cytoplasm, bland, elongated, oval to round nuclei with uniform nuclear morphology. Scattered pale myxoid foci were noted around vessels. Mitotic count was 7 per 20 HPF. There was no necrosis, cytological atypia, or inflammation. A fragment of curetted bony tissue showed spicules of lamellar bone encircled by tumor cells with bland nuclear morphology [Fig. 3d, yellow asterix].

Tumor cells showed strong cytoplasmic expression of vimentin [Fig. 4a], smooth muscle actin [SMA, Fig. 4b], and muscle-specific antigen (MSA), and were negative for desmin, ALK-1, and S-100. CD34 decorated the vessels [Fig. 4c], but not the tumor cells. Ki-67 showed a labeling index of 1–2% [Fig. 4d]. The morphological and immunohistochemical features were consistent with myofibroma. A systemic evaluation was unremarkable, and a diagnosis of solitary orbital myofibroma was made.

Discussion

Myofibroma has a diverse clinical presentation varying from aggressive multicentric to benign localized forms. It is rarely self-regressing and was historically identified by several names, such as infantile myofibromatosis (IM), congenital-infantile hemangiopericytoma (CIH), and congenital-infantile fibrosarcoma (CIF). A systematic review of similar lesions had led to the introduction of the term juvenile fibromatoses. The disease was described to have solitary, multiple, and generalized forms.[5] Subsequently, the skin tumors and tumors of soft tissue and bone working groups advocated usage of the terms "myofibroma", "solitary myofibroma", or "solitary cutaneous myofibroma" to denote solitary lesions of IM and the term "myofibromatosis" for the multicentric forms.[6]

Solitary and multicentric forms can involve the skin, subcutaneous tissue, muscle, and bone. In solitary myofibroma, visceral involvement is very rare.

Multicentric forms with visceral involvement can involve heart wall, pulmonary parenchyma, pleura, thyroid gland, adrenal gland, kidney, pancreas, gastrointestinal tract, mesentery, liver, and rarely the central nervous system. Multicentric forms may demonstrate familial inheritance, present earlier in life, and the number of lesions may widely vary (even upto 100). Mortality due to mass effect and visceral involvement, recurrence, and spontaneous regression on observation has been reported in multicentric forms.

Orbital myofibromas are a group of rare, benign, but locally infiltrative tumors of infants with only a few case reports described in children.[3] Kodsi *et al*. reported only a single case of myofibroma in a review of 340 orbital tumors in children accumulated over a period of sixty years.[5]

A review of English literature revealed 25 cases of orbital myofibroma [Table 1] of which one case had limited information and was excluded from tabulation. There was male preponderance with a M:F ratio of 2.12:1.00. One-third of the cases were congenital, with 60% presenting at the age of less than two years. In total, 72% of the cases had involvement on the left side.

The commonest presentation was a gradually progressive unilateral (100%) orbital mass with soft tissue and bony involvement [47.8%, Table 1].[3,7-25] The presence of bony erosion can make the clinicoradiological diagnosis of myofibroma more challenging. None of the orbital myofibromas demonstrated recurrence at a mean follow up of 20 months (range 6–84 months).

The major differential diagnoses include fibrous histiocytoma, nodular fasciitis, fibromatosis, infantile

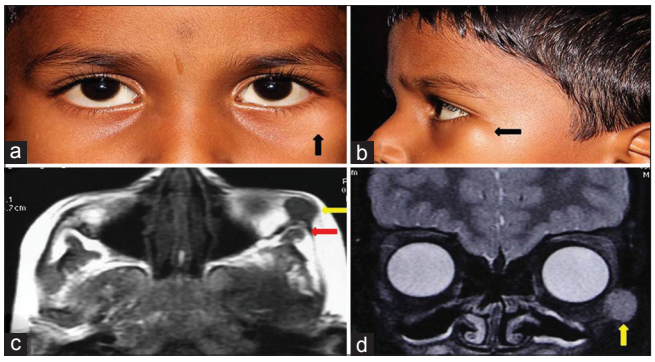

Figure 1: Clinical and radiological features: Well-defined, firm, non-tender soft tissue mass present on the inferolateral orbital rim (black arrow, a, b). Magnetic resonance imaging shows a well-circumscribed soft tissue mass noted to be hypointense on the T1W image (yellow arrow, c) and changes evident in the adjacent zygomatic bone (red arrow, c). The mass shows an increase in signal intensity on the T2W image (yellow arrow, d)

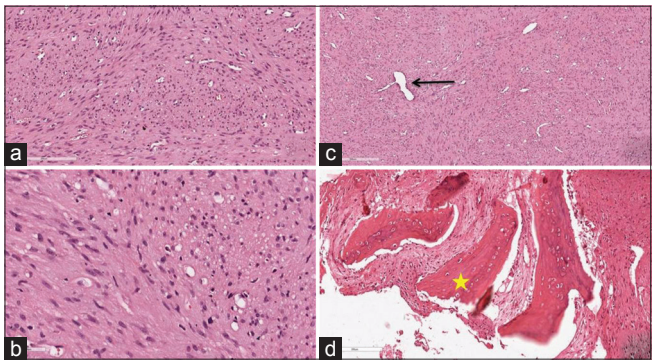

Figure 3: Morphology of Myofibroma: (a) Spindle cell tumor, cells arranged in a fascicular and whorled pattern (a: 10×). Individual cells are spindly with ill-defined cell membrane, abundant eosinophilic fibrillary cytoplasm, bland, elongated, oval, spindly to round nuclei with uniform nuclear morphology (b: 40×). Tumor is richly vascular with thin-walled slit-like to branching vessels imparting a staghorn appearance (c: 6×, black arrow marked). Tumor cells with bland nuclear morphology are seen surrounding fragments of cancellous bone (d, asterix marked, 10×; Haematoxylin and Eosin stain)

fibrosarcoma, solitary fibrous tumor, neurofibroma, and hemangiopericytoma.[3,9] Rarely, malignant lesions like Ewings sarcoma family of tumors and rhabdomyosarcoma may need exclusion. Fibrous dysplasia and non-ossifying fibroma may also need exclusion in cases with predominant osseous involvement. In recent years, studies have revealed consistent pathological findings with a typical fascicular pattern of a rich vascular spindle cell tumor with occasional staghorn-like channels. The centre of the lesion might display more cellularity. Review of published literature showed that all the cases in which IHC was performed (15/25) expressed SMA [Table 1]. These tumors were also found to have strong expression of vimentin, muscle-specific actin, and were negative for desmin, CD34, ALK-1 and S-100.

Though recurrences in myofibroma are extremely rare in spite of positive surgical margins,[4] complete surgical excision should still be the goal.[12] Conservative debulking followed by close observation can be considered in cases with difficulty

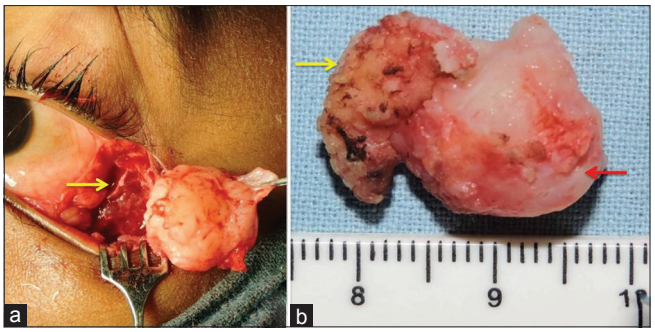

Figure 2: Intra-operative and Gross tumor morphology: intra-operatively, the portion of the zygomatic bone underlying the lesion was eroded (yellow arrow, a). On gross examination, the mass was found to be partially circumscribed (red arrow, b) with an irregular base that was abutting the underlying bone (yellow arrow, b)

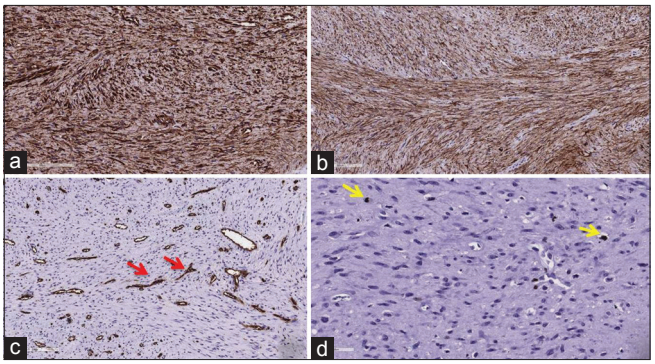

Figure 4: Immunohistochemical staining of Myofibroma: tumor cells strongly express Vimentin (a) and SMA (b); CD 34 decorated the vessel walls (c), but was not expressed in tumor cells. Ki -67 shows 1–2% (d), yellow arrow marked) of proliferative activity

in total excision. There is no conclusive evidence supporting the benefit of adjunctive radiotherapy or chemotherapy in solitary orbital myofibromas.[11] Therefore, a regular follow-up is probably the best-recommended policy.

Conclusion

In conclusion, myofibroma can rarely present in children as a progressive orbital mass with bony erosions simulating a malignant tumor. A conservative/complete excision may cure the tumor with a very low rate of recurrence. A definitive diagnosis and differentiation from other tumors are dependent upon the microscopic findings and immunohistochemistry.

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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.

Table 1: Review of cases of pediatric orbital myofibroma

Author, Yr.	Age (m); Sex	Presenting complaints	Duration of complaints (m)	Site of involvement/Laterality	Other sites	CT/MRI	Tt	FU (yrs)	Outcome, recurrence
Wiswell TE *et al*., 1985	neonate; F	Mass	Birth	LE Lower eyelid/UL	Upper lip, nose on left side	NM	NM	NM	NM
Waeltermann JM *et al*., 1988	infant; M	Proptosis	Birth	LE Orbit/UL	Intracranial extension	Orbital mass with intracranial extension	Incision and close follow up	NM	NM
Nasr AM *et al*., 1986	5m; M	Proptosis	Birth	RE Superolateral orbit/UL	Cranial cavity, Soft tissue-ear, axilla, buttock	Well defined homogeneous intraorbital mass with bone erosion	Excision	0.5	Good, Nil
Stautz CC *et al*., 1991	neonate; M	Proptosis	Birth	LE Orbit/UL	Cranium	Ill defined homogeneous hyperdense mass in orbit with SOF dilatation and intracranial extension	Incision and close follow up	4	Stable, Nil
Campbell RJ *et al*., 1991	30m; M	Ptosis and inferior displacement of globe	6	RE Superolateral orbit/UL	NM	Well defined homogeneous enhancing mass, sclerosis and bone remodelling	Excision	NM	Good, Nil
Linder JS *et al*., 1996	<1m; M	Lower eyelid and medial canthal mass	Birth	LE Lower eyelid and medial canthus/UL	Not involved	NM	Subtotal resection	1.5	Stable, Nil
Duffy M T *et al*., 1997	48m; F	Lower eyelid mass	<1	RE Inferolateral orbit/UL	Not involved	Well defined homogeneous enhancing mass, loss of bone and surrounding hyperostosis	Excision	0.5	Good, Nil
Shields CL *et al*., 1998	3m; F	Proptosis	2	LE Sphenoid bone/UL	Not involved	Well defined intraosseous mass with lytic lesion	Subtotal resection	NM	Good, NM
Tokano H *et al*., 2001	120m; M	Not mentioned	NM	LE Lateral orbital floor/UL	NM	Orbital mass with bony destruction	Incision	0.5	Good, Nil
Westfall AC *et al*., 2003 (2 cases)	neonate; M	Lower eyelid mass	Birth	LE Lower eyelid/UL	Not involved	Heterogeneous mass with variable soft tissue densities and small areas of calcification. There was no extension of the tumour into the orbit	Excision	7	Good, Nil
	72m; M	Upper eyelid mass	1	LE Superonasal orbit/UL	Not involved	Homogeneous well defined isodense mass in superonasal orbit, no bone changes	Incision	1	Stable, Nil

Contd...

Table 1: Contd...

Author, Yr.	Age (m); Sex	Presenting complaints	Duration of complaints (m)	Site of involvement/Laterality	Other sites	CT/MRI	Tt	FU (yrs)	Outcome, recurrence
Larsen AC *et al*., 2003	12m; F	Proptosis and swelling of eyelid	<1	LE Superolateral orbit/ UL	Not involved	Well defined homogeneous	Excision	NM	Good, Nil
Cruz AA *et al*., 2004	6m; M	Not mentioned	5	LE Superolateral orbit/ UL	Head and neck	Well defined lesion with erosion of superolateral bony rim	Excision	0.5	Good, Nil
Nam DH *et al*., 2005	36m; M	Lower eyelid mass	2	LE Inferolateral orbit/ UL	Not involved	Well defined homogeneous mass with bone erosion and hyperostosis	Excision	1.3	Good, Nil
Koujok *et al*., 2005	<1m; M	Not mentioned	NM	LE Infraorbital region/ UL	Lumbosacral plexus neuropathy, left psoas, left iliac bone, left forearm	NM	Incision	NM	NM
Persaud TO *et al*., 2006	29m; M	Proptosis	<1	LE Superior orbit- Greater wing of sphenoid/UL	Middle cranial fossa, adherent to dura	Well defined homogeneous mass with bone erosion and hyperostosis	Excision	NM	Good, Nil
Rodrigues EB *et al*., 2006 (4 cases)	72m; M	Lower eyelid fullness	2	RE Orbital floor/UL	Not involved	Well defined homogeneous Intraosseous mass with thinned bony margins	Excision + bone removal	3	Good, Nil
	11m; M	Proptosis	9	LE Superotemporal intraosseous mass/UL	Not involved	Well defined superotemporal intraosseous mass with bone destruction	Excision	0.5	Good, Nil
	7m; F	Lower eyelid mass	NM	RE Orbit, maxillary and zygomatic bone/UL	Not involved	Well defined mass with bone infiltration and erosion	Excision	3	Good, Nil
	3m; F	Proptosis	2	LE Orbit, maxillary and zygomatic bone/UL	Not involved	Left sphenoid bone with osteolytic lesion	Excision	3	Good, Nil
Galassi E *et al*., 2008	17m; F	Strabismus and ptosis	1	RE Eyelid/UL	Ethmoid sinus, maxillary sinus, anterior skull base with intracranial extension	Inhomogeneously enhancing mass with partial calcification	Excision	0.5	Good, Nil
Mynatt CJ *et al*., 2011	36m; M	Mass	3	LE Intraosseous mass of superolateral margin of orbit/UL	Not involved	Osteolytic expansile intraosseous lesion	Bone curettage	1	Stable, Nil

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Table 1: Contd...

Author, Yr.	Age (m); Sex	Presenting complaints	Duration of complaints (m)	Site of involvement/Laterality	Other sites	CT/MRI	Tt	FU (yrs)	Outcome, recurrence
Bloom RI *et al*., 2013	neonate; F	Proptosis	Birth	RE Retrobulbar mass/UL	Not involved	Retrobulbar mass with mass effect on frontal bone	Debulking	2	Stable, Nil
Bahram Eshraghi *et al*., 2017	60m; M	mass	1.5	LE Superolateral orbit/ UL	Not involved	Well defined isodense homogeneous mass with bone erosion	Excision	1	Good, Nil
Present case, 2018	72m; M	Lower eyelid mass	1.5	LE Inferolateral orbit-zygomatic bone/ UL	Not involved	Well defined homogeneous mass with bone erosion	Excision	0.5	Good, Nil

Yr- year; m-month; F/M- female/male; LE- left eye; RE- right eye; UL- unilateral; Tt- treatment; FU- follow up; NM- not mentioned

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Conflicts of interest

There are no conflicts of interest.

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