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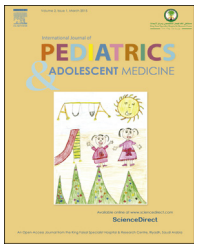

IMAGES IN PEDIATRICS

Infantile orbital myofibroma

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1. Case summary for images in pediatrics

A one-month old baby girl was referred for pediatric oculoplastic assessment for a small right supero-medial angle orbital swelling, without inflammation, which was observed at a postnatal visit (Fig. 1). Ultrasonography and MRI of the head was performed, and these results confirmed a superficial well-circumscribed, cystic orbital lesion and an

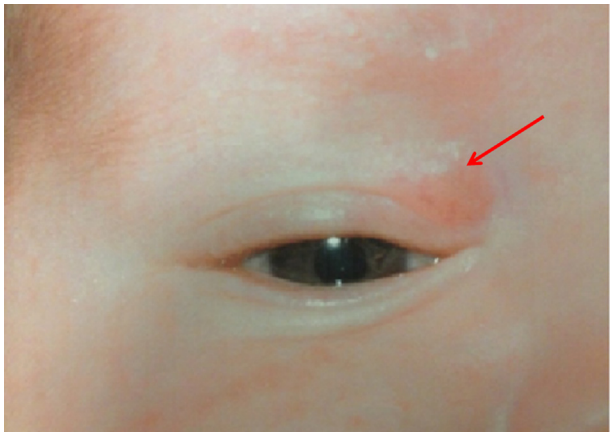

Figure 1 One-month old baby girl with a right upper supero-medial angle cystic lesion.

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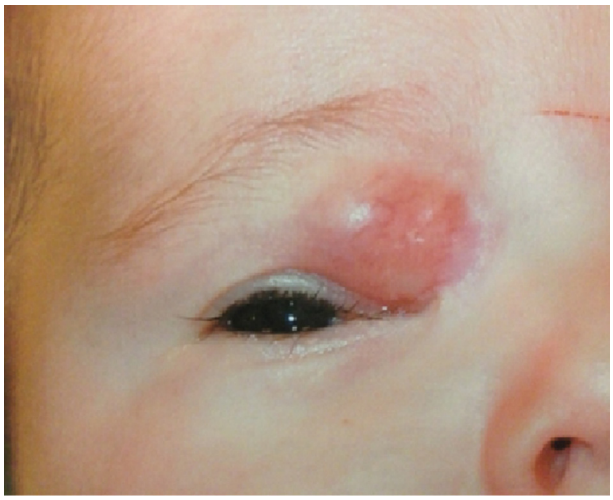

Figure 2 Three-month old baby with a dramatic increase in the supero-medial lesion, with inflammation, lid distortion and threatened occlusion of the visual axis.

absence of a trans-ethmoidal meningocele. The decision was made to observe the patient. At follow-up, at 3 months old, the lesion had dramatically enlarged, with associated inflammation and mechanical deformation of the upper lid, threatening the visual axis (Fig. 2). The rest of her examination was normal with no other swellings. Due to the lesion's increasing size, the presence of inflammation and the threat of visual axis occlusion as well as consequent poor visual development (occlusion amblyopia), we elected to excise the lesion. Orbital exploration and surgical resection were performed, and a solid white tumor was extracted. The mass was superficial to the pulley sheath of the superior oblique muscle. Intra-operative frozen-section histopathology revealed a pattern consistent with a myofibromatous tumor. Subsequent histopathology and immunohistochemistry confirmed this diagnosis, making rhabdomyosarcoma unlikely. The tissue was very vascular with a biphasic cellular pattern in a myxoid matrix, consisting of multi-nodular fascicles of eosinophilic spindles or 'myoid'cells with elongated nuclei amongst the clusters of small round cells with large round hyperchromatic nuclei. Pseudorosettes were observed amongst the smaller cells, and both cell populations exhibited rare mitoses or necrosis (Fig. 3a &b). The highly vascular nature and spindle-shaped cellular clusters were suggestive of hemangiopericytoma. Immunohistochemistry studies revealed diffuse positive smooth muscle actin (Fig. 3c) and focal desmin stains. Other cellular markers (pancytokeratin, CD68, protein S100, CD34, ALK1, HMGA2, caldesmon and myogenin) were negative. The histological pattern was compatible with orbital myofibroma. Genetic testing for the ETV6 gene mutation observed in malignant infantile fibrosarcoma was negative, confirming the sporadic form of this clinical manifestation.

Infantile orbital myofibroma is a rare but well-recognized, benign, locally infiltrative tumor. Commonly manifesting in the head and neck regions, myofibroma may also occur in other parts of the body, affecting skin, bone and viscera [1,2]. The isolated form is the most common and when it manifests simultaneously at multiple sites, it is

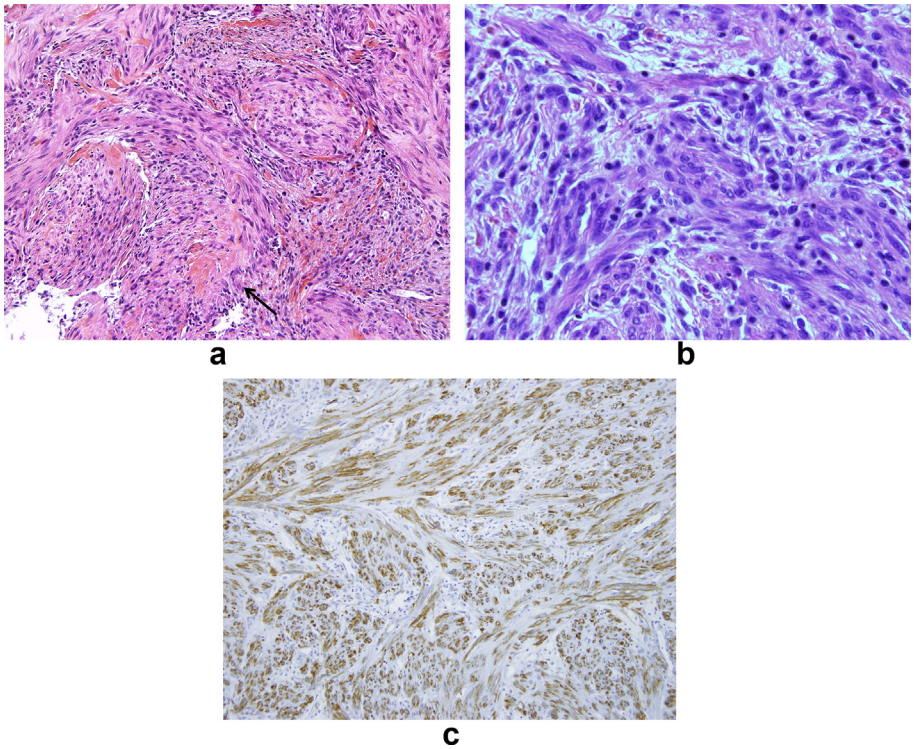

Figure 3 Histopathology of a biopsy specimen: H&E – a (20 ×) & b (40 ×) Robust cellular vascular tissue in the myxoid matrix with a biphasic cellular pattern consisting of fascicles of eosinophilic spindle-shaped 'myoid' cells and small hyperchromatic cells with pseudorosettes (arrow) and rare mitoses. Immunohistochemistry showed diffuse staining for smooth muscle actin c (20×).

referred to as myofibromatosis [3]. The generalized variety is associated with higher morbidity and mortality [2].

Cases of spontaneous involution of infantile orbital myofibroma have been reported [2]. However, this patient's clinical presentation suggested a malignant tumor, and the rarity and lack of specific findings for orbital myofibroma make clinical diagnosis difficult. Furthermore, with such precipitous growth, excision biopsy was essential not only to establish the diagnosis but also to clear the visual axis. Frozen section and basic histology alone are usually insufficient to distinguish myofibroma from specific malignant orbital tumors, such as rhabdomyosarcoma; additional immunohistochemistry and genetic testing afford some assurance of benign myofibroma [4,5]. Although orbital myofibroma are usually isolated, periodic complete pediatric surveillance for multiple myofibromatosis is still required and will be performed. However, despite suspicious initial growth, our patient likely has a good prognosis and will not require further surgery.

Declarations

Drs. Mbekeani and Nezzar and Prof. Kemeny have no financial interests or funding to disclose.

Conflict of Interest

The authors have no conflicts of interest to report.

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