Infantile solitary orbital myofibroma: A Case Report

Ligang Jiang[1], Wencan Wu[2], and Fangzheng Jiang[1]

[1]Quzhou People's Hospital
[2]Wenzhou Medical College Eye Hospital

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1 INTRODUCTION

Infantile myofibroma is a rare benign tumor of myofibroblasts, primarily affecting infants under the age of two, with a notable prevalence in newborns (54%)[1]. These tumors typically manifest in the head and neck region, presenting a wide clinical spectrum ranging from spontaneous regression to severe complications, including multi-organ involvement and mortality. Due to its diverse clinical presentation, infantile myofibroma is often misdiagnosed. The tumors are most frequently located in the scalp, forehead, parotid region, and oral cavity.

Infantile myofibromas are classified into solitary and multicentric forms, with solitary lesions accounting for approximately 74% of cases. Solitary myofibromas predominantly occur in the skin, muscle, subcutaneous tissue, and bone, gastrointestinal and laryngeal involvement is rare while orbital involvement is uncommon. The histological characteristics of orbital myofibroma typically include tumor cells composed of spindle-shaped fibroblasts and myofibroblasts, which grow in an infiltrative manner[2].

This case report discusses a solitary orbital myofibroma found in a 3-year-old child. Initially, the clinical signs, imaging studies, and frozen section suggested a diagnosis of an orbital nerve sheath tumor. However, subsequent immunohistochemical analysis revealed that the correct diagnosis was indeed infantile solitary orbital myofibroma.

2 CASE HISTORY/EXAMINATION

We present a case of a 3-year-old male patient who presented with progressive swelling of the left lower eyelid over the past month. The child reported no redness, pain, diplopia, or ocular motility disturbances, and there was no significant family or past medical history.

Ocular examination revealed visual acuity of 0.5 in the right eye and 0.6 in the left eye. There was noticeable swelling in the right facial area and lower eyelid, with a palpable, firm mass that was mobile and non-tender. The position of both eyes was normal, and ocular motility was intact. Anterior segment examination and fundoscopy showed no abnormalities.

Ultrasound examination (Figure 1) indicated a mass in the anterior wall of the right maxillary sinus, raising the suspicion of a neurogenic tumor. A coronal CT scan of the paranasal sinuses (Figure 2) revealed a mass in the area of the right maxillary sinus anterior wall, with differential diagnoses including an infraorbital nerve sheath tumor or a hemangioma. An enhanced orbital MRI (Figure 3) confirmed the presence of a mass in the same region, suggesting a neurogenic tumor, most likely a nerve sheath tumor.

3 DIFFERENTIAL DIAGNOSIS, INVESTIGATIONS, AND TREATMENT

Based on the clinical findings and imaging results, with no history of ocular trauma or surgery, the patient was tentatively diagnosed with a right-sided orbital nerve sheath tumor, and preparations for surgical intervention were initiated. Pre-operative evaluations were conducted to exclude any contraindications for surgery.

The patient underwent an extensive excision of the orbital mass via a transcutaneous approach to the right lower eyelid under general anesthesia, along with an artificial bone grafting procedure. Intraoperative frozen section analysis (Figure 4、Figure 5a) identified the mass as a spindle cell tumor, primarily considering a nerve sheath tumor, with definitive diagnosis pending routine histological and immunohistochemical evaluation.

4 OUTCOME AND FOLLOW-UP

Postoperatively, immunohistochemical analysis revealing positive staining for smooth muscle actin (SMA) and calponin, confirming the diagnosis of infantile solitary orbital myofibroma not orbital nerve sheath tumor(Figure 5b).Fortunately,the child recovered well, and at the one-month follow-up, he reported no ocular discomfort, with no signs of systemic or visceral involvement.

5 DISCUSSION

Differentiating infantile solitary orbital myofibroma from orbital nerve sheath tumor can be particularly challenging, as both tumors share several clinical, radiological, and histopathological similarities, making misdiagnosis possible without thorough examination.

In our case, we reported a 3-year-old child who presented with right lower eyelid swelling for one month. Initial imaging studies, including ocular ultrasound, orbital CT, and MRI, strongly suggested a diagnosis of nerve sheath tumor. Subsequent surgical treatment and intraoperative frozen section pathology also supported this, identifying the lesion as a spindle cell tumor, most likely a nerve sheath tumor. However, final confirmation came after immunohistochemical analysis, which showed positivity for smooth muscle actin (SMA) and calponin, leading to the accurate diagnosis of infantile orbital myofibroma.

The misdiagnosis in this case initially arose due to the overlapping clinical features and imaging characteristics between myofibroma and nerve sheath tumors. Both are spindle cell tumors commonly found in the orbit, and both can present with painless proptosis. Patients with either tumor may seek medical attention due to symptoms such as eyelid swelling, proptosis, or vision disturbances. Orbital masses from either tumor type can cause ocular motility restrictions and share similar imaging appearances on ultrasound, CT, and MRI. Both can appear as well-defined intraorbital masses with clear borders. On imaging, the masses may be oval, round, or irregular in shape, sometimes showing heterogeneous internal echo or signal intensity, which may reflect cystic degeneration or necrosis within the tumor[3].

However, there are subtle but important differences. Nerve sheath tumors are often located along the path of the nerves and can present with characteristic features such as a "dumbbell shape" or the presence of a small "tail" sign on imaging, reflecting the involvement of a nerve pathway[4]. In contrast, myofibromas may have a different distribution in the orbit. On MRI, nerve sheath tumors tend to have more uniform signal intensities, while myofibromas may show mixed signals due to their fibrous and myofibroblastic components.

Despite these imaging clues, definitive diagnosis relies heavily on histopathology and immunohistochemical studies. In this case, the key to differentiation was the immunohistochemical markers. The positive staining for SMA and calponin in our patient's tumor confirmed the diagnosis of infantile myofibroma, as these markers are characteristic of myofibroblastic tumors. Nerve sheath tumors, on the other hand, typically show S-100 protein positivity, which was not observed in this case.

Regarding treatment, surgical excision remains the primary therapeutic approach for infantile myofibroma. Given that the tumor may adhere tightly to surrounding tissues, special care must be taken during surgery

to preserve critical orbital structures. In our case, the excision was performed successfully, and the patient had an uneventful recovery with no post-operative complications.

6 CONCLUSION

Infantile solitary orbital myofibroma is a rare benign soft tissue tumor that typically occurs in the head and neck region of infants. When it presents in the orbit, it can lead to symptoms such as proptosis and restricted ocular motility, impacting the patient's appearance and vision[5]. This case highlights the diagnostic challenges in differentiating infantile solitary orbital myofibroma from an orbital nerve sheath tumor due to their overlapping clinical and imaging features. Although initial signs, imaging, and frozen section pathology suggested a nerve sheath tumor, immunohistochemistry was essential for the correct diagnosis of myofibroma. Careful attention to histopathological details and immunohistochemical markers is crucial for accurate diagnosis and appropriate management. Surgical excision remains the mainstay of treatment, with a favorable outcome as demonstrated in this case.

AUTHOR CONTRIBUTIONS

Ligang Jiang: writing – original draft; writing – review and editing.**Wencan Wu:** writing – review and editing;Conceptualization.**Fangzheng Jiang** :project administration;supervision

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CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

CONFLICT OF INTEREST STATEMENT

There is no conflict of interest.

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