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Case Report

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Congenital immature orbital teratoma with sarcomatous transformation presenting as exophthalmos: A case report

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KEYWORDS

Surgical oncology, Orbital neoplasms, Teratomas, Immature

ABSTRACT

Background: Congenital immature orbital teratomas are rare neoplasms, with intraorbital cases presenting unique diagnostic and therapeutic challenges.

Case Presentation: This report presents a patient who has exhibited rapid-growing right exophthalmos since birth. Imaging studies revealed a progressively expanding lesion in the right orbital cavity and optic nerve. Following partial resection and subsequent orbital exenteration, histopathological and immunohistochemical analyses confirmed the diagnosis of immature teratoma with sarcomatous transformation. The patient underwent chemotherapy and remains under surveillance with no signs of disease recurrence.

Conclusion: This case highlights the complexities associated with diagnosing and treating rare orbital teratomas, contributing valuably to the existing literature.

INTRODUCTION

Teratomas are rarely found in the orbital region, and congenital cases often manifest in neonates as rapidly proliferating, firm, and fluctuant masses, resulting in pronounced unilateral proptosis accompanied by chemosis, exposure keratopathy, and elongation of the palpebral fissure [1]. True congenital orbital teratomas represent only 0.8–1.3% of pediatric orbital tumors and are categorized as mature or immature based on the differentiation status of their cellular tissues [2].

Extragonadal teratomas exhibit variable sizes, occasionally exceeding 15 mm [3]. Congenital cases typically lack a (pseudo)capsule, which contributes to the challenges of achieving complete resection [2]. Herein, we report a case of a newborn presenting with exophthalmos secondary to a progressively expanding immature orbital teratoma with sarcomatous transformation.

CASE REPORT

A full-term male newborn with a gestational age of 40 weeks, delivered via cesarean section due to reduced amniotic fluid identified during routine ultrasound, had a birth weight of 3015 g and Apgar scores of 9-9. No prenatal complications were reported.

At birth, the patient presented with exophthalmos of the right eye. At 16 days of age, he was admitted to the oncology service in April 2022 due to edema and protrusion of the right eye. Magnetic resonance imaging (MRI) revealed an expansive lesion with high signal intensity on T2/FLAIR, measuring 30 × 21 mm, predominantly occupying the intra-conal space. The lesion displaced the optic nerve medially and compromised the extraocular musculature and the eyeball, resulting in proptosis.

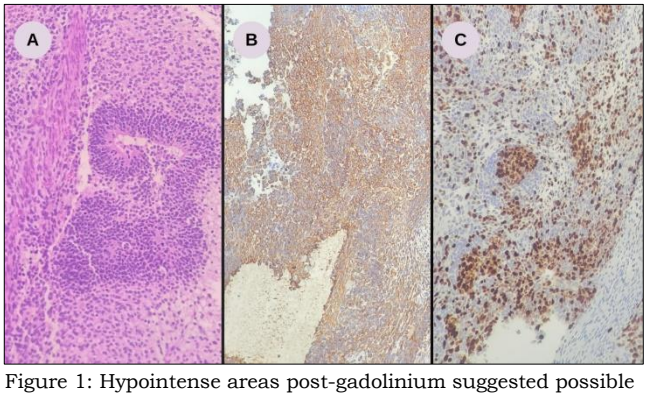

Figure 1: Hypointense areas post-gadolinium suggested possible cystic changes or necrosis.

A subsequent MRI performed 16 days later demonstrated an increase in the lesion's size, now measuring 48 × 34 mm, completely filling the right orbital cavity (Fig. 1). Hypointense areas post-

gadolinium suggested possible cystic changes or necrosis.

The presence of signs indicative of neoplasm with orbital compartment syndrome, combined with the surgical challenges of treating newborns, led the medical team to recommend partial resection of the lesion. Histological analysis of the surgical specimen revealed a neoplasm with an embryonic pattern, consistent with an immature teratoma.

Following the initial surgery, the patient developed a surgical site infection and was readmitted five days post-discharge. His condition deteriorated with symptoms including fixed gaze, cyanosis, inconsolable crying, convulsive status, and episodes of bradycardia, which suggested tumor progression. Consequently, the patient was transferred to the ICU on May 3rd.

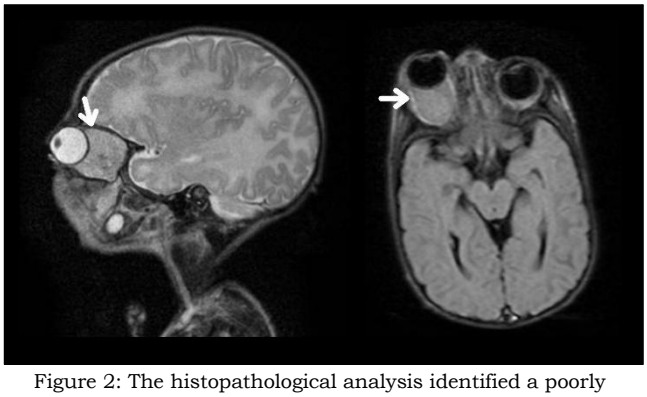

Figure 2: The histopathological analysis identified a poorly differentiated neoplasm

The following day, the patient underwent right orbital exenteration and complete orbital cavity reconstruction. Gross examination revealed a heterogeneous brown solid mass with soft consistency interspersed with stony areas. The optic nerve was enveloped by the lesion. The histopathological analysis identified a poorly differentiated neoplasm (Fig. 2A). While no necrosis or optic nerve involvement was detected, there was evidence of angiolymphatic emboli, and the surgical margins were positive for the neoplasm.

Complementary immunohistochemical studies were conducted to characterize the poorly differentiated areas. These showed negative results for epithelial, germ cell, neuroendocrine, and glial markers. However, the neoplasm expressed vimentin and CD99 (Fig. 2B), indicating mesenchymal histogenesis with a high proliferative index (Ki67 > 80%, Fig. 2C). Based on these findings, the final diagnosis was an immature orbital teratoma with undifferentiated high-grade sarcomatous transformation.

Five months after the surgical intervention, imaging studies indicated disease progression. The patient underwent chemotherapy with a regimen including etoposide, mesna, ifosfamide, and carboplatin. The treatment was completed in March 2023. The patient remains under ongoing surveillance, with no discernible evidence of new lesions on imaging studies.

DISCUSSION

Immature teratomas are neoplasms that typically contain varying amounts of poorly differentiated cell types, primarily neuroectodermal tubules and rosettes. They often exhibit a soft to firm heterogeneous consistency, frequently presenting a nodular, solid, and cystic appearance. The cysts are typically filled with serous, mucoid, or keratinous material. In some cases, cartilage, bone, and even teeth may be present [5]. Intracranial extension is extremely rare and has been documented only in isolated instances, either at presentation or subsequent to initial identification [2].

MRI studies often show teratomas as well-circumscribed lesions with hyperintense signals on T1- and T2-weighted sequences due to sebaceous content. The presence of fat attenuation within a cystic area is a diagnostic feature of mature cystic teratomas. Ultrasound imaging may reveal echogenic foci due to cartilage, calcification, and/or fibrosis [6]. In contrast to congenital cases, orbital teratomas in adults are predominantly unilateral and present as slow-growing, non-tender, mobile masses that are easily dissectable from surrounding tissue [6].

Histopathologically, ectodermal derivatives may include squamous epithelium, cutaneous adnexal structures, hair, and dental enamel. Neuroectodermal tissues may comprise glia, ependyma, and cerebellum. Mesodermal components often include adipose tissue, bone, cartilage, and smooth muscle, while endodermal elements may present as gastrointestinal and respiratory/bronchial epithelium, thyroid, and salivary glands. These tissues can vary significantly in maturity and differentiation, ranging from well-differentiated to poorly differentiated or undifferentiated [2].

The etiopathogenesis of extragonadal teratomas is not fully understood. However, it is believed that they originate from primordial germ cells that become incorporated into structures in the head, mediastinum, or sacrococcygeal region. Intraorbital teratomas are often present at birth, causing severe and progressive unilateral proptosis [7]. Due to their rarity, diagnosing these teratomas is challenging, leading to increased morbidity. Given their location in the orbit and rapid growth, they can cause facial deformities and may involve intracranial structures [8].

Most orbital teratomas are histologically well-differentiated and benign [9]. Regardless of the type of teratoma, the treatment of choice is surgical

resection, aiming for the complete tumor removal with clear surgical margins whenever possible to reduce the risk of recurrence. The relationship between tissue maturity and the risk of recurrence in head and neck teratomas is not well-established [7]. Unfortunately, complete resection of immature tumors is often challenging and may result in significant functional sequelae, including vision loss and compromised globe preservation [9]. In cases of immature teratomas with malignant transformation, the prognosis following surgery and adjuvant therapy is typically poor [9].

Imaging techniques, including computed tomography and MRI, are essential for differentiating orbital teratomas from other potential diagnoses such as dermoid cysts, hemangiomas, neuroblastoma, and retinoblastoma [8]. These imaging modalities also play a critical role in tumor resection planning. Post-surgery, efforts are needed to restore symmetry to the hemiface affected by the tumor and to ensure proper orbital development in the absence of local stimulation [9].

In conclusion, Given the rarity of orbital teratomas and the rapid expansion observed in less differentiated cases, prompt diagnosis and appropriate treatment are critical. This case underscores the complexities involved in diagnosing and treating rare orbital teratomas, making a valuable contribution to the existing literature.

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REFERENCES

1. Tsoutsanis PA, Charonis GC. Congenital orbital teratoma: a case report with preservation of the globe and 18 years of follow-up. BMC Ophthalmol. 2021; 21(1):456.
2. Harms D, Zahn S, Göbel U, Schneider DT. Pathology and molecular biology of teratomas in childhood and adolescence. Klin Padiatr. 2006; 218(6):296-302.
3. Barksdale EM Jr, Obokhare I. Teratomas in infants and children. Curr Opin Pediatr. 2009;21(3):344-9.
4. Sohrabi C, Mathew G, Maria N, Kerwan A, Franchi T, Agha RA, et al. The SCARE 2023 guideline: updating consensus Surgical CAse REport (SCARE) guidelines. Int J Surg. 2023;109(5):1136-1140.
5. Heerema-McKenney A, Harrison MR, Bratton B, Farrell J, Zaloudek C. Congenital teratoma: a clinicopathologic study of 22 fetal and neonatal tumors. Am J Surg Pathol. 2005;29(1):29-38.
6. Rao R, Honavar SG, Mulay K. Primary lacrimal gland teratoma in an adult: A clinicopathologic and radiological correlation. Indian J Ophthalmol. 2019 ;67(7):1238-1240.
7. Alkatan HM, AlObaidan OS, Kfoury H, Al-Faky YH. Orbital immature teratoma: A rare entity with diagnostic challenges. Saudi J Ophthalmol. 2018;32(1):75-8.
8. Yang Z, Du L, Zhang H. Congenital orbital teratoma: A clinicopathologic case report. J Obstet Gynaecol Res. 2021;47(6):2225-9.
9. Dharmarajan H, Rouillard-Bazinet N, Chandy BM. Mature and immature pediatric head and neck teratomas: A 15-year review at a large tertiary center. Int J Pediatr Otorhinolaryngol. 2018;105:43-47.