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Congenital Immature Orbital Teratoma with Sarcomatous Transformation Presenting as Exophthalmos: A case report

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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 that 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 diagnosis and treatment of rare orbital teratomas, contributing valuably to the existing literature.

Keywords: *Surgical Oncology; Orbital Neoplasms; Teratomas, Immature.*

1. INTRODUCTION

Teratomas are scarcely found in the orbital region and congenital cases often manifest in neonates as rapidly proliferating, firm, and fluctuant masses, culminating 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 its cellular tissues[2].

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

CASE REPORT

A full-term male new-born with a gestational age of 40 weeks, with no previous prenatal complications, was delivered via caesarean section due to a reduction in amniotic fluid identified during a routine ultrasound. The birth weight was 3015g, and Apgar scores were 9-9.

At birth, the patient exhibited exophthalmos in the right eye. He was admitted to oncology service in April/2022 due to edema and protrusion of the right eye, at sixteen days of age. A magnetic resonance imaging (MRI) revealed an expansive

lesion with high signal intensity on T2/FLAIR measuring 30 x 21 mm, predominantly occupying the intra-conal space, dislocating the optic nerve medially and compromising the extraocular musculature and the eyeball, resulting in proptosis.

A subsequent MRI, performed sixteen days after the first one, demonstrated an increase in the lesion's dimensions, now measuring 48 x 34 mm, completely filling the right orbital cavity (FIGURE 1) with hypointense areas post-gadolinium, indicating possible cystic changes or necrosis.

The presence of signs suggestive of a neoplasm with orbital compartment syndrome coupled with the challenges involving surgical procedures in new-borns, led the medical team to recommend partial resection of the lesion. The surgical specimen's histological analysis identified a neoplasm with an embryonic pattern, suggesting an immature teratoma.

After the initial surgery, the patient developed a surgical site infection and was readmitted five days after discharge and his condition worsened in the subsequent days, with the presence of a fixed gaze, cyanosis, inconsolable crying, convulsive status and episodes of bradycardia which suggested tumor progression and prompted the patient's transfer to the ICU (May, 3rd).

The following day, the patient underwent right orbital exenteration and complete reconstruction of the orbital cavity through excision and reconstruction. Grossly a heterogeneous brown solid mass with a soft consistency intercalated with stony areas. The optic nerve was enveloped by the lesion. Histopathological diagnosis indicated a poorly differentiated neoplasm (FIGURE 2A). No necrosis or involvement of the optic nerve was detected, but there was evidence of angiolymphatic emboli. The surgical margins coincided with the neoplasm.

Complementary immunohistochemical study was conducted to properly characterize a poorly differentiated area and showed negative epithelial, germ cell, neuroendocrine and glial markers. However, there was expression of vimentin and CD99 (FIGURE 2B), favoring areas of mesenchymal histogenesis with a high proliferative index, with Ki67 exceeding 80% (FIGURE 2C). Therefore, the final report was of an Immature Orbit Teratoma with undifferentiated high-grade sarcomatous transformation.

Five months following surgical intervention, imaging findings indicated disease progression and the patient underwent chemotherapy incorporating etoposide, mesna, ifosfamide, and carboplatin. The treatment was finalized in March 2023, and the patient is presently under on-going surveillance, exhibiting no discernible evidence of novel lesions in imaging studies.

2. DISCUSSION

Immature teratomas are neoplasms that typically contain varying amounts of poorly differentiated cell types, primarily neuroectodermal tubules and rosettes. They often display a soft to firm heterogeneous consistency and may present nodular and frequently display a solid and cystic appearance. The cysts are 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 documented in isolated instances at presentation or subsequent to initial identification[2].

MRI studies often show teratomas as well-circumscribed, showing hyperintense signals on T1 and T2-weighted sequences due to the sebaceous content present in these tumors. The presence of fat attenuation within a cystic area is a diagnostic feature of mature cystic teratoma. Ultrasounds can show echogenic foci due to presence of 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 easily dissectable from the surrounding tissue[7].

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

Extragonadal teratomas' etiopathogenesis is not fully understood, but 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 with severe and progressive unilateral proptosis[8]. Due to their rarity, diagnosing these teratomas can be challenging, leading to increased morbidity. Given their location in the orbit and rapid growth, they can cause facial deformities and may involve intracranial structures[9].

The majority of orbital teratomas are histologically well-differentiated and benign[10]. Regardless of the type of teratoma, the treatment of choice is surgical resection aiming for the removal of the tumor entirely with clear surgical margins whenever possible, reducing the risk of recurrence. The relationship between tissue maturity and the risk of recurrence in head and neck teratomas is not well-established[8]. Unfortunately, complete resection of immature tumors is often challenging and can result in significant functional sequelae, both related to vision and globe preservation[10]. In cases of immature teratomas with malignant transformation, the prognosis following surgery and adjuvant therapy is typically poor[10].

The use of imaging techniques such as computed tomography and MRI is essential in differentiating this entity from other

potential differential diagnosis, which include dermoid cysts, hemangiomas, neuroblastoma, and retinoblastoma[9], and plays a crucial role in tumor resection planning. Following surgery, there remains a need to restore balance to the hemiface affected by the tumor and to develop the orbit after the lack of local stimulation[10].

3. CONCLUSION

Given the rarity of orbital teratomas and their rapid expansion in less differentiated cases, prompt diagnosis and appropriate treatment is emphasized and this case highlights the complexities associated with diagnosis and treatment of rare orbital teratomas, contributing valuably to the existing literature.

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