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Case Report

Primary atypical rhabdoid orbital tumor: An entity with aggressive behavior

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ABSTRACT

Malignant rhabdoid tumors are rare, poorly differentiated tumors which usually affect children under the age of three. These tumors have a predilection for the kidney, central nervous system and soft tissue. The definition classically relies on a characteristic morphology and the inactivation of the hSNF5/INI1 tumor suppressor gene. The diagnosis is based on radiological explorations, as well as anatomopathological and immuno-histochemical studies.

Whatever the location of the tumor, the therapeutic protocol is only decided after multidisciplinary consultation meeting, while resorting to a triad of chemotherapy, surgery and radiotherapy. The prognosis remains poor and the survival rate is below 30%. We report a rare case of retro-orbital malignant rhabdoid tumor of a new born girl.

1. Introduction

Rhabdoid tumors are an extremely rare malignant tumor entity. They are most often diagnosed in the pediatric population, with a median age of 17 months.[1,2]

In addition to the renal site (rhabdoid tumors of the kidney), extra-renal localizations have been reported, notably in the central nervous system (CNS), and soft tissues (atypical teratoid/rhabdoid tumors).[3] We report a rare case of retro-orbital malignant rhabdoid tumor in new born that presented with exophthalmos.

2. Case Report

A newborn girl, without any antenatal abnormality, presented us10 days after her birth with history of gradually increasing forward protrusion of the left eye.

On examination there was left eye axial proptosis, firm on retropulsion with no orbital thrill. Ocular motility was restricted in all direction.

The examination of the anterior segment showed diffuse conjunctival hyperemia and chemosis with exposer keratopathy and abolished photo-motor reflex. Fundus examination was within normal limit. A normal right eye with no other malformations noted on inspection.

Magnetic resonance imaging (MRI) orbit revealed left retro-orbital mass of 14 mm size with a triple component (tissue, cystic, calcifications), a grade III exophthalmos, with a right cerebellar vermis metastasis.

The infant was managed by multidisciplinary approach. A temporary tarsorrhaphy was performed after a biopsy of the tumor. Histopathological examination revealed a teratoid/rhabdoid tumor.

Palliative chemotherapy was given. The infant died few days after second course of chemotherapy.

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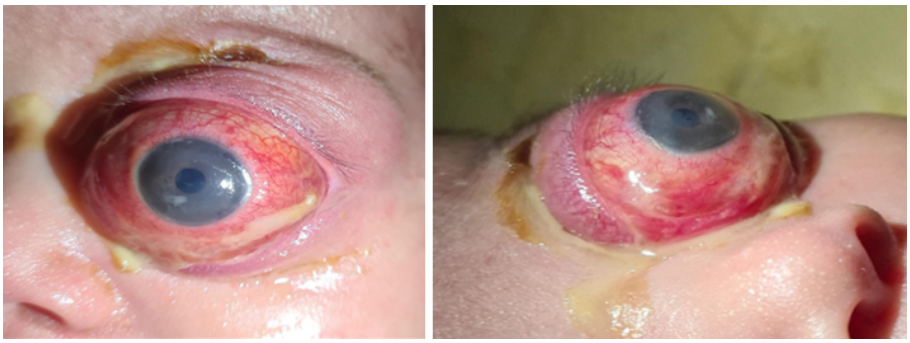

Fig. 1: Clinical photograph taken on day 10 of life, showing a huge mass in the left orbit and proptosis

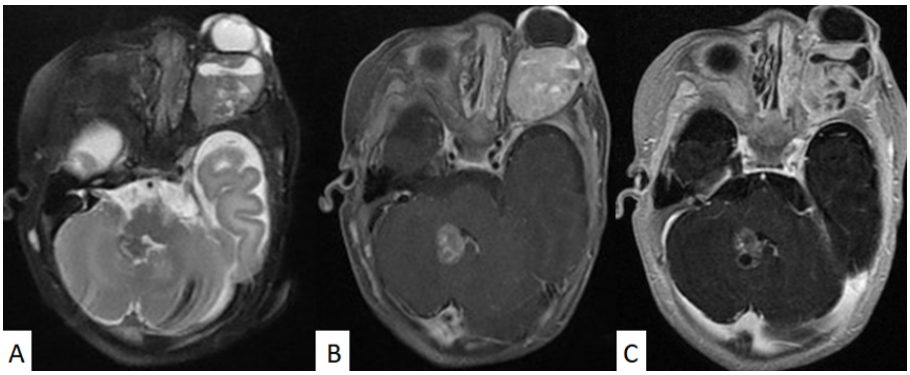

Fig. 2: Magnetic Resonance Imaging of the orbit at admission: Axial T2 **(A)** Axial T1 FAT SAT **(B)** Gadolinium injection **(C)**

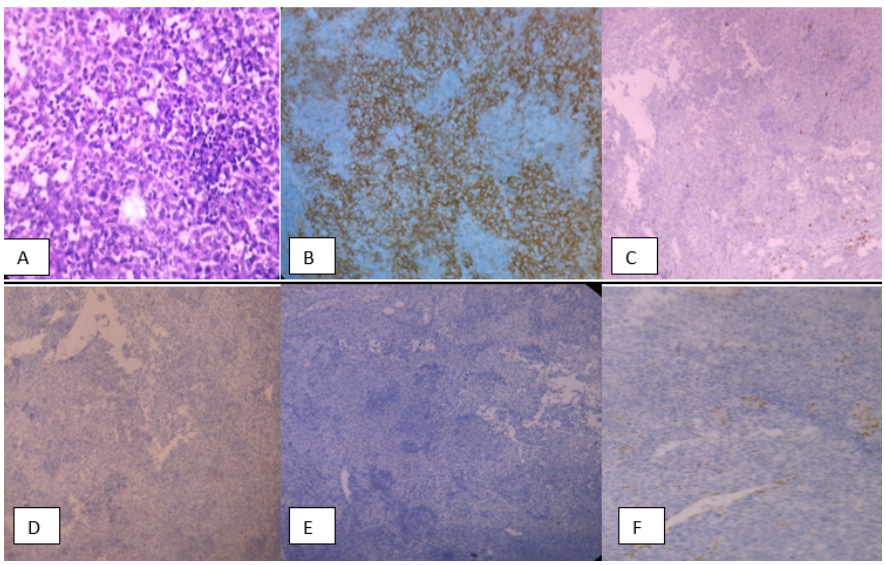

Fig. 3: Microphotographs of immunohistochemical profile of the lesion: **A**: Round cell tumor proliferation (H & E × 20) **B:** Tumor cells positive for MIC-2 (CD99) **C:** tumor cells negative for CD3 **D**: tumor cells negative for CD20 **E**: tumor cells negative for myogenin **F:** loss of expression of INI1 gene

Table 1: Clinical features of congenital orbital rhbdoid tumor cases

Source /year	Age	Sex	Localization	Management	Evolution
Aashim Bhatia (2021)[4]	8years	F	Cranial nerve III	Surgical mass resection	Not available
Mahdi.Y et all (2015)[5]	20 days	F	Optic nerve	chemotherapy	died
Seeringer.A et all(2009)[6]	1 day	F	Periorbital mass infiltrating the periorbital musculature	Total resection of the tumor+ chemotherapy	Complete remission for $5\frac{1}{2}$ years
Verma et all (2008)[7]	3 years	M	Optic nerve	Tumor resection	Not available
Koung Hoon K et al (2007)[8]	1 day	F	Intraconal tumor	Incisional biopsy+ exenteration after biopsy +chemotherapy	died
Jeffrey C. A et all(2006)[9]	3years	M	Optic Pathway tumor	Radiotherapy+ chemotherapy	Died
Gunduz et all (1998)[10]	36months	F	intraconal tumor	chemotherapy +radiotherapy+ orbital exenteration	Died
D. Brian et all (1998)[11]	1day	M	Intraconal tumor	Subtotal-excision+ chemotherapy	Not available
Rootman et all(1989)[12]	6 weeks	M	Intraconal tumor	Incisional biopsy +radiotherapy+ chemotherapy	Not available

3. Discussion

The malignant rhabdoid tumor was first described in 1978 by Beckwith and Palmer, in the kidney.[13] They described characteristics of rhabdoid tumors, namely that they are highly malignant and that they occur mainly in infants and young children.[14]

Malignant rhabdoid tumor has been reported in the ocular region secondarily as an ocular metastasis from the renal tumor or primarily in the orbit, cranial and optic nerve.[4,5,7]

TheTable 1 lists all cases of congenital orbital rhabdoid tumor found in the literature and accessed through the PubMed search engine.

The rarity and morphological pleomorphism of these tumors make their diagnosis often difficult and the use of pathological and immunohistochemical study seems justified for the confirmation of the rhabdoid nature of these neoplasms, despite the fact that they are histologically polyphenotypic.[15] The consistency of this entity is supported by the recent demonstration via multiple translational studies of the inactivation of the tumor suppressor gene SMARCB1(hSNF5/INI1) in 80% of cases.

The discovery of the role of the INI1 gene (22q11.2) has allowed the grouping of various entities under a single term and to define the predisposition syndrome of rhabdoid tumors, which opens perspectives for new targeted therapies aimed at controlling the cancer pathway, but also at prenatal diagnosis of this neoplastic entity.[16]

The therapeutic management of these tumors is based on a combination of surgery, radiotherapy and aggressive conventional chemotherapy. As for prognosis, overall survival is still estimated to be less than 30% regardless of tumor location, despite the progress that pediatric oncology has recognized in the last decades.[15]

4. Conclusion

Malignant rhabdoid tumors are rare specially in a primary orbital location. The diagnosis of this tumor entity remains difficult to establish because of pleomorphism of their clinical presentation. The diagnosis is based on radiological explorations, as well as pathological and immuno-histochemical studies.

5. Conflict of Interest

None.

6. Source of Funding

None.

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