Ocular Manifestations of Primary and Secondary Orbital Rhabdomyosarcoma in Children – Retrospective Analysis of 30 cases

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Introduction

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children, accounting for 4% of all solid tumors in children. It is also the most common primary orbital malignancy. It can arise in the orbit or adjacent structures with secondary spread. About a third (35%) of all rhabdomyosarcomas arise in the head and neck region.

Purpose: To describe the ocular manifestations of rhabdomyosarcoma (RMS) in a cohort of pediatric patients.

Methods

Study Design: Retrospective, single-center cohort study from a tertiary care pediatric hospital.

Patient Population: The medical charts of 30 patients with rhabdomyosarcoma in the head and neck region seen between 2000 and 2019 were analyzed.

Main outcome: Incidence and prognostic role of ocular findings in primary and secondary RMS cases.

Ocular manifestations (n = 30)	At presentation n (%)	Developed during course n (%)
Number of patients	16 [a] (53.3)	15 (50.0)
Orbital involvement [b]	14 (46.7)	8 (26.7)
Ptosis	6 [c] (20.0)	5 (16.7, all transient) [d]
Eyelid swelling	4 (13.3)	0
Eyelid erythema	1 (3.3)	0
Globe displacement	1 (3.3)	0
Proptosis	6 (20.0)	0
Restriction of extraocular motility / strabismus	9 (30.0)	1 (3.3)
Orbital mass	10 (33.3)	1 (3.3)
Optic neuropathy	6 (20.0)	4 (13.3, one case bilateral)
Venous tortuosity / congestion	2 (6.7)	0
IOP	1 (3.3)	0
Hematoma	1 (3.3)	0
Tearing	1 (3.3)	0
Superficial Keratopathy / Dry Eye Disease	3 (10.0)	7 (23.3, 3 were transient [e])
Cranial nerve palsy	9 (30.0)	1 (3.3)
Cataract / Intraocular lens	0	8 (26.7)
Exenteration	0	2 (6.7)
Enophthalmos	0	3 (10.0)
Lagophthalmos	1 (3.3)	1 (3.3)
Infections (orbital cellulitis / conjunctivitis)	0	3 (resolved)
Reduced visual acuity	3 (10.0)	10 (33.3)

Table 1: Ocular manifestations of rhabdomyosarcoma from a cohort of pediatric patients with orbital involvement. [a] One patient was lost to follow up. [b] Patients could present with one or more findings. [c] 2 cases are related to oculomotor palsy. [d] vincristine neurotoxicity [e] radiation related.

Results

There were 30 patients with rhabdomyosarcoma. Median age at diagnosis was 5.0 years and 60% were male. After a median follow-up of 5.2 years, there was disease remission in 67%, partial remission in 3.3%, 26.7% were deceased and one case was lost to follow up (3.3%). The histology was embryonal in 25 cases, alveolar in 5 cases, and spindle cell in 1 case. A total of 19 patients (63.3%) were found to have ocular involvement of rhabdomyosarcoma, 16 at presentation.

Primary orbital RMS was seen in 7 patients and 12 cases had secondary orbital or ocular involvement. The most common findings were proptosis (n=3), swelling (n=4) and orbital mass (n=7) in primary orbital RMS cases, whereas cranial nerve palsies (n=9) and restriction of extraocular movements (n=6) was primarily seen in cases of secondary orbital involvement.

On survival analysis, a favorable prognosis was found for cases with primary orbital RMS (deceased=0/7, 0%), while a worse prognosis was found for cases with secondary orbital/ocular involvement (deceased=7/12, 58%).

Ocular long-term complications were common, the most common were bony hypoplasia (33.3%), cataract (30.0%), changes to visual acuity (30.0%) and keratopathy (23.3%). The most common systemic long-term complications were chemotherapy related ototoxicity with hearing impairment (20.0%), cosmetic changes to the face (33.3%), hormonal deficiency (30.0%) and dental sequelae (16.7%).

	Primary orbital RMS n (%)	Secondary orbital RMS n (%)
Patients (n=19)	7 (36.8)	12 (63.2)
VA >= 20/40	4 (21.1)	6 (31.6)
VA > 20/200 to <20/40	1 (5.3)	0
VA <= 20/200	2 (10.5)	5 (26.3)
Cataract	5 (26.3)	4 (21.1)
Keratopathy	3 (15.8)	4 (21.1)
Changes to VA	5 (26.3)	7 (36.8)
Bony hypoplasia/facial asymmetry	5 (26.3)	5 (26.3)
Need for plastic surgery	1 (5.3)	0
Facial palsy	0	4 (21.1)
Exenteration	2 (10.5)	0
Cardiac toxicity	0	1 (5.3)
Oto toxicity	1 (5.3)	5 (26.3)
Secondary malignancy	0	1 (5.3)
Cosmetic impact	5 (26.3)	5 (26.3)
Dental health	1 (5.3)	4 (21.1)
Anxiety / depression	2 (10.5)	1 (5.3)
Hormonal deficiency	3 (15.8)	6 (31.6)

Table 2: Ocular and systemic outcomes from a total cohort of 30 rhabdomyosarcoma patients. In one patient visual acuity (VA) of the affected side could not be assessed.

Conclusions

- In this cohort of children with RMS, approximately two thirds had ocular manifestations, either at presentation or during the disease course.
- Orbital involvement was common and was associated with a poor prognosis if secondary.
- Ocular long-term sequelae were common, mainly cataract formation, keratopathy or facial asymmetry in this cohort of patients.
- Systemic long-term sequelae included hearing deficits, impact on dental health and cosmetic appearance.

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None of the authors have any financial interests or relationships to disclose

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