IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 22, Issue 6 Ser.15 (June. 2023), PP 01-03
www.iosrjournals.org

Inflammatory Proptosis Revealing Bilateral Retinoblastoma: About A Case

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Abstract
Retinoblastoma Is The Most Common Malignant Ocular Tumor In Children. It Is Usually Revealed By Leukocoria Or Strabismus. We Report The Case Of A Little Girl Followed For Bilateral Retinoblastoma Revealed By Unilateral Proptosis. We Will Discuss The Clinical, Paraclinical And Therapeutic Aspects.

Key Words : *Retinoblastoma, Proptosis, Enucleation, Chemotherapy, Transpupillary Thermotherapy*

Date of Submission: 19-06-2023 Date of Acceptance: 29-06-2023

I. Introduction

Proptosis is the increase in the normal protrusion of the eyeball in front of the orbital bony framework. It is the clinical sign revealing an orbital mass, tumour, infection, orbital cellulitis or trauma. Retinoblastoma can manifest as proptosis.

The aim of our work is to analyze the clinical, paraclinical and therapeutic profiles of a unilateral proptosis revealing a bilateral retinoblastoma.

II. Observation

This is a 19-month-old female infant, with no particular pathological history, who has had proptosis of the left eye for 2 weeks.

The ophthalmological examination under sedation of the right eye found normal adnexa, ocular tone and anterior segment; the fundus shows a single tumor of 4 papillary diameters located pre-equatorial (figure 1)

Examination of the left eye found inflammatory proptosis with significant chemosis, corneal edema with central opacity and 360° corneal neovascularization. Ocular tone is increased on bidigital palpation, the fundus is not accessible (figure 2)

The cranio-orbital MRI performed found a left intraorbital process infiltrating the oculomotor muscles, intra and extraconical fat and the optic nerve without intracranial extension (figure 3).

The extension assessment (abdominal ultrasound and chest X-ray) returned to normal.

In total, it is a bilateral, hereditary retinoblastoma in a 19-month-old infant, classified group B in the right eye and group E in the left eye according to the international classification of retinoblastoma, with orbital and optic nerve extension.

The patient underwent 3 sessions of transpupillary thermotherapy in the right eye, and courses of chemotherapy with 3 molecules (carboplatin, vincristine, etoposide) to facilitate enucleation of the left eye. Thermotherapy was effective and follow-up examination found a calcified atrophic tumor with peritumoral atrophy (figure 4).

The patient was then able to benefit from enucleation, post-operative chemotherapy but also radiotherapy since she presented high histoprognostic factors.

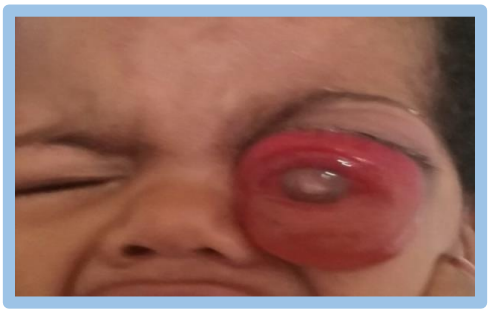

Figure 1 : Inflammatory left proptosis with significant chemosis

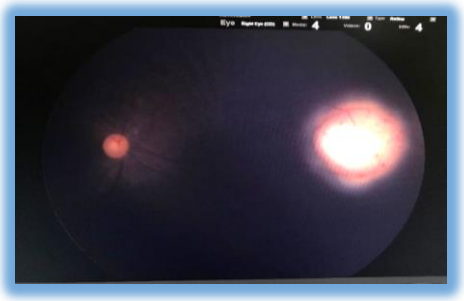

Figure 2 : retinal tumor in preequatorial right eye

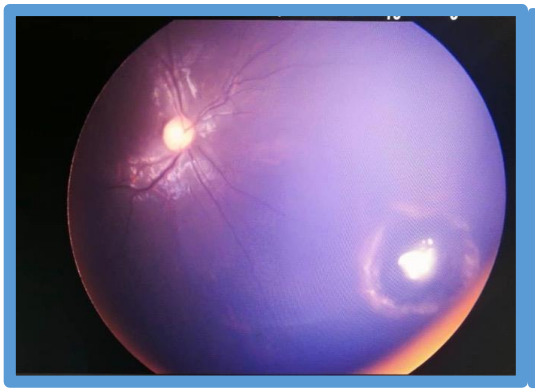

Figure 3 : MRI aspect of a left intraorbital process infiltrating the oculomotor muscles, intra and extraconical fat and the optic nerve without intracranial extension

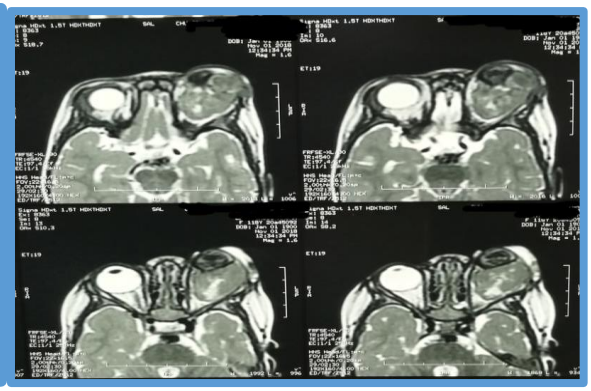

Figure 4 : appearance after transpupillary thermotherapy with calcified atrophic tumor and peritumoral atrophy

III. Discussion

Retinoblastoma is the most common ocular malignancy in children. Its incidence is 1/15,000 to 1/20,000 births, with a sex ratio of 1.5/1. In approximately 60% of cases, the tumor is unilateral with a median age at diagnosis of 2 years and most of these forms are non-hereditary; in 40% of cases, the tumor is bilateral and always hereditary, discovered at a median age of 1 year [1-3]. Retinoblastoma is a tumor of genetic origin. The RB1 predisposition gene, involved in the control of cell division in retinoblasts, is located in the chromosomal region 13q14. According to Knudson's theory, the occurrence of RB requires 2 gene mutations with inactivation of the 2 alleles [4]. Two forms are possible:

• the often bilateral and multifocal hereditary form: ◦ the 1st mutation is germinal, transmissible to offspring according to an autosomal dominant mode with a high penetrance of 90% (risk of transmission to offspring of 45%), ◦ the 2nd mutation is somatic in level of the retinoblast, acquired during fetal life or in the first months of life;

• the sporadic form, always unilateral unifocal: the 2 mutations are somatic and occur in the same retinal cell. This form is not transmissible to offspring [4, 5].

Two telltale signs are particularly common. Leukocoria and strabismus.

Other much less frequent signs can be observed: rubeosis iris, hypopyon, hyphema, buphthalmos, uveitis, orbital cellulitis, proptosis [6]. These signs reflecting a delayed diagnosis are still frequent in certain developing countries, as shown by studies carried out in Morocco[7], Burkina Faso[8] and Nigeria[9] in which exophthalmos was found respectively in 47%, 59.37%, and 84.6% of cases. The delayed diagnosis could be explained by the use of traditional therapy, the low socio-economic level but also the lack of information and awareness of general practitioners and pediatricians and the general public on the revealing signs of retinoblastoma.

Examination of the other eye is essential, as it may reveal a very asymmetrical bilateral shape as in our observation. Ultrasound is useful, especially when the fundus is not accessible, to detect retinoblastoma calcifications. MRI confirms the diagnosis and makes it possible to study the extension to the optic nerve [10]. The management of retinoblastoma is complex, multidisciplinary and is done in a specialized center.

IV. Conclusion

The management of retinoblastoma has improved thanks to the development of conservative treatments. The prognosis is essentially conditioned by early diagnosis and adequate treatment as soon as possible. Benefit of carrying out a bilateral ocular fundus in all children seen in an ophthalmology consultation.

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