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Bilateral Retinoblastoma: Management and Impact on the Lives of Survivors

Banane NK*, Saadi F, Hanafi J and El Houdzi

Department of Pediatric Hematology Oncology, Mohammed VI University Hospital Center Marrakech, Morocco

ABSTRACT

Introduction: Retinoblastoma is a malignant tumor of neuroepithelial origin occurring in infants and young children. Despite its rarity, it is the most common intraocular malignancy in children. It is unilateral in 2/3 of cases and bilateral in 1/3, and may represent a genetic predisposition to cancer. RB survivors are at risk of disease- and treatment-related adverse effects, including loss of vision or one eye, second cancer, genetic predisposition to cancer and reduced quality of life. The aim of our study is to investigate the management Bilateral Retinoblastoma and its impact on the lives of survivors.

Method: We report in this retrospective study of 15 cases collected at the Department of Hematology and Pediatric Oncology at CHU Mohammed VI in Marrakech over a 12-year period, from January 2012 to June 2O24.

Result: The mean age was 1 year and 5 months with a slight male predominance (60%). No patients had a family history of retinoblastoma. Consanguinity was present in 26%. The most frequent reasons for consultation were: leukocoria (86.6%), strabismus (33.3%) and exophthalmos (13.33%). The majority of patients (80%) were seen within 6 to 14 months, with an average delay of 9 months. Diagnosis was based on ophthalmoscopic examination, ultrasound and CT scan data, while histological examination of the surgical specimen enabled confirmation of the diagnosis and prognostic classification. Optic nerve invasion was found in 3 cases, and choroidal involvement in 3 patients. Therapeutically, enucleation was required in most cases, given the advanced stage of the tumours (86.66%): enucleation of the right eye in 4 patients, enucleation of the left eye in 2 patients, and bilateral enucleation in 8 patients, given the advanced stage of the tumours. Neoadjuvant chemotherapy was indicated in 46.6% of cases, to facilitate enucleation. Eleven 11 patients (73.3%) received post-operative chemotherapy, 3 patients received unilateral conservative treatment (thermotherapy, cryotherapy and intravitreal chemotherapy) and 1 patient received bilateral conservative treatment (radiotherapy).

In our series, 11 patients achieved complete remission, two died of severe bone marrow failure after chemotherapy, 1 was lost to follow-up and one girl died of osteosarcoma of the left shoulder. Only one case of tumor recurrence in the optic disc was noted, subsequently put on palliative treatment, 72% benefited from an ocular prosthesis.

The parents of the survivors in our study perceived their children's general health and emotional quality of life as inferior (70%), and the participation of children who had undergone unilateral enucleation in school and the community was significantly higher than that of children who had undergone bilateral enucleation.

Conclusion: Enucleation and chemotherapy remains the gold standard in the management of bilateral retinoblastoma. Enucleation continues to encounter a range of psychosocial problems.

Lifelong follow-up and advice on a healthy lifestyle are necessary for retinoblastoma survivors.

***Corresponding author**

Banane NK, Department of Pediatric Hematology Oncology, Mohammed VI University Hospital Center Marrakech, Morocco.

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Introduction

Retinoblastoma is a malignant tumor of neuroepithelial origin occurring in infants and young children. Despite its rarity, it is the most common intraocular malignancy in children. It is bilateral in 1/3 of cases [1]. Once fatal, this tumor is now curable in 90% of cases, thanks to therapeutic advances over the last 10 years and improved screening and diagnostic techniques, notably medical imaging. It is a subject of great interest and international relevance for several reasons: firstly, its frequency, the contribution of molecular genetics to antenatal diagnosis in families at risk, and above all the therapeutic advances made. Treatment must be multidisciplinary to guarantee patients the best possible outcome. The therapist's aim is to ensure the patient's survival, preserve visual function and improve aesthetics by fitting an ocular prosthesis in patients who have undergone surgery [1]. RB survivors are at risk of disease- and treatment-related adverse effects, including loss of vision or one eye, second cancer and reduced quality of life. And they require adequate care and follow-up to detect side-effects and sequelae, as well as for future social and professional integration Over the past decade, the World Health Organization (WHO) has recognized participation as one of the most important outcome measures of medical interventions. Participation is defined as a person's involvement in the activities of daily living in different areas such as education, social participation and personal care, and

is a central concept in the International Classification of Function, Disability and Health.

Materials and Methods

We report from 15 cases collected at the Department of Hematology and Pediatric Oncology (SHOP) at CHU Mohammed VI in Marrakech during a 12-year period, from January 2012 to June 2O24, always followed in SHOP. Our study consisted in analyzing data from the initial care modalities and, through follow-up and interviews with the child and parents, determining the survival rate and impact on social, school and professional life.

Results

Over a 12-year period, 15 files of patients with bilateral retinoblastoma were collated, representing an annual incidence of 1 case/year. The mean age was 17 months, with a slight male predominance (60%). Six girls and nine boys with a sex ratio of 1.5. No patients had a family history of retinoblastoma. Consanguinity was present in 26% (n = 4). The majority of patients (80%) consulted their doctor within 6 to 14 months, with an average delay of 9 months, reflecting a lack of awareness of the pathology on the part of parents and doctors in some cases.

The most frequent reasons for consultation were: leukocoria (86.6%), strabismus (33.3%) and exophthalmos (13.33%) of cases.

Symptômes	Nombres
la leucocorie	8 cas
la leucocorie+ strabisme le	5 cas
l'exophtalmos	2 cas

Figure 1: Circumstances of Discovery of Retinoblastoma in Patients

The diagnosis was made on the basis of ophthalmoscopic examination, ultrasound and scans, while histological examination of the surgical specimen enabled confirmation of the diagnosis and prognostic classification. Optic nerve invasion was found in 3 cases, and choroidal involvement in 3 patients. Involvement was predominantly G-sided in 7 cases, and right-sided in 8.Therapeutically, enucleation was required in most cases, given the advanced stage of the tumors (86.66%). Surgery was performed within an average of 10 days: enucleation of the right eye in 4 patients, enucleation of the left eye in 2 patients, and bilateral enucleation in 8 patients, given the advanced stage of the tumors. Neoadjuvant chemotherapy was indicated in 46.6% of cases, to facilitate enucleation. These patients received carboplatin, VP16 and Vincristine in 2 courses. Eleven patients (73.3%) received post-operative chemotherapy, 3 patients received unilateral conservative treatment (thermotherapy, cryotherapy and intravitreal chemotherapy) and 1 patient received bilateral conservative treatment (radiotherapy).

The outcome was favorable in 11 patients, with complete remission. Two patients died of severe medullary aplasia following chemotherapy, 1 was lost to follow-up and one girl died of osteosarcoma of the left shoulder. Of the 11 surviving patients, 6 are still being followed up and none have secondary cancer or sequelae, with 36% having a favourable functional prognosis.

The parents of the survivors in our study perceived their children's general health and emotional quality of life as inferior (70%), More than half the survivors (n =8) reported physical problems associated with the loss of one or more eyes, which refer to problematic and practical physical symptoms that impact on quality of life, and include seven sub-themes: vision-related difficulties (37.5%), limited options (25%), prosthesis-related problems (25%), appearance-based (62%), pain and irritation (25%), The most frequently reported physical problems were vision- or eye-related and included visual difficulties, limited peripheral vision and lack of depth perception. Visual difficulties referred to general difficulties with vision (37.5%) participants. of depth perception, or the visual ability to perceive the world in three dimensions and the ability to determine distances between objects, was also noted as a vision-related problem Fifty percent of survivors reported intrapersonal problems, which included two sub-themes: increased insecurity (25%) and regular/ recurring thought processes (n =12.5%). The most frequent type of intrapersonal problem encountered by participants was related to increased insecurity, which was further subdivided into appearance-related self-consciousness, loss of self-esteem/ self-confidence and personality change. Appearance-related self-consciousness was described as discomfort, embarrassment and/ or self-consciousness (n =25%). Survivors' parents reported no difficulties in maintaining a friendship, despite their perception of other children's reluctance to befriend them.

Results also showed that parents reported behavioral problems in their children (25%) they reported behavioral problems as internalizing (anxiety, depression, withdrawal and somatic complaints) rather than externalizing (aggression, rule-breaking behavior).

The participation of children who had undergone unilateral enucleation, at school and in the community was significantly higher than that of children who had undergone bilateral enucleation.

Discussion

Retinoblastoma is a rare malignant tumor (2%) but represents the most common intraocular tumor (30% of ocular tumors). In Morocco, the incidence is estimated at 50 new cases per year, most of which are sporadic. The average age at diagnosis of bilateral forms is 1 years [2] . In our series, the average age of patients at diagnosis was 17 months. The diagnosis of retinoblastoma occurs at an earlier age in developed countries than in developing ones, thanks to good socioeconomic levels and efficient healthcare systems with easy and rapid access to care [2,3].

Most authors report that the disease affects both sexes about equally [4,5]. In our study, male participation predominated, with a M/F sex ratio of 1.5, 6 girls (40%) and 9 boys (60%). There are also differences in the incidence of retinoblastoma between developed and developing countries, probably due to the consanguinity common in developing countries [6]. The search for a family history of ocular pathology or other cancers is crucial. In our series. The two most frequent revelatory signs found in the literature are leukocoria and strabismus, which is consistent with the results of our study.

The diagnosis of retinoblastoma is made by fundus examination under general anaesthetic, revealing a whitish mass. Ocular ultrasound is often performed at the same time as the objective FO [7]. Orbito-cerebral CT has its place in the evaluation of intra- and extra-ocular tumour extension, but is now increasingly being replaced by MRI, especially if access to the latter is rapid [7]. In our experience, orbito-cerebral MRI was performed in 60% of cases. Imaging also enables differential diagnosis with

other pseudotumor lesions (retinal dysplasia, Coats' disease), and MRI is more effective in analyzing optic nerve infiltration and cerebral extension [7, 8]. In our study, 46.6% of cases received initial chemotherapy, which led to a reduction in tumour mass. It is indicated in forms with locoregional extension followed by enucleation, and finds its indication in developing countries, unlike industrialized countries where it is increasingly rarely indicated [7].

Surgery (enucleation) is still the norm in our context, whereas it is discussed or replaced by conservative treatments in developed countries [7]. The use of postoperative chemotherapy was 73.3% in our study, close to the 83% reported by A.S. Sow et al. in a Senegalese study [9]. The indication for adjuvant treatment, aimed at limiting the risk of extra-ocular relapse, remains debated, based on the results of histopathological analysis of the enucleation specimen [10].

Although the impact of treatment on objective measures such as overall survival (Yu et al. 2009; Tamboli et al., 2015), subsequent malignancy risk (Chauveinc et al., 2001; Fletcher et al., 2004; Kleinerman et al., 2005; Marees et al., 2008, 2010; Liu et al., 2011; MacCarthy et al., 2013; Fidler et al., 2018) and even ocular aesthetics (Aggarwal et al., 2013; Mourits et al., 2018) has been extensively studied in retinoblastoma patients, there are far fewer data on subjective and psychosocial measures, particularly with regard to the impact of enucleation on long-term quality of life. There are quantitative studies on quality of life with variable results. For example, a study of 65 child and adolescent retinoblastoma survivors showed that survivors reported very good health-related quality of life. However, in a similar study of 87 adult retinoblastoma survivors, survivors reported a slightly lower quality of life, particularly with regard to mental health (anxiety, feelings of depression and loss of control). Similarly, van Dijk et al. 2007a reported that 37% of children with RB in their sample had not attended mainstream education. This may explain the children's lower perception of quality of life at school. This hypothesis is supported by studies on normative samples as well as on children with cancer, which have shown that teachers play a crucial role in children's perception of quality of life at school.

To fill this gap in the literature, we assessed survivors' perceptions of the impact of treatment on quality of life. Overall, the study revealed three main findings. First, a qualitative analysis of retinoblastoma survivors revealed that they continued to report physical problems, intrapersonal problems, social and relational problems and emotional problems.

Importantly, participants discussed their physical difficulties with their appearance, as well as their difficulties at school.

Conclusions

Enucleation and chemotherapy remain the gold standard in the management of bilateral retinoblastoma. Enucleation continues to encounter a range of psychosocial problems.

Lifelong follow-up and advice on a healthy lifestyle are necessary for retinoblastoma survivors.

References

1. Elkhaoua M (2016) Management of Retinoblastoma (ABOUT 38 CASES), Rabat Specialty Hospital, Journal of the Moroccan Society of Ophthalmology.
2. Koné S (2015) Epidemiological and anatomo-pathological aspects of retinoblastoma at the Pathological Anatomy and Cytology Department of CHU du Point G. Thèse médecine Bamako 14: 25422.
3. Ilhan Günalp, Kaan Gündüz, Yasemin Arslan (1996) Retinoblastoma in turkey: diagnosisand clinical characteristcs. Ophthalmic Genet 17: 21-27.
4. Ozdemir H, Tacyildiz N, Unal E, Gulsan Yavuz, Handan Ugur, et al. (2007) Clinical and epidemiological characteristics of retinoblastoma: correlation with prognosis in a Turkish pediatric oncology center. PediatrHematol Oncol 24: 221-231.
5. Sarraux H, Lemasson C, Offret H, Renard G (1982) Anatomie et Histologie de l'oeil. 2nd Ed, Paris, Masson 218.
6. Sidibé H: Study of the epidemio-clinical aspects of retinoblastoma in the pediatric department of the Gabriel Touré University Hospital. Medical thesis, Bamako, 2008-2009.
7. Urbieta M. New therapeutic strategies for retinoblastoma. Dissertation for the university diploma in pediatric oncology, 1998-99, University of Paris XI. https://toubkal.imist.ma/bitstream/handle/123456789/17023/MS2222022.pdf?sequence=1.
8. Aerts I, Lumbroso-Le Rouic L, Gauthier-Villars M, Brisse H, Doz Retinoblastoma News F, et al. (2015) Archives of Pediatrics Elsevier Masson SAS. All rights reserved 1: 31.
9. Helen Dimaras, Timothy W (2019) Corson. Retinoblastoma, the visible CNS tumor: A review. J Neuro Res 97: 29-44.
10. Sow AS, Ndoye Roth PA, Moreira C, Diagne Akonde FB, Ka AM, et al. (2014) Thérapeutique du rétinoblastome: expérience sénégalaise 37: 381-387.
11. Khelfaoui F, Validire P, Auperin A, Quintana E, Michon J, et al. (1996) Histopathologic risk factors in retinoblastoma: a retrospective study of 172 patients treated in a single institution. Cancer 77: 1206-1213.