Contents lists available at www.innovativejournal.in

INNOVATIVE JOURNAL OF MEDICAL AND HEALTH SCIENCE

Journal homepage: http://innovativejournal.in/ijmhs/index.php/ijmhs

ISSN No. 2277-4939

Case Report

PEDIATRIC TUMOUR- RETINOBLASTOMA AND ITS CLINICOPATHOLGICAL CORRELATION

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ARTICLE INFO

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Key words: retinoblastoma, pathology, Proptosis

DOIhttp://dx.doi.org/10.15520/ijmhs.2017.vol7.iss4.175.

ABSTRACT

Retinoblastoma is the commonest childhood primary malignant intraocular neoplasm that is often characterized by spontaneous regression. They display photoreceptor differentiation. This study provides the clinical presentations and histological profiles of retinoblastoma PT JNM Medical college Raipur CG.. A retrospective study of clinically and histologically verified retinoblastoma from January 2010 to December 2011 was undertaken. The clinical and histological features were analyzed using the patient's case folder and surgical pathology records. There were 20 patients, 9 males and 11 females (M:F ratio 1.7: 1), age range from 5½ months to 6 years with 23 eyeball tumours histologically confirmed retinoblastoma during the study period. Proptosis with chemosis was the most common clinical presentation .Bilaterality was 15% in this study. Enucleation and Exenteration combined with chemotherapy were offered to patients . A moderately differentiated type with extensive areas of tumour necrosis was the commonest histological pattern.

INTRODUCTION

Retinoblastoma (RB) is a rare embryonic tumour but commonest primary malignant intraocular tumour of childhood [1, 2]. In the United States of America, retinoblastoma occurred in 1 of every 15000 live births[3], whereas in developing countries of Africa and Asia reports showed that it occurred in 1:18000 live births [4, 5, 6]. Retinoblastoma accounted for more than 50% of all ocular and orbital malignancies of all age groups in African series [5, 6, 7, 8]. However in Caucasians it is the second most common tumour following malignant melanoma [3, 9]. Retinoblastoma occurs in sporadic (60%) – usually unilateral or germline [hereditary] (40%) forms; the later manifesting either as unilateral or bilateral disease and characterized by early onset and predisposition to developing secondary non-ocular, intracranial malignancies termed Trilateral Retinoblastoma [10, 11]. . The RB gene is located in the long arm of chromosome 13 (13q14) and the gene is believed to be a recessive suppressor; thus, it requires the loss, deletion, mutation or inactivation of both copies of the gene at the 13q14 locus for RB to develop which correlates with the two "hits" theorized by Knudson [12,13,14]. Prenatal and postnatal prediction of susceptibility to inherited RB using recombinant DNA markers is now possible [15]. This would make genetic counselling for familial (hereditary) RB more accurate, leading to an earlier tumour detection and effective therapy. The clinical presentation of RB includes leucocoria, strabismus, conjunctivalchemosis, proptosis, and even blindness, which are attributed to late presentation and diagnosis [2, 16]. In spite of the treatment modalities such as enucleation, radiotherapy, photocoagulation, chemotherapy and even exenteration as in extra-ocular involvement, the mortality from RB in our environment is very high [2, 5, 6, 16, 17, 18].

EXPERIMENT WORK

A total of 42 ocular surgical specimens from 50 patients with clinical diagnosis of retinoblastoma at the ophthalmology department of Pt.JNM Medical college Raipur ,Chhattisgarh from January 2010 to December 2011 and registered at the pathology department for histological verification were retrospectively studied. The age, sex, presenting symptoms, duration of symptoms, site, management and other adjunct treatment were obtained from the patient's case notes and analyzed. Paraffin sections stained by standard haematoxylin and eosin (H&E) were retrieved and reviewed by one of the Authors. Special stains, Methyl green pyronnin (MGP) for calcium deposition and pearl blue reaction for haemosiderin pigments were employed where necessary. Each optic nerve was examined for involvement in tumour spread up to the line of transection. . The histological diagnosis was based on cell morphology, degree of differentiation, presence of Flexner-Wintersteiner rosettes, optic nerve infiltration by tumour cells to the cut end of the optic nerve and tumour necrosis. Clinical photographs of some

of the patients as well as photomicrographs were also included for illustration.

RESULTS

42 cases of retinoblastoma were diagnosed from 50 patients during the 2 - year interval which accounted for 33.3% of the total number of ocular and orbital tumour cases seen at the eye clinic and 10.5% of childhood malignancies during the same period at Pt JNM Medical college. There were 32males and 18 females, giving a male to female ratio of 1.7:1. The ages of the children ranged from 5½months to 6years with a mean age of 29months (SD 0.78). The commonest clinical presentation in our experience was proptosis with associated conjunctivalchemosis (76.1%). This was followed by leucocoria 61.9%, and hypopyon 42.8%. Eyelid swelling and blindness were observed in approx 30 % of the cases each - Table 1.

DISCUSSION

In this study, Retinoblastoma accounted for 33% of all cases of ocular and orbital malignancies in our centre that compares well with the findings in other studies where the incidence varies from 34 – 55 percent [6, 19, 20]. Similarly, our present experience of Retinoblastoma accounting for 10.5% of all childhood cancer and the commonest childhood intraocular tumour corroborates findings in African series [19, 20,21, 22] but high when compared with developed countries where 2.4 – 4% has been reported [2, 23]. We reported a male to female sex ratio of 1.7:1. This differs with the observation of no sex predilection in other studies [14, 17, 23, 24]. Our patients appeared to present very late as the average age of the children at the time of diagnosis was 29 months This is comparable to similar findings in the developing countries [6, 8, 17, 19], it is high when compared with the reports of western countries [14, 23, 24,25]. The late presentation in our study could be due to among others, lack of awareness of the disease, delayed referral prompted by consultation with traditional medical practitioners and poor affordability and accessibility to available medical facilities. No single case of RB was reported in an adult in this series which agrees with the findings of Olurin et al [19]. Proptosis with conjunctivalchemosis was the commonest clinical presentation manifesting in 76.1% of our patients. This is at variance with findings in other studies where leucocoria and strabismus were observed [16, 17, 23]. This picture is attributed to delay in presentation of the patients for diagnosis and treatment. This opinion is supported by the claim that proptosis is the result of orbital involvement [16]. Also Erwenne et al concluded in their study of 158 consecutive cases of RB that extra ocular disease was strongly dependent on the age at diagnosis and lateness of referral [25]. The incidence of bilateral RB in this series is 15% that agrees with the findings in some African series [17, 19]. In Zambia it is 4 % whereas in the western countries such as Germany, England and Wales, the incidence is higher [22, 23]. The disparity may be associated with incomplete data or due to higher incidence of unilateral (sporadic) RB over bilateral cases in India.. Histologically the two characteristic arrangements of cells that are pathognomonic of RB are Flexner- Wintersteiner rosette and Fleurrette representing an attempt to differentiate into photoreceptor cells. Well differentiated tumours are characterized by the presence of Flexner-Wintersteiner rosettes. In this communication, 22 eyes (52.3%) were of moderately differentiated type -compared with other studies the above findings are comparable [17, 23].

CONCLUSION

Retinoblastoma is the commonest malignant intraocular tumour of childhood, mostly of moderately differentiated type histologically and characterized by high mortality in our center. There is need for improved accessibility to health facilities and the importance of genetic counseling should be emphasized

Figure 1:

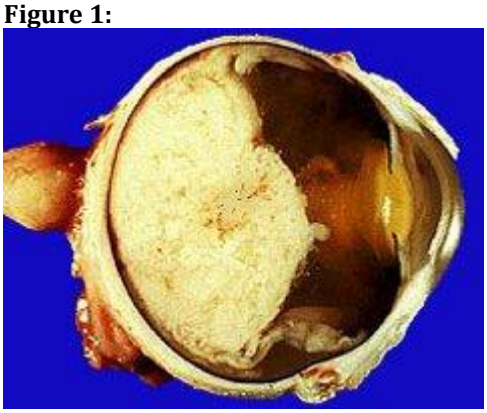

Figure 2 :

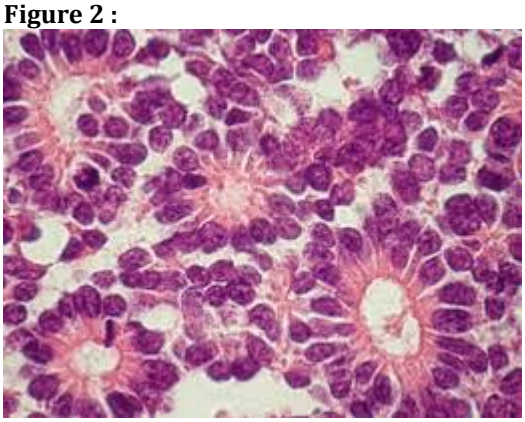

Table 1 Clinical signs and symptoms

signs and symptoms	no of cases	percentage
proptosis and conjuctivalchemosis	32	76.1
Leucocoria	26	61.9
Hypopyon	18	42.8
eyelid swelling	13	30.9
Blindness	12	28.5
eye discharge	6	14.2
Pain	4	9
globe perforation	4	9

Table no 2. characteristic histological features of retinoblastoma differentiation

histological features	no of cases	percentage
poorly differentiated	15	35.7
moderately differentiated	22	52.3
well differentiated	5	11.9
rosette		
flexnerwintersteiner	13	30.9
pseudo rosette	29	69.0
Microscopic spread		
optic nerve	16	38
skeletal muscle	1	2.3
lacrimal gland	2	4.7
vascular emboli	2	4.7
others		
necrosis	34	80.9

dystrophic calcification	11	26.1
haemorrhage	10	23.8
Siderosis	4	9.5
cholesterol slits	3	7.1
basophillia	3	7.1

ACKNOWLEDGEMENT

I would like to acknowledge the department of ophtalmology, pathology for allowing me to conduct my study.

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