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Case report

RETINOBLASTOMA IN A 12 YEAR OLD GIRL: A CASE REPORT

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ABSTRACT

Retinoblastoma, although a common ocular childhood tumour is rare in the older age groups. It present commonly as leucocoria and strabismus especially in the younger age groups. However in older children, its presentation may be atypical. We report a case of retinoblastoma in a 12 year old girl who initially presented with features of non specific inflammatory external ocular disease and later with proptosis and weight loss. Non response to ealier treatment prompted an enucleation whose specimen reveal a histological diagnosis of retinoblastoma.

Keywords: Retinoblastoma, Enucleation, Proptosis

INTRODUCTION

Retinoblastoma is the commonest intraocular malignancy of childhood.[1] It's incidence varies between 1 in 3300to1 in 20,000 live birth.[2,3] Average age at diagnosis is 12 months for unilateral cases and 24 months for bilateral cases.[4] However it has been reported in older children up to the age of 16years.[5,6]Cases of retinoblastoma have also been reported in adults up to the age of 37 years.[7,8]The tumour have been found to be due to a mutation in the retinoblastoma gene(RB1),located in the 14 band of chromosome 13.[9,10]Being a tumour suppressor gene,it's loss is said to lead to tumour formation. Clinically, the tumour commonly presents as leucocoria(60%) and strabismus(20%) and rarely as secondary glaucoma, pseudouveitis,orbital inflammation, proptosis, metastatic features and raised intracranial pressure in trilateral cases among others (20%).[11,12]We report one of these rare presentation in a 12 year old Nigerian girl.

CASE REPORT

A 12 year old girl, presented in the eye clinic of Diete Koki Memorial Hospital, Yenagoa Bayelsa State accompanied by her mother. She complained of an inability to see with her right eye for the past 2 years, dull intermittent and non radiating pain and the eye being more prominent than the fellow eye. Symptoms started gradually and there was no family history of similar ocular problems. She was the only child of a single mother, whose occupation was petty trading. They had visited other health care providers including faith healers, traditional healers and patent medicine stores, prior to presenting to the eye clinic.

Examination revealed an emantiated moderately pale girl with peripheral lymphadenopathy. On ocular examination, visual acuity in the right eye was no perception of light. There was mild lid retraction, moderate temporal sclera injection and clear cornea in same eye. The anterior chamber was of normal

depth, pupils were fixed and dilated and there was a white pupillary reflex. Fundal view was not possible. Intraocular pressure was 2mmHg .The left eye was normal.

A working diagnosis of chronic scleritis [scleromalacia] was made, with differentials of possible non specific orbital inflammatory disease, toxocariasis and late onset retinoblastoma.

An urgent ocular B-scan showed an unknown solid ovoid mass with regular border and a central calcified lesion measuring 3.4 x 2.9cm.Serology for toxocariasis was not done on account of non availability. Other investigation requested included a Full blood count, Blood film analysis and Erythrocyte sedimentation rate which were all normal.

At follow up it was noted that she had deteriorated clinically, and also not responded to the medication given earlier, and had developed a non-axial proptosis.

At this point, the diagnosis was reviewed, with the possibility that it may be an intraocular mass. The family was counseled and options discussed including enucleation of the eye. With informed consent, an enucleation was done under general anaesthesia.

Histopathological assessment macroscopically revealed a tan white tumour mass measuring 3.5cm by 2cm covered by opaque fibrous conjunctiva with a haemorhagic posterior surface.The cut surface showed focal necrosis

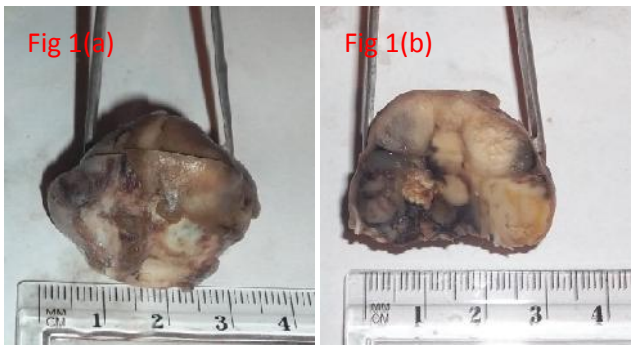

Fig 1: (a) Eyeball in retinoblastoma showing distinct tumour nodules. (b) Cut section of eyeball showing solid tumour nodules with areas of haemorrhage and necrosis.

Microscopically, it is highly cellular and composed of dense masses of small round cells with hyperchromatic nuclei and scanty cytoplasm. There were wide irregular areas of tumour necrosis and associated areas of calcification.There were areas showing loose trabeculae and nesting formations. Also seen were many rosettes (Flexner-wintersteiner type) within the tumour. Tumour seedings within the optic nerve was also noted. A histopathological diagnosis of retinoblastoma was made.

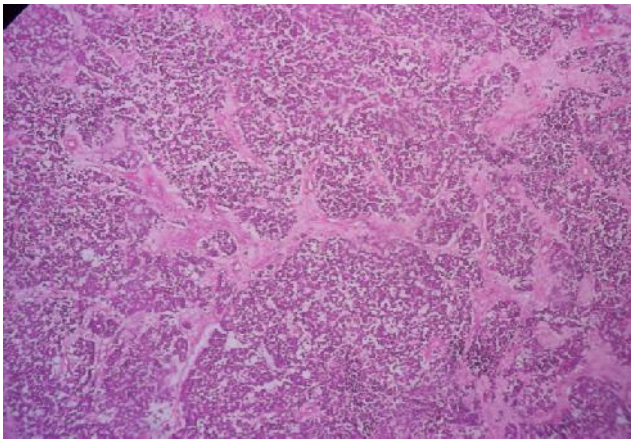

Fig 2: Retinoblastoma showing highly cellular neoplasm with scanty stroma (H/E., X100)

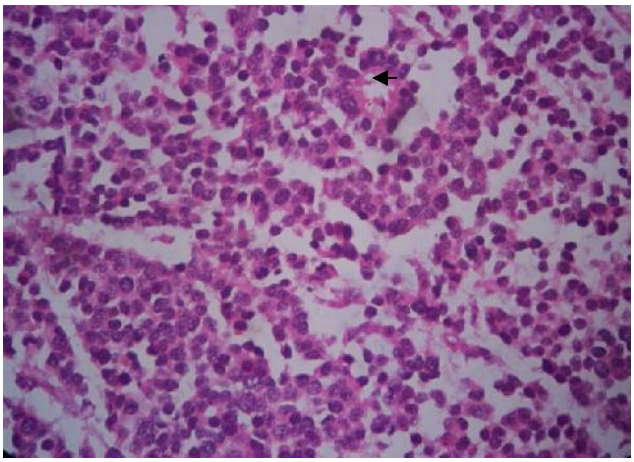

Fig 3: Retinoblastoma showing malignant small round cell tumour with typical rosettes (arrows). (H/E. X400)

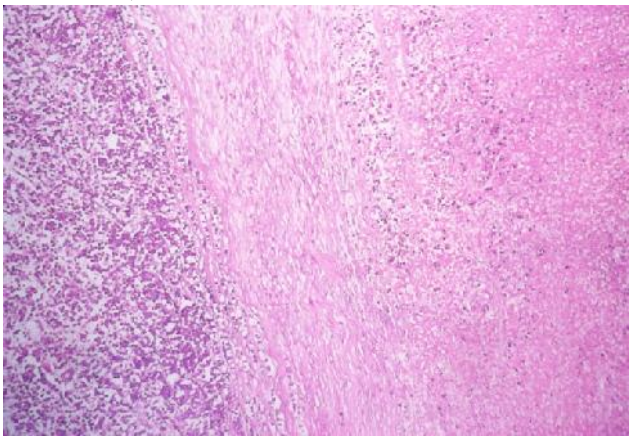

Fig 4: Irregular areas of necrosis (right half of slide), a common finding in retinoblastoma as the tumour tends to outgrow its blood supply. (H/ Estain. X100)

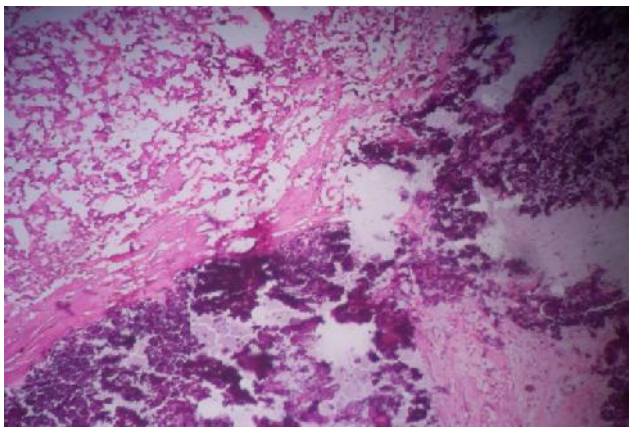

Fig 5: Wide areas of dystrophic calcification in retinoblastoma (H/E stain Mag. X100)

The child was referred for chemotherapy and/or radiotherapy but was not commenced on financial grounds. She developed severe anorexia and vomiting 1 month after surgery and died.

DISCUSION

When seen in older children, retinoblastoma is typically unilateral and sporadic[13].It occurs in a single somatic cell which becomes malignant. Comparative genomic studies in older children have shown more frequent and complex genetic abnormalities than in younger children[14].

Karciogluetal[15] found both clinical and histopathologic features were atypical in older children as were the findings in this patient which showed Flexner wintersteiner differentiation albeit fewer than would in younger patients.

In all patients with retinoblastoma the risk of metastases is increased in patients with advanced tumour, retro-laminar optic nerve invasion, anterior chamber involvement, late presentation and orbital spread. This patient presented late, and histology showed evidence of optic nerve invasion.

Sometimes as in this case retinoblastoma can present a confusing clinical scenario[16].Features of localized external ocular inflammation as in this case may add to the confusion. This external localized ocular inflammation may be due to a subtle orbital invasion as evidenced by B scan Ultrasonography findings.A high index of suspicion is therefore required to diagnose retinoblastoma in the older age group.

CONCLUSION

Retinoblastoma should be considered in the differential diagnosis of an external ocular inflammation associated with visual blurring and proptosis in the older age group.

REFERENCES

1. Abdul-salam AA, Adenipekun AA, Akinlade BJ, ElumeluTN. Pattern of Paediatric malignancies seen at the Radiotherapy department of University College Hospital,Ibadan,Nigeria.Nig QJ Hosp Med. 2007:152-4.
2. Freedman J, Goldberg L. Incidence of retinoblastoma in the Bantu of South Africa.Br J Ophthalmology. 1976;60:655-56.
3. Suckling RD, Fitzgerald PH, Stewart J, Wells E. The incidence and epidemiology of retinoblastoma in New Zealand: a 30 year survey.Br J cancer 1982;46:729-36.
4. American Academy of Opthalmology series
5. Karcioglu ZA, Abboud EB, Al-Mesfer SA, Al-Rashed W, Pilapil DH. Retinoblastoma in older children. JAAPOS 2002;6:26-32.
6. Park JJ, Gole GA, Finnigan S, Vandeleur K. Late presentation of unilateral sporadic retinoblastoma in a 16 year old girl. Aust NZJ Ophthalmol 1999;27:365-8.
7. Shrestha A, Adhikari RC, Saiju R. Retinoblastoma in a 37 years old man in Nepa: a case report. Kathmandu univ Med J 2010;8:247-50.
8. Biwas J, Mani B, Shanmugam MP, Patwardhan D, Kumar KS, Badrinath SS. Retinoblastoma in adults. Report of three cases and review of the literature. Surv Ophthalmol 2000;44:409-14.
9. Dryja TP, Rapaport JM, Joyce JM , Peterson RA. Molecular detection of delections involving band q14 of chromosome 13 in retinoblastoma. Proc Nati Acad Sci USA.1986;83:7391-94.
10. Cavenee WK, Dryja TP, Phillips RA. Expression of recessive allele by chromosomal mechanism in retinoblastoma. Nature 1983;305:779-84.
11. Abrahamson DH, Frank CM, Susman M. Presenting signs of retinoblastoma. J Paediatr. 1998;132:505-8.
12. Kanski JJ. Clinical Ophthalmology: a systematic approach. Butterworth Heinemann. Elsevier, 2007.6th ed.Edinburgh.P545.
13. Shields CL, Shields JA, Shah P. Retinoblastoma in older children: ophthalmology 1991;98:395-9.

14. Sussanne Herzog. Marked differences in unilateral isolated Retinoblastoma from young and older children studied by comparative genomic hybridization. Human genetics. 2001:108(2): 98-104.
15. Karcioglu ZA, Emad Abboud MD, Saleh A , Al-Mesfer, Waleed Al-Rashed, Delia Pilail.Retinoblastoma in older children.Journal of American Association of Paediatric Ophthalmology and Strabismus 2002;6(1):26-32.
16. Park JJ, Gole GA, Finnigan S, Vandeleur K.Late presentation of a unilateral sporadic retinoblastoma in a 16year old girl. Aust NZJ Ophthalmology. 1999; 27: 365-8.