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A RARE CASE OF BILATERAL RETINOBLASTOMA WITH DURAL METASTASIS – CASE REPORT

Radio-Diagnosis

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ABSTRACT

Retinoblastoma is the most common primary intraocular malignancy in childhood.[1] It is thought that retinoblastoma (RB) arises from retinoblasts, which are retinal precursor cells.It occurs due to mutation of the Rb1 gene on chromosome 13q14 [2] Ultrasonography (USG) can be used as screening modality for suspected retinoblastoma cases. However Computed tomography (CT) or Magnetic resonance imaging (MRI) are used for the diagnosis and extension of the retinoblastoma and for preoperative planning[5]. Herein we report a case of 2 year old child diagnosed as bilateral retinoblastoma with dural metastasis.

KEYWORDS

Retinoblastoma, MRI, metastasis, calcifications.

INTRODUCTION:

Retinoblastoma is the most common primary intraocular malignancy in childhood.[1] The frequency of retinoblastoma ranges from 1 in 14,000 to 1 in 20,000 live births and about 90% of patients are diagnosed under 3 years of age.[1].

It is thought that retinoblastoma (RB) arises from retinoblasts, which are retinal precursor cells. It occurs due to mutation of the Rb1 gene on chromosome 13q14 [WB Saunders et all 2016]

RB is often a highly calcified tumor, typically presents with leukocoria or strabismus, and in later stages, the most common sign may be proptosis [H. Dimaras et all 2019]. Delayed diagnosis of more than 6 months from the first clinical sign leads to spreading of RB from the eye and a 70% mortality rate in developing countries[R. Balasubramanya et all2004].

Ultrasonography (USG) can be used as screening modality for suspected retinoblastoma cases. However Computed tomography (CT) or Magnetic resonance imaging (MRI) are used for the diagnosis and extension of the retinoblastoma and for preoperative planning[P. de Graaf 2012].

Herein we report a case of 2 year old child diagnosed as bilateral retinoblastoma with dural metastasis.

Case Report:

Child of 1.5 years who presented to Gadag institute of medical science, Gadag, for ophthalmology department with complaints of white reflex in right eye since birth. With 15 days history of swelling, pain, watering in right eye. On examination there was hazy cornea on right side and white reflex at pupil. (figure 2) Left eye ball was normal.

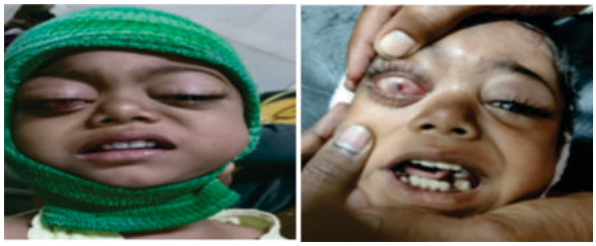


Figure 1- Right eye ball showing hazy cornea with leukocoria.

B-scan of right eye showed ill defined soft tissue mass lesion occupying near complete posterior segment with hyper reflective echoes in the mid vitreous may represent intralesional calcification. The lesion showed vascularity on color Doppler. Lens was not separately seen. The lesion was seen to extending to anterior segment. (figure -2).

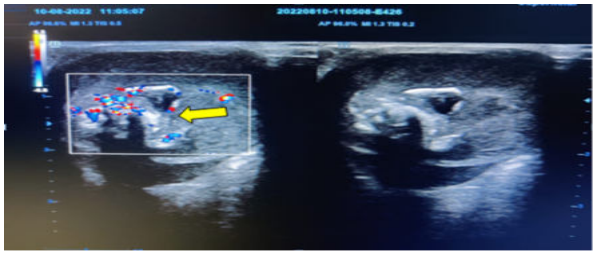


Figure 2- USG right globe reveals soft tissue mass lesion with intralesional calcification. Calcifications shows twinkling artifacts within.

Screening of left eye revealed small well defined hyperechoic triangular shaped lesion with base towards medial wall of the posterior segment extending to vitreous, minimal vascularity seen within the lesion. No obvious calcifications seen. (Figure 3)

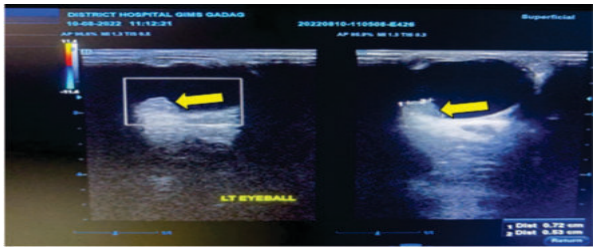


Figure 3 - USG left globe reveals triangular soft tissue mass lesion along the medial wall of posterior segment. No obvious calcifications within.

The patient was further evaluated with CT which showed ill defined iso to hyperdense soft tissue lesion seen involving near complete right globe (Posterior and anterior segment) with few foci of calcifications within. The soft tissue lesion is seen extending to extra conal space. (Figure 4)

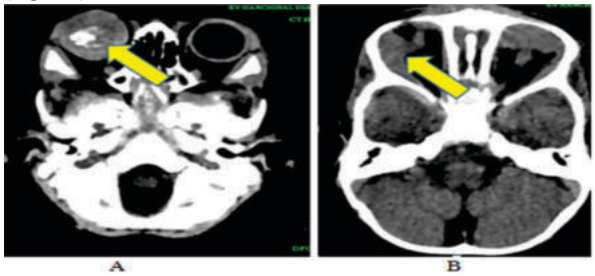


Figure 4 – Soft tissue lesion in the right lobe with intralesional calcification seen. B- there is extension of soft tissue lesion to the extra conal space.

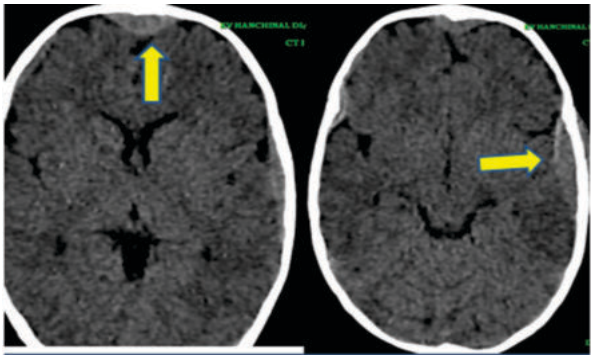
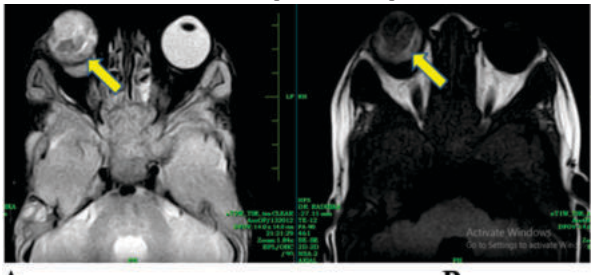


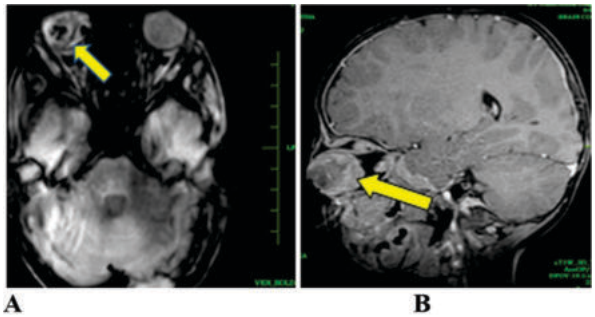
Figure 5 – Soft tissue extra axial lesion seen along the frontal and left temporal convexity – possibly dural metastasis.
Similar appearing soft tissue lesions seen in the subcutaneous region of frontal region.

Then patient was further evaluated by MRI orbits with contrast, which revealed, large heterogenously enhancing mass lesion filling almost whole of the right eyeball, including posterior and anterior segments (figure 6). There was no differentiation of iris, ciliary bodies, lens and other intra ocular structures. The lesion is heterogenous containing hypo and hyperintense areas on T1/ T2 & FLAIR images. Few areas of blooming in SWI representing dense calcifications are seen within the mass lesion (FIguer 7). Extra scleral extension of tumour mass into retrobulbar fat. Involvement of optic nerve at optic nerve head noted.



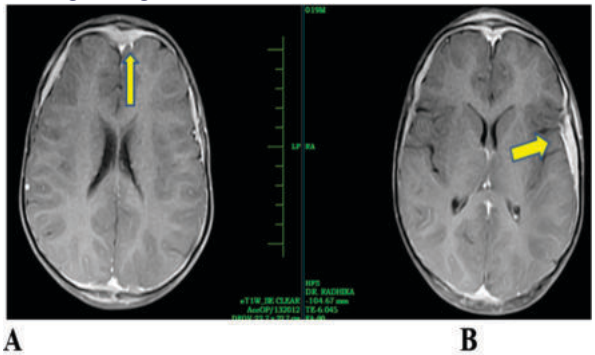
A B

Figure 6 – (A) axial T2 WI, (B) axial T1 WI showing heterogenous soft tissue in the right globe.



A B

Figure 7 – (A) axial SWI, showing blooming within the lesion representing calcifications. (B) Post contrast T1 WI images showing heterogeneous enhancement.



A B

Figure 8 – (A) and (B) axial post contrast T1 WI showing enhancing dural mets

There is similar small endophytic mass lesion in the left eyeball at infero-medial aspect measuring about 6.5x3.5 mm. No obvious extra scleral extension of this lesion seen.

Radiological features were consistent with bilateral retinoblastoma. There are multiple intracranial extra axial / dural based T1/ T2 isointense enplaque lesions in the left fronto-temporal, midline frontal and right frontal regions – features are consistent with dural metastasis (figure 8). Patient was further treated with series of chemotherapy.

DISCUSSION:

A retinoblastoma is a rare malignant intra-ocular tumor that arises in the retinal neuro-ectodermal cells of infants and children [Lupo G, Motta C et all 2014, Weissleder R et all 2007].

Retinoblastoma is estimated to represent 3% of all childhood cancers [Shields CL et all [20148,] de Jong MC et all 2015].

A heritable retinoblastoma commonly seen in younger than the age of one year and inheritable retionoblastoma usually presents after one year of age group. [Khan S et all 2014]

At early stages, most patients present with leukocoria and strabismus. At later stages, patients may exhibit proptosis, buphthalmos, or hypopyon [1].

Rb is often a highly calcified tumor. Calcification is more often presented in older children and advanced disease, as a response to necrosis or tissue damage.

There can be different types of tumor growth patterns, either endophytic, exophytic or diffuse or a combination of both. Endophytic tumors arise from the inner layers of the retina and grow into the vitreous body. Exophytic tumors originate in the outer layers and grow in the subretinal space, which causes retinal detachment. Diffusely infiltrating retinoblastoma is a rare histological form characterized by diffuse infiltration of the retina without a tumor mass.

In retinoblastoma, US (ultrasonography) demonstrates an irregular mass, more echogenic than the vitreous body, with fine calcifications and with internal vascularity [P. de Graaf et all 2012].

On CT, retinoblastoma is typically a mass of high density compared with the vitreous body, usually shows calcification and enhancement on post contrast study.

CT detection of calcifications in retinoblastoma has a sensitivity of 81–96%, and an even higher specificity The gold standard of Rb evaluation is the MRI of the orbits and brain, even though CT gives better evaluation of calcifications, because of risk of ionizing radiation, MRI is preferred over CT.

Compared to the vitreous body, retinoblastoma has moderately higher signal intensity on T1- and lower on T2-weighted images. Laterality and growth pattern should be mentioned as well as the location of the tumor, with respect to the equator of the eye (anterior, posterior or combined) and with respect to the papilla (the optic nerve disk) and the macula. One should in particular identify tumor close to the optic disk, because this may invade the nerve. In normal-size optic nerves, the abnormal enhancement of (enhancement $\geq$ 2 mm in diameter) in the distal nerve represents of metastatic extension of the tumor.

Careful analysis of midline structures should be performed to depict trilateral retinoblastoma (i.e. pineal gland tumor) or rarely lesion in the suprasellar area depicting quadrilateral RB. Leptomeningeal spread in the form of dural thickening or enhacement or dural based masses should be evaluated to rule out metastasis, as it was seen in our case.

CONCLUSION:

Together with US, high-resolution MR imaging has emerged as the most important imaging modality in the assessment of retinoblastoma—for diagnostic confirmation and for determination of local tumor extent. CT should be preferred less for evaluation of children with leukocoria because of ionizing radiation and no added diagnostic values as compared to MRI.

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