

IMAGING SPECTRUM OF RETINOBLASTOMA ON MRI. A PICTORIAL REVIEW

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ABSTRACT

Retinoblastoma (RB) is the most common malignant intraocular tumor of childhood. It is small round-cell tumor arising from neuroepithelial cells. The incidence ranges from 1 in 14,000 live births to 1 in 34,000. The average age at diagnosis is 18 months with most of the cases occurring before 3-4 years. The diagnosis may be made from shortly after birth until 7 years of age. Approximately 30% are bilateral and are typically diagnosed earlier than unilateral cases. Lesions may be synchronous, metachronous, unifocal, or multifocal. Diagnosis is typically by ophthalmologic examination, prompted by leukocoria or white reflex seen in 60% of patients. Leukocoria can be seen with large tumors or total retinal detachment. With advent of high resolution imaging techniques such as MRI, the extent and stage of the tumor can be very well depicted. Herein we concentrate on the pictorial review of different presentation of retinoblastoma concentrating on the MRI ability to stage the disease. Further pathological correlation has also been done in cases of enucleation.

Key words: Retinoblastoma, MRI, Leukocoria

Introduction

Retinoblastomas (RB) are the most common intraocular neoplasm in pediatric patients, in most cases, curable with modern treatment. Retinoblastomas may be sporadic or inherited. Bilateral cases are diagnosed early and almost always have a germline mutation. Unilateral tumors can also be due to germline mutation however majority cases (85 %) are sporadic. The germline mutation is inherited in an autosomal dominant fashion with ~90% penetrance.^{1,2} Children with germline mutations are also at increased risk of developing trilateral retinoblastoma and osteosarcoma^{3,4,5} and usually present at an early age.² Retinoblastoma may metastasize via direct spread into the orbit, optic nerve and into brain. Leptomeningeal, bone, bone marrow and liver metastasis can also be seen.⁶ Different imaging modalities can be used for diagnosis, extension and staging of RB. CT can depict calcifications better as compared to

MRI, however MRI is more accurate for disease extension and staging and hence is modality of choice in pretreatment staging and follow up.⁴ RB is intermediate signal intensity on T1, hypointense on T2WS compared to the vitreous. The mass usually enhances relatively homogeneously when small but larger tumors often have heterogeneous enhancement. As RB grows, it may cause retinal detachment. The growth pattern is either endophytic or exophytic. Calcification commonly seen in RB as signal void on MRI. The tumor shows restricted diffusion. DWI is also valuable in evaluating the response to eye-preservation treatment.^{7,8} Calcifications and lesions smaller than 2 mm are unreliably revealed with MR imaging. The vitreous may be abnormally bright on T1-weighted sequences because of increased globulin content and a decreased ratio of albumin to globulin that occurs with malignancy.^{1,9}

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Retinoblastoma needs a thorough clinical evaluation and complete ophthalmic evaluation including a dilated fundus examination under anesthesia. Further investigation modalities includes B scan, CT and MRI. We herein discuss the pictorial demonstration of RB on dedicated orbital MRI. The multiplanar capabilities of MR imaging provide detailed examination of tumors and retrobulbar and intracranial structures.

Discussion of cases

CASE 1:

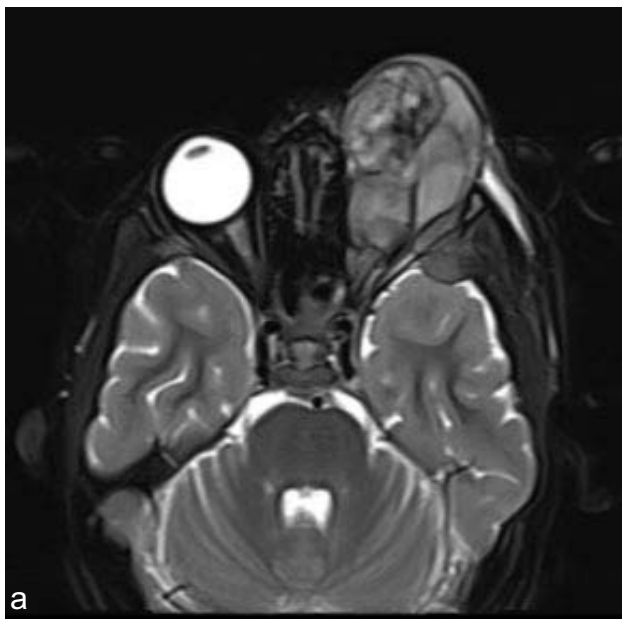
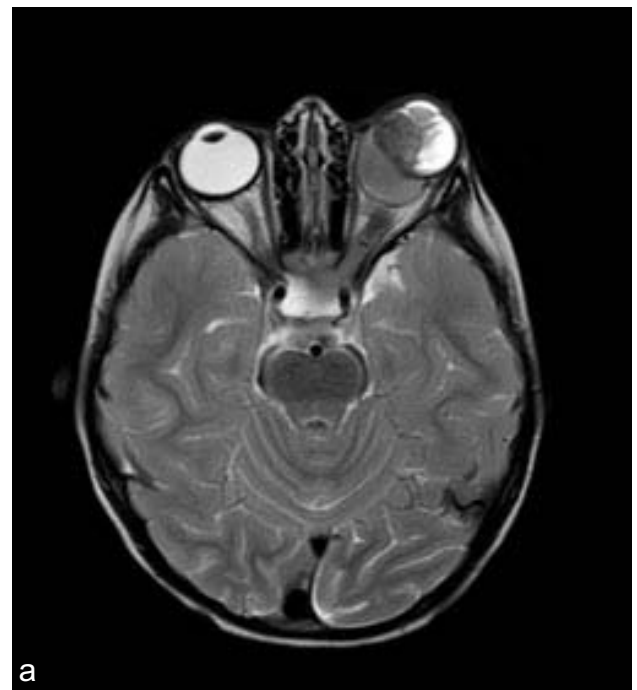


Figure 1: Axial T2 (a), CE T1W (b) sequences in 2 years old male. Left globe is deformed and replaced by heterogeneously enhancing mass (RB) which is involving retrobulbar space, extraocular muscles and intraconal part of optic nerve. Left lateral orbital wall is also involved with intracranial and extraxial soft tissue along anterior temporal lobe. Large nodal metastatic mass (c) also noted with involvement of left parotid gland.

CASE 2:



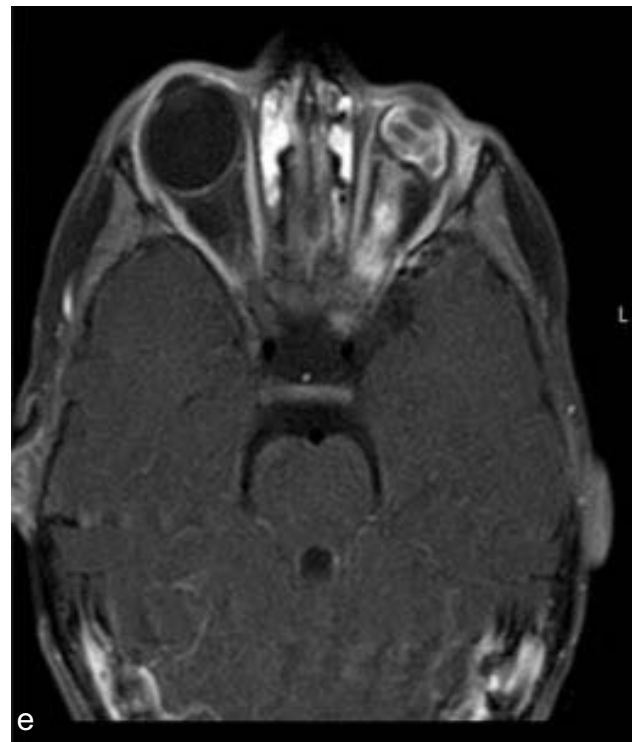
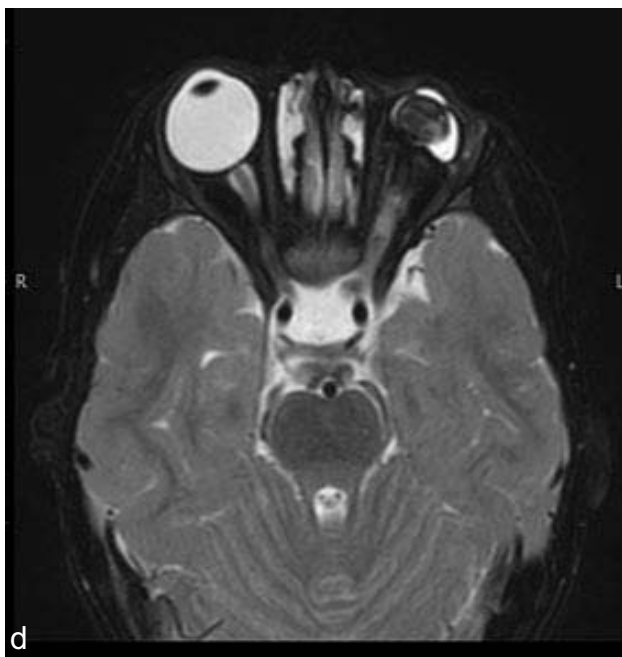
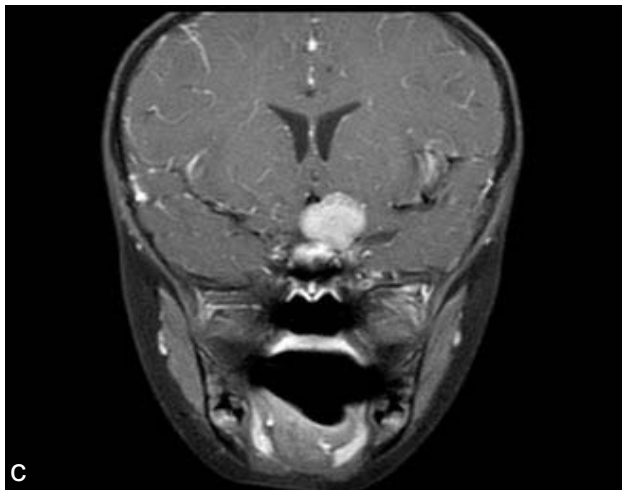
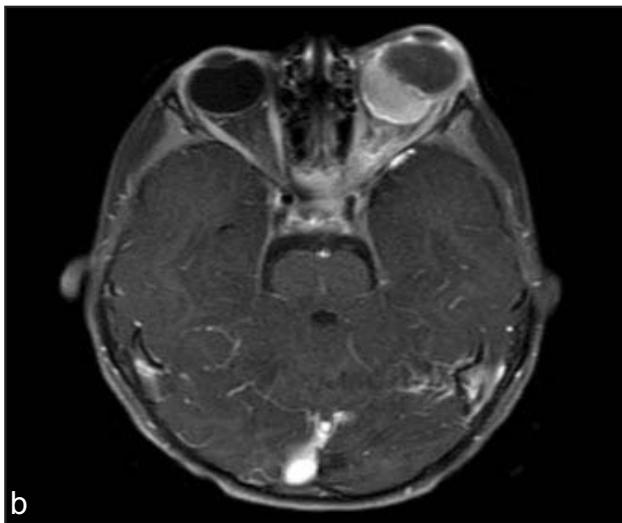


Figure 2: Axial T2 (a) and CE T1WS (b) shows enhancing hypointense mass along nasal aspect of the left globe. Transcranial spread is noted with intraconal soft tissue. Coronal T1WS (c) Contiguous extension along optic nerve to brain is noted with left sided parasellar component.

Figure 2: Axial T2 (d), axial (e) and coronal CE T1WS (f). Subsequent MRI after chemotherapy showed complete resolution of intraconal and intracranial component with enhancing residual mass in left globe (d,e). Persistent perineural enhancement is redemonstrated extending to optic chiasm (e,f).

CASE 3:

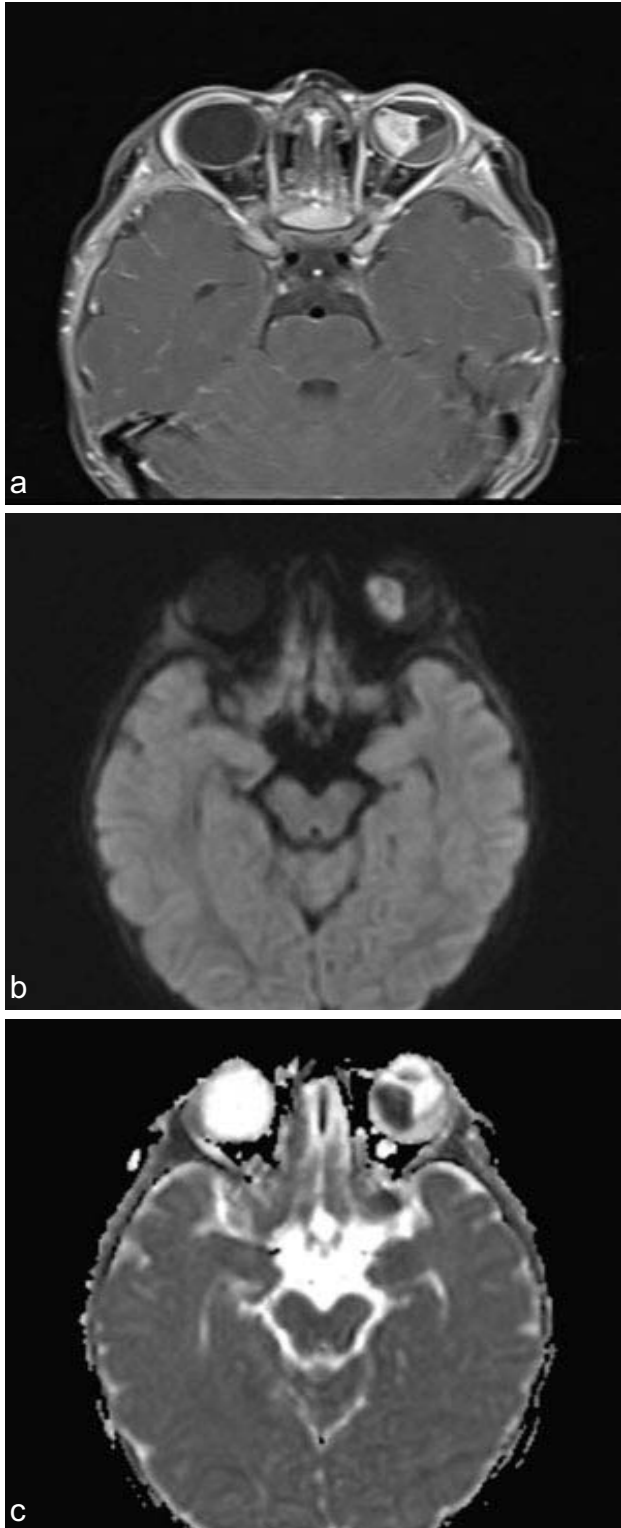


Figure 3: Axial CE T1 (a), DWI (b) and ADC (c). Diffusion restricting (b,c) enhancing vitreous chamber endophytic mass with retinal detachment and subretinal hemorrhage (a) in 1 year old male patient.

CASE 4:

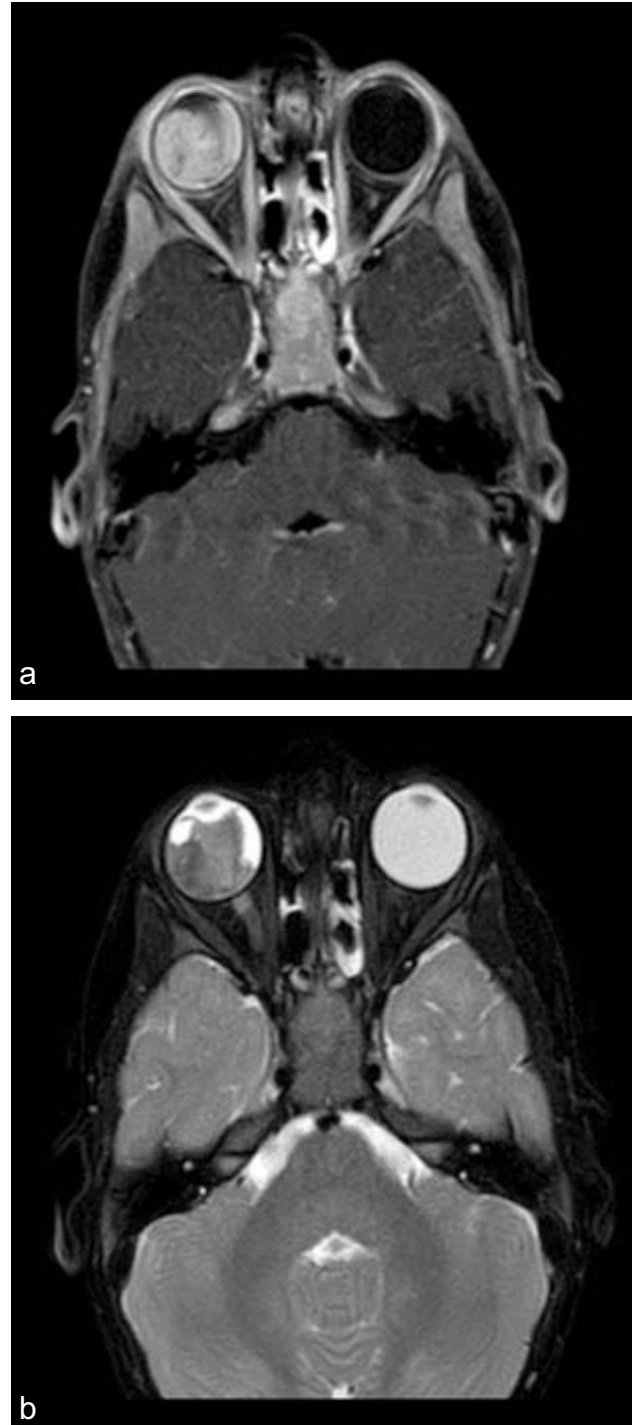


Figure 4: Axial post contrast T1 (a) and T2W (b) sequences in 2 years old female. Showing enhancing mass in vitreous chamber almost completely filling the chamber. Subsequent enucleation was done. Histopathology showed retinoblastoma, moderately differentiated (grade 2), with massive choroidal invasion (pT3a). Optic nerve margin were free of tumor. After multidisciplinary tumor board meeting, it was decided that s its intermediate risk histology. 4 cycle of JOE chemotherapy was planned.

CASE 5:

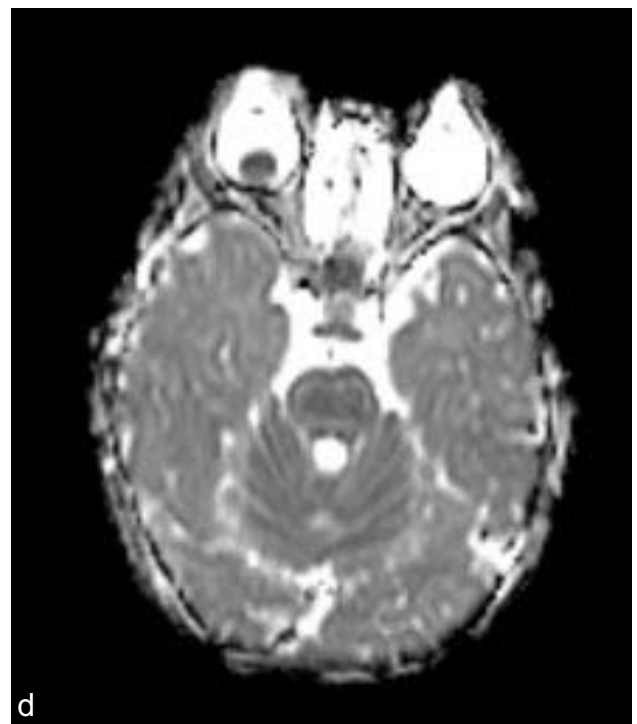
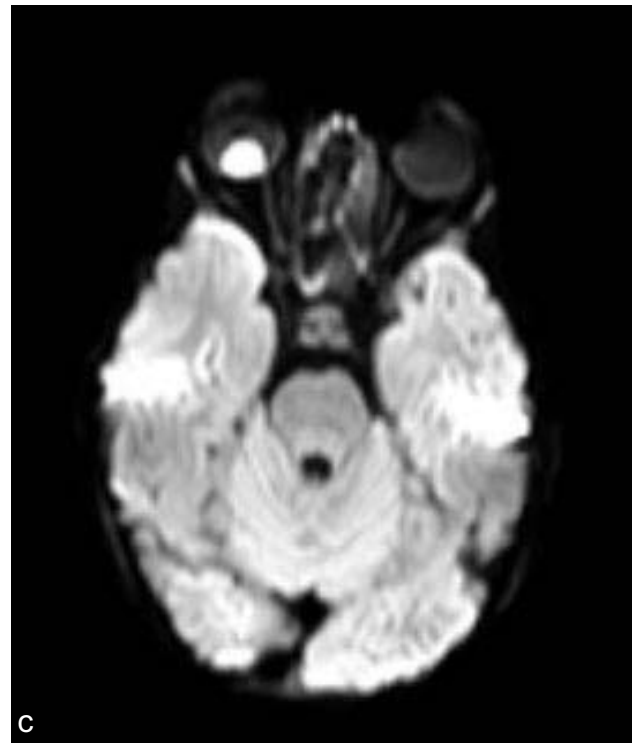
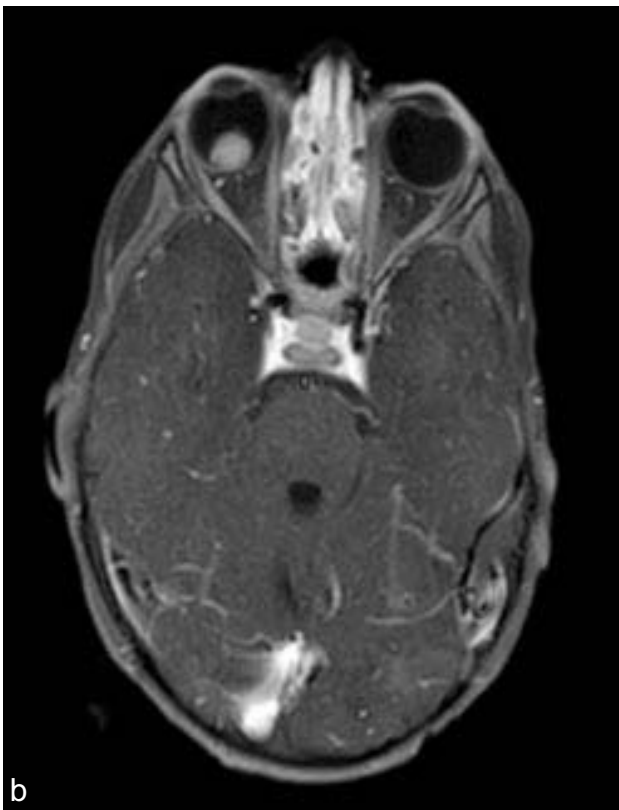


Fig 5. Axial T2 (a), CE T1 (b), DWI (c) and ADC (d). 3 years 9 months old child with white pupillary reflex since 2 months. MRI showed low signal intensity (a) diffusion restricting (c, d) enhancing retinal based tumor close to the optic disc. EAU (evaluation under anesthesia) also showed large lesion from inferior retina involving 50 % of vitreous cavity. CSF and bone marrow aspirates were negative for malignant cells and patient was started on chemotherapy.

CASE 6:

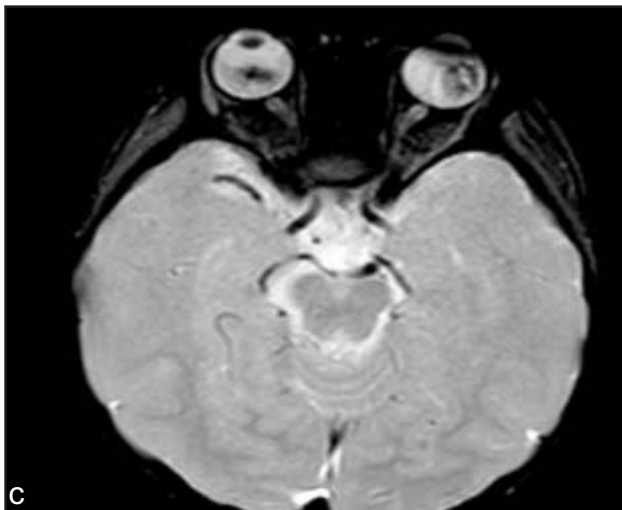
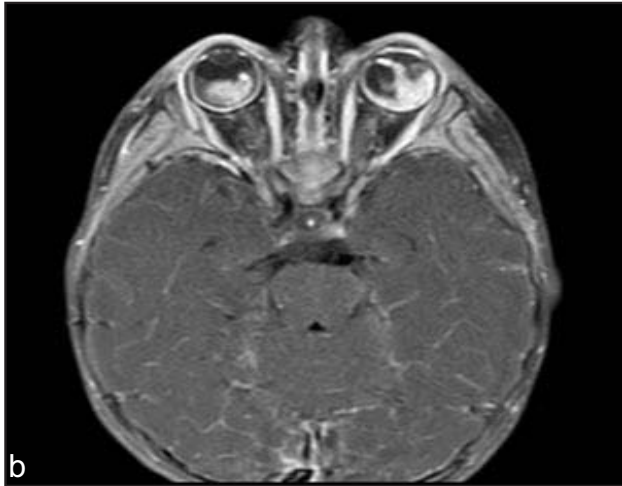
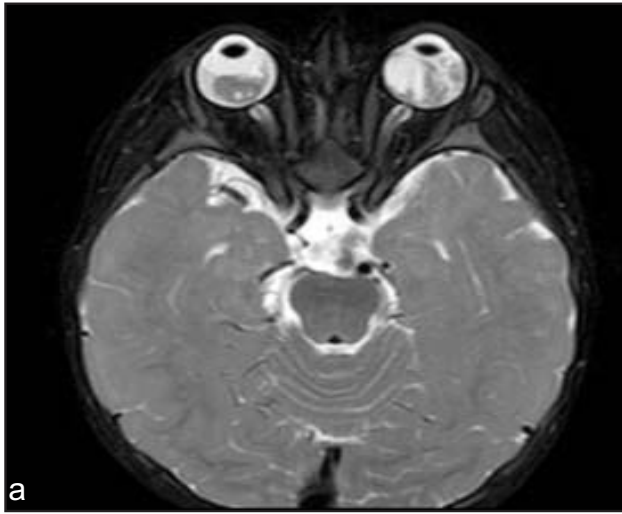
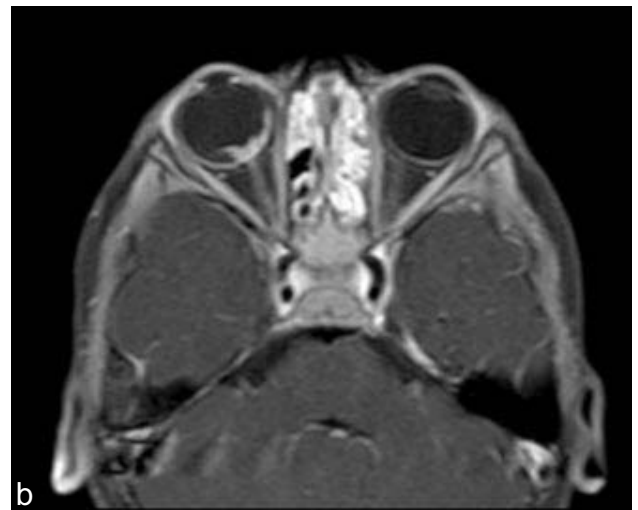


Figure 6: T2 (a), CE T1 (b) and GRE (c) images in 1 year old male baby showing bilateral enhancing masses with retinal detachment and internal signal void areas on GRE (c) suggesting calcification. EUA also showed bilateral retinoblastomas.

CASE 7:



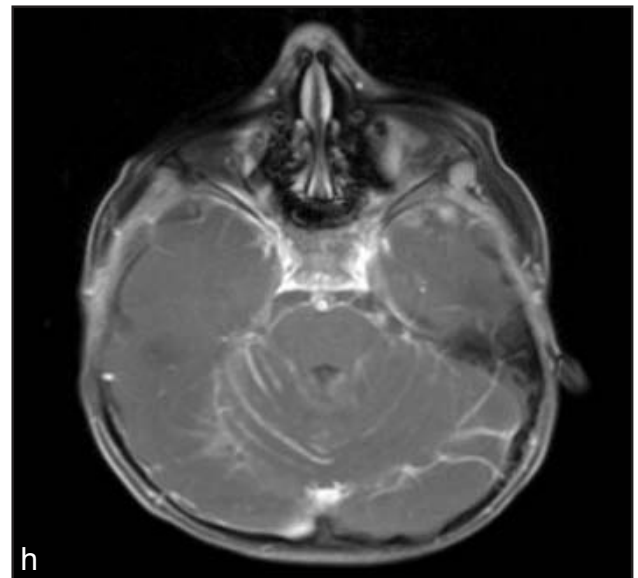
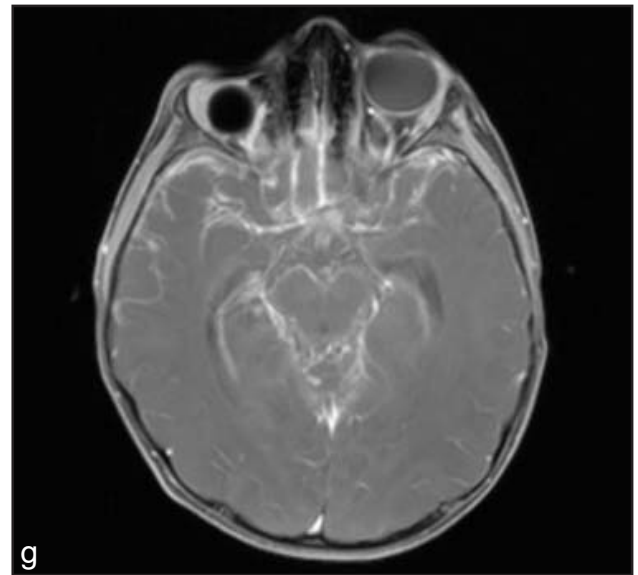
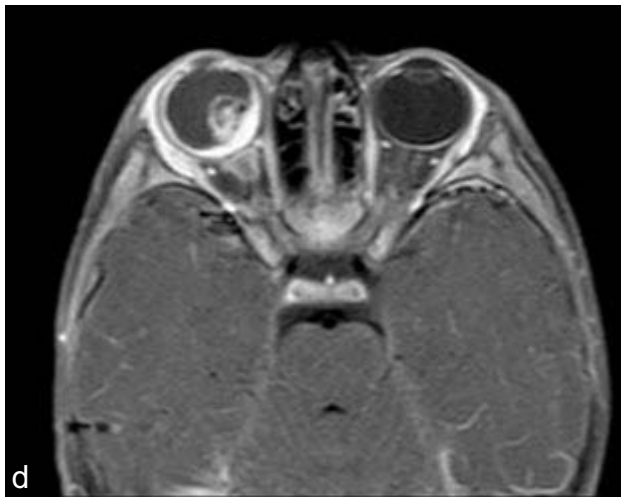


Figure 7: Axial T2 (a) and CE T1WS (b). MRI performed in April 2019 in 2-year-old male child with plaque-like retinal mass and retinal detachment.

Figure 7: T2 (c) and CE T1WS (d). MRI performed after 3 months for reassessment as the patient delayed his treatment. MRI showed progression with increase in size of the lesion and optic nerve involvement (d). CSF and bone marrow aspirates were negative for malignant cells.

Figure 7: T2 (e) and CE T1WS (f). Post enucleation MRI performed on 1st Dec 2019 which showed enhancement along the right optic nerve stump (f) and follow-up was suggested. Histopathology showed retinoblastoma pT3b. Tumor showed invasion of lamina cribrosa and optic nerve however mass was 8 mm from the optic nerve resection margin. Patient chemotherapy was planned.

Figure 7: (g) and (h). Subsequent MRI performed on 25th Dec. 2019 as child presented with vomiting. Clinical suspicion was high for disease progression. MRI showed diffuse nodular leptomeningeal enhancement along the cerebral convexities and cerebellar folia more pronounced in the basal cisterns and right orbital apex and was consistent with leptomeningeal metastasis.

CASE 8:

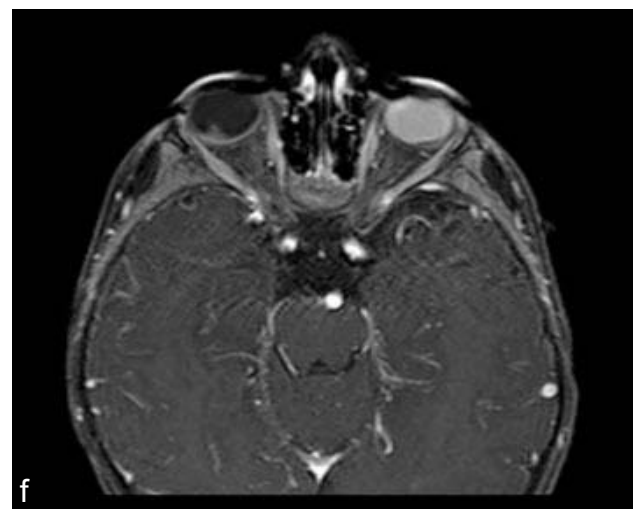
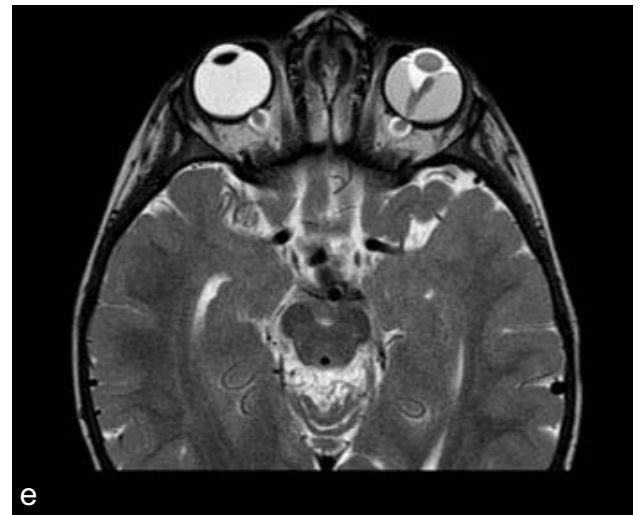
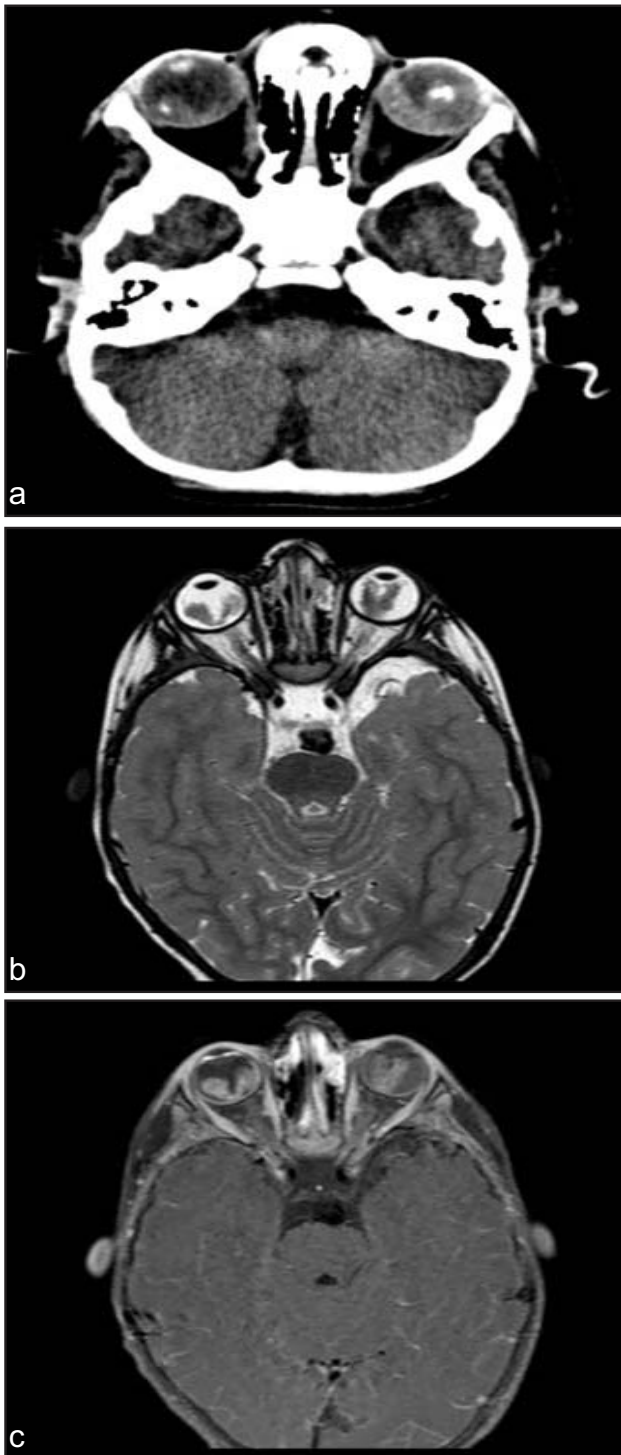


Figure 8: Axial CE CT (a) and axial MRI T2 (b) and CE T1WS (c). CT scan performed in nov 2015 showed bilateral enhancing masses in vitreous chamber with calcifications in 2 years old male (a). MRI showed bilateral enhancing retinoblastomas with retinal detachment (b,c). EUA was also consistent with bilateral RB. CNS / bone marrow aspirates were negative for metastasis. Patient was started on chemotherapy.

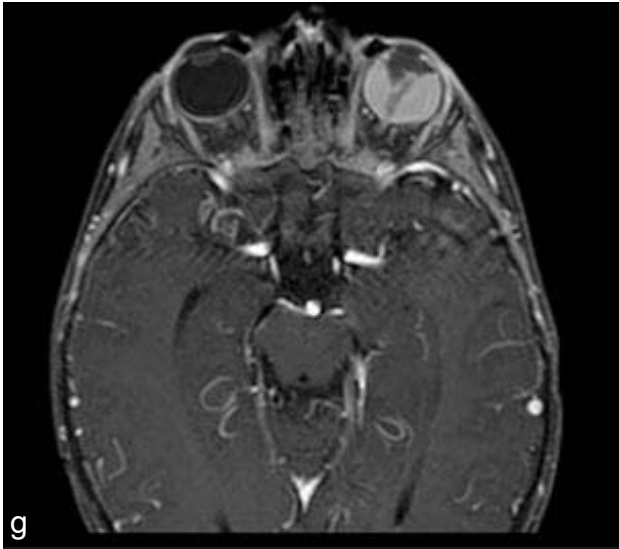


Figure 8d, e, f, g: Feb 2016 mid treatment MRI, axial T2 and CE T1WS. Showed reduction in size of bilateral retinoblastoma. Intraorbital haemorrhage and persistent retinal detachment on left (e,f,g). No intracranial abnormality was noted. Patient continued treatment.

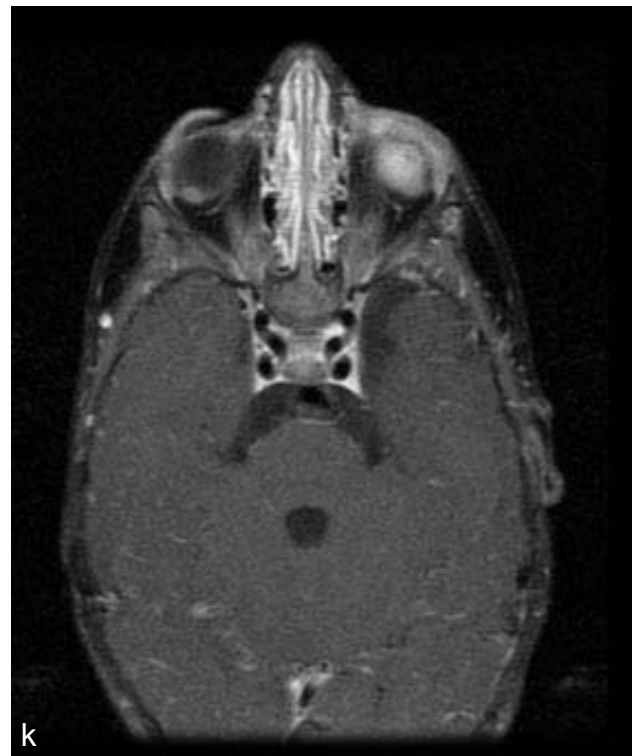
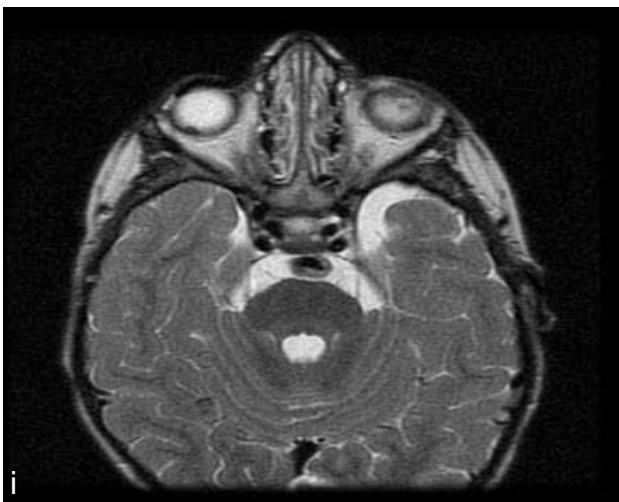
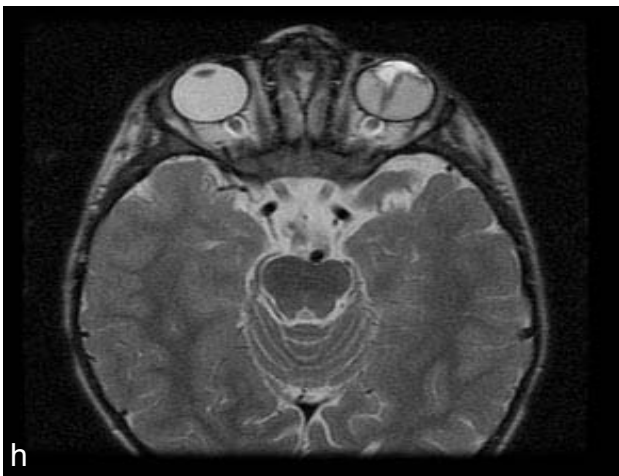
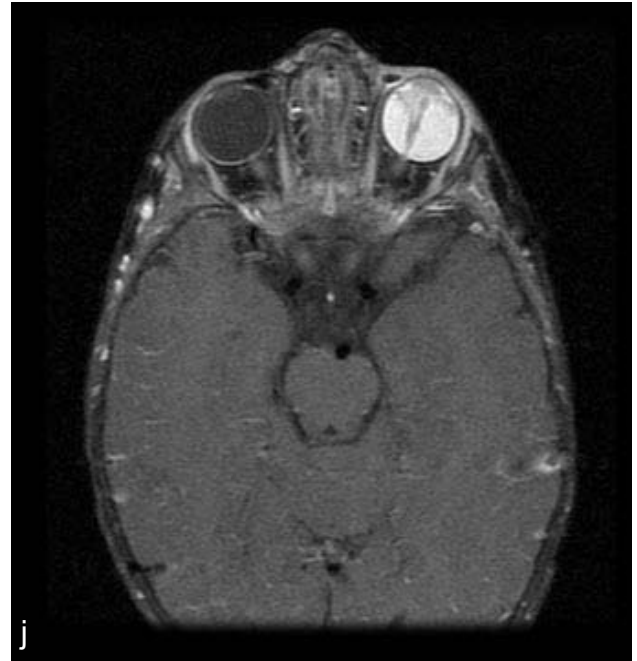


Figure 8h, i, j, k: MRI performed in may 2016 after completion of 8 cycles of JOE chemotherapy. Reduction in size of right mass which appears as nodular lesion (i,j). Left sided changes of RD remained stable. No diffusion restriction within left globe.

Aug.2016. Left sided enucleation performed. HP of left globe showed retinoblastoma invading lamina cribrosa. Optic nerve resection margins were free of tumor. No choroidal or scleral invasion was seen. MRI showed stable changes of small plaque like thickening in right vitreous chamber.

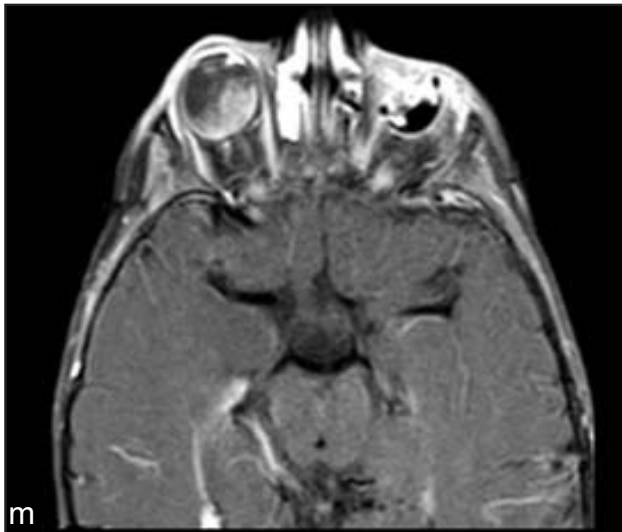
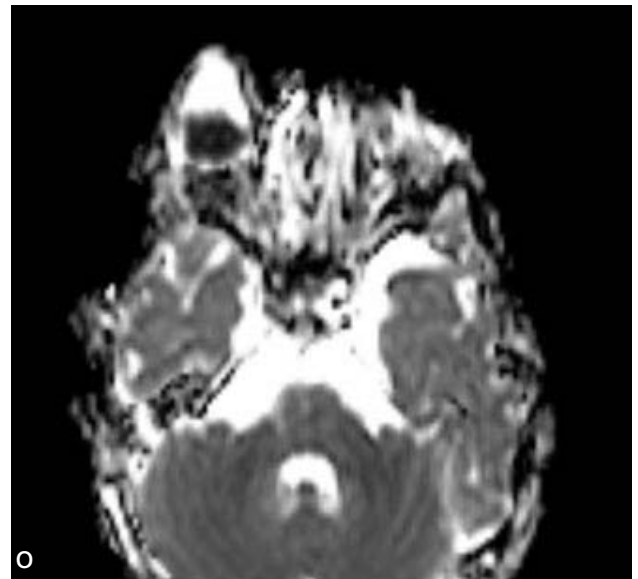
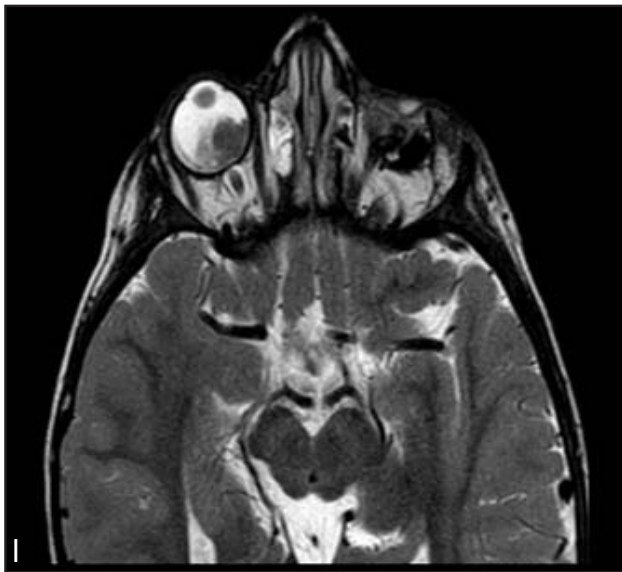


Figure 8l, m, n, o: June 2017. Subsequent surveillance MRI stable enucleation changes on left without recurrence and right sided progressive disease with enlargement of RB (**l,m**). Laser photocoagulation treatment / IV melphalan and Cryotherapy for right globe lesion was performed. Subsequent second line chemotherapy was also given. EUA still showed persistent disease in right globe. Patient workup was negative for intracranial / bone marrow metastasis. Finally due to persistent disease right enucleation was performed in sep 2018. HP of right enucleation showed residual 20mm retinoblastoma. Lamina cribrosa and optic nerve were free of tumor. Primary Tumor was pT1. Microscopic evaluation showed neoplasm composed of round blue cell arranged in sheets forming rosettes.





Figure 8p, q: Nov 2019 MRI showed bilateral enucleation with prosthesis without disease recurrence.

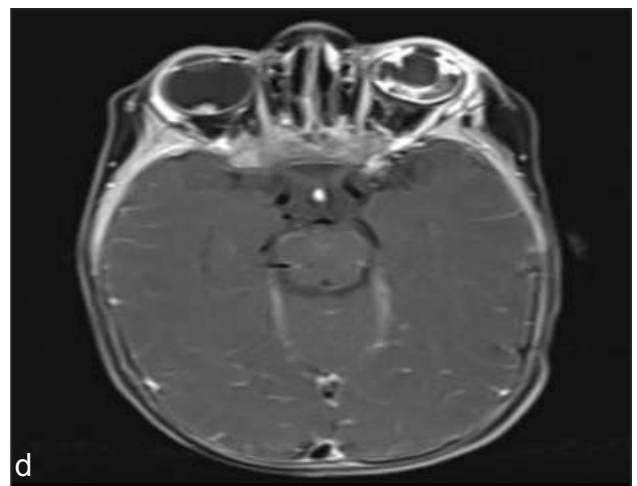
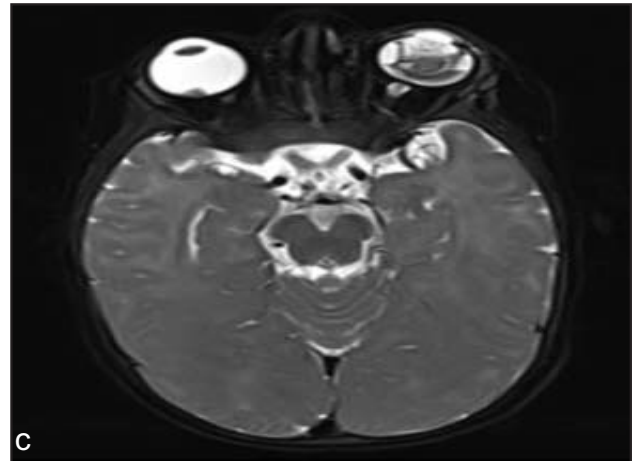
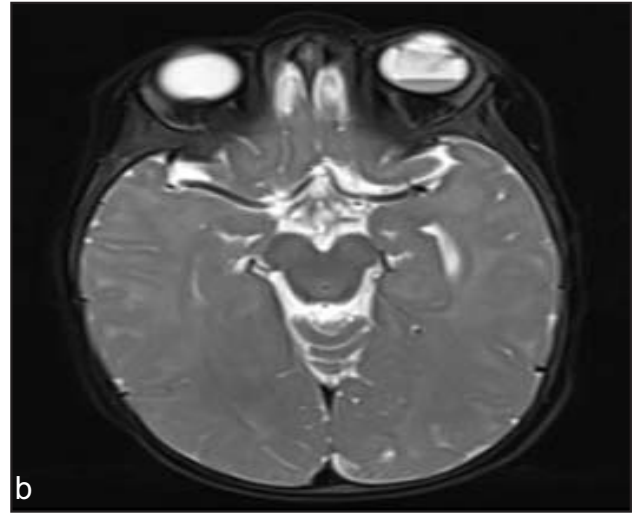


Figure 9: Sagittal (a) & Axial T2 (b,c) and CE T1WS (d) in 2 years 2 months old male with trilateral retinoblastoma. Small cystic lesion in pineal gland (a), small enhancing nodular RB in right globe (b,c) and retinal based mass in left vitreous chamber with retinal detachment and hemorrhage forming fluid level (b). Lumbar puncture and bone marrow aspirates were negative for malignant cells. Upfront enucleation of left globe was decided with 6 cycles of chemotherapy.

CASE 9:



Conclusion

MRI is the modality of choice to detect intra-ocular, extra-ocular and intracranial extension of the tumour. MRI is very helpful in determining pretreatment extent and monitoring treatment response. The tumour extension is not well delineated with other imaging modalities and MRI should be used to answer the key clinical questions that help in the selection of an appropriate line of treatment.

Conflict of interest: None

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