OPTIC DISC MELANOCYTOMA- A RARE CASE

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PRESENTATION OF CASE

A 22-year-old gentleman from Assam presented to Ophthalmology Department with complaints of squinting of right eye since 5 years of age. He had no significant illness in the past or in family.

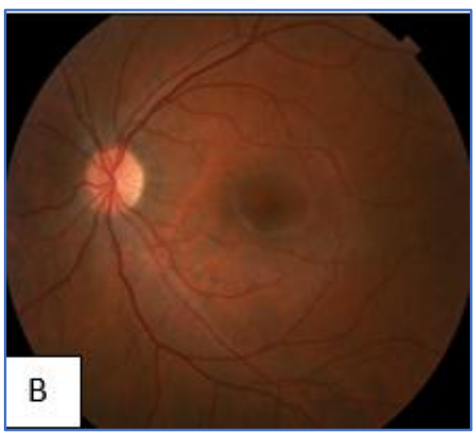
ON EXAMINATION

He had a right divergent squint of 30 degrees. Best corrected visual acuity was 6/18 in the right and 6/6 in the left. Anterior segment examination was normal in both eyes except for grade IV relative afferent pupillary defect (RAPD) in the right eye. Fundus examination of right eye showed a large pedunculated brownish-black mass protruding into the vitreous and almost covering the entire optic disc sparing a small superonasal disc border with peripapillary choroidal extension inferior to disc more than superior. Vessels over disc were obscured by lesion. Surrounding retina showed no haemorrhages, oedema or lipofuschin deposits.

Figure 1. Right Divergent Squint

Figure 2. Fundus Picture

A) Right Eye Showing Optic Disc Melanocytoma



B) Normal Left Eye

DIFFERENTIAL DIAGNOSIS

Though a clinical diagnosis of optic nerve melanocytoma was made, it is necessary to distinguish it definitely from more dangerous close mimickers like optic nerve melanoma, juxtapapillary choroidal melanoma and choroidal haemangioma.

CLINICAL DIAGNOSIS

Ultrasonography (USG) showed 2 x 2 mm dome-shaped optic nerve mass with a scan showing moderate-to-high internal reflectivity with no internal vascularity.

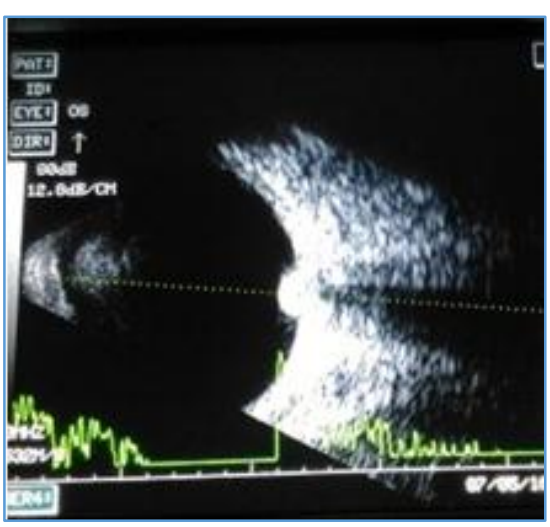


Figure 3. USG B-Scan showing Elevated Lesion with a Scan showing High Reflectivity

Automated perimetry with Humphrey field analyser (HFA) 30-2 showed near total inferior field defect with evolving superior arcuate scotoma in the right eye.

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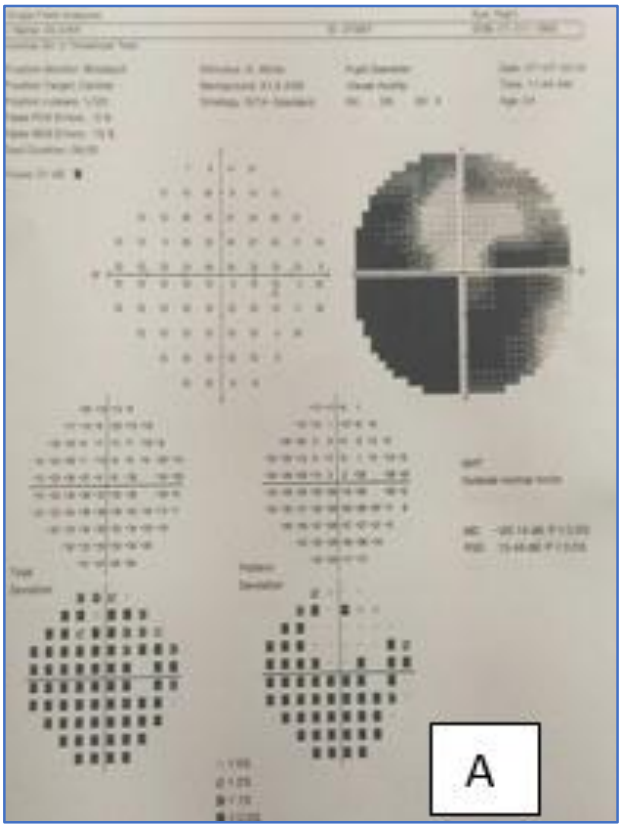
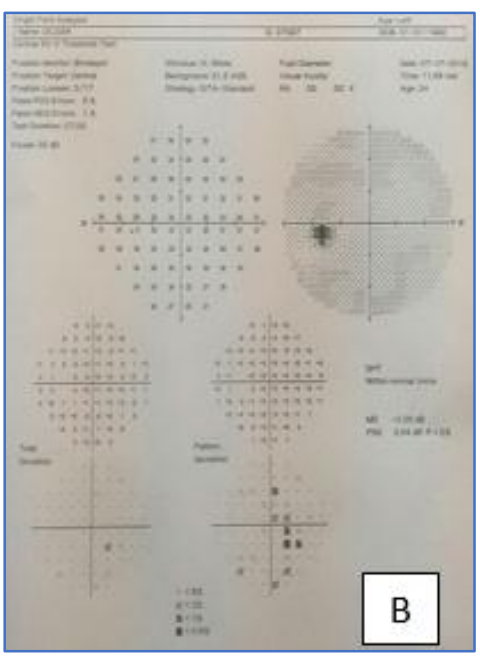
Figure 4.



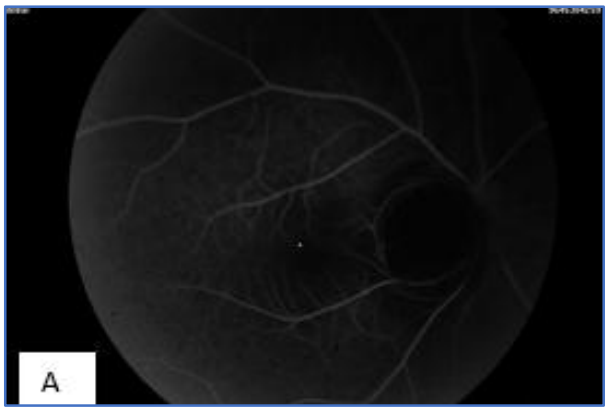
A) Field Defect in Right Eye



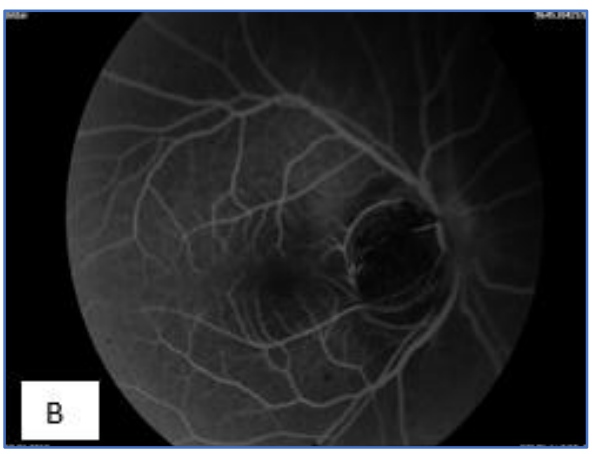
B) Normal Field in Left Eye

Fundus photo was taken and showed no autofluorescence. Fluorescein Angiography (FA) showed persistent hypofluorescence in all phases with fine vessels over the lesion. Late staining was seen all around the disc.

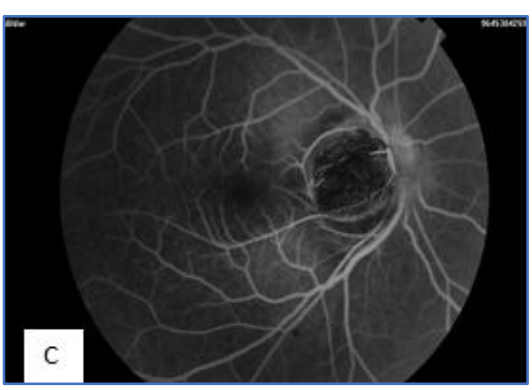
Figure 5. Fluorescein Angiography



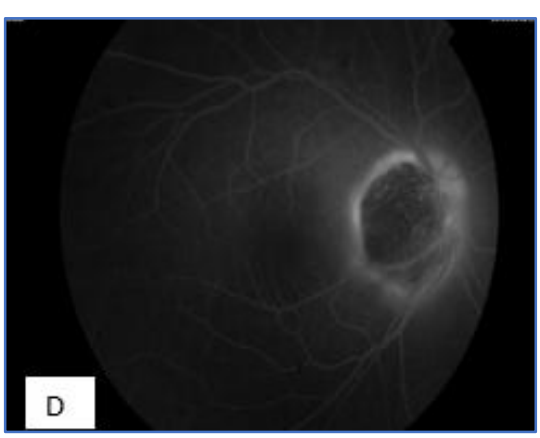
A) Hypofluorescent Lesion



B) Persistent Hypofluorescence with Early Filling of Fine Vessels over Lesion



C) Persistent Hypofluorescence with Complete Filling of Fine Vessels over Lesion



D) Late Staining around Disc

3D Topcon Optical coherence tomography (OCT) showed dome shaped lesion in the anterior portion of optic nerve with echogenic line delineating the anterior border of lesion and back shadow obscuring underlying details of optic nerve.

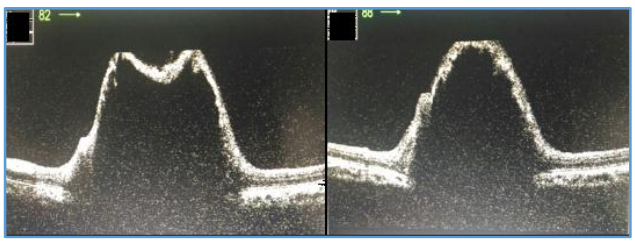


Figure 6. OCT showing Dome-Shaped Lesion with Back Shadowing

Magnetic resonance imaging (MRI) showed T1 intermediate and T2 hypointense signal, nodular lesion of 2mm size at the anterior end of the optic nerve with no post contrast enhancement or extraocular extension.

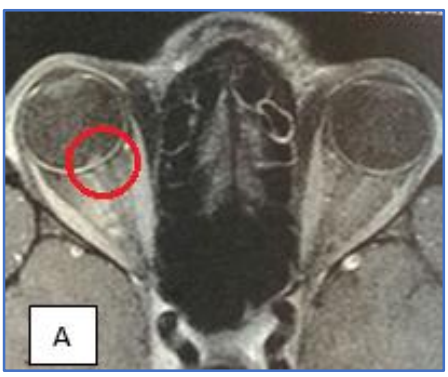


Figure 7. MRI. (A) ***Axial Image with T1 Intermediate Intense Lesion***

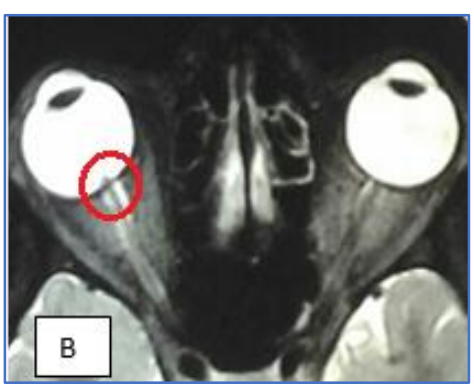


Figure 7. (B) T2 Hypointense Lesion

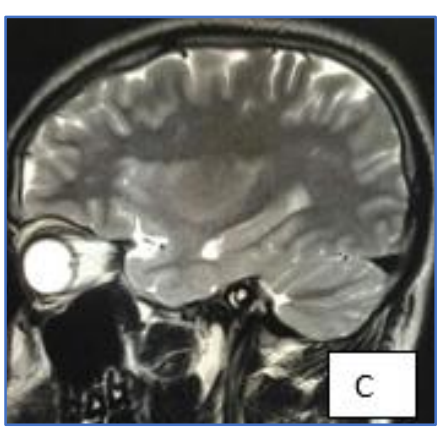


Figure 7. (C) Sagittal Image with No Post Contrast Enhancement

PATHOLOGICAL DISCUSSION

Pigmented lesion of optic nerve have always raised suspicion of malignant melanoma. Melanocytoma is described as a benign dark brown or black lesion, flat or slightly elevated with feathery margins extending over disc. It may be confined to disc (15%), involve adjacent choroid (54%) and retina (30%). Complications may be associated with melanocytoma like disc oedema (25%), intraretinal oedema (16%), subretinal fluid (14%), intraretinal exudation (12%), focal haemorrhages (5%), vitreous seeds (4%) and retinal vein obstruction (3%).[1] Our patient had a large brownish-black pedunculated lesion protruding into the vitreous covering the entire optic disc with choroidal extension. Absence of subretinal fluid, retinal oedema and lipofuschin pigment are against the diagnosis of malignant melanoma.

Most cases of melanocytoma are asymptomatic and detected incidentally. Slight visual loss can occur (26%) due to retinal oedema or subretinal fluid. Severe visual loss can occur due to retinal vein occlusion, tumour necrosis or malignant transformation.[2-4] Visual field defects include blind spot enlargement related to tumour extension beyond disc margin and arcuate defects related to compression of axons in disc.[5] Even with excellent visual acuity 10% - 30% of eyes with melanocytoma show RAPD due to compression of optic nerve fibres.[2,6] Affected eye of our patient had divergent squint with best corrected vision 6/18, RAPD and severe field defect corresponding to tumour.

Shield et al review on 116 eyes using Kaplan-Meier estimates showed that minor tumour enlargement occur in 11% by 5 years and 32% by 10 years.[2] This mild growth should not be misconstrued as malignant transformation.[7] There are reports that progressive enlarging tumours have undergone enucleation and biopsy proved benign. Analysis showed that initial thickness > 1.5 mm and nodular configuration is a predictive risk factor for growth.[2] Our patient had tumour of size 2 mm with nodular configuration, therefore at high risk of growth. But malignant transformation occurs in 1% - 2%. It is not always possible to distinguish two pathological processes clinically.

Melanocytoma can be assessed by ultrasound when elevation > 0.5 mm. Cervantes et al described that internal reflectivity in melanocytoma range from medium to high, unlike choroidal melanoma with low reflectivity. Avascularity of the tumour points towards benign nature of lesion.[8] The melanocytoma of our patient was avascular and had high internal reflectivity again pointing towards the benign nature.

OCT shows dome-shaped peripapillary elevation with dense back shadowing and occasionally vitreous seeds.[9] Thin echogenic line delineates the anterior aspect of the lesion. Retinal involvement appears as dense area in nerve fibre layer and choroid involvement as elevation of retinal pigment epithelium. In this case, OCT showed typical features as described. OCT angiography (OCT-A) allows visualisation of vascularisation using motion contrast principle, thus can show choroidal involvement and neovascularisation. Hence, OCT-A could be a non-invasive alternative to assess the depth of tumour and choroidal involvement without contrast medium.[10]

Fluorescein angiography of melanocytoma shows dense hypofluorescence throughout angiogram, because cells are

pigmented and compact with less vascularity. In disc oedema, hyperfluorescence is seen adjacent to tumour.[11]

Typical description was noted in our case. Increased surface vascularity as in our case has been found to correlate with tumour growth.[12]

MRI grossly detect the retrolaminar extension of melanocytoma using gadolinium enhancement.[13] Extensive involvement of optic nerve with severe visual loss may suggest malignant transformation. MRI cannot help distinguish melanoma and melanocytoma, as both appear hyperintense on T1 and hypointense on T2 images.

FINAL DIAGNOSIS AND MANAGEMENT

As all investigations confirmed benign nature of lesion, final diagnosis of optic nerve melanocytoma was made. Melanocytoma in this case has high chance of growth due to characteristics like increased thickness, elevated lesion and surface vascularity. Chance of malignant transformation is low, but requires careful yearly follow-up including all the investigation modalities mentioned. If there is progressive growth with severe visual loss, biopsy confirmation followed by enucleation should be considered. He was followed up after 3 months and since the status of tumour remained same a cosmetic squint surgery was done explaining the necessity for constant yearly followup.

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