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Case Report

Incidental detection of teratoid benign medulloepithelioma of cilliary body of eye- A rare case report & literature review

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ABSTRACT

Intraocular medulloepithelioma of cilliary body is a nonhereditary neoplasm of childhood arising from primitive medullary epithelium characterized by cords of multi-layered neuroepithelial cells resembling medullary epithelium of the optic cup. It often present as loss of vision, painful eye & leucocoria. Common signs include secondary glaucoma & dislocated lens. About half of patients undergo surgery before discovery of this tumor. We report a case of cilliary body teratoid benign meduloepithelioma in a 4 year female child, presented as phthisical eye, detected incidentally & relevant literature.

1. Introduction

Intraocular medulloepithelioma is a congenital tumor, which arise from the nonpigmented ciliary epithelium of the eye. Medulloepithelioma is the second most common primary intraocular neoplasm of childhood after retinoblastoma, median age group of 2 -5 years, female preponderance(63%).[1–3] Although rare cases have been described in adults. Due to extreme rarity of medulloepithelioma, very few case report/series published in English literature till date.

Most common site of origin is inner surface of ciliary body, but rarely arise from optic nerve & retina. Clinically it may remain as asymptomatic leads to delay in diagnosis, common presentations are poor vision or blindness (27%), cataract, leucocoria(20%), strabismus(12%), red painful eye(20%) & high intraocular pressure(44%).[4] According to histopathologic findings, intraocular medulloepithelioma is further classified as nonteratoid or teratoid, and benign or malignant.[5]

We report an incidental detection of teratoid benign medulloepithelioma presenting with a mass in a phthisical eye in a 4-year female child.

2. Case Report

A 4-year old female child presented in outdoor patient department as gradually dimness of vision in the right eye. She was admitted with painful blindness in her right eye that had developed after a trauma. On ultrasound biomicroscopy & slit lamp examination was non-contributory. In view of the above clinical findings, differentials like retinoblastoma or organized hematoma were considered. Birth, family & sibling history were unremarkable. Her left eye was unremarkable. Right eye was phthisis bulbi & planned for enucleation.

An informed consent was taken, enucleation of right eye was done. On gross examination of the sectioned globe revealed a white firm mass arising from the ciliary body of the eyeball meas. 1.8x1.5 cm. without extraocular extension. Histopathological examination revealed interlinking bands and cords of neuroepithelial cells with scant cytoplasm

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in loose mesenchymal tissue & foci of glial tissue & rosettes were seen (Figures 1 and 2). No malignant features was observed. No evidence of extraocular extension of tumour was seen. The diagnosis of teratoid medulloepithelioma was made & confirmed by positive stain of synaptophysin, chromogranin & neuron specific enolase of immunohistochemistry (IHC). No further genetic analysis was done. After 4 years, follow up is uneventful.

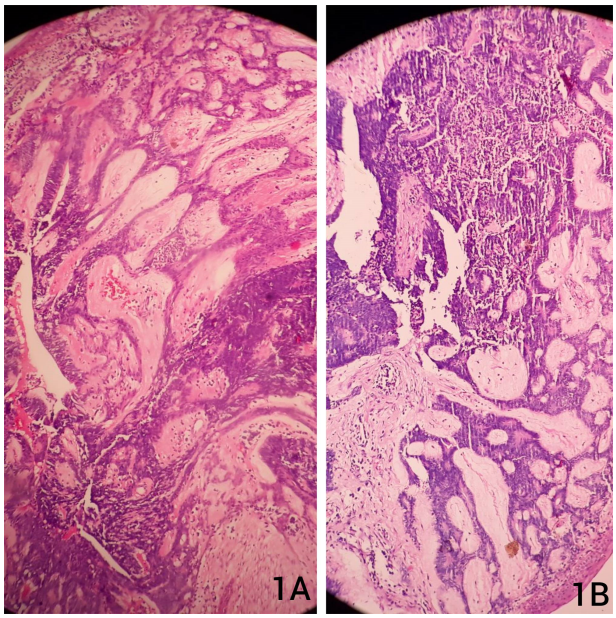

Fig. 1: A & B): H & E stain(10x) shows cords & loops of medullary neuroepithelial cells separated by pale mesenchymal stroma

3. Discussion

Medulloepithelioma is a rare embryonal neuroepithelial neoplasm of the eye arising from the non-pigmented ciliary epithelium. Fuchs renamed the tumor as "diktyoma" means a network of medullary epithelial bands. In 1931, Grinker introduced the terminology of "medulloepithelioma". It has been accepted and favoured since its discovery.[5] DICER1 is required for the final production of miRNAs; the gene encodes a ribonuclease that participates in miRNA formation.[6] There has been systemic association of ciliary body medulloepithelioma and related pleuropulmonary blastoma due to germ line mutation of DICER-1.[4] On the basis of these findings, it seems that <1% of patients with PPB manifest ciliary body medulloepithelioma, and 5% of patients with medulloepithelioma have a history of PPB.[5]

Ciliary body medulloepithelioma clinically present as secondary glaucoma, cataract, staphyloma, anterior uveal cyst/ uveitis.[3] Ultrasound bio microscopy and Optical coherence tomography(OCT) reveal the medium to highly reflective ciliary body mass with intratumoral cystic spaces.[7]

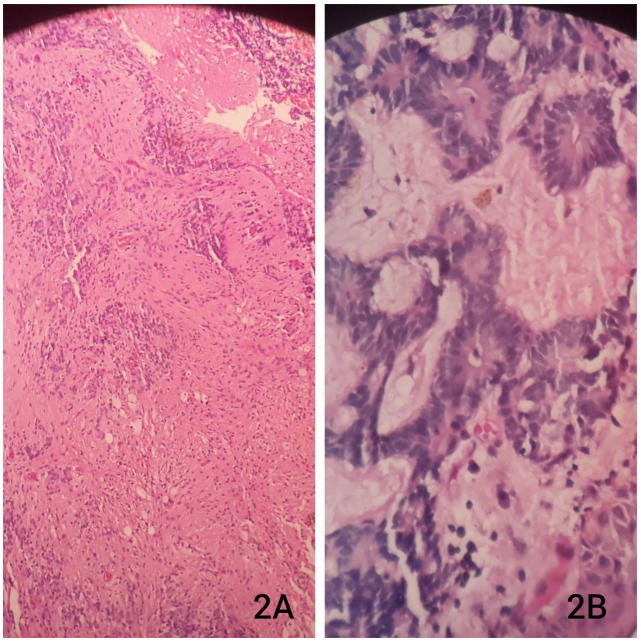

Fig. 2: A): H&E stain(40x) shows pink fibrillary glial brain like tissue; **B):** H&E stain (40x) shows pseudo stratified medullary epithelium forming rosettes of varying size

On histopathology, meulloepithelioma is characterized by poorly differentiated neuroepithelial cells arranged in cords & sheets, surrounded by hyaluronic acid rich mesenchymal tissue & associated with Homer Wright & Flexner–Wintersteiner rosettes.[2]

The non teratoid medulloepithelioma shows proliferation of primitive medullary epithelium, whereas teratoid medulloepithelioma shows, hyaline cartilage (most common), brain like tissue, rhabdomyoblasts in addition to the proliferation of the neuroepithelial elements.

Broughton and Zimmerman proposed the histopathological criteria for malignancy in ocular medulloepithelioma.[2] According to these criteria, approximately 66%–80% of cases are classified as malignant.[2,5] However, clinical outcome has not been found to correlate with the classification using these criteria.According to WHO classification tumour of eye, proposed three tiered grading system in place of benign and malignant medulloepithelioma.[4] As per Grade I tumour corresponds to current WHO designation-benign, Grade II tumours are progressive characterized by pleomorphism,increased mitosis & local invasion. Grade III tumours have malignant transformation, extrascleral extension & metastasis spread.[8] Our case would be classified as grade- I as per new grading system.

Kaliki et al, found the median age group was 5 years and 78% of cases occurred in the 0-10 years of life, large number of cases were initially misdiagnosed (88%)

and 39% of cases had an additional history of treatment. Systemic metastasis occurred in 3 cases (8%) over a mean follow-up of 49 months.[5]

Retinoblastoma may be the most challenging differential diagnosis of medulloepithelioma, when typical medullary epithelium is sparse.[1] LIN28A staining is specific for medulloepithelioma, uniformly negative in retinoblastoma. Persistent fetal vasculature, cilliary body cyst & cataract are also comes in differential diagnosis. Therapeutic inhibitors of the LIN28/let-7/mTOR pathway such as rapamycin, or agents that interrupt DNA methyl transferase (5-azacytidine and vorinostat) may serve as potential useful therapies in the future.[9,10] Intraoccular medulloepithelioma often shows cytogenetic aberration in chromosome 1p,4,8 & 16p.[11] Recently, somatic mutations of KMT2D along with DICER1 have been identified in intraocular medulloepitheliomas, though understanding of involvement of genes & pathogenesis of the disease have not been clearly outlined.[12]

Treatment modality for medulloepithelioma include cryotherapy(smaller and recurrent tumour), local resection, plaque radiotherapy, external beam radiotherapy and enucleation. Enucleation has proved to be an effective treatment with very good outcome in the tumour localised to the globe.[2–4]

Ciliary body medulloepithelioma is uncommonly metastasize. It may lead to death in cases with extraocular extension, orbit involvement & intracranial extension.[2,3]

To conclude, In our case shows female child presenting as phthsical eye with prior history of trauma & enucleated for painful blind eye, clinically not suspected for any tumour, incidentally discovered ciliary body teratoid benign medulloepithelioma, grade- I, no extra ocular extension & uneventful follow up till date.

Ciliary body medulloepithelioma most commonly presented as cataract & glaucoma in pediatric age group.[5] Patients are often treated for the secondary complications before the actual mass is discovered leading to a delay in diagnosis & appropriate management. Awareness among ophthalmologist & pathologist of such tumours is necessary to avoid delays in referral or mismanagement of cases as glaucoma or uveitis. As ultrasonography is an efficient additional diagnostic tool & characteristic histopathology features will help to arrive early diagnosis.

4. Source of Funding

None.

5. Conflict of Interest

None.

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