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Case Report

Multimodal Imaging in a Case of Atypical Choroidal Osteoma

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Abstract

A 19 year old male presented with low vision in right eye since 3 years. Clinical examination and investigations revealed a peripapillary, depigmented choroidal osteoma. The presence of pseudo-disc edema and retinal nerve fiber layer (RNFL) edema, and male sex were unusual for choroidal osteoma. However, multimodal imaging in this case helped to arrive at a diagnosis.

Keywords: Choroidal Osteoma; Secondary CNVM; Pseudo-Disc Edema

Introduction

Choroidal osteoma is an ocular tumour which is benign in nature and of unknown etiology, characterized by the presence of cancellous bone within the choroid, first described by Van Dykin (1975) [1]. It affects females in two-thirds of the cases, in their second or third decade of life, and is unilateral in 75% of cases [2-6]. The pathogenesis of the choroidal osteoma remains unknown. The speculated pathogenesis includes possible traumatic, inflammatory, choristomatous, metabolic, hormonal, hereditary or environmental etiologies [3].

Choroidal osteomas are typically flat orange-yellow lesions with distinct margins that are often located in the juxta-papillary or peripapillary area, with or without extension into the macula. It can rarely be confined to the macular region without the involvement of the juxta-papillary area [5-7].

Patients with choroidal osteoma are often asymptomatic; however, the development of secondary choroidal neovascular membranes (CNVM) remains the most common cause of low vision in these patients. Symptoms include metamorphopsia, visual loss, and visual field defects corresponding to the location of the tumor [3,5-7].

Case Report

A 19 years old male was referred to the outpatient department (OPD) with gradual diminution of vision in the right eye since 3 years. There was no relevant previous ocular or medical history. Additionally, the patient had no history of trauma. The patient did not have any systemic illness.

The best-corrected visual acuity at the time of presentation in the right eye was 20/50, N12, and 20/20, and N6 in left eye for distance and near, respectively.

Right eye fundus examination revealed a well-defined, yellowish- white choroidal lesion below the optic nerve head and around the peri-papillary region, measuring approximately 6 disc diameter (DD) in the area. Most of the lesion appeared yellowish white in colour with a small area near the supero-nasal margin

with orange-yellow pigmentation. There was an outer retinal atrophy surrounding the lesion, with hyper-pigmentation at its temporal edge. Temporally, the lesion extended to the edge of the fovea with sub-retinal fluid (SRF). Clinically, no CNVM complexes were identified (Figure 1a). The disc margin was blurred and was elevated from the retinal surface by approximately five dioptres.

Fundus autofluorescene (FAF) showed an area of central hypo-autofluorescence due to an optic disc with surrounding hyper-autofluorescence corresponding to the osteoma lesion. Patchy hypo-autofluorescence in the periphery is due to atrophy of the retinal pigment epithelium (RPE) and decalcified lesion (Figure 1b).

Fundus Fluorescein Angiography (FFA) revealed early, mild, patchy hyperfluorescence (Figure 1c) of the tumor that evolved into diffuse intense late staining (Figure 1d). Hyperfluorescence was observed throughout the lesion. No evidence of leakage from the optic disc was found.

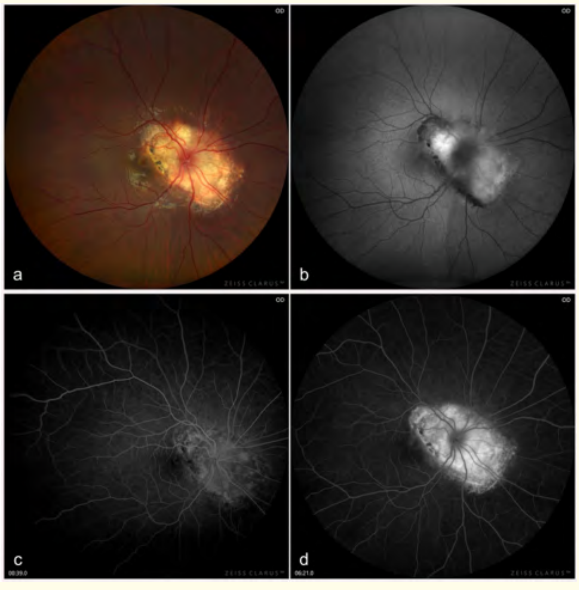

Figure 1: (a-d): (a) – Fundus photograph of Right Eye showing a well-defined yellowish white choroidal lesion below the optic nerve head and the parapapillary region measuring approximately 6 DD in area with disc and RNFL edema. (b) FAF shows area of central hypo autofluorescene due to optic disc and area of surrounding hyper autofluorescence corresponding to osteoma lesion. Peripheral hypo autofluorescene representing RPE atrophy and decalcified lesion. (c) Fundus Fluorescein angiogram showing early phase of FFA showing mild, patchy hyper-fluorescene and late diffuse intense staining of the tumour. (d) No leakage from the disc.

Spectral domain optical coherence tomography (SD-OCT) of the fovea revealed a smooth vitreoretinal interface (Figure 2a). There was presence of intra-retinal fluid (IRF) along with SRF nasal to the fovea. The Inner Segment-Outer Segment (IS-OS) junction was disrupted, and the presence of a hyper-reflective sub-retinal CNVM complex was noted.

Ultrasound A-scan revealed a high-intensity echo spike (100% reflectivity) from the inner surface of the tumor, with a decreased amplitude of the orbital soft tissue echoes immediately posterior to the tumor (Figure 2b).

The B-scan demonstrated a slightly elevated, linear membranous high-amplitude echogenicity with acoustic shadow (Figure 2c), giving an appearance of "pseudo-optic nerve".

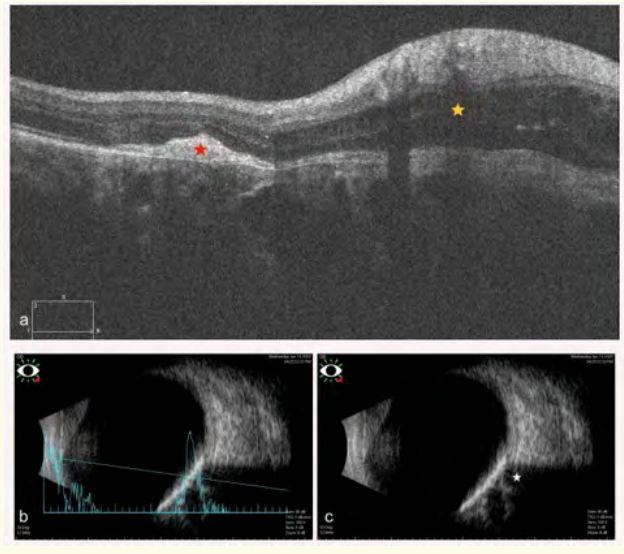

Figure 2: (a-c): (a) Spectral Domain OCT of Right eye showing hyper-reflective CNVM complex (yellow star), with intra-retinal fluid (red star). (b,c) A scan ultrasound scan showing a high intensity echo spike from inner surface of the tumor; B scan showing elevated, linear membranous high amplitude echogenicity with post acoustic shadowing that giving an appearance of "pseudo-optic nerve" (white star).

The patient was advised intra-vitreal Bevacizumab, to receive on monthly basis for 3 months.

Discussion

Choroidal osteoma has a female preponderance, affecting one-third of males. RPE alterations at the margin of the lesion are

observed in approximately 74% of patients. Secondary CNVM was associated with 31% of patients, while decalcification was seen in over 46% (at the end of 10 years of follow-up) [6]. None of the previous reports have mentioned about the occurrence of pseudo-disc edema in cases of choroidal osteoma with secondary CNVM in a male patient.

The current case report presents a relatively unusual presentation of a choroidal osteoma in a male patient with a depigmented lesion. The presence of RNFL and pseudo-disc edema, male sex, and absence of typical pigmentation are unusual features of choroidal osteoma. However, typical ultrasound and fluorescein angiographic features helped to arrive at a diagnosis.

Conclusion

Choroidal osteoma (CO) predominantly affects young females and is a rare, benign ossifying tumor of the choroid. This case report highlights an atypical presentation of choroidal osteoma in a young 19-year-old male, characterized by pseudo-disc edema and a secondary CNVM. The unusual clinical features posed a diagnostic challenge, necessitating a comprehensive multimodal imaging approach to confirm the diagnosis and guide management.

Multimodal imaging played a pivotal role in identifying and characterizing the choroidal osteoma and its associated complications. Fundus photography and fundus autofluorescence (FAF) revealed well-defined yellowish subretinal lesions with heterogeneous autofluorescence patterns, correlating with different degrees of ossification and atrophic changes. OCT provided detailed cross-sectional imaging, demonstrating a hyperreflective bony plate, subretinal fluid, and an associated secondary CNVM. OCT further delineated the extent of the lesion and its impact on the RPE and choriocapillaris. Fluorescein angiography (FA) confirmed active CNVM and choroidal osteoma. Additionally, B-scan ultrasonography helped confirm the calcified nature of the lesion, ruling out alternative diagnoses such as choroidal hemangioma or sclero-choroidal calcifications.

The presence of pseudo-disc edema in this case adds a layer of complexity to the diagnosis, as optic disc edema is more commonly associated with inflammatory, infectious, or vascular conditions than choroidal osteoma. Careful differentiation from true disc edema is necessary to avoid unnecessary interventions and to focus on managing the underlying pathology. Secondary CNVM, a known complication of CO, requires timely intervention to preserve the visual function. In this case, intravitreal anti-vascular endothelial growth factor (anti-VEGF) therapy was initiated, demonstrating a favorable response, with regression of CNVM and improvement in visual acuity, which has been previously shown in studies and case reports [8-10].

This case underscores the need and importance of multimodal imaging in diagnosing and managing atypical choroidal osteomas, particularly when presenting with uncommon features such as pseudo-disc edema and secondary CNVM. Early recognition and targeted treatment can help mitigate vision loss and improve the long-term outcomes. As choroidal osteomas can exhibit progressive changes over time, regular follow-up with multimodal imaging is essential for monitoring lesion evolution, detecting CNVM recurrence, and guiding further management. Further studies and case series are needed to expand our understanding of the diverse clinical manifestations of choroidal osteoma and optimize treatment strategies.

Conflict of Interest

The authors declare no conflict of interest.

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