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Case Report

Navigating Diagnostic Challenges: An Isolated Optic Nerve Head Astrocytic Hamartoma: A Case Report

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Abstract

An isolated astrocytic hamartoma of the optic nerve head (ONH) is a rare entity that can present without associated systemic manifestations or other retinal findings. We report a unique case involving a 36-year-old Yemeni male who presented with rapid and progressive right eye vision loss. Comprehensive ophthalmic and systemic evaluations were performed. Fundus examination revealed an ONH mass with calcification, while fundus fluorescein angiography (FFA) demonstrated late leakage from the optic disc lesion. B-scan ultrasonography and optical coherence tomography (OCT) further characterized the ONH calcifications. The diagnosis was made presumptively based on clinical findings and exclusion of other conditions, including tuberous sclerosis complex (TSC). Informed consent was obtained from the patient prior to documenting this case.

Keywords: Astrocytic Hamartoma; Optic Nerve Head; Multimodal Imaging; OCT; FAF; AHONH

Abbreviations

AHONH: Astrocytic Hamartoma of Optic Nerve Head; ONH: Optic Nerve Head; OCT: Optical Coherence Tomography; TSC: Tuberous Sclerosis Complex; FFA: Fundus Fluorescein Angiography; FAF: Fundus Auto-Fluorescence; MRI: Magnetic Resonance Imaging; CT- Scan of the Brain: Computed Tomography of the Brain

Introduction

Hamartomas are local malformations of cells that demonstrate abnormal proliferation in the area where they are normally present. Retinal and optic disc hamartomas include astrocytic hamartoma, congenital hypertrophy of the retinal pigment epithelium (CHRPE), simple congenital hamartoma of the retinal pigment epithelium (CSHRPE), combined hamartoma of the retina and retinal pigment epithelium (CHRRPE), retinal hemangioblastoma (retinal capillary hemangioma), and retinal cavernous hemangioma. Retinal and optic disc hamartomas can present sporadically as well as with systemic associations [1]. This is most frequently linked to tuberous sclerosis complex (TSC), a hereditary disease with neurocutaneous symptoms and multisystem hamartomas [2]. Astrocytic hamartoma of optic nerve head (AHONH) can be sporadic or in combination with other systemic or ocular diseases such Leber's hereditary optic neuropathy, or retinitis pigmentosa [1,2].

The hamartoma of the eye can include the optic nerve head or the surrounding retina and can manifest as a single or numerous, flat or raised, translucent or opaque, white or yellowish lesion [3]. AHONH typically has no symptoms and is discovered accidentally during normal ocular examinations; nonetheless, it can result in optic atrophy, visual field problems, or secondary consequences such as vitreous haemorrhage, neovascular glaucoma, or retinal detachment [3,5].

The clinical characteristics and the patient's history of TSC or other related disorders are the primary factors used in the diagnosis of AHONH. Histopathological evaluation necessitates surgical excision or enucleation of the eye [2,5].

A Case Description

We report a case of a 36-year-Yemeni male patient who presented with a painless sudden and progressive defective vision in his right eye over one year. He had no history of trauma, systemic disease, or ocular surgery.

On examination, his right eye had an uncorrected visual acuity of counting fingers at 1.5 meter in nasal field, a relative afferent pupillary defect (RAPD), and a normal intra-ocular pressure of 16 mmHg.

Fundus examination revealed a bulging brownish soft tissue with calcification within the optic disc which obscured its' edge, gave it an irregular appearance, measuring 2*2*1.5 mm with perivascular subretinal exudates, and multiple peripheral retinal calcifications.

His left eye had a vision of 2/60, which improved to 6/12 with glasses, a normal anterior segment, a normal intra-ocular pressure of 14 mmHg, and a myopic fundus changes.

Magnetic resonance imaging (MRI) of the brain was normal unless abnormal mild increase in anteroposterior diameter of both eyes which suggesting myopia, whereases Computed tomography (CT) of the brain showed a mass with calcification at the right optic disc. A prior fundus image showed an obscured right optic disc and mild myopic left fundus, figure (1). We requested a B-scan ultrasound which showed optic disc mass with calcification with subretinal exudates, figure (2), and Optical coherence tomography (OCT) of the optic nerve head in the right eye showed an elevated

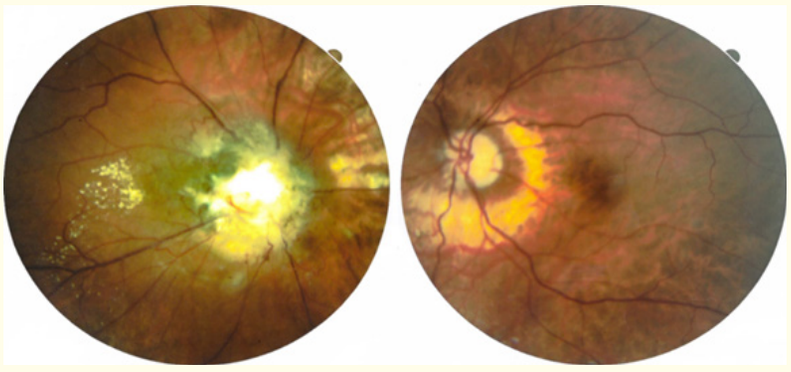

Figure 1: Fundus image showed OD obscured optic disc margin, and OS mild myopic changes.

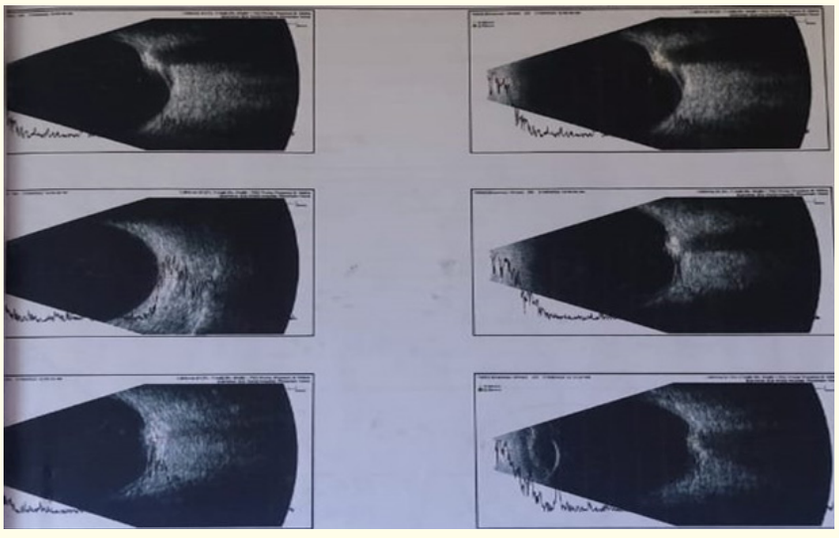

Figure 2: B-Scan OD showed an optic disc mass with subretinal exudate.

optic disc with a thickness of 1.2 mm. Serum calcium, phosphate, parathyroid hormone, and calcitonin levels were requested and were normal.

A wide field-coloured fundus image of the right eye shows a yellowish grey calcific optic nerve head lesion (mulberry-like), obscuring disc edge, altered foveal reflex, attenuated retinal vasculature with subretinal exudation along vessels and haemorrhage, and peripheral RPE changes seen inferiorly figure (3).

Multimodal imaging, Fundus auto-fluorescence (FAF) of the right eye shows small hyper-auto-fluorescence calcific lesions temporal and within the ONH, and black non-calcified lesions within the ONH and in the periphery, figure (4).

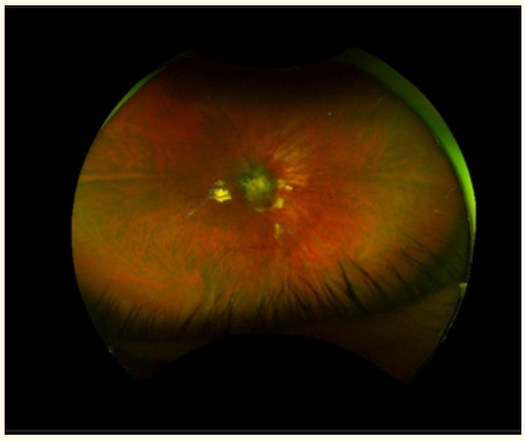

Figure 3: A wide field color photo (Opto Map) OD shows a grayish calcific optic nerve head lesion (mulberry-like) with overlaying telangiectatic vessels.

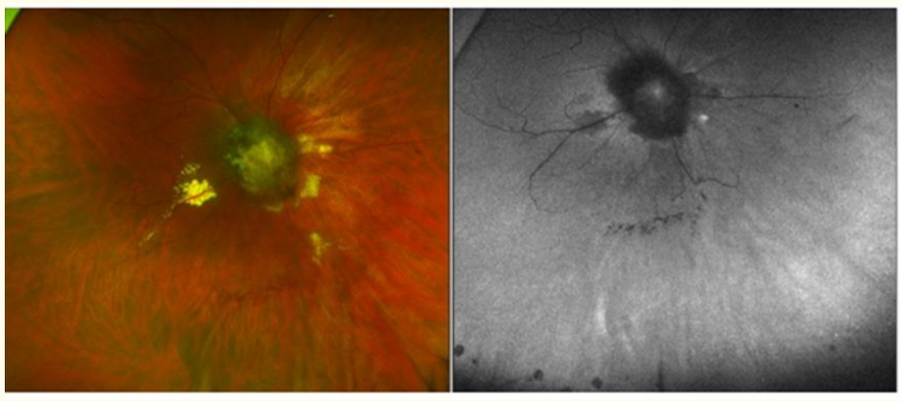

Figure 4: Fundus auto-fluorescence image (FAF) reveals a small hyper-auto-fluoresce calcific lesions temporal and within the ON, and hypo-auto-fluorescence non calcified lesions within the ONH as well in the periphery with areas of subretinal exudate.

Fundus fluorescein angiography (FFA) of the right eye showed a normal arm for retinal circulation time and a delayed arteriovenous filling time.

The optic disc lesion showed late hyper-fluorescence throughout the angiogram. There was evidence of blocked choroidal fluorescence coinciding with the calcification seen in the coloured photo, figure (5).

The patient was diagnosed with presumed sporadic astrocytic hamartoma of the optic nerve head (AHONH), depending on negative family history, no major or minor TSC features, CT showed an optic disc mass with calcification, FAF and FFA suggestive findings of AHONH.

The patient was advised to have regular follow-up visits to monitor the progression of the tumor and the development of any complications. He was also counseled about the possibility of having a similar tumor in his offspring, as astrocytic hamartoma of optic nerve head can be inherited in an autosomal dominant manner.

A low-vision aid was prescribed for the right eye and a refraction correction for his left eye. At the last follow-up visit, 18 months after the initial presentation, his visual acuity and fundus findings were stable, and he did not report any new symptoms.

Discussion and Conclusion

Astrocytic hamartomas of the optic nerve head are frequently observed in eyes suffering from retinitis pigmentosa and are linked

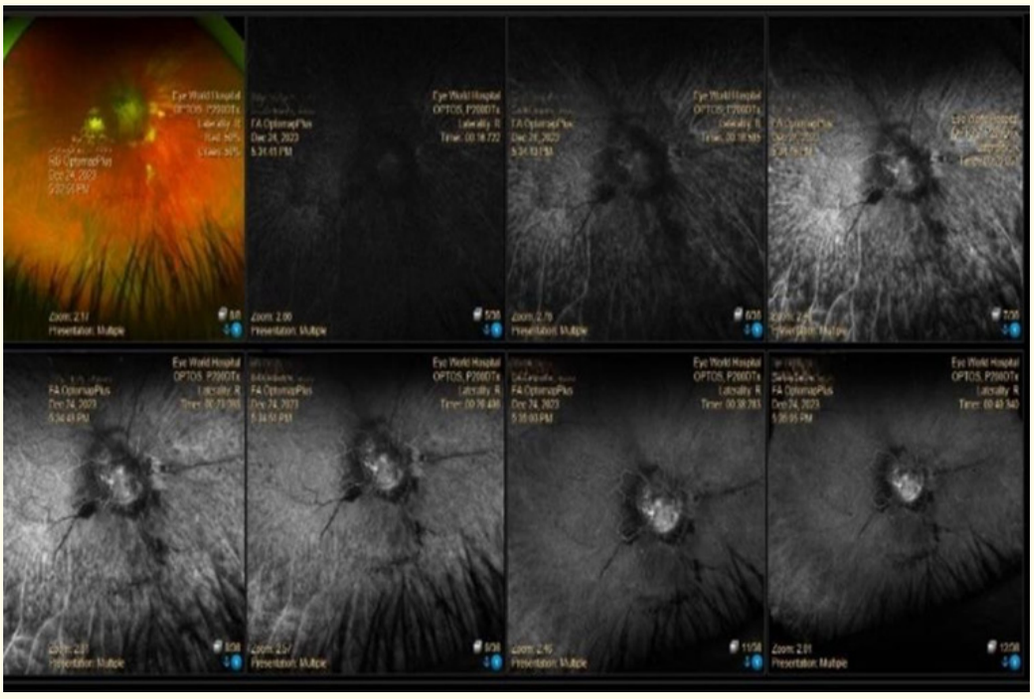

Figure 5: Fundus fluorescein angiography (FFA) of the right eye shows a normal rim of retinal circulation time and delayed arteriovenous filling time. The optic nerve lesion showed late hyper-fluoresceine through the angiogram. There was evidence of blocked choroidal fluorescence coinciding with the calcification seen in the colored photo and subretinal exudate.

to several phakomatoses, including neurofibromatosis and tuberous sclerosis. As demonstrated in this case, optic nerve head drusen can be challenging to differentiate from an astrocytic hamartoma, despite being more common in retinitis pigmentosa patients [5]. In Drusen, calcific deposits accumulate within the nerve's substance, but disc hamartomas are made up of both astrocytic cell proliferations and calcifications that cover the disc and subsequently affect the peripapillary region.

Differential diagnoses for a suspected case of astrocytic hamartoma includes advanced papilloedema, optic disc hemangiomas, retinoblastomas, meningiomas, metastatic tumours, optic nerve head drusen, combined hamartomas of the retina and retinal pigment epithelium and granulomas of different aetiologies. A multimodal imaging, FAF, FFA, OCT, B- scan, and brain MRI, and CT all are helpful in diagnoses of Sporadic AHONH [3]. Perimetry helps in assessing the effect of the extent of retinal astrocytic hamartoma.

The management of AHONH depends on the visual acuity, the extent of the tumor, and the presence of complications. Most AHONH cases are stable and require no intervention, except for periodic observation and monitoring.

Treatment options for progressive or complicated AHONH include laser photocoagulation, cryotherapy, radiotherapy, or surgery. However, the efficacy and safety of these modalities are limited by the risk of damage to the optic nerve or the surrounding retina, including vitreous haemorrhage, macular exudation, retinal detachment, macular hole, epiretinal membrane, and choroidal neovascularization require treatment [4].

In our case, the patient was diagnosed as a case of Sporadic AHONH by exclusion, after discussing possible differential diagnosis and utilizing several ophthalmic imaging modalities. three years after the initial presentation, he is well and his vision is stable, and accepting his glasses. A wide-field fundus photography (Optomap) does not show any increase in hamartoma size or increased calcification. And advised to be on regular follow up.

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