Combined Hamartoma of Retina and Retinal Pigment Epithelium Masquerade as Choroidal Melanoma in Young Patient

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ABSTRACT

Introduction: Combined hamartoma of retina and retinal pigment epithelium is a rare benign ocular tumour resulting from abnormal proliferation of glial cells, vascular tissue and pigment epithelial cells

Case: A 29-year-old male presented with complaint of diminution of vision in his right eye, noticed incidentally during screening.

Observation: On examination, a flat, unilateral, solitary, greyish brown peri-papillary lesion with irregularly pigmented border with an epiretinal membrane in right eye. The Optical Coherence Tomography showed a thickened retina with disorganisation of retinal layers, a hyper-reflective inner retinal layer with fine retinal traction peaks, and presence of an epiretinal membrane. Histopathological examination was not done.

Conclusion: Combined hamartoma of retina and retinal pigment epithelium is a rare benign tumour. Proper diagnosis is important as it can be confused with more severe pathologies like choroidal melanoma, Retinal pigment epithelium adenocarcinoma.

Key words: Combined hamartoma; epiretinal membrane; ocular hamartoma; retina; retinal pigment epithelium.

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INTRODUCTION

Combined hamartoma of retina and retinal pigment epithelium (CHRRPE), first came to prominence in 1970s when Gass (1973) published a case series describing an uncommon entity and described it as an elevated, charcoal grey mass involving the retinal pigment epithelium (RPE), retina, and overlying vitreous, extending in a fanlike projection toward the periphery and blending imperceptibly with surrounding RPE. Gass (1973) mentioned the lesions were usually covered by a thickened grey-white retinal and pre-retinal tissue while showing contraction of the inner surface. There was a notable absence of RPE or choroidal atrophy at the margin along with absence of retinal detachment, haemorrhage, exudation, and vitreous inflammation. The CHRRPE is a rare benign lesion in the macula, juxta-papillary, or peripheral retina involving the pigment epithelium, glial cells and vascular tissue of the retina. These lesions frequently get misdiagnosed as malignant neoplasms, such as retinoblastoma and choroidal melanoma, with the eyes eventually being enucleated.

CASE REPORT

A 29-year-old male was referred to the ocular oncology unit, Tilganga Institute of Ophthalmology with a presumed choroidal melanoma. The patient initially presented with diminution of vision in the right eye, which was noted incidentally during screening. His BCVA was 6/9 on the right eye and 6/6 on the left. On slit lamp examination, the anterior segment was unremarkable. On fundus evaluation (Figure 1), there was a slightly raised, unilateral, solitary, charcoal grey peri-papillary lesion approximately two-disc diameters in length and three-disc diameters in width, with a fanlike projection towards the periphery in the right eye. The mean lesion diameter was approximately 4693.5 µm, based on vertical (4514 µm) and horizontal (4873 µm) measurements. An epiretinal membrane was also noted in the same eye. No haemorrhage, exudation, retinal detachment or vitreous inflammation were observed. The patient had no significant systemic findings.

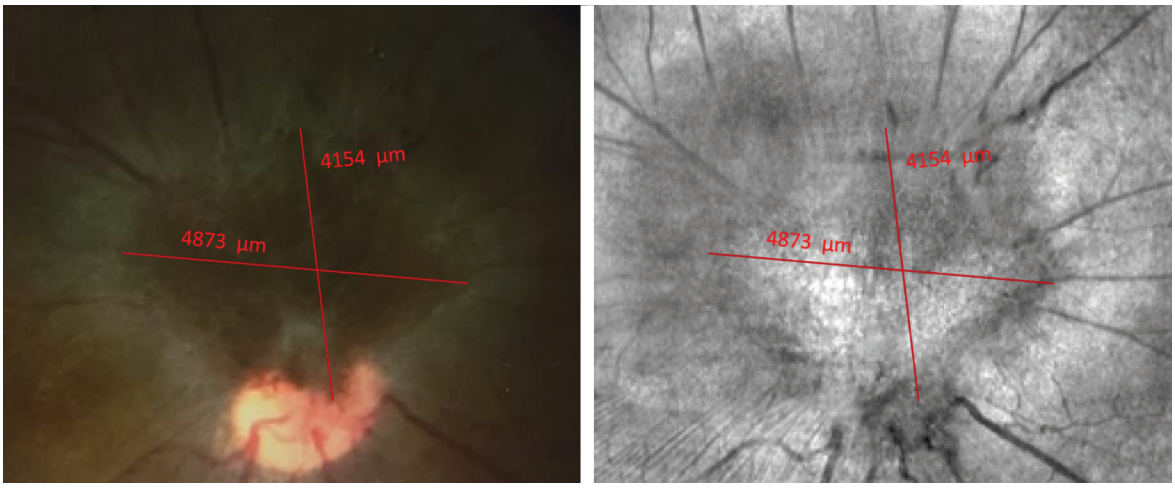

Figure 1: Fundus photograph showing a flat, unilateral, solitary grey brown peri-papillary lesion extending superior to the disc, with irregular pigmentation, approximately 2 DD × 3 DD in size, corresponding to actual measurements of 4514 µm (vertical) × 4873 µm (horizontal), Mean diameter = 4693.5 µm.

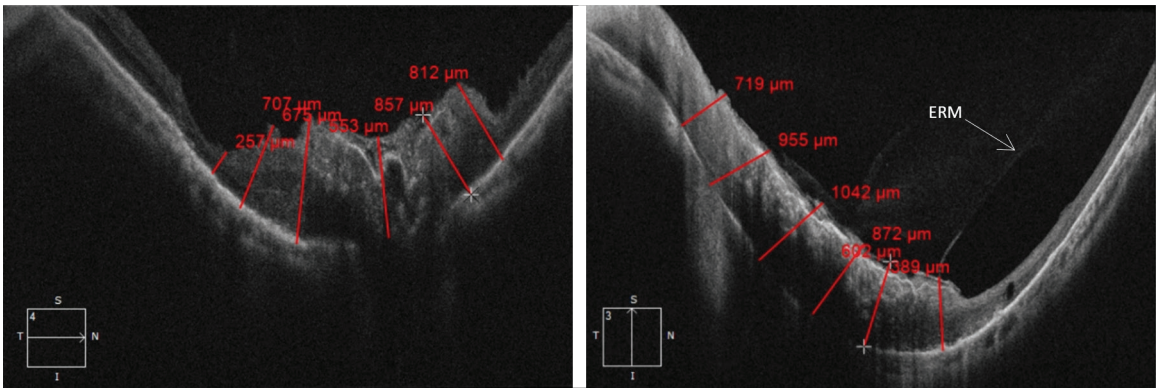

Figure 2: Five-line raster scan showing temporal-to-nasal and inferior-to-superior configurations with irregular retinal contour, increased thickness, distorted architecture, and presence of an epiretinal membrane. Retinal thickness ranged from 257–857 µm(temporal–nasal) and 389–1042 µm (inferior–superior), Mean Thickness = 703.35 µm.

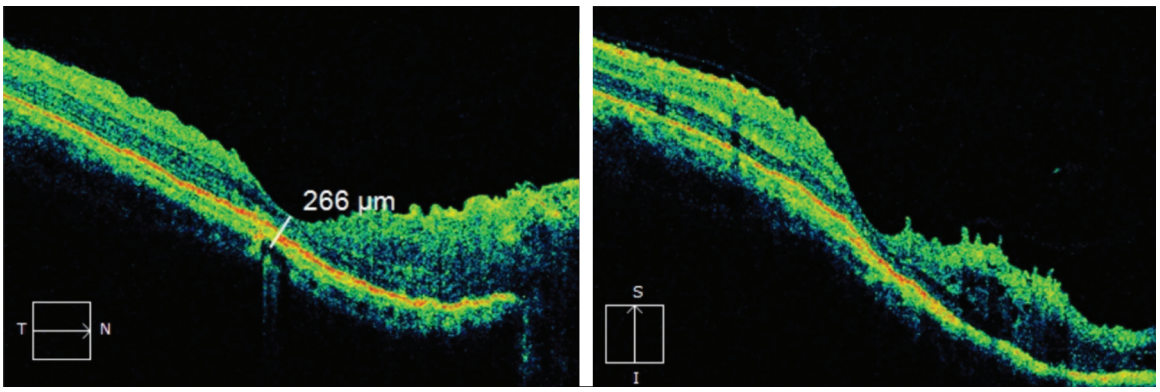

Figure 3: Macular cube temporal to nasal and superior to inferior OCT scan showing an undulated ILM, increased retinal thickness, disturbed retinal architecture with some intra-retinal fluid.

An Optical Coherence Tomography (OCT) (Figure 2; Figure 3) of the patient showed a thickened retina with disorganisation of retinal layers and a hyper-reflective inner retinal layer with fine retinal traction peaks, and presence of an epiretinal membrane. The mean retinal thickness was approximately 703.35 µm, with values ranging from 257–857 µm (temporal–nasal) and 389–1042 µm (inferior–superior). The patient was diagnosed as right eye combined hamartoma of the retina and retinal pigment epithelium. He was advised close follow-up every three months or *Si Opus Sit* (SOS = if there is need) as he had good vision in the affected eye.

DISCUSSION

The CHRRPE is an uncommon but benign neoplasm of the retina. It is usually asymptomatic, non-progressive lesion. Aetiologically CHRRPE are thought to be congenital lesions, although they have not been reported at the time of birth (Schachat et al., 1984). The youngest patient in a study by Shields et al., (2008) was two weeks old. The patient in this case report presented later, aged 29 years. This could be due to lack eye screening in a developing country like Nepal, where a lot of patients see an ophthalmologist for the first time only in their adulthood when

they have symptoms. Schachat et al., (1984) reported no gender predilection in their study. However, Font et al., (1989) and Shields et al., (2008) reported a male preponderance in their studies.

Although initially thought to be an isolated ophthalmic condition, CHRRPE have been associated with other systemic entities. It has been most strongly associated with neurofibromatosis type II, particularly when the tumour is bilateral (Kaye et al., 1992). They have also been associated with Gorlin syndrome, Juvenile nasopharyngeal angiofibroma, Poland anomaly, etc., (De Potter,1960; Fonseca,2001; Stupp, 2004). However, patient in this case report did not have any findings suggestive of these features and his systemic examinations were normal.

The CHRRPE is mostly asymptomatic. Patients may also present with symptoms like diminution of vision, strabismus and floaters. Patient in this case report presented with diminution of vision in his right eye, which he noticed when participating in a heath camp where they checked his vision.

Diagnosis of CHRPE depends on thorough history, fundoscopic exam and OCT findings. Gass (1973) first described the characteristic features of CHRRPE which helped distinguish them from other more sinister lesions. Schachat et al., (1984) and the Macula Research Society Committee further described clinical difference between combined hamartomas in the 1980s. They noted several clinical differences among the two. Combined hamartomas mainly affect the retina and retinal pigment epithelium whereas melanomas arise in the choroid. Associated retinal detachment, subretinal haemorrhage, vitreous haemorrhage, inflammation is absent in combined hamartoma as noted by Gass (1973), but all can be present in patients with choroidal melanomas. Schachat et al., (1984) noted that pre-retinal "gliosis" and vitreoretinal interface alterations, which are the hallmarks of combined hamartomas, are rarely present with malignant melanoma. They also mentioned that since both the lesions could be elevated, it could be an area of confusion. However, the degree of elevation is not as significant in combined hamartomas as in melanomas and the elevation is in front of the retina and not underneath it. On fundus evaluation of patient in this case report, slightly raised, unilateral, solitary, charcoal grey peri-papillary lesion with a fanlike projection towards the periphery and blending imperceptibly with surrounding RPE in the right eye was noted. There was a macular epiretinal membrane in the same eye. No haemorrhage, exudation, retinal detachment or vitreous inflammation were noted. These findings were highly suggestive of a diagnosis of CHRRPE. Despite distinguishing features described in literature by Gass (1973) and Schachat et., (1984), CHRRPE can be confounding entity for a lot of ophthalmologists, particularly with the grave nature of other similar lesions. Thus, with the advent of non-invasive investigative tools like OCT and ED-OCT, efforts have been made to study CHRRPE with these tools. They have proved to be of great use in further helping clinicians differentiate melanoma and combined hamartomas. Shields et al., (2008) have described OCT findings of 11 patients with combined hamartoma of retina and retinal pigment epithelium. They noted an epiretinal membrane and striae with associated retinal disorganisation in all patients. Arepalli et al., (2014) in their study using ED-OCT have mentioned combined hamartoma of retina and retinal pigment epithelium characteristically show prominent epiretinal membrane with vitreoretinal traction in either a sawtooth (mini-peak) appearance, folded (maxi-peak) appearance, or both. This traction

leads to increased retinal thickness and reduced underlying choroidal thickness. The OCT of the patient in this case report showed thickened retina in the right eye with disorganisation of retinal layers and a hyper reflective inner retinal layer with fine retinal traction peaks similar to those described by Shields and Arepalli, which further corroborated our diagnosis.

Management can be non-surgical with observation and amblyopia therapy if necessary, or surgical with vitrectomy and membrane peeling for epiretinal membrane associated with CHRRPE. If systemic features are present as in Neurofibromatosis II, a multidisciplinary approach in conjunction with specialists like neurologists etc. is recommended. Patient in this case report was managed conservatively and was kept under regular follow-up.

With careful clinical examination of the lesion, and with the availability of modern imaging technologies like the OCT and ED-OCT scans, diagnosis combined hamartoma of the retina and retinal pigment epithelium can be made with increased accuracy sparing patients from invasive procedures.

CONCLUSION

CHRRPE is a rare benign tumour which can mimic grave pathologies like choroidal melanoma. Proper clinical examination and use of modern diagnostics can save patients from undue destructive surgeries.

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