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Case Report

Congenital hamartoma of RPE - The tumour that isn't

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Abstract

Congenital simple hamartoma of the retinal pigment epithelium (CSHRPE) is a rare disease-causing vision loss due to a solid pigmented lesion within the macula. It is a benign condition that can cause significant visual loss and the entity is limited to retinal tissue. The aetiology is unknown, and there is no standard guideline. Patients should have regular eye examinations to monitor changes in the lesion or vision. Hence, we describe a case of 18-year-old female with diminished vision in left eye since childhood and on fundus examination, clear media with size of 4–5-disc diameter elevated subretinal greyish yellow lesion noted in posterior pole and peripapillary area with epiretinal membrane causing apparent traction of retina, with overlying dilated and tortuous vessels. Optical coherence tomography showed highly reflective elevated lesion with hyporeflective shadowing of underlying tissue. Patient was under observation, then discharged with regular follow-up advice.

Keywords: Congenital simple hamartoma of the retinal pigment epithelium, Macula, Visual acuity, Benign, Optical coherence tomography.

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1. Introduction

Congenital simple hamartoma of the retinal pigment epithelium (CSHRPE), a rare disease presents with solid pigmented lesion within the macula.[1] It is a benign condition that can cause significant vision loss.[2] With varying degrees of diffusion to adjacent surfaces, the entity is limited to the retinal tissue. Although the lesion give the impression to be congenital, its aetiology is yet unknown.[3] The correlation between the embryology of the retinal pigment epithelium and the neurosensory retina could be the possible mechanism.[1] Also, the histopathological evaluation of tissue is considered to be rare in this case, hence the related features are not well-established to identify the CSHRPE.[3] This urges a larger study with histopathological correlation to further explore the exact origin of such lesions.

With the availability of evidence in the literature, it is suggested that CSHRPE should be monitored regularly, especially in cases where the lesion is larger in size or has a more diffuse border.[1,3,4] Treatment options for CSHRPE include laser photocoagulation, cryotherapy, or surgical excision.[5] However, the decision to treat should be made on a case-by-case basis, taking into account factors such as the location and size of the lesion, as well as the patient's age and overall health.[6] Though, there are no standard guideline for the treatment of CSHRPE. Patients with CSHRPE should also be advised to have regular eye examinations to monitor any changes in the lesion or vision. Hence, we describe a case of 18-year-old female with diminished vision in left eye since childhood and on fundus examination, patient had clear media with size of 4 – 5-disc diameter elevated subretinal greyish yellow lesion noted in posterior pole and peripapillary area with epiretinal membrane (ERM) causing apparent traction of retina, with overlying dilated and tortuous vessels. Optical coherence tomography (OCT) showed highly reflective elevated lesion with hyporeflective shadowing of underlying tissue. Patient was under observation, then discharged with regular follow-up advice.

2. Case Report

An 18-year-old-female presented with the complaints of diminished vision in left eye (LE) since childhood with no

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other significant history. No significant family history also present. For the past 5 years, the patient noticed further diminution of vision in LE which was painless and progressive in nature. On presentation her Snellen's best corrected visual acuity (BCVA) in right eye (RE) was 6/6 and in LE it was HM+PL+PR (Hand movements, perception of lights, projection of rays) were accurate in all quadrants. Both eyes had normal intraocular pressure (IOP). On anterior segment examination in LE she had Grade-1 Relative afferent pupillary defect (RAPD). Other examinations were found to be normal in both eyes.

Fundus examination revealed normal fundus in the RE, while in LE **(Figure 1)** we found a clear media with size of 4 – 5 DD (disc diameter) elevated subretinal greyish yellow lesion noted in posterior pole and peripapillary area with ERM causing apparent traction of retina, along with overlying dilated and tortuous vessels. There was no evidence of macular edema, or subretinal fluid. OCT **(Figure 2)** showed highly reflective elevated lesion with hyporeflective shadowing of underlying tissue & obscuration of normal retinal layers with an ERM. This made the diagnosis of LE CSHRPE. This was further explained to the patient and her parents. She was followed by observation.

Figure 1: Fundus picture of left eye

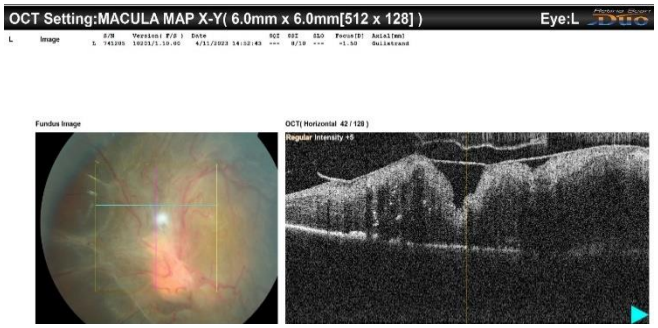

Figure 2: OCT picture of left eye

3. Discussion

Congenital simple hamartoma of the retinal pigment epithelium (CSHRPE), a rare condition was first described by Laqua H in 1981,[7] and reported by Gass JD,[8] as the congenital anomaly of RPE. It is a benign tumor that can be found incidentally as in ours. It is characterized as heavily pigmented, nodular benign tumor with well-circumscribed usually located adjacent at foveola or rarely at the foveal center.[9] It seems that there is a limited amount of literature available on this topic, with only a few cases being documented.[1,3,4,9,13] In India only few cases were reported on CSHRPE.[2,14,15] In only one study done by Shields et al,[11] where he reported the five cases of CSHRPE and classified based on the clinical features. The common symptoms that patients experience is the reduction of vision over time, which was similar to our case.

Regarding the progression of the lesion, Shields et al,[11] observed that in all five cases they maintained a stable pattern during the follow-up period with no progression of lesion and even the VA has not changed. Also, other literature implied the same with no progression.[3,9,10,13,16,17] But as in our case, at the first visit itself she had the progressive vision loss in LE which was considered rare, when compared with the other cases found in the literature.[3,9-11,13-17] While, in case of associated choroidal neovascularization which may cause vitreous hemorrhage, retinoschisis, and macular hole, can lead to further diminution of vision.[18] While, in our case there is no such associated condition but the diminution of vision was noted.

In our case, fundus examination and OCT revealed abnormalities such as hyperpigmentation, vascular tortuosity, elevation, and ERM. Subretinal fluid, choroidal neovascularization, full thickness retinal holes with or without schisis, and vitreous hemorrhage—which was also observed in other cases that have been reported—are additional less common characteristics.[5,9–11] The original Macula Society investigation on CSHRPE recognised the tumour's similar characteristics as elevation, pigmentation, vascular tortuosity, and ERM.[5]

OCT is considered golden investigation to assess CSHRPE till now based on the literature.[12] There have been conjectures that OCT may offer valuable insights into potential visual benefits, since a more intact retina may yield better visual results than a completely disorganized, thickened retina. Fluorescein angiography (FA) also plays an essential role in diagnosis of this condition.[1,3,9-11,13-17]

Also, retinal traction, which was commonly not seen in CSHRPE, but this can be occurred in the combined RPE with retinal abnormalities. To differentiate that, in our case only traction of retina was present without any other involvement of retinal structure, which made this case differ from combined type. Also, Shields et al,[11] described the association of CSHRPE with retinal traction, dilated feeding and draining retinal vessels, were seen in our patient. Additionally, macular lesions are linked to reduced VA (37% versus 43%), strabismus (25% versus 31%), and younger presentation (mean age 14.2 months versus 9.5 months). These associations are also true when compared to

extramacular lesions.[6] But unfortunately in our case, there was no macular edema that distinguished from other cases of CSHRPE.

Studies reported multiple association including NF 1&2, Gorlin-Goltz syndrome,[18] Poland anomaly,[19] and branchio-oculo-facial syndrome.[20] Chawla et al in a case series of CHRPE reported that the true nature of CHRPE should be re-analysed because majority of cases were limited posteriorly by the outer plexiform layer without any involvement of the outer retinal layers and RPE.[2]

On treatment there were no standards guideline for CSHRPE. The role of epiretinal membrane peeling surgery is controversial as for CSHRPE.[6,14-17] Only long-term follow-up of the case were required for further assessment.

4. Conclusion

Because of its rarity and variable clinical appearance, CHRRPE can be misdiagnosed as choroidal melanoma, retinoblastoma, congenital hypertrophy of the RPE, chorioretinal scar, vascular anomaly, or even retinal detachment. Periodic monitoring is recommended for treatment and over-the-long-term follow-up is required to rule out the any suspicious growth.

5. Source of Funding

None.

6. Conflict of Interest

None.

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