Foveal Congenital Simple Hamartoma of Retinal Pigment Epithelium: A Report of Two Cases

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Abstract:
We report two cases with foveal congenital simple hamartoma of the retinal pigment epithelium (CSHRPE), as both patients presented to our retina services complaining of a unilateral decreased vision. Full ophthalmic examination and multimodal imaging were performed including fundus photography, fundus autofluorescence, optical coherence tomography, fluorescein angiography, and electrophysiological testing. Both patients presented with 20/80 vision in the affected eyes. Foveal CSHRPE was found in both eyes, along with parapapillary hyperpigmented rim, multiple pinpoint macular lesions, and few posterior pole hyperpigmented lesions. Multifocal electroretinogram showed diminished central amplitude in both eyes, with three-dimensional topography map showing blunted foveal peaks in one eye and the absence of a central peak in the other patient. Both patients had a stable vision and clinical examination of the CSHRPE during 5 and 6 years follow up, respectively. Foveal CSHRPE is usually symptomatic and results in a decline in visual acuity. Follow-up of these patients showed stable vision and clinical examination.

Keywords:
Congenital simple hamartoma of retinal pigment epithelium, multifocal electroretinogram (mfERG), retinal tumors, retinal pigment epithelium

Introduction
Congenital simple hamartoma of the retinal pigment epithelium (CSHRPE) is a rare tumor of the retinal pigment epithelium (RPE) which was initially identified by Laqua. CSHRPE differs from the classic congenital hypertrophy of the RPE and RPE hyperplasia. Later, Gass provided a further characterization of the tumor with photographic documentation. It is usually asymptomatic and nonprogressive. Upon fundoscopy, it appears as a small, dark, elevated lesion within the macular area. It can be associated with feeder vessels, macular exudation, and vitreous cells. Causes of decreased vision in CSHRPE include epiretinal membrane (ERM) formation with macular traction, macular edema, or macular hole. The differential diagnosis includes combined hamartoma of the retina and RPE, congenital hypertrophy of the RPE, adenoma or adenocarcinoma of the RPE, RPE hyperplasia, and retinal invasion from a choroidal nevus or melanoma. Here, we report two CSHRPE cases with multimodal imaging and multifocal electroretinogram findings.

Case Reports
Case 1
A 28-year-old male presented 5 years ago to King Khaled Eye Specialist Hospital (KKESH) with a unilateral decreased vision in the right eye for 1 year. His best-corrected visual acuity (BCVA) was 20/80 in the right eye and 20/20 in the left eye.

How to cite this article: Badawi AH, Magliyah M, Allam K, Alzahrani YA. Foveal congenital simple hamartoma of retinal pigment epithelium: A report of two cases. Middle East Afr J Ophthalmol 2020;27:128-30.
Anterior segment examination of both eyes, as well as the fundus examination of the left eye, were within normal limits. Fundus examination of the right eye showed a small, elevated dark lesion at the fovea area [Figure 1a] measuring 1051 µm. There was no macular edema, retinal exudation, or RPE atrophy. Spectral domain-optical coherence tomography (SD-OCT) showed an elevated hyperreflective lesion at the fovea with an absence of the normal foveal pit and a complete shadowing, preventing visualization of details underneath the lesion [Figure 1e]. Fundus autofluorescence (FAF) revealed a sharply demarcated hypoafluorescent area corresponding to the site of the lesion [Figure 1b]. Fundus fluorescein angiography (FFA) revealed nonfluorescence throughout the angiogram, and in the late phase, a mild hyperfluorescence was noted around the lesion [Figure 1f]. Full-field electroretinogram (ffERG) was normal in both eyes. Subjective refraction did not improve the visual acuity in the affected eye. After 5 years of follow-up, the patient had a stable vision and CSHRPE lesion.

**Case 2**
A 58-year-old male presented to KKESH 6 years ago with a unilateral decreased vision in the right eye for 4 years before presentation. His BCVA was 20/80 in the right eye and 20/20 in the left eye. Anterior segment examination of both eyes, as well as the fundus examination in the left eye, was within normal limits. Fundus examination of the right eye showed a foveal hyperpigmented lesion measuring 1042 µm, along with parapapillary hyperpigmented rim, multiple pinpoint macular lesions, and few posterior pole hyperpigmented lesions [Figure 2a]. More peripherally, there were diffuse hypopigmented retinal lesions indicating RPE atrophy [Figure 2a]. There was no macular edema and retinal exudation. SD-OCT showed sharply elevated hyperreflective lesion at the fovea with complete shadowing, preventing visualization of details underneath the lesion [Figure 2e]. There was an absence of the normal foveal pit as well [Figure 2e]. FAF revealed a sharply demarcated hypoafluorescent area corresponding to the site of the hyperpigmented lesions, along with peripheral hyperautofluorescent areas corresponding to the hypopigmented lesions [Figure 2b]. FFA revealed nonfluorescence throughout the angiogram and, in the late phase, a faint hyperfluorescence was noted around the lesion [Figure 2c and d]. mfERG revealed markedly diminished amplitude centrally in the right eye, and 3D topography map showed absence of the central peak [Figure 2f]. ffERG was normal in both eyes. Subjective refraction was not beneficial for visual improvement in the right eye. Through 6 years of follow-up, the vision, as well as the CSHRPE, was stable.

**Discussion**

CSHRPE is a rare and benign hamartoma of the RPE.
As has been described previously in the literature, a small hyperpigmented lesion at the macular area was found in both patients. The SD-OCT findings of elevated hyperreflective lesions completely shadowing the underneath structures were typical to what has been described in CSHRPE, apart from the absence of the normal foveal pit in our patients. The fluorescein angiographic findings in our patients were similar to what was reported by Shields et al. which is characterized by persistent nonfluorescence and late staining at the borders of the lesion. The FAF in CSHRPE shows sharply demarcated areas of hypofluorescence at the site of the lesions.

When full-field and multifocal electrophysiological testing was performed, mfERG was found useful for the assessment of foveal function in both patients.

While juxtafoveal CSHRPE is almost always asymptomatic unless associated with further retinal pathologies including ERM, macular edema or macular hole formation. The foveal location of the CSHRPE has made both patients in our study visually symptomatic with 20/80 vision. Multimodal imaging revealed no macular edema, ERM, or macular hole in either patient. Consequently, the best explanation of visual symptoms and decreased vision in our patients is the location of both tumors in the foveal area. The abnormal foveal response on mfERG in both patients further confirms the foveal location as the cause of decreased vision.

Recently, Stavrakas et al. have reported a CSHRPE which was located at the fovea in a 14-year-old girl who presented with 20/100 vision. After 4 years of follow-up, she presented with counting fingers vision and developed a full-thickness macular hole (FTMH) at the site of the CSHRPE with probable migration of the lesion to the vitreous cavity. Before the development of the FTMH, her vision was relatively similar to our two cases with foveal CSHRPE. The development of FTMH in the previous case after 4 years indicates that long-term regular follow-up is necessary for these patients. The younger age of the girl might indicate a stronger adhesion between the vitreous and the lesion as well as more tractional forces which have led to the FTMH formation.

The preferred approach for foveal CSHRPE is a watchful observation for extended periods. The central location, as well as lack of further complications in our patients, limits treatment options in these patients despite abnormal retinal function at the site of the lesion. Ophthalmologists need to be aware of the possibility of a FTMH in these patients, especially if a further drop in vision is noted.

**Financial support and sponsorship**
Nil.

**Conflicts of interest**
There are no conflicts of interest.

**References**

1. Laqua H. Tumors and tumor-like lesions of the retinal pigment epithelium. Ophthalmologica 1981;183:34-8.
2. Gass JD. Focal congenital anomalies of the retinal pigment epithelium. Eye (Lond) 1989;3 (Pt 1):1-8.
3. Shields CL, Shields JA, Marr BP, Sperber DE, Gass JD. Congenital simple hamartoma of the retinal pigment epithelium: A study of five cases. Ophthalmology 2003;110:1005-11.
4. Shukla D, Ambatkar S, Jethani J, Kim R. Optical coherence tomography in presumed congenital simple hamartoma of retinal pigment epithelium. Am J Ophthalmol 2005;139:945-7.
5. Barnes AC, Goldman DR, Laver NV, Duker JS. Congenital simple hamartoma of the retinal pigment epithelium: Clinical, optical coherence tomography, and histopathological correlation. Eye (Lond) 2014;28:765-6.
6. Bach A, Gold AS, Villegas VM, Wildner AC, Latiff A, Ehlies FJ, et al. Simple hamartoma of the retinal pigment epithelium with macular edema. Optom Vis Sci 2015;92:S48-50.
7. van de Moere A, Clark JB. Congenital simple hamartoma of the retinal pigment epithelium with macular edge. Retin Cases Brief Rep 2009;3:80-2.
8. Stavrakas P, Yachtsevanos A, Karakosta E, Kozeis N, Triantafylla M, Transos P. Full-thickness macular hole associated with congenital simple hamartoma of retinal pigment epithelium (CSHRPE). Int Ophthalmol 2018;38:2179-82.
9. Shields JA, Shields CL. Tumors and related lesions of the pigmented epithelium. Asia Pac J Ophthalmol (Phila) 2017;6:215-23.