Congenital Simple Hamartoma Of Retinal Pigment Epithelium In Premature Baby

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ABSTRACT

Congenital simplex hamartoma of retinal pigment epithelium is a rare retinal pigment epithelial tumor. We report a case of congenital simplex retinal pigment epithelium hamartoma in a 4-week-old premature boy. The bilateral anterior segment examination was normal in the patient. There was no pathology in the left eye in fundus examination. The fundus examination of his right eye revealed a foveal pigmented lesion which was half of optic disc size. In the patient, the optical coherence tomography showed hyper-reflectivity in the inner retinal layers and shading due to lesion in the other layers. B-scan sonography showed a small echogenic nodular mass.

KeyWords: Congenital simple hamartoma of retinal pigment epithelium, premature, optical coherence tomography.

INTRODUCTION

Congenital simplex hamartoma of retinal pigment epithelium (CHRPE) is a rare retinal pigment epithelial (RPE) tumor.[1] In general, it is asymptomatic and is recognized incidentally. They generally show no progression during follow-up. It is frequently located adjacent to fovea and pigmentary.[2] They involve full-thickness of retina. Macular edema, retinal traction, exudation and dilated, tortuous vessels may also present.[3] The differential diagnosis includes retinal pigment epithelium hypertrophy, adenoma, nevus and melanoma. The optical coherence tomography is helpful in the diagnosis.

Case Report

A 4-weeks old boy referred to our clinic for retinopathy of prematurity. The patient was born with birth weight of 2000 g at 32-weeks of gestation. In fundus examination by Retcam (Massie Research Laboratories, Including, Dublin, California), no retinopathy of prematurity was detected. The bilateral anterior segment examination was normal in the patient. There was no pathology in the left eye in fundus examination. The fundus examination of his right eye revealed a foveal pigmented lesion which was half of optic disc size (Figure 1). In the patient, the optical coherence tomography (iVue portable Spectral domain optical coherence tomography, Optovue, Fremont, California) showed hyper-reflectivity in the inner retinal layers and shading due to lesion in the other layers. This was compatible with umbrella sign seen in congenital simplex hamartoma of retinal pigment epithelium (CHRPE) (Figure 2). B-scan sonography revealed a small echogenic nodular mass. The patient was diagnosed as CHRPE. The patient is attending regular follow-up visits.

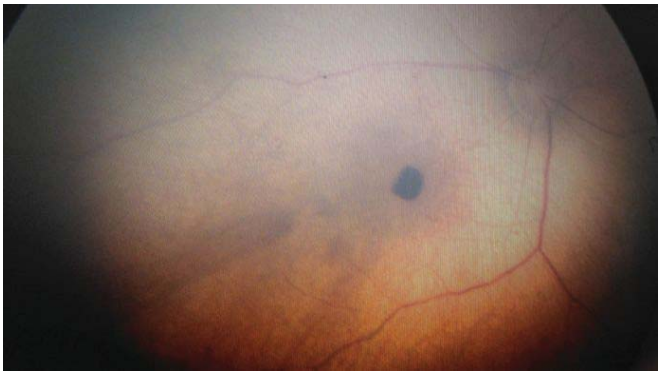

Figure 1: *Fundus image, foveal pigmented lesion (half-disc in size) (RetCam, Massie Research Laboratories, Inc., Dublin, CA).*

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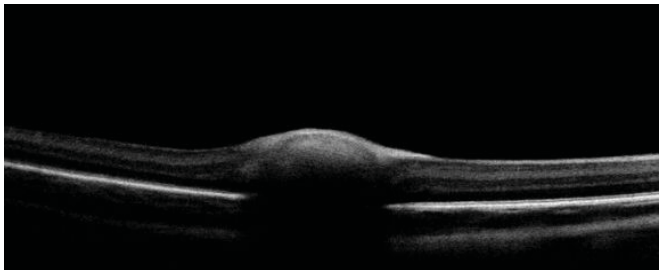

Figure 2: *Optical coherence tomography, hyper-reflectivity in the inner retinal layers and shading due to lesion in the other layers (iVue portable Spectral domain optical coherence tomography, Optovue, Fremont, California).*

DISCUSSION

Congenital simplex hamartoma of retinal pigment epithelium was first defined in a case series of 2 patients by Laqua in 1981. In these cases, there were pigmented lesions at proximity of fovea and across retina.[5] Subsequently, Gass was the first author suggested diagnosis of retinal pigment epithelial hamartoma in 1989.[6] Shield published largest case series (5 patients) of CHRPE and reported OCT findings for the first time.[7]

Congenital simplex hamartoma of retinal pigment epithelium is a rare RPE tumor. Its size is generally varies from one-half or one optic disc; it is hyper-pigmented and involves full-thickness of retina.[4, 8] In our case, CHRPE was half-optic disc in size with hyper-pigmentation and full-thickness retinal involvement.

It generally shows extra-foveal localization with good vision prognosis. The lesions show no tendency towards growth.[3, 9] In our case, CHRPE was located at fovea. Macular edema, retinal traction, exudation, increased dilatation and tortuosity as well as macular hole may also present.[3, 8, 10, 11]

It is asymptomatic in most cases and recognized incidentally.[8,9] In our case, CHRPE was detected during screening for retinopathy of prematurity.

The sonography, fundus fluorescein angiography (FFA) and OCT are helpful in the diagnosis. A small, nodular, echogenic mass lesion is observed in B-mode sonography. It appears as hypo-florescence on FFA while a slight hyper-florescence can be present at temporal to lesion in advanced stage.

On optical coherence tomography, hyper-reflectivity in the inner retinal layers and shading due to lesion in the other layers can be seen, which is termed as umbrella sign.[7, 9, 10, 11]

In our case, no FFA was performed since there was no indication for FFA such as retinopathy of prematurity. There were similar findings on sonography and OCT.

The differential diagnosis includes retinal pigment epithelium hypertrophy, adenoma, nevus and melanoma.[8,10] The congenital hypertrophy of retinal pigment epithelium is generally located at retina and lesions appears as plain on OCT. Adenoma and adenocarcinoma of retinal pigment epithelium involve full-thickness of retinal with a supplying vessel and exudative retinopathy and vitreal hemorrhage can accompany to these cases. They appear irregular on OCT.

Choroidal nevi are generally benign lesions localized at choroid. Choroidal melanoma is one of the intraocular malignancies. Choroidal excavation and orbital shading are present on sonography. In our case, these diagnoses were excluded by fundus examination, sonography and OCT. In the literature, Coat's disease and CHRPE was reported in the same eye in one patient.[12]

Congenital simplex hamartoma of retinal pigment epithelium can be seen in different age groups. The youngest case was 6-years old.[13] Our case was 4-weeks old and youngest case published. In addition, it is first premature case in the literature.

In general, it has an asymptomatic course and is recognized incidentally.[8, 9] The cases cannot be recognized as they are asymptomatic. CHRPE can be diagnosed early if fundus examination and OCT can be performed in younger ages. However, fundus examination and OCT is challenging in infants and requires experience. In our patients, OCT (iVue portable Spectral domain optical coherence tomography, Optovue, Fremont, CA) was performed as described previously.[14] In our case, fundus examination and OCT were performed in a 4-weeks old patient.

Congenital simplex hamartoma of retinal pigment epithelium develops due to migration of RPE cells towards vitreoretinal junction. In histopathological examination, it has been elucidated that these lesions, in fact, are gliotic retinal and nodular formation of RPE cells attached to internal limiting membrane.[15, 16]

In lesions with extra-foveal localization, vision prognosis is generally good and follow-up is recommended. In lesions with foveal localization and accompanying findings such as traction, macular edema, vision prognosis is poor. The potential complications should be treated.[8, 17] In our case, follow-up was recommended as he was 4-weeks old.

In conclusion, we presented a CHRPE case with foveal localization in a premature patient. This is the first

premature case with CHRPE in the literature, comprising youngest patient. The sonography and OCT are helpful in the diagnosis. The patient is attending to follow-up visits.

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