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Combined hamartoma of the retina and retinal pigment epithelium: A case report

Ren Q *et al*. CHRRPE: A case report

Abstract

BACKGROUND

Combined hamartoma of the retina and retinal pigment epithelium (CHRRPE) is a rare congenital benign tumor which is commonly monocular. Typical CHRRPE comprises slightly raised lesions at the posterior pole, with proliferation membrane often leading to vascular distortion. In severe cases, macular edema, macular hole, retinal detachment or vitreous hemorrhage may occur. Some other fundus diseases such as ocular toxoplasmosis or uveitis may have similar clinical manifestations to CHRRPE, which may be misdiagnosed by inexperienced ophthalmologists.

CASE SUMMARY

A 33-year-old man reported onset of right eye blurred vision for one week prior. Anterior segment and intraocular pressure were normal in both eyes. Left eye fundus photography was normal. Right eye ophthalmoscopy showed vitreous hemorrhage and off-white raised retinal lesions below the optic disc. Proliferative membranes on the lesion surfaces resulted in superficial retinal detachment and tortuosity and occlusion of peripheral blood vessels. A horseshoe-like tear in the temporal periphery was surrounded by retinal detachment. Optical coherence tomography revealed retinal thickening at the focal site with structural disturbance indicated by high reflectance. Right eye ultrasound showed retinal thickening at the lesion, stretching and uplifting of the proliferative membrane, with moderately patchy echo at the optic disc edge. The initial diagnosis was toxoplasmosis. Vitreous fluids were taken during the surgery for cytokine and antibody detection. Fundus fluorescein angiography (FFA) at postoperative follow-up led to final diagnosis of CHRRPE.

CONCLUSION

FFA is helpful in diagnosing retinal and retinal pigment epithelial combined hamartoma. In addition, other cytokine and etiological tests facilitate further differential diagnosis to rule out other suspected diseases.

Key Words: Combined hamartoma of the retina and retinal pigment epithelium; Ocular toxoplasmosis; Fundus fluorescein angiography; Vitreous hemorrhage; Retinal tears; Case report

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Core Tip: Combined hamartoma of the retina and retinal pigment epithelium(CHRRPE) is a rare benign retinal tumor, especially in atypical cases with vitreous hemorrhage and retinal hole, and is likely to be misdiagnosed by inexperienced young doctors. Fundus fluorescein angiography is instructive when diagnosis is difficult, and some etiological tests can also help to identify other inflammatory diseases. This article provides an example of diagnosis to aid young doctors' reflection and learning.

INTRODUCTION

Combined hamartoma of the retina and retinal pigment epithelium (CHRRPE) typically occurs in retinopathy with a very mild elevation of the optic disc or with a tightly attached hyperplastic membrane. Retinal blood vessels are often distorted or even occluded by traction of fibrous membrane, and the macula may be displaced through a macular hole[1]. Ocular toxoplasmosis (OT) is a common cause of posterior uveitis. It can be characterized by thickening of posterior vitreoretinal cortex, adhesion of vitreoretinal retina, and highly reflective deposits at the vitreoretinal interface with retinal vascular occlusion[2,3]. These two diseases are rare but can both be accompanied by macular hole, macular epiretinal membrane, vitreous hemorrhage, or retinal tear. We report a case of CHRRPE with vitreous hemorrhage masquerading as toxoplasmosis.

CASE PRESENTATION

Chief complaints

A 33-year-old male presented to our hospital reporting impaired vision in his right eye.

History of present illness

The patient reported no accompanying symptoms such as headache or eye pain and had no known disease. He denied rheumatic immune disease; infectious diseases such as tuberculosis and acquired immunodeficiency syndrome were excluded by routine examination after admission.

History of past illness

Unremarkable.

Personal and family history

The patient denied a family history of genetic disease, had no history of pet feeding, eating raw meat, working in a farm or slaughterhouse, and no family history of Toxoplasma infection. His parents were not consanguineous.

Physical examination

No anterior segment abnormalities were found on slit lamp examination. Under the ophthalmoscope, vitreous hemorrhage and off-white raised retinal lesions were observed below the right optic disc. Proliferative membranes on the lesion surfaces resulted in superficial retinal detachment and tortuosity and occlusion of peripheral blood vessels. A horseshoe-like tear in the temporal periphery was surrounded by retinal detachment.

Laboratory examinations

On ophthalmic examination, the patient's best corrected visual acuity was 20/40 in the right eye and 20/20 in the left eye, and intraocular pressure was 14 mmHg in the right eye and 15 mmHg in the left eye (1 mmHg = 0.133 kpa). The patient underwent vitrectomy on his right eye on January 5, 2022. During the procedure, vitreous fluid was

collected to test for the following pathogenic microorganisms: bacteria, viruses, fungi, parasites and other pathogens (including mycobacterium and mycoplasma/chlamydia). Test results were negative. Other cytokines and antibodies were detected in vitreous fluids (Table 1). Interleukin (IL)-10 and IL-6 were in the normal range, with only a slight increase in IL-8, excluding uveitis. Basic fibroblast growth factor (which can stimulate cell mitosis and promote collagen production in fibrocytes) and its receptors are present in proliferative vitreoretinopathy, proliferative membranes and vascular endothelial cells. In this case, increased BEGF content in vitreous fluid corresponds to the formation of proliferative film on the lesion surface. Vascular endothelial cell adhesion molecules are associated with blood-eye barrier breakdown, and moderately elevated titers indicate moderate intraocular tissue edema.

Imaging examinations

Fundus photography of the right eye showed vitreous hemorrhage and off-white elevated retinal lesions just below the optic disc, with irregular edges protruding into the vitreous cavity. Proliferative membranes observed on the surface of the lesions had resulted in superficial retinal detachment and tortuosity and occlusion of peripheral blood vessels. A horseshoe-like tear about 1 disc diameter in size was seen in the temporal peripheral retina, surrounded by a small area of retinal detachment (Figure 1A). Optical coherence tomography revealed retinal thickening at the focal site with retinal structural disturbances indicated by areas of high and low reflectance (Figure 1B). Color ultrasound of the right eye showed retinal thickening at the lesion, stretching and uplifting of the proliferative membrane, moderate strength patchy echo at the edge of the optic disc, and sound shadow behind the echo (Figure 1C). Twelve days after the surgery, fundus fluorescein angiography (FFA) was performed and revealed vascular lesions of about 1.5 disc diameters below the optic disc of the right eye, with surrounding pigment disorder and atrophy, twisted blood vessels below the optic disc and fluorescence leakage. Indocyanine green angiography (ICGA) showed persistently

low fluorescence below the optic disc (Figures 1D and 1E). These features led to the diagnosis of CHRRPE.

FINAL DIAGNOSIS

CHRRPE.

TREATMENT

CHRRPE is a congenital benign retinal tumor which grows slowly and generally does not require treatment in the absence of ocular symptoms. In the present case, due to the superficial retinal detachment caused by the formation of anterior retinal membrane, accompanied by vitreous hemorrhage and retinal tears, right eye vitrectomy combined with silicon oil tamponade was performed on January 5, 2022. Intraoperatively, the proliferative membrane on the surface of the lesion was removed and the final stage of the procedure was to fill with silicone oil, which is essential to allow time for chorioretinal adhesions to form after laser treatment. There were no adverse events during or after surgery.

OUTCOME AND FOLLOW-UP

The patient was reexamined two weeks after surgery and examination showed good retinal reduction and sealing of the original retinal tears. Regular reexamination was arranged and silicone oil was removed from the eye as appropriate.

DISCUSSION

CHRRPE was first reported by Gass[4] in 1973 and tends to occur in children, but has also been reported in young people or the elderly[5,6]. It usually occurs in one eye and is rarely associated with systemic disease, but a few cases may be associated with type II or type I neurofibromatosis[7]. The common symptoms of CHRRPE are painless vision loss and strabismus, but it may be symptomless and detected only on fundus examination, depending on the lesion location[8]. In the present case, the patient was

hospitalized due to painless vision loss caused by vitreous hemorrhage, and an off-white lesion was found below the optic disc during specialist physical examination. Typical CHRRPE commonly manifests as mild elevation of the retina at the optic disc or posterior pole, with a layer of proliferative membranes tightly attached to the surface. Retinal blood vessels are often distorted by traction of the fibrous membrane, and the macula may be displaced through macular holes. Vascular leakage, also caused by traction of the fibrous membrane, is a rare occurrence which results in macular edema, retinal detachment, and even vitreous hemorrhage[9]. Retinal tears are an important cause of vitreous hemorrhage and are mainly horseshoe shaped and located in the superior temporal quadrant[10,11], consistent with the present case findings. During surgery in this case, we observed that vascular occlusion was caused by traction of the proliferative membrane on the surface of the lesion, no bleeding was observed during the removal of the membrane, and there was no significant correlation between the temporal retinal tear and the lesion below the optic disc. The patient was considered to have spontaneous retinal tears complicated by CHRRPE, and vitreous hemorrhage originated from the former. The FFA manifestations were typical of CHRRPE. In the early stage of angiography, the choroidal background fluorescence was weak and obscured, particularly in highly pigmented areas. The retinal vessels in the lesion area were clearly tortuous and deformed, while other vessels appeared normal. Due to telangiectasia, microhemangioma and vascular permeability changes in the lesion area, fluorescein leakage may occur in the late stage of angiography. As a result, the lesion area shows high fluorescence, while the pulled vessels around the lesion show no apparent fluorescein leakage[8]. In the present case, FFA after surgery showed vascular lesions with a diameter of about 1.5 disc below the right optic disc, peripheral retinal pigment disorder and atrophy, twisted blood vessels under the optic disc, with obvious fluorescence leakage in the late stage. ICGA showed persistently low fluorescence below the optic disc. OT and uveitis were excluded by the detection of cytokines and antibodies in vitreous fluids during the surgery.

CONCLUSION

In the differential diagnosis of atypical CHRRPE cases and those easily misdiagnosed as OT. FFA and ICGA are also important auxiliary means in addition to examining antibodies in vitreous fluid and aqueous humor, and inquiring about past living and working environment in detail to exclude the diagnosis of OT. In this case, the vascular occlusion around the lesion was determined to be related to anterior retinal membrane traction, the retinal hiatus in the temporal side was considered to be spontaneous, and the vitreous hemophore was also caused by the hole.

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