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Case Report

Multimodal Imaging of Pigmented Paravenous Retinochoroidal Atrophy in a Pediatric Patient with Cystoid Macular Edema

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Abstract

The aim of this case report is to present the multimodal imaging characteristics of pigmented paravenous retinochoroidal atrophy (PPRCA) in a pediatric patient with cystoid macular edema (CME). A 7-year-old girl was admitted to our clinic with complaints of mild blurred vision and poor night vision. Best corrected visual acuity was 10/10 in both eyes. Fundus examination showed atrophic areas around the optic nerve and along the retinal vessels in both eyes. A few small dot-shaped paravenous pigmentations were observed in the mid-peripheral retina. Fundus autofluorescence was consistent with PPRCA. Spectral-domain optical coherence tomography (OCT) revealed the presence of CME and loss of the outer retinal layers outside the macula, with intact retinal layers in the macula. OCT angiography revealed normal choriocapillaris vasculature and flow. The patient was followed up for 6 months but showed no change in CME or clinical appearance. CME without ocular inflammation is an unusual finding of PPRCA and may suggest the involvement of chronic or latent inflammation in the etiology of PPRCA.

Keywords: Cystoid macular edema, optical coherence tomography angiography, pigmented paravenous retinochoroidal atrophy

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Introduction

Pigmented paravenous retinochoroidal atrophy (PPRCA) is characterized by pigment accumulation along the retinal vessels and retinal pigment epithelium (RPE) as well as atrophy of the choriocapillaris bilaterally. The diagnosis is usually based on a typical and characteristic appearance of the fundus. The etiology of PPRCA is unknown, but it is believed to be hereditary or associated with an initial inflammatory cause.[1,2,3,4,5]

PPRCA is a non-progressive or slowly progressive ocular disease and the visual prognosis is generally good. Macular involvement is rare, but macular changes such as macular RPE atrophy, choroidal thinning, pigmentary macular degeneration, epiretinal membrane, and lamellar holes can be seen.[1,2] To date, the presence of cystoid macular edema (CME) in PPRCA with accompanying active inflammation has been reported in only one case.[3] Herein, we present the multimodal imaging characteristics of PPRCA in a pediatric patient with CME but no signs of ocular inflammation.

Case Report

A 7-year-old girl was admitted to our clinic with complaints of mild blurred vision and poor night vision. There was no family history of hereditary retinal disease. The patient had a medical history of hospitalization for an unidentified viral illness at 15 months of age. Written informed consent was obtained from the patient's parents.

Best corrected visual acuity was 10/10 and intraocular pressure was 12 mmHg in both eyes. Slit-lamp examination of the anterior segment was unremarkable and there were no cells or flare in the vitreous. Fundus examination showed the presence of atrophic RPE areas around the optic nerve and along the retinal vessels in both eyes. A few small, dot-shaped paravenous pigmentations were observed in the mid-peripheral retina.

Color fundus photography (Visucam 524 Fundus Camera, Carl Zeiss Meditec AG, Jena, Germany), fundus autofluorescence (FAF), spectral-domain optical coherence tomography (SD-OCT), fluorescein angiography (FA) (Heidelberg Spectralis HRA + OCT, Heidelberg, Germany), and OCT angiography (OCTA) (RTVue-XR Avanti AngioVue OCTA, Optovue Inc., Fremont, CA) were performed.

Color photographs clearly showed multiple areas of changes in the RPE along the retinal vessels and the spots of paravenous pigmentation in the mid-peripheral retina. FAF demonstrated areas of hypoautofluorescence along the retinal vessels consistent with the observed RPE changes in both the central and peripheral retina. SD-OCT showed significant retinal thinning with loss of all outer retinal layers including the outer nuclear layer and external limiting membrane outside of the macula, as well as the presence of CME in both eyes. However, all retinal layers were intact within the macula. Subfoveal choroidal thickness and choriocapillaris were normal on enhanced depth imaging (EDI) with SD-OCT. FA revealed areas of hyperfluorescence along the vessels in both the central and peripheral retina, with no leakage in any of the phases. En face OCTA showed normal flow of superficial and deep retinal layers and choriocapillaris (Figures 1 and 2). The clinical examination was consistent with PPRCA.

The patient was referred to a pediatric infectious disease specialist for etiological investigation that included both clinical examination and laboratory tests. However, no clinical or serological evidence of bacterial, viral, or parasitic disease such as tuberculosis, syphilis, toxoplasmosis, herpes simplex virus, herpes zoster virus, cytomegalovirus, rubella, or measles could be identified. The patient was given topical nonsteroidal anti-inflammatory eye drops and followed up 6 months later. No change in the CME or clinical findings were detected.

Discussion

In this study, we demonstrated PPRCA in a pediatric patient with CME but no signs of ocular inflammation, and documented its characteristics in multimodal imaging including color fundus photography, FAF, SD-OCT, FA, and OCTA.

The diagnosis of PPRCA is usually based on a typical and characteristic fundus appearance such as paravenous pigment accumulation and areas of RPE atrophy around the optic disc and along the vessels. However, it has been reported that the fundus appearance may vary and retinal changes can be mild, moderate,

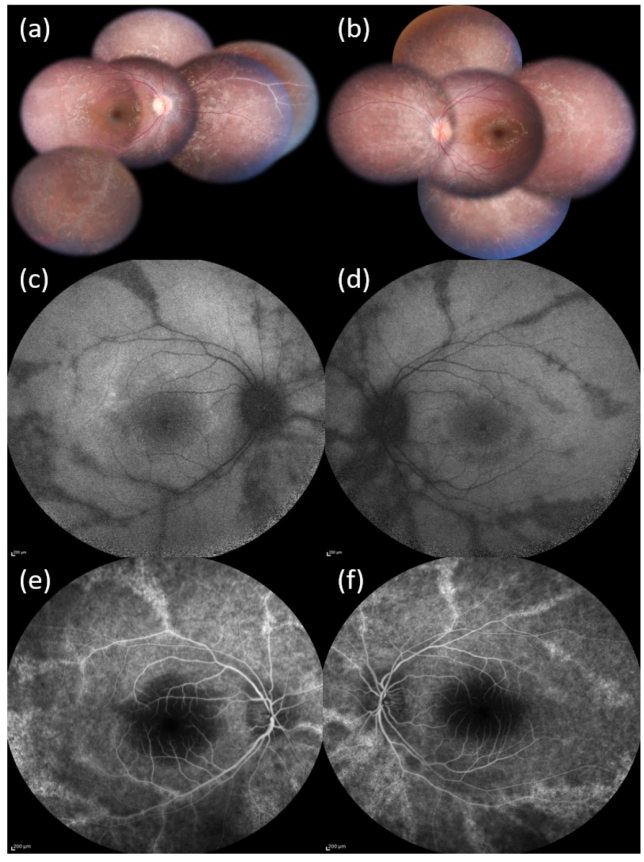

Figure 1. Multimodal imaging of the patient. a,b) Composite color fundus photography shows areas of retinochoroidal atrophy along the retinal veins without pigmentation. c,d) Fundus autofluorescence reveals perivenous hypoautofluorescence and cystoid macular edema. e,f) Fundus fluorescein angiography shows perivenous hyperfluorescence

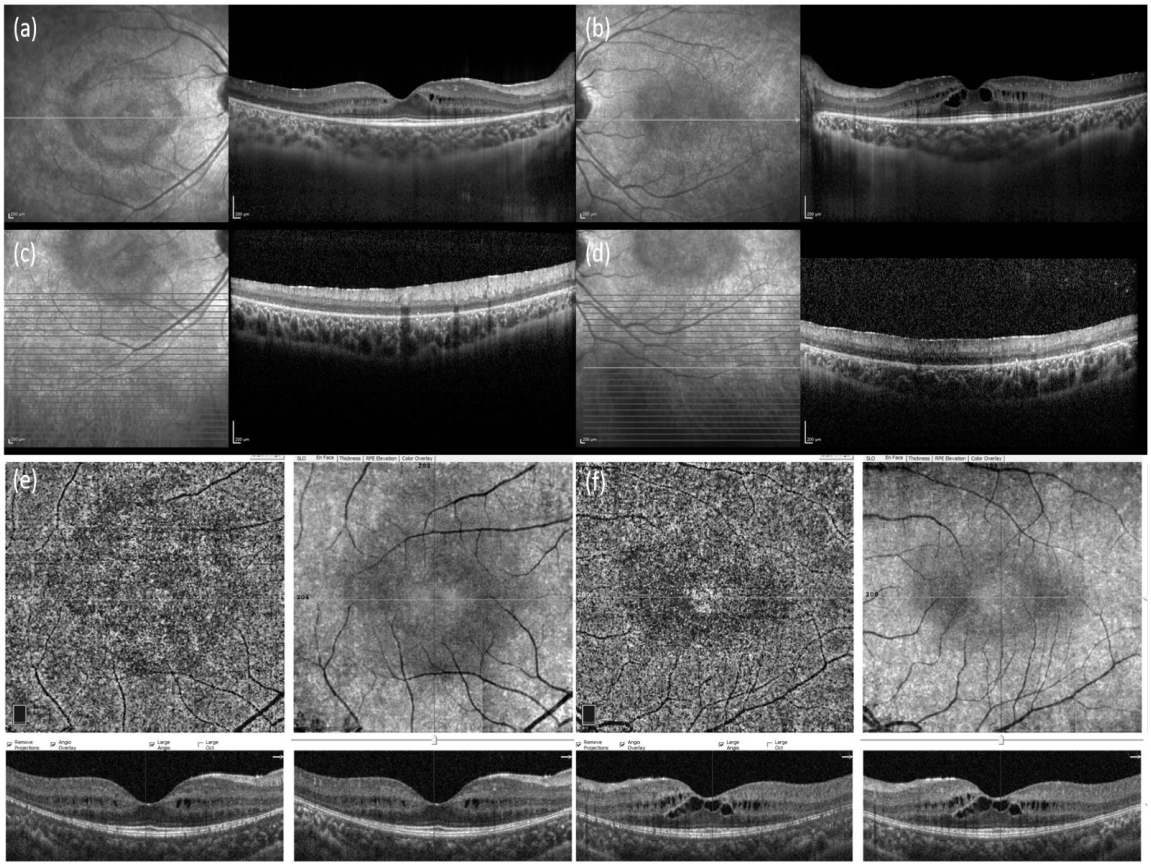

Figure 2. Optical coherence tomography (OCT) and OCT angiography images. a,b) Macular OCT shows cystoid macular edema. c,d) OCT of the perivenous atrophic areas reveals outer retinal layer loss. e,f) OCT angiography images of the macula

or very severe in PPRCA. FAF, SD-OCT, and FA are helpful tools to confirm the diagnosis of PPRCA.[2] The pediatric patient described in this case report had minimal paravenous pigment deposition and large areas of RPE atrophy around the optic disc and along the vessels. FAF, SD-OCT, and FA findings were also consistent with the diagnosis of PPRCA.

Visual acuity is generally good and minimally adversely affected in PPRCA patients without macular involvement. Only a small number of PPRCA cases with macular involvement have been reported in the literature.[3,5] Shona et al.[1] reported in a series of 23 cases that two-thirds of the patients were asymptomatic and the most common macular changes were mild or severe disruption of the outer retina and RPE, and/or choroidal thinning. They found mild macular intraretinal cysts in only one patient with extensive PPRCA. Batioglu et al.[3] reported the case of a 54-year-old woman with the typical fundus appearance of PPRCA accompanied by active inflammation with CME. To the best of our knowledge, the current case is the first reported case of PPRCA with CME but without obvious signs of ocular inflammation.

PPRCA is defined as a non-progressive or slowly progressive disease with an unknown etiology. Several infectious diseases that cause inflammation have been associated with PPRCA, including tuberculosis, sarcoidosis, syphilis, measles, rubella, and Behçet's disease. However, no systemic disease has yet been identified as the cause of PPRCA.[2] We could not find any clinical or serological evidence of disease in our etiological investigations of the patient in the current study. However, the presence of CME and a history of an unidentified viral illness at 15 months of age may support the contribution of chronic or latent inflammation in the etiology of PPRCA.

Shen et al.[6] described areas of choriocapillaris hypoperfusion on OCTA and suggested that it may also result from RPE/outer retinal loss. Recently, Ranjan et al.[4] reported that swept-source OCTA may show a relatively normal choriocapillaris structure, which they noted may be due to a milder form of the disease in their young patient. We demonstrated significant retinal thinning with loss of all outer retinal layers in the patient in the current study, although subfoveal choroidal thickness and choriocapillaris were normal on EDI SD-OCT. The flow in the superficial and deep retinal layers and choriocapillaris was also normal in the 6x6 mm^2 central macular area on OCTA.

PPCRA seems to be an acquired rather than inherited retinal disorder that is generally non-progressive. The underlying

basis of PPCRA is controversial and may include genetic and postinflammatory etiologies. In the literature, PPCRA was reported to be associated with a heterozygous CRB1 variant of uncertain significance identified in a family with apparently dominantly inherited PPCRA with variable expressivity. Others have proposed degenerative, developmental, vascular, or congenital etiologies. Differential diagnoses include chorioretinal degeneration in addition to inflammatory diseases that cause chorioretinal atrophy, including retinitis pigmentosa (pericentral, sector, and typical), helicoid peripapillary chorioretinal atrophy, serpiginous choroidopathy, angioid streaks, cone dystrophy or degeneration, Stickler syndrome, gyrate atrophy choroideremia, and Wagner's dominant vitreoretinal degeneration, and these aspects should be discussed including the patient's family history.[1,7,8,9,10]

In conclusion, by describing the multimodal imaging characteristics of an early/mild form of PPRCA in a pediatric patient with CME, we provide helpful insights into the stages and etiology of the disease.

Ethics

Informed Consent: Written informed consent was obtained from the patient's parents.

Peer-review: Externally peer reviewed.

Authorship Contributions

Surgical and Medical Practices: J.M., C.D., Concept: J.M., C.D., Design: J.M., C.D., Data Collection or Processing: J.M., C.D., Analysis or Interpretation: J.M., C.D., Literature Search: J.M., C.D., Writing: J.M., C.D.

Conflict of Interest: No conflict of interest was declared by the authors.

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