Case Report

Pigmented Paravenous Chorioretinal Atrophy and Mucopolysaccharidosis: A Case Report

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Abstract

Purpose: To report the atypical case of a patient with mucopolysaccharidosis type II (MPS II) in whom bilateral pigmented paravenous chorioretinal atrophy (PPRCA) was found.

Methods: An observational case report.

Results: We present the case of a 31-year-old male patient who presented with decreased visual acuity and in whom fundus examination and multimodal imaging revealed a typical appearance of bilateral symmetrical PPRCA. Our patient also had thick facies with hypertelorism and stubby hands, which prompted an enzyme assay revealing MPS II.

Conclusions: PPRCA is an uncommon condition with an uncertain origin that continues to be a subject of debate. Our case represents an unusual association that points to a potential genetic origin; however, further studies are needed to understand this condition.

Keywords: Mucopolysaccharidosis, Paravenous chorioretinal atrophy, Pigmented retinopathy, Retinal degeneration

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Introduction

Pigmented paravenous chorioretinal atrophy (PPRCA) is a rare disease of unknown etiology, characterized by the presence of pigment clusters arranged along the retinal veins associated with atrophy of the pigment epithelium, outer layers of the retina and the choroid.[1]

Its diagnosis is clinical, based on the characteristic appearance of the fundus, whereas its etiology remains controversial to this day.

Due to its rarity, the available literature on this disease is limited. In the present article, we provide a detailed description of this condition, using multimodal imaging, in a patient with PPRCA in whom we diagnosed mucopolysaccharidosis type II (MPS II).

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Case Report

We report the case of a 31-year-old male patient who presented with progressive decreased visual acuity.

On general examination, it was observed that the patient had a short stature, measuring 158 cm in height, a thick face with hypertelorism, flattened nasal bridge, stubby hands, and a slight intellectual deficit [Figure 1].

Ophthalmological examination revealed a distance visual acuity of 10/10 with correction, and near visual acuity of 1.5 on the Parinaud scale, in the both eyes.

Examination of the anterior segment was unremarkable, with normal intraocular pressure. The fundus revealed bony spicule pigmentations scattered around the venous trunks, associated

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Figure 1: (a) Dysmorphic features with a short neck, large head, and a short stature. (b) Prominent forehead, short nose with a flattened bridge, and thickened lips. (c) Large, thick hands with stubby fingers

with perivenous atrophy. The optic disc and the retinal vessels, on the other hand, were normal.

Fluorescein angiography revealed diffuse hyperfluorescence of the choroidal background by window defect, reflecting diffuse atrophy of the retinal pigment epithelium (RPE), and a masking effect of perivascular pigments in the both eyes. The fundus autofluorescence showed hypoautofluorescent areas along the large vessels [Figure 2a-d].

Spectral-domain optical coherence tomography by Zeiss Cirrus 5000 showed a disruption of the integrity of the ellipsoidal and external limiting membrane in the perifoveal region [Figure 2e].

On the full-field electroretinogram, the responses were not discernible [Figure 2f]. Finally, the visual field was uninterpretable because of the lack of cooperation of the patient.

Given this presentation, the diagnosis of PPCRA was made.

Further questioning revealed a history of inguinal hernia surgery in childhood, with hearing loss that had benefited from a hearing aid.

In addition, no history of consanguinity was found, and the ophthalmological examination of the siblings and parents was without anomalies.

Given the overall clinical presentation, the diagnosis of MPS was suspected and the patient was referred to the geneticist for further management.

Total glycosaminoglycan (GAGs) levels were 33.93 mg/mmol (normal range: 1.19–7.17 mg/mmol); with decreased levels of iduronat 2-sulfatase 0 umol/L/h (cut-off value: >2.5), other enzyme activities were normal, which is consistent with MPS II.

Follow-up at 6 months and 1 year showed preserved visual acuity and stationary lesions.

The patient's informed consent was obtained for the publication of this case report.

Discussion

PPCRA is a rare disease of unknown etiology, characterized by the presence of pigment clusters arranged along the retinal veins associated with atrophy of the pigment epithelium, the outer layers of the retina and the choroid.

Brown described the first case of this disease in 1937 as retinochoroiditis radiata,[1] and some 100 cases have since been reported,[2] with a clear male predominance.

It is bilateral and symmetrical in the majority of cases, as in our patient; however, some unilateral presentations have been described in the literature.[3]

Patients are generally asymptomatic, and discovery is often accidental during a routine ophthalmological examination.[4] Visual acuity is preserved in most cases, depending on the degree of retinal atrophy.

Diagnosis is made based on fundus findings, which show an accumulation of retinal pigments in the form of spicules, spreading out along the retinal veins, associated with perivenous chorioretinal atrophy. This differentiates it from retinitis pigmentosa, where pigments are dispersed throughout the retina.

The macula, optic nerve, vessel caliber, and unaffected retina are usually normal.[5] However, some authors have reported rare cases of macular involvement, such as coloboma, cystoid edema, or macular hole.[6,7]

In mild forms of the condition, there are only a few scattered areas with minimal signs of retinochoroidal atrophy and paravenous pigmentation. Marked cases, on the other hand, are associated with extensive pigmentary deposits and diffuse areas of retinochoroidal atrophy arranged in patches.[2]

In our patient, choroidal atrophy was not very marked, unlike pigment epithelial atrophy.

The etiology of PPRCA remains unknown; however, genetic, developmental, and inflammatory causes have been proposed.

The inflammatory origin remains debated, with case reports suggesting the presence of PPRCA in patients with a history of tuberculosis, congenital syphilis, and other inflammatory diseases such as Behcet's disease, measles, or rubella.[8]

Other authors suggest a genetic origin for this condition. McKay *et al*. reported a mutation in the CRB1 gene located on chromosome 1q31-q32.1 of uncertain significance identified in a family with PPRCA.[8]

Its occurrence in children with macular coloboma or congenital glaucoma would suggest a developmental theory.[4,7]

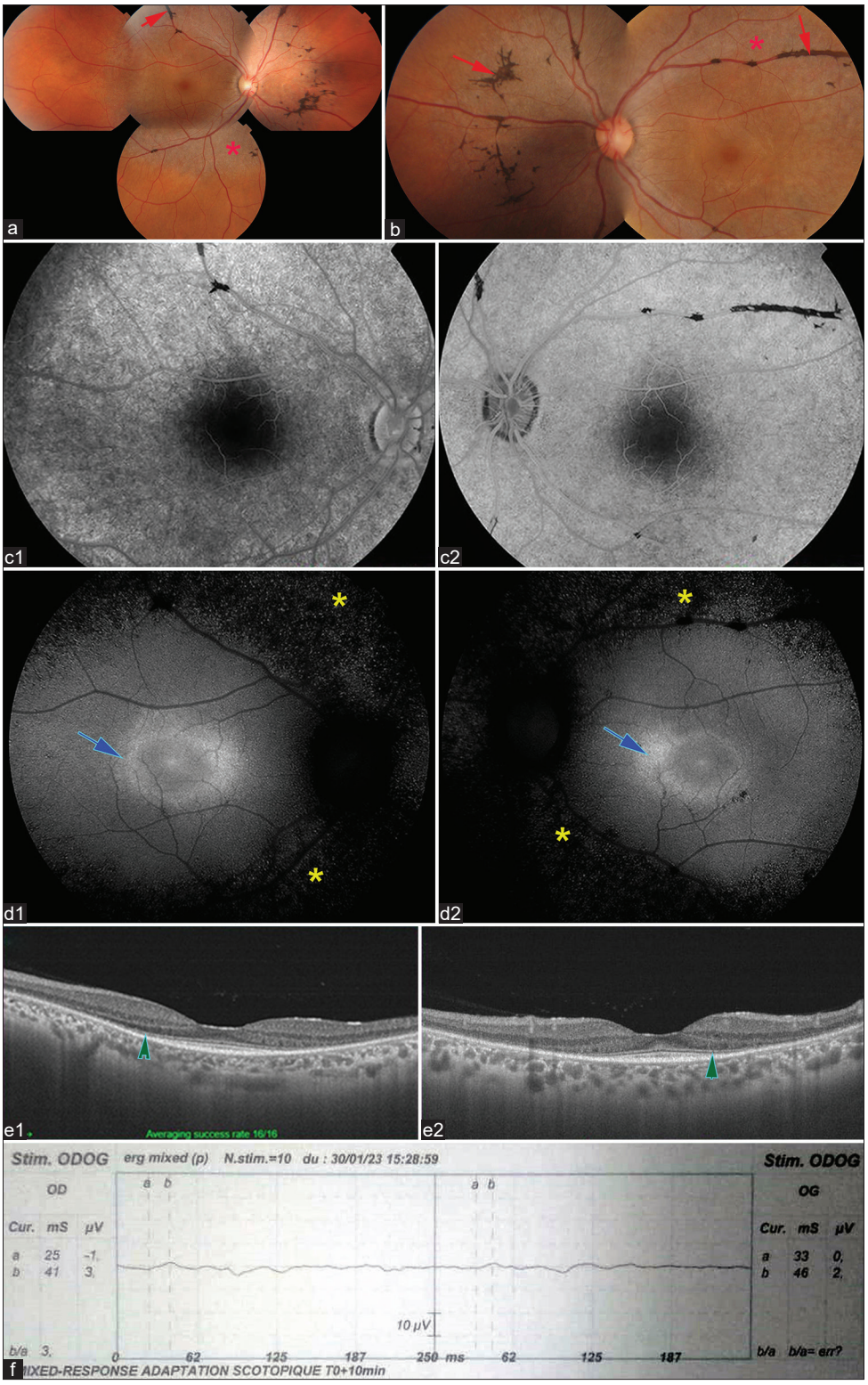

Figure 2: (a) Retinography of the right eye showing pigmentary deposits along venous trunks (red arrow) and atrophy along the retinal vessels (red star). (b) Retinography of the left eye showing pigmentary deposits along venous trunks (red arrow) and atrophy along the retinal vessels (red star). (c) 1, 2: Fluorescein angiography showing masking of the fluorescein due to the pigment deposits with diffuse hyperfluorescence indicating pigment epithelial atrophy ODG. (d) 1, 2: Autofluorescence images showing hypoautofluorescence along the large vessels (yellow star), and hyperautofluorescence of the macular zone due to dysfunction of retinal pigment epithelium (RPE) and accumulation of lipofuscin in RPE (blue arrow). (e) 1, 2: Macular optical coherence tomography showing disruption of the integrity of the ellipsoidal and external limiting membrane in the perifoveal region (green arrow). (f) Mixed response electroretinogram showing altered responses in both the eyes with almost flat tracings

In this case, our patient presented with MPS II or Hunter syndrome.

These conditions constitute a heterogeneous group of genetic diseases caused by a systemic accumulation of GAGs resulting in coarse facial features, skeletal deformities, growth retardation, respiratory and cardiac disorders, inguinal and umbilical hernias, organomegaly, deafness, and neurological involvement in at least 2/3 of cases.[9,10]

The most common ocular manifestations in MPS II are hypertelorism and exophthalmos, optic nerve abnormalities, and retinopathy.[11,12]

Retinal degeneration is a result of the deposition of GAGs in the RPE and in the inter-photoreceptor matrix. Symptoms may include nyctalopia and sensitivity to light. The fundus examination reveals mild changes in the RPE. Whereas, ERG shows more severe depression in rod-mediated responses than cone-mediated responses. However, signs and symptoms are not necessarily correlated to the ERG changes, and mild RPE modifications may be accompanied by important ERG alterations.[13,14]

To the best of our knowledge, this is the first reported case associating MPS with PPRCA. This observation is an additional argument in favor of a genetic origin. However, gene sequencing remains essential, which represents a limitation to our study.

Pigmented perivenous chorioretinal atrophy is an uncommon condition with an uncertain origin that continues to be a subject of debate. We present the case of a patient with MPS II, who presents with bilateral PPRCA, an association reported for the first time in the literature. Whether a mere coincidence or evidence of a genetic origin, further researches are essential to clarify the etiology and guide the management of this condition.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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