CASE REPORT

Case Report: Pigmented paravenous retinochoroidal atrophy: a case report [version 1; peer review: 2 approved with reservations]

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Abstract

This article, to the best of our knowledge, reports the youngest typical case of pigmented paravenous retinochoroidal atrophy (PPRCA) reported to date. A 27-month-old girl presented with exodeviation in her right eye. She had normal birth and development with unremarkable family history. There were no inflammatory signs. In funduscopy, typical bilateral radial paravenous pigmentary changes and retinochoroidal atrophy were noticed in both eyes. The pigmentations consisted of coarse black pigmentations and fine subretinal yellowish round flecks. They arborized into the peripheral retina along the veins. Unaffected areas between the lesions seemed to be normal. Electroretinogram (ERG) responses showed mild to moderate reductions in both scotopic and photopic tests. Based on retinal examination and ERG findings PPRCA was diagnosed. On 16-month follow up, clinical and ERG findings were the same as the initial presentation. This case showed no progression during 16 months of follow up, which may indicate that primary congenital PPRCA with no inflammatory association may be a non-progressive disease.

Keywords

pigmented paravenous retinochoroidal atrophy, Retinochoroidal atrophy

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Introduction
Pigmented paravenous retinochoroidal atrophy (PPRCA) is a bilateral and symmetrical condition, which is characterized by atrophy of choriocapillaris and retinal pigment epithelium (RPE), and pigmentation along the retinal veins. Patients are usually asymptomatic and diagnosis is made during routine examination based on typical fundus appearance and non-progressive nature of disease. Here we present, to the best of our knowledge, the youngest typical case of PPRCA reported to date.

Case report
A 27-month-old girl presented with exodeviation in his right eye to retina clinic, at Farabi eye hospital in April 2016. She experienced a normal birth and development, and had unremarkable family history. She used no medications. Visual acuity testing was not feasible due to patient’s young age. Her parents had noticed outward deviation in her right eye. The cyclorefraction of both eyes were +2.25 diopters. On 16-month follow up, fundoscopy and cycloscopic examination were normal and there were no inflammatory signs. In funduscopy, typical bilateral radial paravenous pigmentary changes and retinochoroidal atrophy were noticed in both eyes (Figure 1). The pigmentations consisted of coarse black pigmentations and fine subretinal yellowish round flecks. They arborized into the peripheral retina along the veins. Unaffected areas between the lesions seemed to be normal.

The fundus examination of her parent and newly born brother showed no abnormality. Electroretinogram (ERG) responses showed mild to moderate reduction in both scotopic and photopic responses, equally. Based on retinal examination and ERG findings, PPRCA was diagnosed. At 9 months later, with no treatment, the fundus findings showed no changes and cyclorefraction was +1 diopter. On 16-month follow up, fundoscopy and ERG findings were the same as those at the initial presentation.

Discussion
To the best of authors’ knowledge, this patient is the youngest with PPRCA ever reported. The patient showed typical characteristics of PPRCA and considerable stability over the course of a 16-month follow up. This presentation is in concordance with The congenital origin hypothesis of PPRCA. In 1937, Brown described this condition for the first time in a 47-year-old man with alopecia areata and named it as retinochoroiditis radiata. The patient had already been under treatment for a disseminated choroiditis at the age of 26 years and had symptoms of tuberculous spondylitis. Considering that his close relatives had died of tuberculosis, the condition was presumed to be a form of tuberculous periphlebitis. The hypothesis of inflammatory origin was further confirmed by other case reports with congenital syphilis, rubella, measles and Behcet’s disease. However, in the years after, a hypothesis of congenital origin was developed and the condition was considered a hereditary disease.

Later in 1962 Franceschetti, changed the name of this condition from retinochoroiditis radiata to a more generally accepted term of PPRCA. So far, there are more than 100 case reports in literature and most of the affected patients are men. In 2003, after lengthy follow ups, Yanagi et al. found that this disease is stationary in younger patients while is slowly progressive in older subjects. The progression in older patients may be attributed to wrong diagnosis of PPRCA and these patients may actually have pseudo-PPRCA.

Although the majority of cases have been sporadic, there are reports of familial occurrence. In 1986, Traboulsi and Maumenee described a mother and her three sons with PPRCA. Every member had a different chief complaint. The youngest son who was 4 years old, had poor fixation, nystagmus, and peripheral pigmentary abnormalities. The 10-year-old son had no signs of pigmentary change and the second son had mild pigmentary changes at age of 7 years. To our knowledge, the members of this family were the youngest cases ever been reported to date.

ERG findings are not the same in all cases and have a wide spectrum. While in some cases, ERG shows noticeable involvement, in others it may be normal or show only mild involvement. Reduction of b wave amplitude is the most common finding, followed by a wave amplitude reduction and prolonged latency. In some cases, rod responses may be affected more than cone responses, while in others cone response reduction is the dominant feature. In our case both responses were almost equally diminished. This variation may reflect the heterogeneous impairment of various cell types in the retina.

Our patient presented with exotropia in right eye and bilateral fundus involvement. Previous studies have reported the association of PPRCA with different ocular problems, including anisometropia, amblyopia, esotropia, exotropia, nystagmus, optic disc drusen, and macular changes, such as cystoid macular edema, pigmentary macular degeneration, lamellar macular holes and macular coloboma. Since secondary PPRCA or pseudo-PPRCA
has been reported, clinicians should be aware of differential diagnoses, which include chorioretinal degenerations, serpiginous choroidopathy, retinitis pigmentosa, tuberculous disseminated choroiditis, helicoid peripapillary chorioretinal atrophy and angioid streaks\textsuperscript{7,8,11,12}.

One of the main advantages of this case report was a relatively long-term follow-up period. This case showed no progression during 16 months of follow up, which may indicate that primary congenital PPRCA with no inflammatory association may be a non-progressive disease. Cases with progressive course or other concomitant findings may be secondary PPRCA and pseudo-PPRCA can be a better term for them.

**Data availability**
All data underlying the results are available as part of the article and no additional source data are required.

**Consent**
Written informed consent for publication of their clinical details was obtained from the parents of the patient.

**Grant information**
The author(s) declared that no grants were involved in supporting this work.

**References**

Open Peer Review

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Version 1

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The review in my opinion will be approved with reservations to improve the paper I will make some suggestions:

1. Picture of fundus of both eyes. Is it possible Autofluorescence or AFG?

2. What kind of Electroretinogram has been made? Picture of it to show the response.

3. Differential Diagnosis, 27-month-old girl is too young to think in PPRA, what kind of systemic or genetic diagnosis has been made to get to that excluded diagnosis?

4. Ask for pregnant infectious illness: Rubeola, Toxoplasma....

I have doubts that the diagnosis will be correct with the information provided in the paper.

*Is the background of the case's history and progression described in sufficient detail?*
Partly

*Are enough details provided of any physical examination and diagnostic tests, treatment given and outcomes?*
No

*Is sufficient discussion included of the importance of the findings and their relevance to future understanding of disease processes, diagnosis or treatment?*
No

*Is the case presented with sufficient detail to be useful for other practitioners?*
Partly

*Competing Interests:* No competing interests were disclosed.
I confirm that I have read this submission and believe that I have an appropriate level of expertise to confirm that it is of an acceptable scientific standard, however I have significant reservations, as outlined above.

Reviewer Report 24 June 2019

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Kim Ramasamy
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Chitaranjan Mishra
Aravind Eye Hospital, Madurai, India

1. What is the name of the imaging device/fundus camera?

2. Can we get the periphery photo? Any history of laser photocoagulation done? The current figure shows 2 pigmented spots at the left margin.

3. Other eye Fundus photo?

4. ERG report available? Please provide the report or at least the important line graphs e.g scotopic, combined response and photopic response.

5. In the case report it is described that: "The pigmentations consisted of coarse black pigmentations and fine subretinal yellowish round flecks. They arborized into the peripheral retina along the veins". However practically, the abnormal pigmentation is a hypopigmentation of the affected area of fundus.

6. In the discussion section, there is no need to start with "the best of authors’ knowledge, this patient is the youngest with PPRCA ever reported." This can be written at the end.

Is the background of the case’s history and progression described in sufficient detail? Partly

Are enough details provided of any physical examination and diagnostic tests, treatment given and outcomes? Partly

Is sufficient discussion included of the importance of the findings and their relevance to future understanding of disease processes, diagnosis or treatment? Partly
Is the case presented with sufficient detail to be useful for other practitioners? 
Partly

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We confirm that we have read this submission and believe that we have an appropriate level of expertise to confirm that it is of an acceptable scientific standard, however we have significant reservations, as outlined above.

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