Unilateral retinitis pigmentosa: Report of two cases

Retinitis pigmentosa unilateral: reporte de 2 casos

Abstract

Retinitis pigmentosa (RP) is the most prevalent form of hereditary retinal dystrophy. Typically, the disease is bilateral and symmetrical. Cases of unilateral RP are very rare and only isolated cases or short series have been reported. We present two cases of unilateral RP. In the first, a 38-year-old woman was diagnosed with unilateral RP in the left eye, and followed up for 10 years. In the second, we describe a 64-year-old male with unilateral RP complicated with cystoid macular edema in the right eye, from the time of diagnosis, followed up for 6 years.

Key words: Unilateral retinitis pigmentosa. Posterior subcapsular cataract. Cystoid macular oedema. Funduscopy. Electroretinography.

Resumen

La retinitis pigmentosa (RP) constituye la distrofia retina hereditaria que presenta una mayor prevalencia. Tipicamente la enfermedad es bilateral y simétrica. Los casos de RP unilateral son muy poco frecuentes, y solo existen en la literatura reportes de casos aislados o series con un número reducido de casos. Presentamos 2 casos clínicos de RP unilateral. En el primer caso describimos una mujer de 38 años diagnosticada de RP unilateral en su ojo izquierdo y un periodo de seguimiento de 10 años. En el segundo caso describimos un varón de 64 años con RP unilateral complicada con un edema macular cistoide desde el momento diagnóstico en su ojo derecho y 6 años de seguimiento.

Introduction

Retinitis pigmentosa (RP) is the most frequent retinal dystrophy. It constitutes a heterogeneous group of clinically and genetically diverse entities. Their common feature is an affection of the photoreceptors, of the rods in the initial stages and of the cones in more advanced stages.1-3

The disease can have a sporadic presentation or follow an autosomal dominant inheritance (AD), recessive or X-linked inheritance pattern. The most common inheritance pattern is AD. The X-linked form is the least frequent, although the most severe.1-5

It typically occurs in the second or third decade of life, with a characteristically bilateral and symmetric loss of visual functions. Patients usually start with nictalopia and a concentric and progressive constriction of the visual field.1,3 Central vision can be altered in advanced phases of the disease due to the occurrence of cystoid macular edema (CME) or posterior subcapsular cataract, among others.1

The typical finding in fundoscopy are intraretinal bone spicules, which initially appear in the middle-peripheral retina, although they may be absent in some cases of the disease, as in RP without pigment. Other characteristic signs are arteriolar stenosis and waxy pallor of the optic disk.

The unilateral form of presentation is extremely infrequent, and most studies report isolated clinical cases or case series with few patients.1-6

There have been studies in which this disease has been treated with vitamin A, although there is no evidence that its use is beneficial.7,8

We present two clinical cases of unilateral RP with several years of follow-up.

Clinical case 1

A 38-year-old woman with no personal or family relevant history, who attended consultation for the first time in 2006 due to blurred vision in her left eye (OS) of several years of evolution.

On examination, the intraocular pressure was 16 mm Hg in the right eye (OD) and 18 mm Hg in the OS. Best-corrected visual acuity (BCVA) was 1.0 in the OD and 0.7 in the OS. The anterior pole was normal in both eyes (OU). Funduscopy of the OD was normal, but in OS, pigment dispersion was observed in the form of spicules that reached the vascular arcades, respecting the macula, with pallor of the optic disc and arteriolar narrowing (Figs. 1 A and B). An electroretinogram (ERG) was requested, which was normal in both scotopic and photopic conditions in the OD. The OS showed an unstructured response both in scotopic and photopic conditions.

Visual fields (VF) were performed, with a normal result in OD. The OS showed an absolute annular scotoma with a small islet of central-inferior vision (Figs. 1 C and D).

Given these findings and the short follow-up period, a possible diagnosis of unilateral RP was made.

The patient was followed up with annual revisions without changes, except for the slow development of a posterior subcapsular cataract in the OS. During the 2016 visit, the patient reported decreased visual acuity in the OS. BCVA was 0.9 in the OD and 0.4 in OS. The OD showed an incipient posterior subcapsular cataract. OS showed an advanced posterior subcapsular cataract (Figs. 1 E and F). Fundoscopic findings were unchanged compared to previous visits. The ERG and the VF remained stable. We agreed with the patient to continue monitoring with annual examinations.

Clinical case 2

A 54-year-old male with no personal or family relevant history, who attended consultation for the first time in 2010 due to diminished vision in OD of 3 days of evolution. The patient reported worse vision in the OD than in the OS for at least 10 years, but he never consulted an ophthalmologist before.

On examination, the intraocular pressure was 9 mmHg in OU. BCVA was 0.3 OD and 1.0 OS. The anterior pole was normal in OU. Funduscopy of the OD showed pigment clusters in the form of bone spicules in the equatorial area of the retina. In addition, papillary pallor and arteriolar narrowing were observed, with an atrophic macula. Funduscopy of the OS was normal (Figs. 2 A and B).

An optical coherence tomography (OCT) was performed, which showed CME with cysts of discrete size and preserved foveal profile with macular atrophy in the OD. OCT of the OS was normal (Figs. 2 C and D).

An ERG showed absence of rod response and a cone response with increased latency and decreased amplitude in OD. OS was normal both in scotopic and photopic conditions.

Visual fields in OD showed an absolute scotoma, whereas the OS was normal (Figs. 2 E and F).

Given these findings and the short follow-up period, a possible diagnosis of unilateral RP was made, complicated by CME.
The patient refused to start treatment for CME, so a conservative attitude with periodic examinations was chosen. The patient is stable after 6 years of follow-up.

Discussion

Unilateral RP is a very rare form of RP presentation. Pedraglia first described it in 1865. The symptoms and fundoscopic findings in the affected eye are identical to the bilateral form; however, the age at presentation is usually higher. Patients with unilateral RP can perform their daily activities almost normally thanks to the healthy eye. This fact could justify a later medical consultation and, therefore, a higher age at presentation (related to the time of diagnosis) compared to the bilateral form. In our first case, the patient showed the typical age of presentation at diagnosis, although she did not consult early, since the involvement in her OS did not prevent her from doing daily activities. Our second case had a very advanced age at diagnosis, despite having started the symptoms in the OD.

Figure 1. A: The retinography of the OD was normal. B: In the retinography of the OS, there are several very confluent spicules that reach the arcades, respecting the macula, with papillary pallor and arteriolar narrowing. C: Humphrey campimetry (24-2) of the OD is normal. D: Humphrey campimetry (24-2) of the OS shows an absolute annular scotoma with a small islet of central-inferior vision. E: After 10 years of follow-up, an incipient posterior subcapsular cataract is observed in OD. F: After 10 years of follow-up, an advanced posterior subcapsular cataract is observed in the OS.
many years ago, because the vision of the OS allowed him to perform his daily activities without disability.

The fact that a disease of genetic origin occurs only in one eye could be explained by two phenomena: on one hand, it could be due to a possible genetic mosaicism in which the mutation would affect only a few cells. On the other hand, the occurrence of a somatic mutation that would only be present in some cells could explain the unilateral presentation of the disease. However, the genetic mechanisms related to this entity are not completely understood.

Figure 2. A: In the retinography from the first visit, OD shows bone spicules, papillary pallor and arteriolar narrowing with an atrophic macula. B: Normal retinography in OS. C: Optical coherence tomography in OD shows small cysts in the middle layers of the retina with a preserved foveal profile and macular atrophy. D: Optical coherence tomography of the OS shows no alterations. E: Humphrey campimetry (24-2) of the OD shows an absolute scotoma. F: Humphrey campimetry (24-2) of the OS is normal.

The diagnostic criteria for unilateral RP were proposed by François et al.: 1) occurrence of typical functional and fundoscopic RP findings in one eye; 2) absence of symptoms of tapetoretinal dystrophy in the contralateral eye with a normal ERG; 3) exclusion of infectious, inflammatory and vascular causes that explain the changes in the affected eye; and 4) a follow-up period long enough to rule out the appearance of RP in the contralateral eye (at least 5 years). Our two cases meet the four criteria, so we can consider them as confirmed unilateral RP.
There are cases in which unilateral RP has been associated with other disorders such as iris heterochromia, pseudoexfoliation syndrome, papillary pit, temporal arteritis or glaucoma. The fact that RP is a very rare disease makes it difficult to know if these associations are due to chance alone or if there is a true association between them\(^6\). During our follow-up, case number 1 developed posterior subcapsular cataracts in OU, more advanced in OS, but an expectant attitude was agreed with the patient, with periodic examinations. Case number 2 presented CME from the time of diagnosis. There are several therapeutic options for the management of this complication, such as topical or systemic carbonic anhydrase inhibitors, or intravitreal injections of antiangiogenic agents or corticosteroids;\(^1\) however, our patient rejected any form of treatment and decided to continue with periodic examinations.

Differential diagnosis should include\(^1,4\):
- Infectious processes: the congenital infection by rubella or Treponema pallidum can cause changes in the fundoscopy that remind RP. Affectation can be unilateral or bilateral.
- Retinopathy due to toxins: phenothiazines typically produce maculopathy, although they can also produce an affectation in the retinal periphery similar to RP. Something similar can happen with chloroquine and hydroxychloroquine.
- Blunt trauma: may lead to changes in the pigment epithelium of the retina, similar to those observed in RP.
- Inflammatory processes: uveitis or autoimmune diseases.

**Conclusion**

Although RP represents the most frequent hereditary retinal dystrophy, its unilateral presentation is extremely rare. In order to diagnose unilateral RP, the four diagnostic criteria must be met, thus requiring a minimum follow-up period of 5 years.

Currently, its pathogenic mechanisms are not well understood, partly due to the small number of cases collected to date.

There are no satisfactory treatment options, so the monitoring and treatment of possible complications that may occur in the course of the disease, such as CME or subsequent subcapsular cataract, would be appropriate.

**Ethical disclosures**

**Protection of human and animal subjects.** The authors declare that no experiments were performed on humans or animals for this study.

**Confidentiality of data.** The authors declare that they have followed the protocols of their work center on the publication of patient data.

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**References**