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Case Report
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Unilateral Pigmentary Retinopathy: A Case Report

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Abstract

Pigmentary retinopathies (PR) are the leading inherited retinal dystrophy and are characterized by threefold heterogeneity: clinical, genetic, and molecular. They are characterized by progressive cell loss involving the photoreceptors and adjacent cell layers leading to atrophy of the retinal tissue. Genetically, all modes of genetic transmission can be seen in pigmentary retinopathy. Like any genetic disease, PR is usually bilateral and symmetrical. The unilateral form remains a rare entity and should only be considered after eliminating differential diagnoses.

Keywords: Retinitis Pigmentosa; Unilateral RP; Photoreceptor loss; Retinal atrophy; Electroretinography (ERG); Visual acuity; Nyctalopia; Pigmented deposits; Optic disc pallor; Genetic transmission

Abbreviations: RP: Retinitis Pigmentosa; ERG: Electroretinography; OCT: Optical Coherence Tomography; VA: Visual Acuity

Introduction

Retinitis pigmentosa (RP) comprises a group of degenerative hereditary retinal conditions characterized by visual dysfunction associated with progressive cell loss affecting photoreceptors and adjacent cellular layers leading to retinal tissue atrophy. The involvement is often bilateral and symmetrical. Unilateral presentation is a rare entity characterized by RP-like changes involving only one eye, for which we report a case.

Case Report

We present the case of a 31-year-old patient, non-consanguineous parents, with no significant medical or family history, who consulted for decreased visual acuity in the right eye over the past month. Furthermore, the patient reported no other associated symptoms such as night blindness, nor any history of ocular trauma, endocular inflammation episodes, or medication or toxic exposure. Examination revealed visual acuity of 3/10 uncorrectable in the right eye, with normal anterior segment and intraocular pressure. Fundus examination revealed pigmented deposits extending throughout the retina sparing only the macular region, a poor macular reflection appearing thickened, rigid and slender retinal arteries and arterioles, and a slightly paler optic disc compared to the contralateral eye (Figure 1). In the right eye, visual acuity was preserved at 10/10, and fundus examination showed a well-colored optic disc, with the retina and retinal arteries appearing normal in all quadrants (Figure 2). Macular OCT showed retinal thinning in the right eye affecting the photoreceptor layer, with macular edema obscuring the foveolar depression. In the left eye, macular OCT was normal (Figure 3). Electroretinography (ERG) revealed significant retinal electrogenesis impairment involving cone and rod photoreceptors in the right eye. In the left eye, retinal electrogenesis was normal (Figure 4). This clinical and electroretinographic presentation suggested unilateral retinitis pigmentosa.

Discussion

Retinitis pigmentosa represents the leading cause of hereditary retinal dystrophies and is characterized by triple heterogeneity: clinical, genetic, and molecular. Clinically, RP manifests in its early stage with nyctalopia and, more pronouncedly, night blindness, reflecting rod dysfunction and contrasting with normal vision under daylight conditions. At this stage, fundus examination is uninformative and does not show pigment deposits; instead, electroretinography detects the involvement. Typically, later in adolescence or young adulthood, progressive peripheral visual field disturbances develop under daylight conditions, with fundus lesions including pseudo-osteoblastic pigment migrations, vascular thinning, and optic disc pallor. Subsequently, foveomacular cones gradually disappear leading to irreversible blindness. Clinical diagnosis of RP is relatively straightforward in advanced stages. Initial involvement with normal fundus examination is diagnosed through ERG, which is altered in the

disease's early stages [1]. This presentation of RP is typically bilateral and symmetrical. Unilateral form remains a rare entity; the patient exhibits characteristic signs of RP in one eye and strictly normal retina clinically and electroretinographically in the contralateral eye. The first case of unilateral RP was described by Pedraglia in 1865 [2]. Other sporadic cases, less frequently familial, have since been reported, giving it an estimated frequency of 2 to 5% of retinitis pigmentosa cases [3]. However, the diagnosis of unilateral RP should only be made after excluding certain retinopathies that may mimic its ophthalmoscopic and/or electroretinographic appearance. François and Verriest proposed four criteria that a presumed case of unilateral RP must meet [4]:

1. Presence of functional changes in the affected eye and an ophthalmoscopic appearance typical of retinitis pigmentosa.

2. Absence of signs of PR in the healthy eye, with a normal ERG.

3. A sufficiently long observation period (over five years) to exclude late-onset disease in the unaffected eye.

4. Exclusion of an inflammatory cause in the affected eye

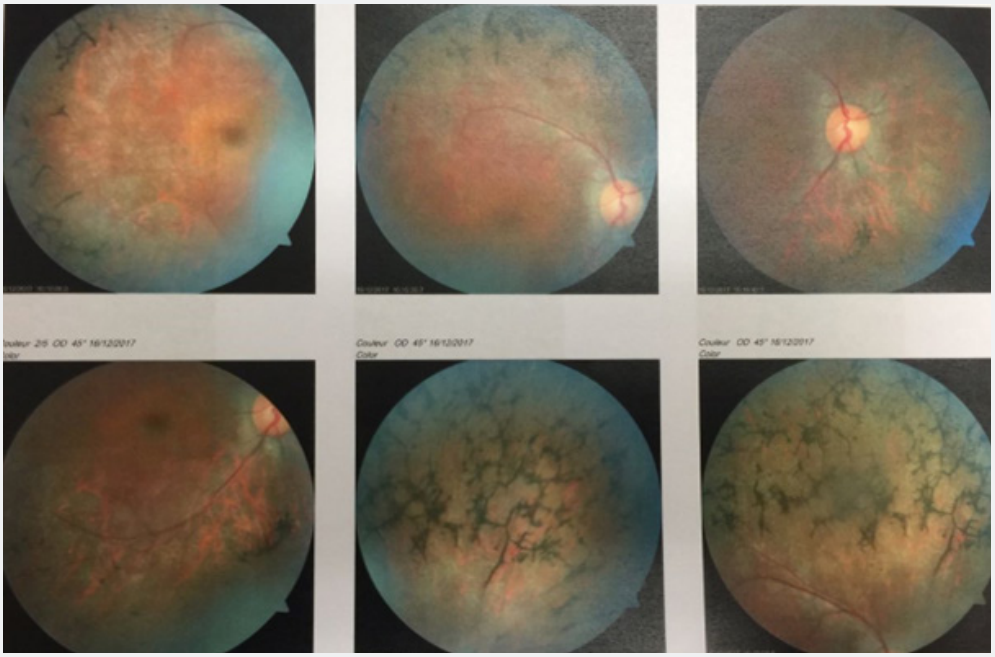

Figure 1: Retinal photographs of the right eye showing pigment deposits resembling osteoblasts extending throughout the retina sparing only the macular region, with rigid and slender retinal arteries and arterioles, and a slightly paler optic disc compared to the contralateral eye.

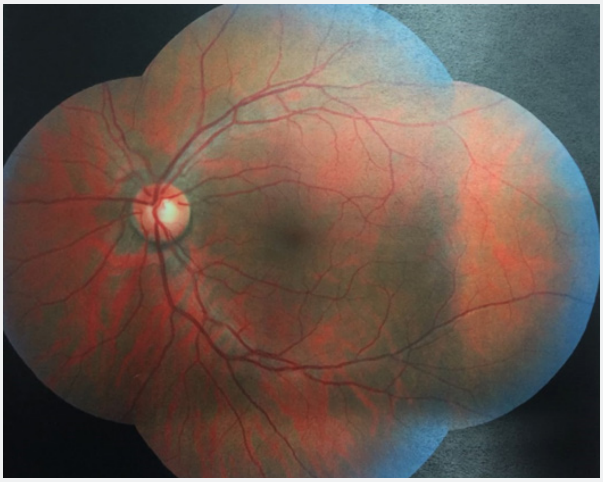

Figure 2: Retinal photograph of the left eye: normal appearance.

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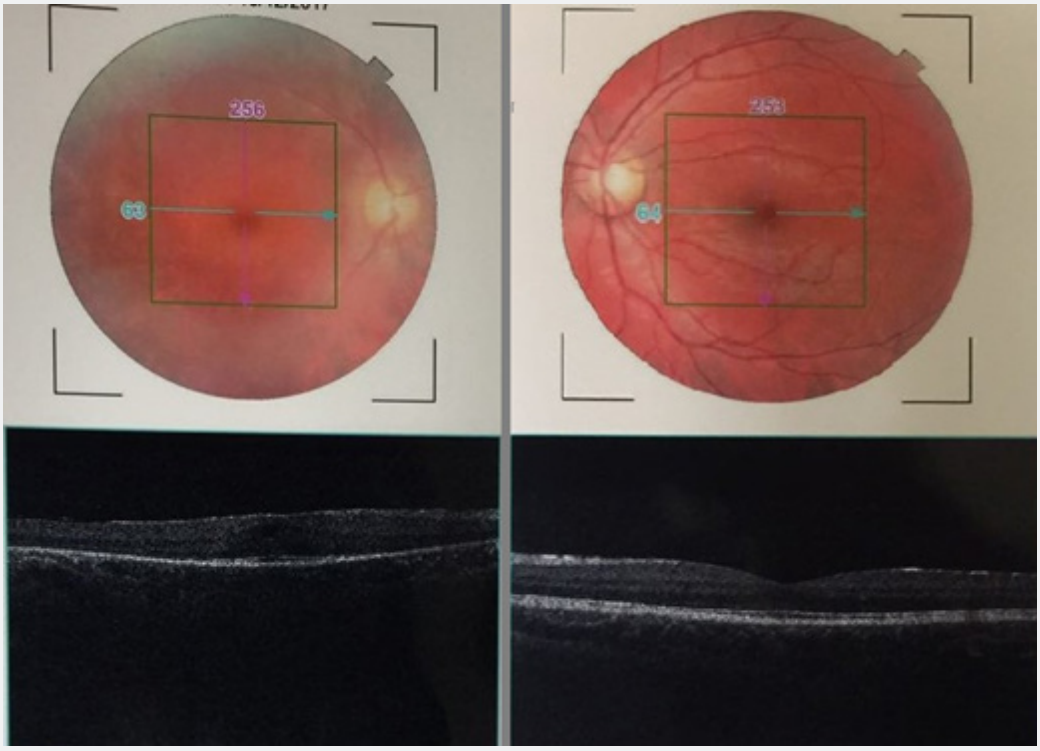

Figure 3: Macular OCT: thinning of the photoreceptor layer with macular edema in the right eye. Normal appearance in the left eye.

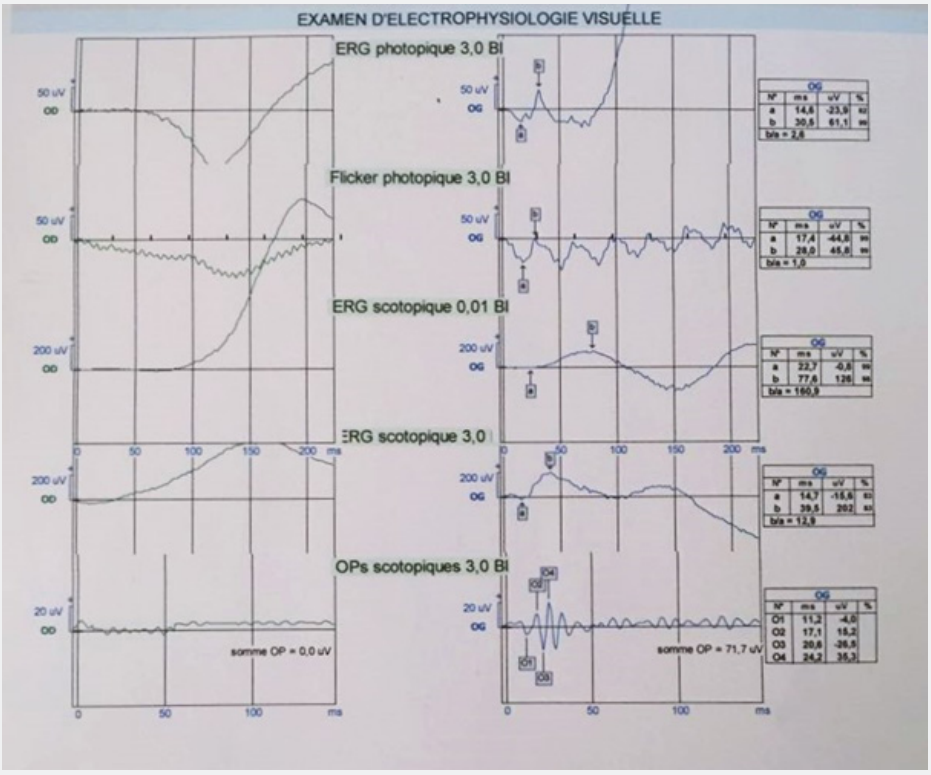

Figure 4: Global ERG: significant impairment of retinal electrogenesis on the right. On the left, retinal electrogenesis is normal.

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The latter criterion should be expanded to include all differential diagnoses to which unilateral RP could be related. Thus, before considering the diagnosis of degenerative unilateral RP, it is essential to exclude other possible aetiologies that can cause ophthalmologic manifestations similar to retinitis pigmentosa, including: congenital ocular infections, notably rubella or syphilitic origin, which can cause retinal changes with pigment irregularities, decreased visual acuity, and visual field constriction, thus mimicking unilateral RP [5,6]; toxic retinopathy related to the intake of antipsychotics from the Phenothiazine group, which can induce not only maculopathy but also peripheral pigmented retinopathy [7]; traumatic retinopathy after blunt trauma can cause pigmentary changes resembling RP by migration of retinal pigment epithelium cells into the retina and formation of pseudo-osteoblastic structures mimicking unilateral RP [8]; finally, some cancers can be associated, as paraneoplastic syndromes, with retinopathy similar to RP with decreased VA, pigment dispersion, and campimetric and electroretinographic deficits [9].

Genetically, all modes of genetic transmission can be encountered in retinitis pigmentosa: autosomal dominant, autosomal recessive, and X-linked, or more rarely non-Mendelian modes of transmission: digenic transmission, maternal transmission (mitochondrial cytopathies) [10]. Increasingly, mutations directly linked to the disease are identified over the years. Currently, the different forms of retinitis pigmentosa can be attributed to mutations in over 250 genes [11]. Genetic diseases rarely affect only one eye. Two genetic mechanisms could explain a unilateral presentation of RP: mosaicism and non-germinal somatic mutations. These two mechanisms can cause asymmetric or unilateral involvement of a genetic disease [12].

Conclusion

Unilateral RP is a rare entity that should only be diagnosed after excluding differential diagnoses. Regular follow-up would be recommended to assess disease progression and search for asymmetric contralateral involvement.

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