CASE REPORT

Retinitis Pigmentosa Sine Pigmento Mimicking a Chiasm Disease

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ABSTRACT

A 75-year-old woman presented to her ophthalmologist complaining of visual loss for several years. The ophthalmic examination was remarkable for a bitemporal visual field defect. Magnetic resonance imaging (MRI) scan of the brain was normal without evidence of chiasm compression. Neuro-ophthalmic examination was consistent with a retinal rather than a chiasmal disease. Retinal multimodal imaging helped in the correct diagnosis of retinitis pigmentosa, later confirmed by genetic testing.

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Introduction

Retinitis pigmentosa is a heterogeneous group of progressive inherited retinal disease associated with visual disfunction caused by degeneration of retinal photoreceptors. The visual field defect caused by retinitis pigmentosa varies widely but is generally represented by peripheral progressive constriction linked to degeneration of rods. However, if the disease is sectorial, the visual field defect may vary and reflect the area of the retina involved. A bitemporal hemianopia is usually caused by damage to the optic chiasm and can occur from the direct or indirect effects of a variety of lesions, almost always of compressive type. We describe a patient with a bitemporal visual field defect caused not by optic chiasmal dysfunction but by retinitis pigmentosa that was diagnosed by electroretinography and autofluorescence.

Case report

A 75-year-old woman with a history of arterial hypertension presented to her ophthalmologist complaining of visual loss for several years. The visit was remarkable for bilateral posterior capsular cataract (more evident in OS) and a best-corrected visual acuity (BCVA) of 16/20 in OU. The computerised 30-2 visual field test showed a

bitemporal hemianopia, and an urgent brain computed tomography (CT) scan was performed. This resulted negative as well as subsequent brain contrast magnetic resonance imaging (MRI) scan. She was sent to us for a second opinion. At our visit, her BCVA was 16/20 OU, slit-lamp examination (SLE) confirmed a mild posterior capsular cataract OU, and pupils were equal and briskly reactive to light with no RAPD. Colour vision was slightly reduced at Ishihara test (10/12 OU). Fundus examination (Figure 1) revealed a normal optic disc and macula appearance in both eyes. Peripheral retina examination was hindered by cataract but revealed atrophy of retinal pigment epithelium (RPE) mostly in the nasal retina. No bone spiculae were present. Retinal artery vessels were slightly reduced in diameter. Repeated computerised visual field was remarkable for a bitemporal visual field defect, with no respect of the vertical midline and a relative central sparing (Figure 2). Optical coherence tomography (OCT) scan showed marked disruption of photoreceptor layer outside the macula in OU (Figure 3). Short-wavelength fundus autofluorescence (SW-AF) was performed and showed a discrete halo of hyperautofluorescence at the posterior pole associated with retinal hypo-autofluorescence especially in the nasal retina, compatible with visual field results (Figure 4). Near-infrared AF (NIR-AF) was also

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Figure 1. Retinography OU is unremarkable except for mild arterial narrowing.

Figure 2. Humphrey computerised 30-2 visual field shows temporal visual field defect OU. Vertical midline is not respected, and there is a relative central sparing.

remarkable for high macular AF in OU (Figure 5) more evident than SW-AF, and hypo-autofluorescence of the nasal retina as well. The results were compatible with retinitis pigmentosa (sine pigmento) confirmed later by electroretinography (ERG) studies (reduced A-wave on full-field ERG). At specific questioning, the patient also admitted to have a worst visual performance in dim light condition, underlying the nyctalopia-related rod

disfunction. Genetic testing was positive for mutation in *USH2A* gene, which usually causes Usher syndrome but also autosomal recessive retinitis pigmentosa. The patient denied hearing problem.

Discussion

Retinitis pigmentosa (RP) is a heterogeneous group of progressive inherited retinal diseases characterised by

Figure 3. OCT scans show marked disruption of photoreceptor layer outside the macula in OU.

a degeneration of photoreceptors with visual dysfunction. The characteristic feature that gives the name to the disease is pigmented epithelial changes in the mid-periphery of the retina. However, there is a high variability in the clinical appearance of the retina in these patients; when the characteristic peripheral bone-like spiculae pigmentary changes are

present, as happens most of the times, diagnosis of RP is straightforward. But in RP sine pigmento, an atypical form of RP, fundus examination may be unremarkable because the characteristic pigmented epitheliopathy is not seen, and the diagnosis may be missed, particularly during the early stages of the disease, when other features suggestive of RP are usually not yet present (e.g., arterial narrowing, cystoid macular oedema, pale disc, and vitreous changes). Since virtually any visual field defect caused by an inner retinal lesion can also be produced by a lesion of the optic nerve, a normal appearance of the retina at a superficial fundus examination may represent a diagnostic dilemma and induce the physician to consider a retrobulbar (in this case an optic chiasm) disorder as the cause of patient's complaint. A few papers were published about hemianopia related to retinitis pigmentosa^{1,2} or other retinal disease,^{3,4} but to our knowledge this is the first case described of retinitis pigmentosa sine pigmento simulating a bitemporal hemianopia from a chiasmal disease. Salgado et al.⁵ underscored the importance of performing an ERG when a bitemporal hemianopia is not associated with a chiasmal disease. Our case, moreover, also highlights the importance and the advantages of performing AF, and in general retinal multimodal imaging, when a genetic retinal disease is suspected. Compared with full-field ERG, which is still the gold standard for the diagnosis of many inherited retinal disorders, autofluorescence is

Figure 4. SW-AF images show a discrete halo of high AF at the posterior pole and a retinal hypo-autofluorescence especially in the nasal retina, compatible with visual field results.

Figure 5. NIR-AF is remarkable for a macular high AF in OU.

faster and more readily available and may give the initial clue to the final diagnosis. Results are easily interpretable, and the test is less invasive. Notably, NIR-AF may have the advantages over SW-AF in unveiling a process that precedes the formation of fluorophores that emit the SW-AF signal.⁶

This case highlights the potential for retinal disorders to produce visual field defects that mimic those produced by chiasmal lesions and the importance of performing autofluorescence of the fundus when a neurologic pattern of visual field defect is not explained by brain MRI scan. The correct diagnosis of a retinal disease may finally avoid unnecessary and expensive radiologic testing.

Declaration of interest

The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the article.

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