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Fundus auto fluorescence imaging and optical coherence tomography analysis in a patient with retinitis pigmentosa inversa

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Abstract: We present a case of retinitis pigmentosa inversa in a 63-year-old female patient. Fundus examination revealed diffuse chorio retinal degeneration with bone-spicule pigmentary alterations in the posterior pole and mid-peripheral retina, whereas pigmentary alteration was not detected in the peripheral retina. Fundus auto fluorescence imaging demonstrated clearly defined hypo fluorescence was observed corresponding to the chorio retinal atrophy at the posterior pole in both eye. On optical coherence tomography, the photoreceptor line was undetectable in both eyes. Our findings may contribute to a better understanding of this rare condition.
Keywords: retinitis pigmentosa, retinitis pigmentosa inversa, fundus auto fluorescence, optical coherence tomography

INTRODUCTION

Retinitis pigmentosa (RP) is a retinal degenerative disease characterized by the loss of photoreceptors and formation of retinal pigment deposits [1]. RP inversa or inverse RP is an atypical variant of RP that remains localized to the posterior pole of the eye [2-5]. The exact prevalence is unknown. Both central and peri central forms of RP inversa have been described, depending on the degree of macular involvement. Pericentral retinopathy is characterized by areas of choroidal degeneration with pigment migration and bony spicules around the macula in a ring-shaped pattern that leave central vision unaffected [6-9]. The central form of RP inversa is characterized by bilateral, symmetric retinal changes limited to the posterior pole of the eye involving the macular area, ultimately leading to severely decreased visual acuity with intact peripheral vision [2-5]. The optic disc may appear pale, with attenuated vessels. Choroidal sclerosis and atrophy usually occur centrally, with funduscopically normal peripheral retina.

Recently, optical coherence tomography (OCT) and fundus auto fluorescence (FAF) imaging have been recognized as useful tools to observe microstructural changes such as loss of the photoreceptor and retinal pigment epithelium (RPE) cells [10, 11]. Herein, we present a case of RP inversa in a 63-year-old female patient.

CASE REPORT

A 63-year-old Japanese female patient was referred to our clinic for progressive vision loss in both eyes. She noticed night blindness since childhood. The patients' medical history was unremarkable and there was no family history of ocular disease. Her parents had a cousin marriage. Best corrected visual acuities were hand motion for the right eye and 0.02 for the left eye, and intraocular pressure was 13 mmHg in both eyes. Fundus examination revealed diffuse chorio retinal degeneration with bone-spicule pigmentary alterations in the posterior pole and mid-peripheral retina (Figure 1), whereas pigmentary alteration was not detected in the peripheral retina. In addition, waxy optic disc pallor in the right eye and retinal arteriolar attenuation were observed in both eyes.

Ultra-wide-field retinal imaging (OPTOS 200Tx; Optos, Scotland, UK) demonstrated above peculiar findings (Figure 2A and B arrows). In addition, clearly defined hypo fluorescence was observed corresponding to the chorio retinal atrophy at the posterior pole (Figure 2C and D arrows) by FAF imaging.

Visual field testing using Goldmann perimetry could not be performed in the right eye (Figure 3A), while the visual field of the left eye showed moderate peripheral constriction with absolute scotoma in the central field (Figure 3B).

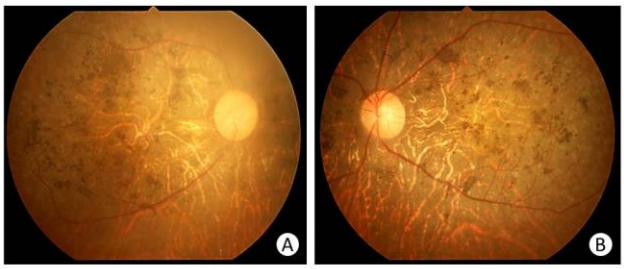

Fig. 1 Fundus photographs of the right (A) and left (B) eyes

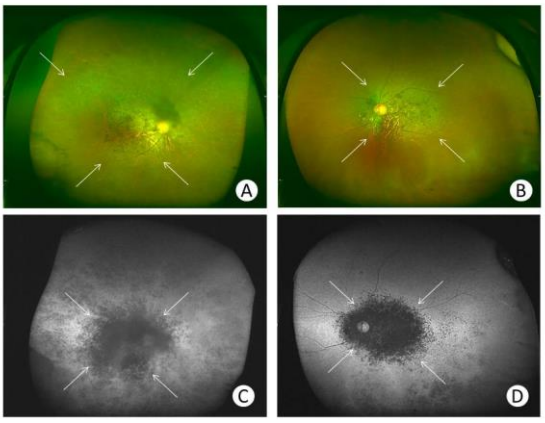

Fig. 2 Ultra-wide field imaging of the right (A) and left (B) retina; Fundus auto fluorescence imaging of the right (C) and left (D) retina

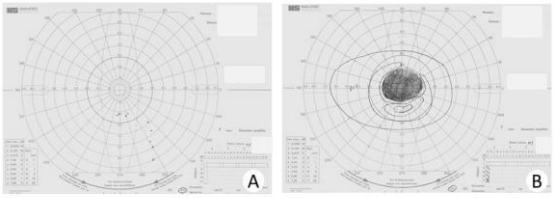

Fig. 3 Goldmann perimetry of the right (A) and left (B) eyes

On OCT (RS-3000, NIDEK, Japan), macular thickness was reduced in the right eye (Figure 4A and C arrows), the photoreceptor line was undetectable, and hyper reflective punctuate drusen-like substances were clearly detected in the atrophic area (Figure 4A-D arrowheads).

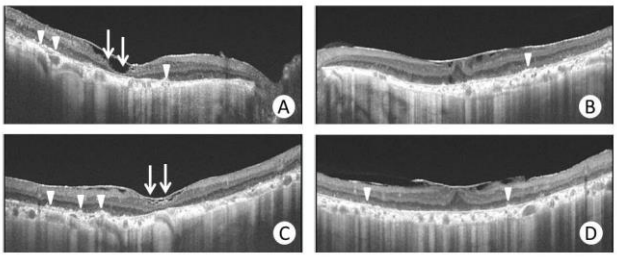

Fig. 4: Optical coherence tomography images of the right (A and C) and left (B and D) eyes. (A and B; horizontal scan, C and D; vertical scan)

Arrows indicate macular atrophy in the right eye. Arrowheads indicate hyper reflective punctuate drusen-like substances. The standard electro retinogram including flicker, flash, rod response and cone response could not be recorded in both eyes (Figure 5A-D).

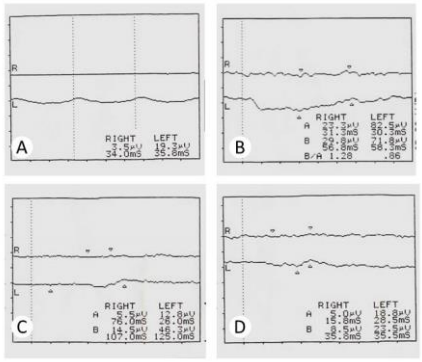

Fig 5: Standard flicker (A), flash (B), rod response (C) and cone response (D) electro retinogram

On the basis of the above findings, we diagnosed RP inversa in this patient. The patient continues to receive low vision rehabilitation.

DISCUSSION

RP inversa or inverse RP is an atypical variant of RP that remains localized to the posterior pole of the eye. FAF and OCT analyses of this rare condition are extremely rare. Generally, FAF is physiologically observed and arises from lipofuscin that accumulated in RPE cells [10, 11]. Hypo auto fluorescence may reflect atrophic or defective RPE cells or blockage by hard exudates or hemorrhages, while hyper auto fluorescence is thought to arise from metabolic or oxidized byproducts in the extracellular space or in RPE cells, macrophages, or other cells. In eyes with RP, hypo auto fluorescence suggests RPE atrophy, accompanying loss of overlying photoreceptors, and a possible association with impaired visual function. Ogura et al. [10] described that the hypo fluorescence was well correlated with the scotoma, while iso fluorescence and hyper fluorescence were consistent with the remaining visual field. Similarly, our patient showed that the FAF images was co-localized with the visual field seen on Goldmann perimetry. Furthermore, OCT findings confirmed the undetectable photoreceptor line. Thus, FAF imaging may be useful to recognize fundus changes topographically as well as functional visual filed changes.

Although our findings were based on a single case of advanced RP inversa, they may contribute to a better understanding of this rare condition.

DISCLOSURE

The authors have no conflicts of interest to disclose.

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