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A Case of Fundus Albipunctatus in a 61-Year-Old Man

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Abstract: We present a case of fundus albipunctatus in a 61-year-old man. The patient had a 2-year history of night blindness. Fundus examination revealed numerous yellow-white flecks throughout the midperipheral retina, sparing the posterior pole in both eyes. The standard flash electroretinogram revealed severely decreased a- and b-waves in both eyes. Although patients with fundus albipunctatus generally complain of night blindness from early childhood, we emphasize that it is important to consider the possibility of fundus albipunctatus even in the elderly with a recent history of night blindness.
Keywords: Fundus albipunctatus, Night blindness, Electroretinogram.

INTRODUCTION

Fundus albipunctatus is a type of congenital stationary night blindness with an autosomal recessive inheritance pattern [1–3]. The fundus of these patients has a characteristic appearance: a large number of discrete, small, round or elliptical yellow-white lesions at the level of the retinal pigment epithelium (RPE) [1–3]. Usually the lesions are concentrated in the midperiphery and not in the center of the macula. Patients with fundus albipunctatus typically complain of night blindness from early childhood [1–3]. In contrast, retinitis punctataalbescens has a progressive course and is classified as a form of retinitis pigmentosa [1, 2, 4]. It was reported that mutations of the 11-*cis*–retinol dehydrogenase (RDH5) gene, which is expressed predominantly in the RPE, cause fundus albipunctatus [2, 3, 5–8]. We present a case of fundus albipunctatus in a 61-year-old man.

CASE REPORT

A 61-year-old Japanese man presented with a 2-year history of a night blindness. His family history was unremarkable. Best corrected visual acuity was 1.0 and 1.2 in his right and left eyes, respectively. Intraocular pressure was 10 mmHg in each eye, and the patient had no specific medical history. No inflammatory cells were observed in the anterior segment or vitreous of either eye. Fundus examination revealed that the posterior pole was spared in both eyes (Fig. 1: A, B), and that numerous yellow-white flecks were present throughout the midperipheral retina (Fig. 1: C,D).

Visual field testing by Goldmann perimetry showed relative scotomain the right eye (Fig. 2: A) and mild inferonasal visual field defect in the left eye (Fig. 2: B).

The standard flash electroretinogram (ERG) showed severely decreased a- and b-waves in both eyes after 30 min of dark adaptation (Fig. 3).

Multifocal ERG response arrays (Fig. 4: A, B) and 3-dimensional plots (Fig. 4: C, D) showed decreased amplitudes in both eyes.

On the basis of the above findings, we diagnosed fundus albipunctatus in this patient.

DISCUSSION

Generally, patients with fundus albipunctatus complain of night blindness from early childhood, and the clinical course has been considered to be stationary with normal visual acuity, visual field, and color perception [1–3]. However, recent studies have shown that the diseaseis not stationary; cone visual function deteriorates; and cone dystrophy may develop in a subset of patients [6, 7, 9]. Nakamura *et al.* [6] described 14 Japanese cases of fundus albipunctatus. According to their report, an average current age was 38.8 years (range, 12–74 years). Further, fundus albipunctatus either with or without cone dystrophy is caused by mutations of the RDH5 gene. Cone dystrophy is observed frequently in elderly patients with fundus albipunctatus. Makiyama *et al.* [7] described 5 Japanese cases in which average current age was 29.6 years (range, 15–64 years). Wada *et al.* [8] described 6 Japanese cases with an average current age of 56.2 years (range, 40–57 years). Sergouniotis *et al.* [5] described 9 cases in which the average current age

was 31.9 years (range, 7–55 years). Littink *et al*. [4] described 13 cases that involved retinitis punctataalbescens and fundus albipunctatus; according to the Littink report, average diagnosed and current ages were 13.8years (range, 4–47 years) and 23.9years (range, 7–65 years), respectively. In the case we describe, the patient was not aware of night blindness in childhood. Moreover, he maintained normal visual function without cone dystrophy.

Although this case lacks novel findings, we emphasize that it is important to consider the possibility of fundus albipunctatus even in the elderly with a recent history of night blindness.

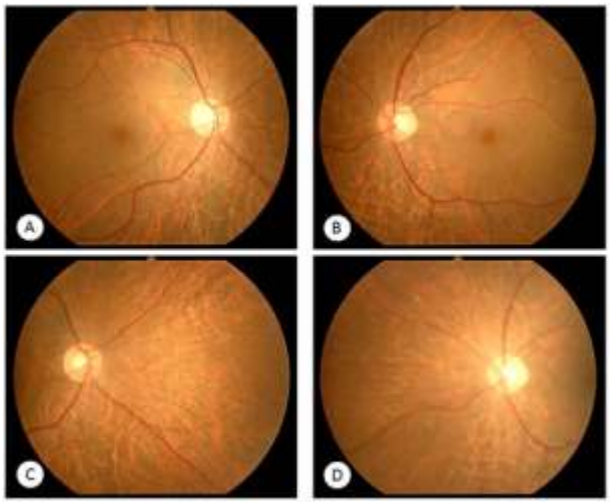

Fig. 1: Photographs of right (A, C) and left (B, D) fundus, showing intact posterior pole in both eyes (A, B), and yellow-white flecks throughout midperipheral retina (C, D)

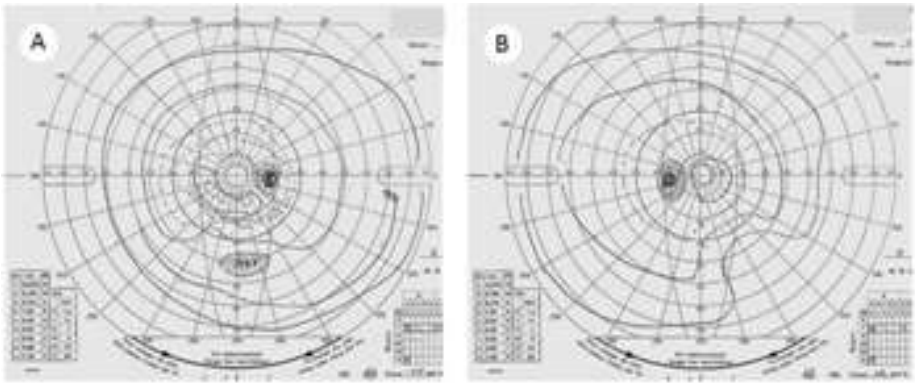

Fig. 2: Goldmannperimetry of right (A) and left (B) eye

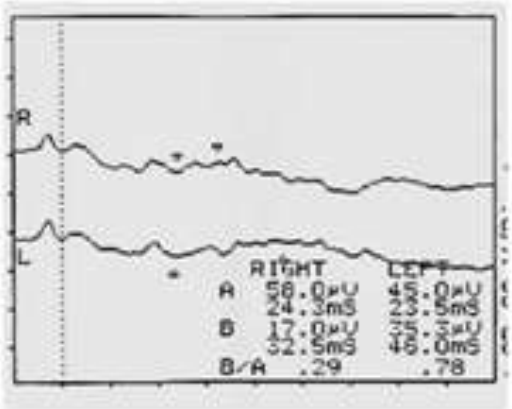

Fig. 3: Standard flash electroretinogram

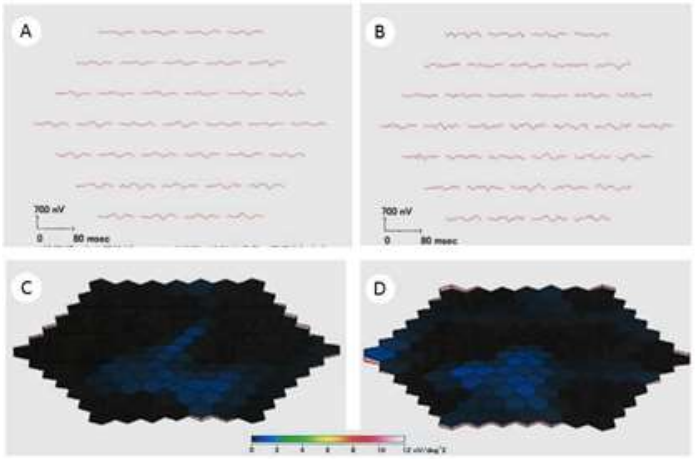

Fig. 4: Multifocal electroretinogram depicting response arrays (A, B) and 3-dimensional plots (C, D).

REFERENCES

1. Marmor MF; Long-term follow-up of the physiologic abnormalities and fundus changes in fundus albipunctatus. Ophthalmology, 1990; 97(3): 380-384.

2. Genead MA, Fishman GA, Lindeman M; Spectral-domain optical coherence tomography and fundus autofluorescence characteristics in patients with fundus albipunctatus and retinitis punctataalbescens. Ophthalmic Genet., 2010; 31(2): 66-72.

3. Wang NK, Chuang LH, Lai CC, Chou CL, Chu HY, Yeung L *et al*.; Multimodal fundus imaging in fundus albipunctatus with RDH5 mutation: a newly identified compound heterozygous mutation and review of the literature. Doc Ophthalmol., 2012; 125(1): 51-62.

4. Littink KW, van Genderen MM, van Schooneveld MJ, Visser L, Riemslag FC, Keunen JE *et al*.; A homozygous frameshift mutation in LRAT causes retinitis punctataalbescens. Ophthalmology, 2012; 119(9): 1899-1906.

5. Sergouniotis PI, Sohn EH, Li Z, McBain VA, Wright GA, Moore AT *et al*.; Phenotypic variability in RDH5 retinopathy (Fundus Albipunctatus). Ophthalmology, 2011; 118(8): 1661-1670.

6. Nakamura M, Hotta Y, Tanikawa A, Terasaki H, Miyake Y; A high association with cone dystrophy in Fundus albipunctatus caused by mutations of the RDH5 gene. Invest Ophthalmol Vis Sci., 2000; 41(12): 3925-3932.

7. Makiyama Y, Ooto S, Hangai M, Ogino K, Gotoh N, Oishi A *et al*.; Cone abnormalities in fundus albipunctatus associated with RDH5 mutations assessed using adaptive optics scanning laser ophthalmoscopy. Am J Ophthalmol., 2014; 157(3): 558-570.

8. Wada Y, Abe T, Fuse N, Tamai M; A frequent 1085delC/insGAAG mutation in the RDH5 gene in Japanese patients with fundus albipunctatus. Invest Ophthalmol Vis Sci., 2000; 41(7): 1894-1897.

9. Miyake Y, Shiroyama N, Sugita S, Horiguchi M, Yagasaki K; Fundus albipunctatus associated with cone dystrophy. Br J Ophthalmol., 1992; 76(6): 375-379.