

## Fundus Autofluorescence Imaging, Optical Coherence Tomography, and Multifocal Electoretinogram Analysis of Congenital Grouped Albinotic Spots

Shinji Makino, Yusuke Arai, Hironobu Tampo

Department of Ophthalmology, Jichi Medical University, Shimotsuke, Tochigi, Japan

### \*Corresponding Author:

Name: Shinji Makino

Email: [makichan@jichi.ac.jp](mailto:makichan@jichi.ac.jp)

**Abstract:** A 7-year-old Japanese girl displayed multiple, variably sized, sharply circumscribed, placoid, white lesions in both eyes, with no additional symptoms. The appearance of these lesions was consistent with congenital grouped albinotic spots of the retina. Fundus autofluorescence imaging showed clearly-defined abnormal autofluorescence, corresponding to these albinotic spots. The outer retinal layer and the photoreceptor inner/outer segment junctions were observed to be irregular and discontinued corresponding to some albinotic spots, by optical coherence tomography. Multifocal electroretinograms of both eyes displayed decreased amplitudes. Therefore, we have described a case of congenital grouped albinotic spots in the eyes with functional deterioration.

**Keywords:** Congenital albinotic spots of the retina, Fundus autofluorescence, Optical coherence tomography, Multifocal electroretinogram

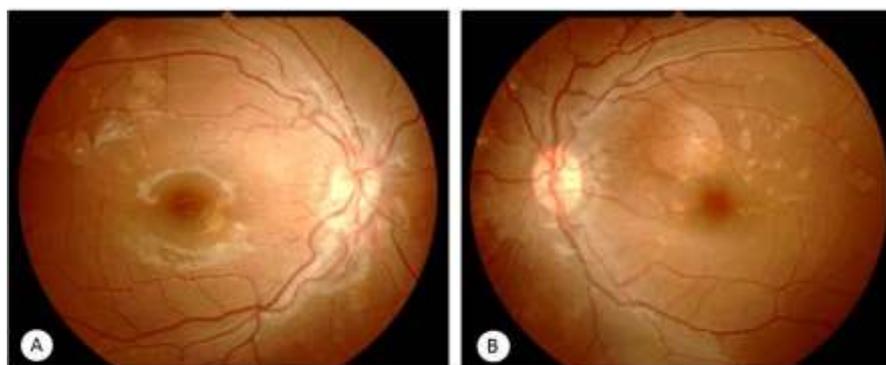
### INTRODUCTION

Congenital grouped albinotic spots (CGAS), or “polar bear tracks”, is an uncommon disorder affecting the retinal pigment epithelium (RPE), and is characterized by multiple, variably sized, sharply circumscribed, placoid, white lesions, organized in patterns often resembling animal footprints [1–7]. The detection is usually coincidental during routine ocular examination. It has been speculated that the spots are focal changes in RPE cells, where a white material, possibly a precursor of melanin, is deposited instead of melanin granules [1, 2]. The disorder is considered to be without functional consequence, and patients typically have normal visual acuity, visual fields, color vision, dark adaptation, electroretinography, and electrooculography findings [2]. Some previous reports describe the use of fundus autofluorescence (FAF), optical coherence tomography (OCT), and multifocal electroretinogram (mfERG) for CGAS identification [2,

4, 5]. In this report, we present a case of CGAS affecting a 7-year-old Japanese girl, with functional deterioration.

### CASE REPORT

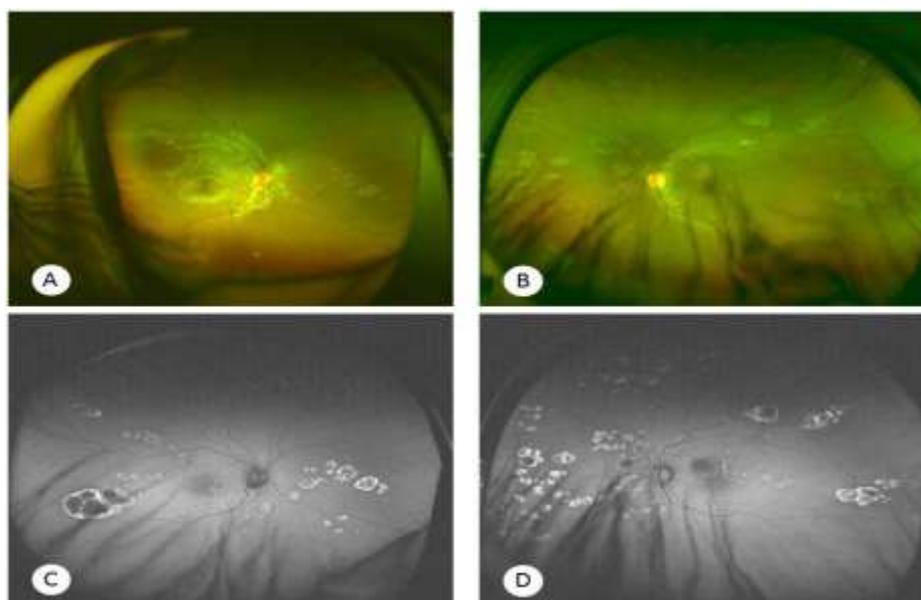
A 7-year-old, asymptomatic, Japanese girl was referred to our clinic for ophthalmological examination of bilateral fundus discoloration. The patient had a surgical history including congenital biliary atresia, vesicoureteral reflux, cleft lip and palate, and intestinal malrotation. She displayed a visual acuity of 1.2 in both eyes, normal anterior segments in both eyes, and normal ocular pressures. Ophthalmoscopy of both eyes revealed multiple, variably sized, sharply circumscribed, placoid, white lesions (Fig. 1A and 1B). Additionally, a slightly excavated lesion was also detected in the left eye, resembling a coloboma, measuring 1.5 × 1.0 disc diameters, near the superonasal fovea (Fig. 1B).



**Fig. 1: Photographs of the (A) right and (B) left fundus**

The size of the lesions was detected to be variable by ultra-wide-field retinal imaging (OPTOS 200Tx; Optos, Scotland, UK), and varied between the diameter of one vessel and 2 times the diameter of the optic disc (Fig. 2A and 2B). Clearly defined abnormal autofluorescence was observed corresponding to the

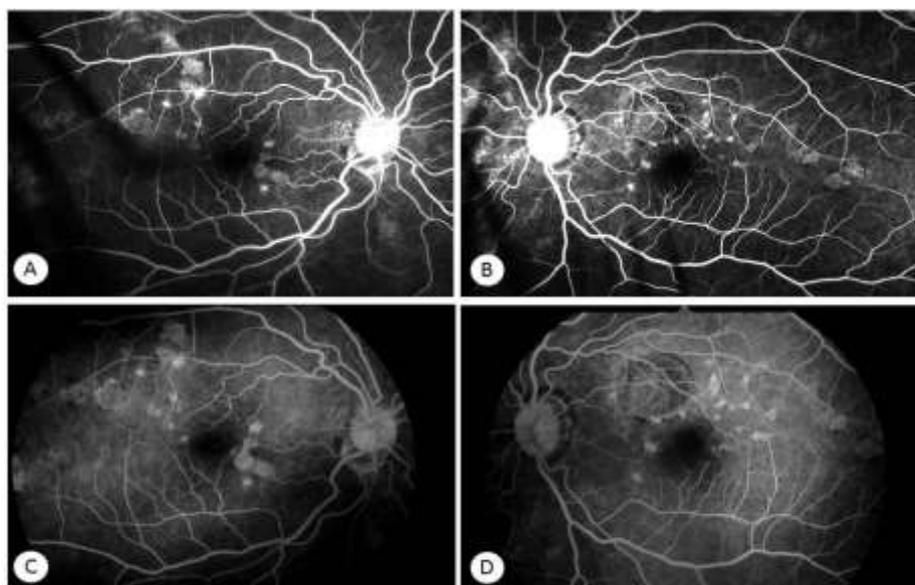
albinotic spots (Fig. 2C and 2D) by FAF imaging (OPTOS 200Tx). While the majority of the spots showed increased autofluorescence, some with reduced autofluorescence were also observed. A colobomatous lesion demonstrated hypoautofluorescence.



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

Hyperfluorescence was observed during the early (Fig. 3A and 3B) and late phase (Fig. 3C and 3D)

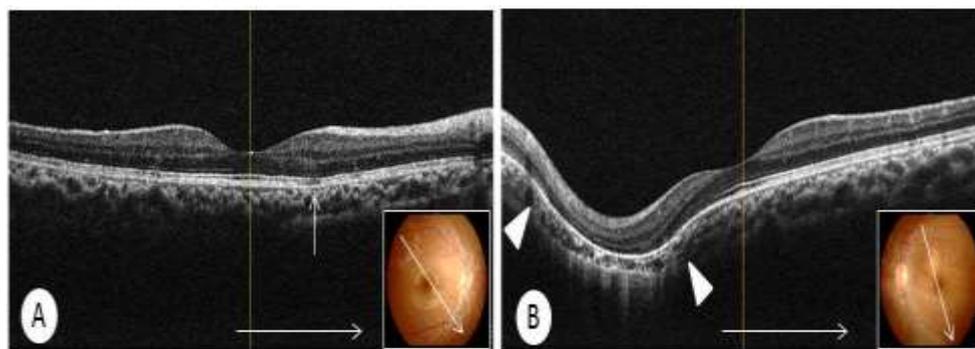
corresponding to the albinotic spots, by fluorescein angiography.



**Fig. 3 Fluorescein angiography of the (A and C) right and (B and D) left eye during the early and late stages**

OCT (RS-3000; NIDEK, Aichi, Japan) revealed the outer retinal layer and the photoreceptor inner/outer segment junctions to be irregular and

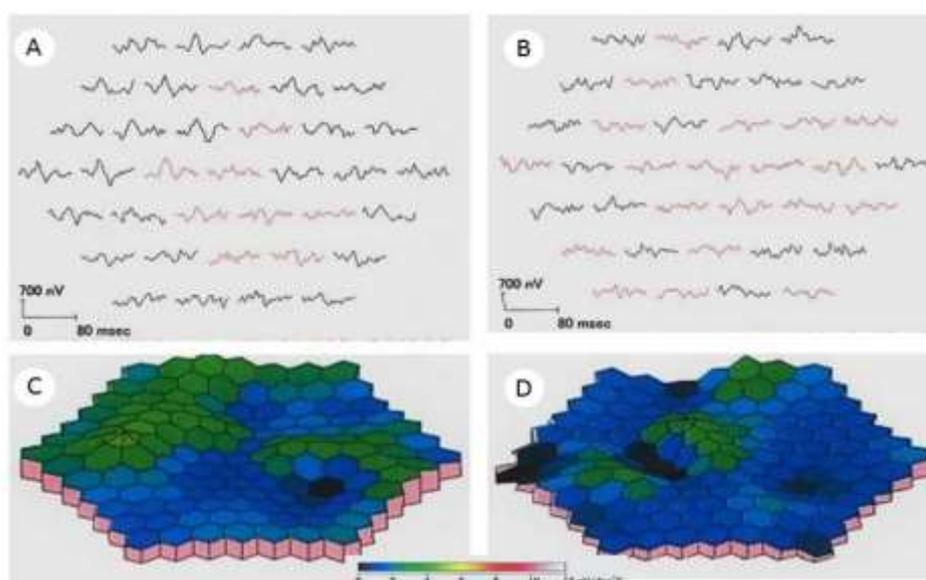
discontinued, corresponding to some of the albinotic spots (Fig. 4A arrow). A colobomatous lesion revealed excavated change (Fig. 4B arrowheads).



**Fig. 4** Optical coherence tomography of the lesions in the (A) right and (B) left eye

Multifocal ERG (LE-4100; Tomey, Aichi, Japan) response arrays (Fig. 5A and 5B) and 3-

dimensional plots (Fig. 5C and 5D) showed decreased amplitudes in both eyes.



**Fig. 5 A & B:** multifocal electroretinogram response arrays and (C and D) 3-dimensional plots

## DISCUSSION

In this report, we have described a case of CGAS with functional deterioration, as detected by mfERG analysis. Patients with CGAS generally display normal visual acuity, visual fields, color vision, dark adaptation, electroretinography, and electrooculography findings [2]. However, very few reports have focused on the functional examination of patients with CGAS using mfERG [2, 5]. Kim *et al.* [2] described the presence of defects corresponding to CGAS in three patients without visual complaints by microperimetry and mfERG. Barbazetto *et al.* [5] described a patient with a solitary albinotic spot. In these cases, the defects observed by mfERG were of decreased amplitude.

CGAS is typically diagnosed by its characteristic appearance during fundus examination. FAF imaging, as opposed to a fundus photograph, was used to show clearly defined abnormal autofluorescence in the eyes. The abnormal autofluorescence patterns could be consistent with the hypothesis that defective

lipofuscin metabolism contributes to varying stages of disease in CGAS [2]. The areas showing increased FAF could indicate RPE dysfunction, while the areas with decreased FAF may represent advanced atrophy in the RPE [2].

Kim *et al.* [2] reported that the OCT of hypoautofluorescent CGAS lesions showed disrupted signals from the hyperreflective layer of the retina, corresponding to the inner and outer segment junction in the region corresponding to the spots. These FAF and OCT findings suggest abnormalities in the RPE and the overlying photoreceptors. We theorized that the functional deterioration may be a result of the above findings.

## CONCLUSION

Our findings were based on a single case, and we could not obtain the OCT and mfERG readings corresponding to all albinotic spots. Therefore, long-term follow-up and additional cases are necessary to

definitively characterize the functional changes in CGAS.

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