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Multimodal Imaging of Retinochoroidopathy in Choroideremia Carrier

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Abstract **Case Report**

A 48-year-old woman was referred for ocular fundus abnormality by medical checkup. She had no subjective complaints. Fundus examination revealed diffuse moth-eaten appearance with chorioretinal atrophy in both eyes. Moreover, patchy areas of depigmentation were observed around the optic disc and in the peripheral retina. Fundus autofluorescence showed mosaic appearance in widespread retina, and patchy hypoautofluorescence areas corresponding to patchy areas of depigmentation. Although optical coherence tomography scan through the fovea of both eyes showed relatively preserved foveal contour and retinal layers, subtle irregularities at the level of the retinal pigment epithelium and ellipsoid zone observed. Based on these collective findings, we diagnosed our patient with retinochoroidopathy in choroideremia carrieir. Choroideremia is rare, occurring even less frequently as a symptomatic disease in female carriers. Our findings may contribute to a better understanding of this rare condition.

Keywords: choroideremia, carrier, fundus autofluorescence.

INTRODUCTION

Choroideremia is an X-linked disorder characterized by degeneration of the retina, retinal pigment epithelium (RPE), and choroid owing to mutations in the *CHM* gene [1-3]. Patients afflicted with the disease typically present with poor night vision followed by a gradual loss of peripheral vision, often culminating in legal blindness in the fourth or fifth decade. With a prevalence of 1 in 50,000 individuals, choroideremia is rare, occurring even less frequently as a symptomatic disease in female carriers [1-3].

Here we report multimodal imaging of retinochoroidopathy in choroideremia carrier.

CASE REPORT

A 48-year-old woman was referred for ocular fundus abnormality by medical checkup. She had no subjective complaints. Her personal history as well as physical examination result was unremarkable. Her father had a severe visual impairment. Her best-corrected visual acuity was 1.2 in the both eyes. The anterior segment was normal in both eyes with no crystals visible at the corneal limbus. Fundus examination revealed diffuse moth-eaten appearance with chorioretinal atrophy in both eyes. Moreover, patchy areas of depigmentation were observed around the optic disc and in the peripheral retina (Figure 1).

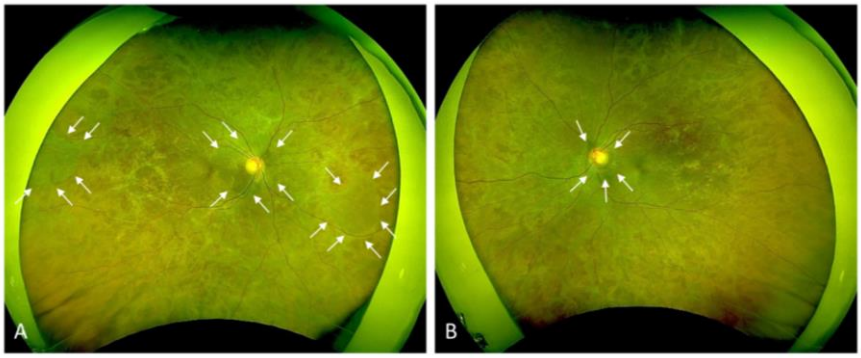

Figure 1: Fundus photographs of the (A) right and (B) left eyes

Note diffuse moth-eaten appearance in both eyes. Patchy areas of depigmentation were observed around the optic disc and the peripheral retina (arrows).

Fundus autofluorescence (FAF) showed mosaic appearance in widespread retina, and patchy hypoautofluorescence areas corresponding to patchy areas of depigmentation (Figure 2).

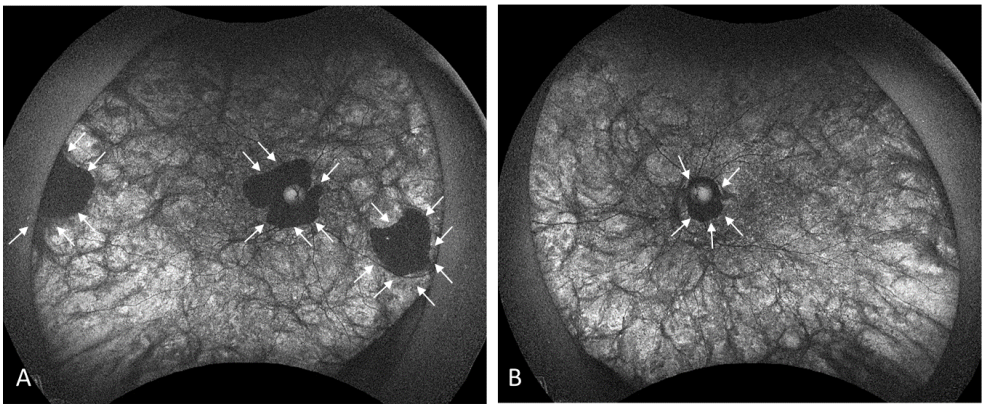

Figure 2: Fundus autofluorescent imagings of the (A) right and (B) left eyes

Note mosaic appearance in widespread retina, and patchy hypoautofluorescence areas corresponding to patchy areas of depigmentation (arrows).

On fluorescein angiography, diffuse moth-eaten appearance with increased visibility of the choroidal blood vessels was more clearly observed in both eyes (Figure 3).

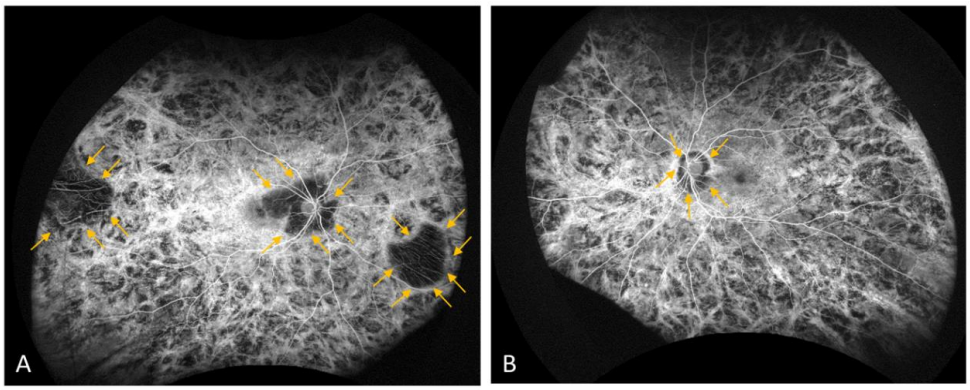

Figure 3: Fluorescein angiography of the (A) right and (B) left eyes

Diffuse moth-eaten appearance was more clearly observed in both eyes.

Indocyanine green angiography demonstrated hypofluorescent areas corresponding to patchy areas of depigmentation (Figure 4).

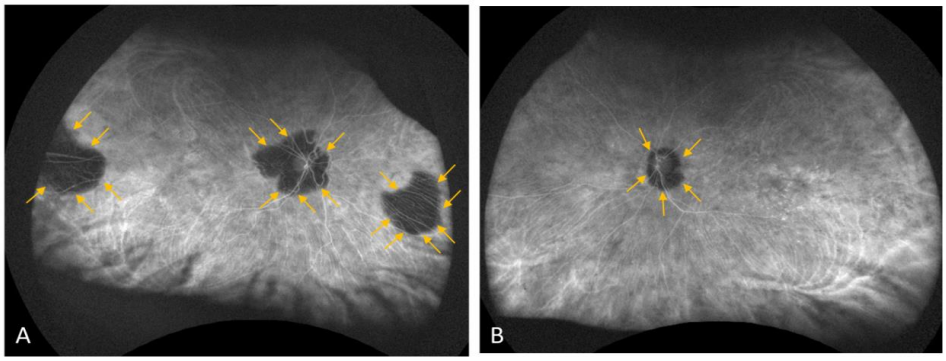

Figure 4: Indocyanine green angiography of the (A) right and (B) left eyes

Note hypofluorescent areas corresponding to patchy areas of depigmentation (arrows).

Although optical coherence tomography (OCT) scan through the fovea of both eyes showed relatively preserved foveal contour and retinal layers, subtle irregularities at the level of the RPE and ellipsoid zone (EZ) observed more in the right eye than in the left eye (Figure 5). In addition, intraretinal microcyst was detected in the right eye.

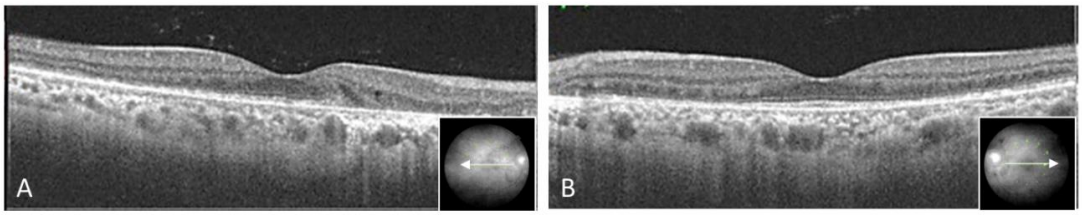

Figure 5: Optical coherence tomographic images of the (A) right and (B) left eyes

Note subtle irregularities at the level of the RPE and ellipsoid zone, intraretinal microcyst in the right eye

Goldmann visual field testing identified enlarged blind spot and relative scotoma corresponding to patchy areas of depigmentation in both eyes (Figure 6).

Figure 6: Goldmann perimetry of the left (a) and right (b) eyes

Note enlarged blind spot and relative scotoma corresponding to patchy areas of depigmentation.

The patient had normal electroretinography responses. Although genetic screening was not available in this case, based on the patient's family history and the aforementioned examinations, a diagnosis of choroideremia carrier was made. The patient was followed without any treatment.

DISCUSSION

In this case, there were no subjective symptoms, even though there was a widespread fundus change.

Jauregui *et al.*, [1] evaluated the disease spectrum of female choroideremia patients. They stated that "Severe disease" when widespread retinal atrophy is observed throughout the posterior pole. On FAF, large areas of hypoautofluorescence were observed throughout the posterior pole, corresponding to areas of extensive chorioretinal atrophy. "Intermediate disease" was defined as presenting with a smaller, localized area of chorioretinal atrophy in proximity or covering the optic disc. "Mild disease" was defined as presenting without areas of chorioretinal atrophy.

Preising *et al.*, [2] describe FAF in carriers of choroideremia, and to compare FAF findings with ophthalmoscopy and electrophysiologic and psychophysical data. They conclude that FAF is a rapid, non-invasive indicator even when other functional tests are unremarkable. In previous reports [1-5], FAF showed speckled autofluorescence as hyperautofluorescence dots alternate with hypoautofluorescence granular areas, but these changes were not observed in this case. FAF is considered as a surrogate of lipofuscin distribution in RPE and has

become an essential biomarker for disease progression [1-5]. FAF reveals regions of chorioretinal atrophy in patients with choroideremia as hypoautofluorescent areas with sharp hyperautofluorescent edges because of remaining degenerating RPE and loss of photoreceptors.

CONCLUSION

Our findings are based on a single case; additional cases and genetic examination are necessary. A minority of female carriers may present with significant retinal and choroidal atrophy that leads to night blindness and visual impairment comparable to that seen in affected male subjects, thus long-term follow up is necessary in this case.

DISCLOSURE

The author declares no conflict of interest.

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