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Clinical features and visual function in a patient with Fish-eye disease: Quantitative measurements and optical coherence tomography

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ABSTRACT

Purpose: We describe a case of fish-eye disease (FED) where the clinical features and visual function were investigated using anterior segment optical coherence tomography (OCT) and quantitative measurements.

Observations: A 36-year-old Japanese woman with FED presented with bilateral corneal opacities and visual complaints. Both contrast sensitivity and straylight were measured and OCT imaging was performed. Contrast sensitivity and straylight measurements revealed remarkably decreased visual function, despite good visual acuity. OCT demonstrated homogenously hyper-reflective corneal opacification, and a normal total corneal thickness.

Conclusions and Importance: The findings from this case suggest that OCT is useful for analyzing the clinical features of FED, and that the quantitative measurement of visual function furthers the understanding of vision-related symptoms in FED, despite good visual acuity.

1. Introduction

Bilateral corneal opacity is a hallmark clinical feature of Fish-eye disease (FED), which was first reported by Carlson et al.[1] The dense corneal opacification is slowly progressive and causes severe visual impairment beginning as early as 15 years of age.[2] FED is a rare autosomal recessive disorder and known as a partial disease of lecithin–cholesterol acyltransferase (LCAT) deficiency.[3,4] It is thought that cholesterol is deposited in the corneal stroma because patients with FED cannot esterify free cholesterol contained in high-density lipoprotein (HDL) particles. Transparency is a fundamental optical property of the cornea, and the accumulation of free cholesterol can decrease corneal transparency and impair visual function. If the visual impairment becomes severe, penetrating keratoplasty may be considered as a treatment option.[5] Thus far, the quantification of decreased visual function in FED has not been reported.

Here we report a case involving a 36-year-old woman with FED whose clinical features and visual function were investigated using anterior segment optical coherence tomography (OCT) and quantitative measurements of contrast sensitivity and straylight.

2. Case report

A 36-year-old Japanese woman with bilateral corneal opacities and a suspected lipid metabolism disorder was referred to the Department of Ophthalmology. Analysis of fasting serum demonstrated an extremely low HDL cholesterol level (1.0 mg/dl), a low low-density lipoprotein cholesterol level (16.0 mg/dl), and a high free cholesterol to esterified cholesterol ratio (74.0 mg/dl to 1.0 mg/dl). LCAT activity was attenuated (99 U; standard value, 235–550 U) when determined using the Sekisui Medical Anasolv LCAT kit (SEKISUI MEDICAL CO., Tokyo, Japan); however, renal function was preserved (no proteinuria). On the basis of these findings, the patient was diagnosed with FED.

The patient reported several vision-related symptoms, including hazy vision, photophobia and visual impairment in darkness. Because of severe photophobia, she often experienced difficulty in opening her eyes. She had pale blue eyes in her early teens. Her maternal great-grandfather also had bilateral corneal opacifications, and there was no relevant history in her paternal family. She reported no other relevant ophthalmological history and was not taking any medications. Her best-corrected visual acuity measured using a standard visual acuity test with Landolt rings was 20/12.5 in the right eye and 20/20 in the left eye. Slit-lamp examination revealed a diffuse, cloudy opacity involving

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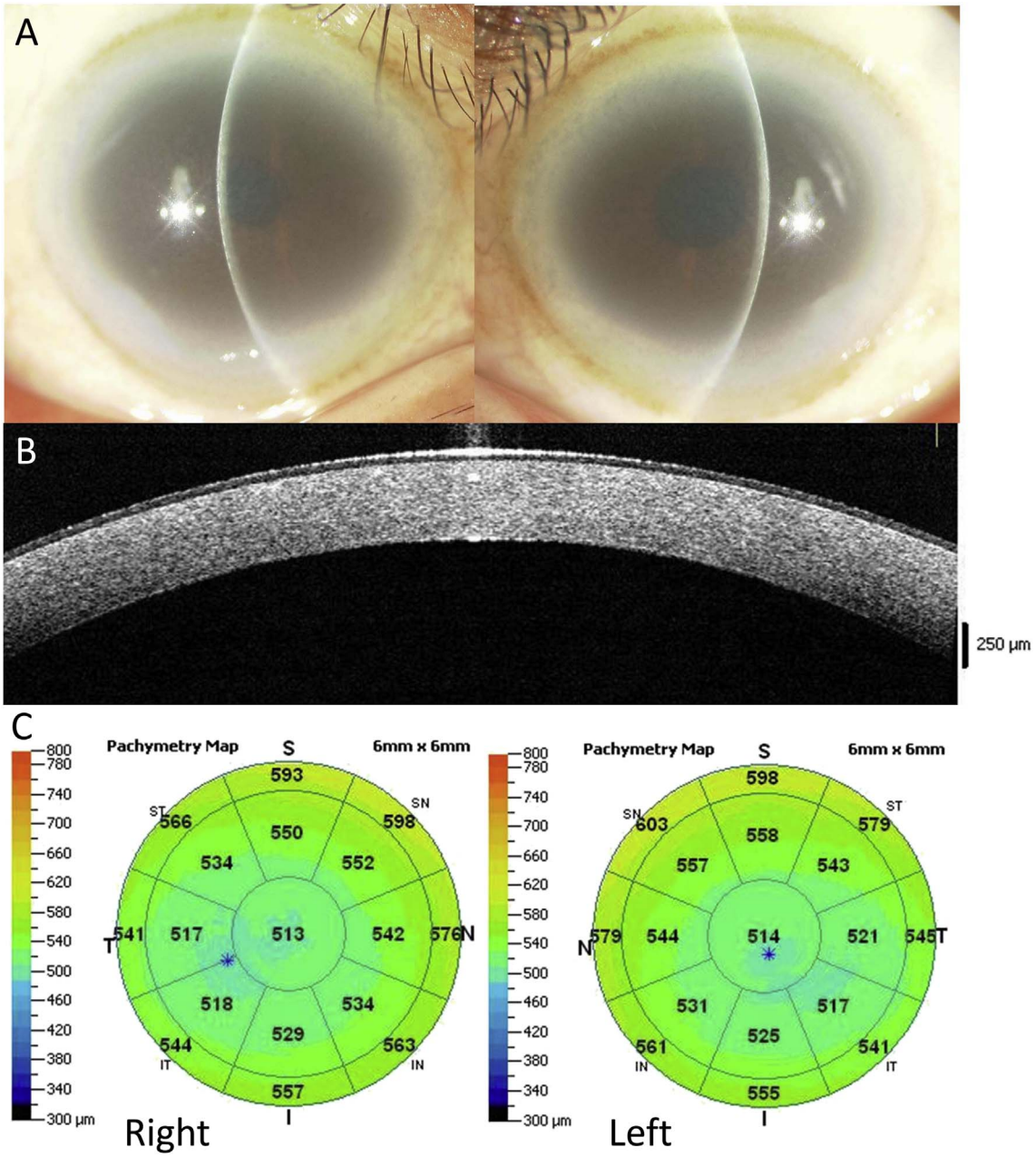

Fig. 1. Findings of ophthalmological examinations for a 36-year-old Japanese woman with fish-eye disease.
(A) Slit-lamp examination shows bilateral, diffuse corneal clouding.
(B) Fourier-domain optical coherence tomography (OCT) image of the right eye shows a homogeneously hyper-reflective corneal stroma.
(C) Corneal thickness mapping on a Fourier-domain OCT image shows a normal total corneal thickness (513 µm; right eye, 514 µm; left eye).

the entire cornea in both eyes. The peripheral cornea showed the maximum involvement, with no distinct lucid interval (Fig. 1A). No corneal vascularization was observed, and there were no other abnormal findings in the anterior or posterior segment of both eyes.

2.1. Visual function assessment

Contrast sensitivity and straylight were measured to quantify visual function. Contrast sensitivity was measured using a standard test chart (CSV-1000E chart; Vector Vision Co., Greenville, OH), which presents vertical sine wave gratings at four spatial frequencies. Each spatial frequency includes eight different contrast levels. In our patient, contrast sensitivity was decreased relative to the normal range in both eyes (Fig. 2). Straylight was measured using a straylight meter (Oculus GmbH, Wetzlar, Germany). Straylight measurements can quantify light scattering that results in a veil of straylight over the retinal image, which can lead to hazy vision or increased glare hindrance. The amount of straylight was expressed as the logarithm of the straylight parameters (log[s]); greater values indicate more straylight and more glare sensitivity. Straylight values for both eyes (2.09 log[s] for right, 2.18 log[s] for left) were remarkably increased by ten times when compared with those reported for normal eyes of age-matched individuals (approximately 0.9 log[s]).[6]

2.2. OCT

Fourier-domain OCT (RTVue-100; Optovue, Inc., Fremont, CA) revealed that the entire corneal stroma in both eyes was homogenously hyper-reflective (Fig. 1B). Thickness mapping for the total cornea demonstrated a central corneal thickness of 513 and 514 µm in the right