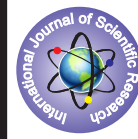


## Anterior segment optical coherence tomography in a patient with lecithin-cholesterol acyltransferase deficiency

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**KEYWORDS:** Lecithin-cholesterol acyltransferase deficiency, Corneal opacity, Anterior segment optical coherence tomography

### ABSTRACT

We present a 70-year-old woman with lecithin-cholesterol acyltransferase (LCAT) deficiency and discuss the potential advantages of using anterior segment optical coherence tomography (AS-OCT). Her eyes had been grayish blue since a young age. She was diagnosed as LCAT deficiency at the age of 50. Slit-lamp examination revealed diffuse bilateral opacity of the corneal stroma. AS-OCT revealed a thinned corneal epithelium and increased homogeneous hyperreflectivity of the entire stroma. We consider that it is important to note anterior segment images using AS-OCT is useful method in LCAT deficiency with corneal opacity.

### INTRODUCTION

Lecithin cholesterol acyltransferase (LCAT) deficiency is a rare autosomal recessive disorder characterized by progressive bilateral corneal clouding and dyslipoproteinemia.<sup>1-3</sup> Corneal opacity is observed in young people and progresses slowly toward the center from the periphery of the cornea.

Anterior segment optical coherence tomography (AS-OCT), recently applied to the diagnostics and preoperative evaluation of the anterior segment diseases including congenital corneal opacities, Wilson disease, and corneal dystrophies.<sup>4-6</sup> To our knowledge, there was only one report describing AS-OCT findings of LCAT deficiency.<sup>2</sup>

Here, we present AS-OCT findings of the LCAT deficiency and discuss the potential advantages of using AS-OCT.

### CASE PRESENTATION

A 70-year-old woman presented with blurred vision in both eyes. Her eyes had been grayish since a young age. She was diagnosed as LCAT deficiency at the age of 50. Her best corrected visual acuity was 0.7 in the right eye and 1.0 in the left eye. Slit-lamp examination revealed diffuse bilateral opacity of the corneal stroma extending to the limbus (Figure 1). The corneal stroma showed diffuse drop-shaped opacity. Nuclear opacity was detected in both lenses.

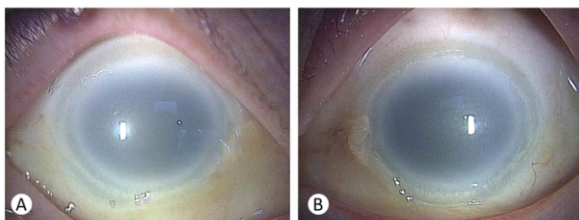


Fig. 1 Gross appearance of the right (A) and left (B) eyes.

Note diffuse bilateral opacity of the corneal stroma extending to the limbus.

Anterior segment optical coherence tomography (OCT) revealed a thinned corneal epithelium (Figure 2; inset arrows) and increased homogeneous hyperreflectivity of the entire stroma (Figure 2). The central corneal thickness was 568μm in the right eye and 569μm in the left eye.

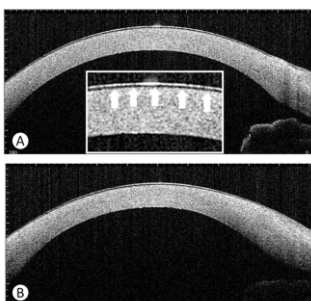


Fig. 2 Anterior segment optical coherence tomography images of the right (A) and left (B) eyes.

Note a homogeneous hyperreflectivity throughout the entire corneal stroma.

### DISCUSSION

To our knowledge, there was only one report describing AS-OCT findings of LCAT deficiency.<sup>2</sup> According to their report, AS-OCT revealed a thinned epithelium, homogeneous hyperreflective stroma, and focal disruptions of Bowman's layer. Similarly, a thinned epithelium and homogeneous hyperreflective stroma were detected in this present case.

Histopathological studies of corneal changes in patients with LCAT deficiency have shown stromal extracellular vacuoles with lipid infiltration.<sup>1-3</sup> Tateno et al.<sup>3</sup> described that the vacuoles were prominently distributed in the anterior corneal stroma. In addition, the corneal epithelium, Bowman's layer, Descemet's membrane and corneal endothelium appeared normal in thickness and structure. In this present case, homogeneous hyperreflectivity seen in the stroma was not detected in a thinned corneal epithelium.

The high-resolution ability of AS-OCT imaging modality allows resolution of structures that is not possible with conventional methods such as slit-lamp biomicroscopy. Therefore, the AS-OCT images may become a helpful adjunct to clinical biomicroscopic evaluation and provide an optical biopsy image.

### CONCLUSIONS

Although our findings were based on single case, we consider that it is important to note anterior segment images using AS-OCT is useful method in LCAT deficiency with corneal opacity. Accumulation of reports may be needed.

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