Scholars Journal of Medical Case Reports **ISSN 2347-6559 (Online)**
Sch J Med Case Rep 2017; 5(4):235-238 **ISSN 2347-9507 (Print)**
©Scholars Academic and Scientific Publishers (SAS Publishers)
(An International Publisher for Academic and Scientific Resources) DOI: 10.36347/sjmcr.2017.v05i04.003

Advantages of Infrared Fundus Photography Evaluation of the Retinal Complications in Lecithin-Cholesterol Acyltransferase Deficiency

Shinji Makino
Department of Ophthalmology, Jichi Medical University, Shimotsuke, Tochigi 329-0498, Japan

***Corresponding author**
Shinji Makino
Email: makichan@jichi.ac.jp

Abstract: We investigate the utility of infrared fundus examination in a case of lecithin cholesterol acyltransferase (LCAT) deficiency. A 57-year-old woman presented with blurred vision in both eyes. 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. Because of the hazy view, fundoscopy was not possible. However, normal fundus appearance was clearly defined as examined by infrared fundus photography. We consider that it is important to note ophthalmic images using infrared fundus photography is useful method for detecting retinal complications in LCAT deficiency with corneal opacity.
Keywords: Lecithin-cholesterol acyltransferase deficiency, Corneal opacity, Anterior segment optical coherence tomography, Infrared fundus photography

INTRODUCTION

Fish-eye disease is a rare autosomal recessive disorder characterized by progressive bilateral corneal clouding and dyslipoproteinemia [1-6]. Patients with fish-eye disease have low lecithin cholesterol acyltransferase (LCAT) activity and fail to esterify free cholesterol in high-density lipoproteins (HDL) so that the cholesterol is deposited in the corneal stroma [1-6]. Corneal opacity is observed in young people and progresses slowly toward the center from the periphery of the cornea. Generally, the ocular fundus is hard to examine because of corneal opacity. Here, we investigate the utility of infrared fundus examination in a case of LCAT deficiency.

CASE REPORT

A 57-year-old woman presented with blurred vision in both eyes. Her eyes had been grayish blue since a young age. She was diagnosed as LCAT deficiency at the age of 50. She had consanguineous marriages. At the first examination, her best corrected visual acuity was 0.9 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 lens was slightly opacified with a cataract. The corneal stroma showed diffuse bilateral drop-shaped opacity (Figure 2).

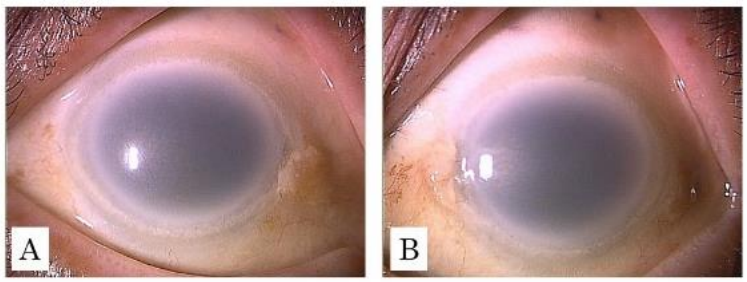

Fig-1: Gross appearance of the right (A) and left (B) eyes

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

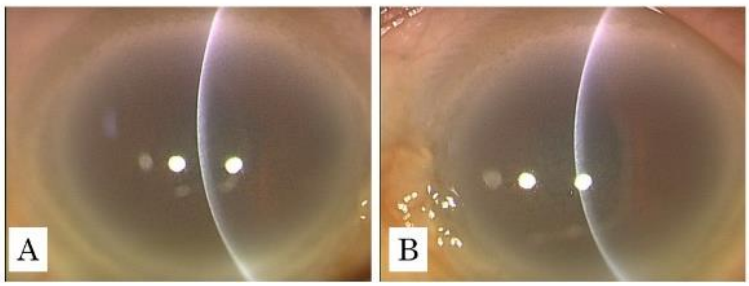

Fig-2: Slit-lamp photographs of the right (A) and left (B) eyes

Note diffuse bilateral drop-shaped opacity involving the entire cornea.

Because of the hazy view, fundoscopy was not possible. However, normal fundus appearance was clearly defined as examined by infrared fundus photography (Figure 3).

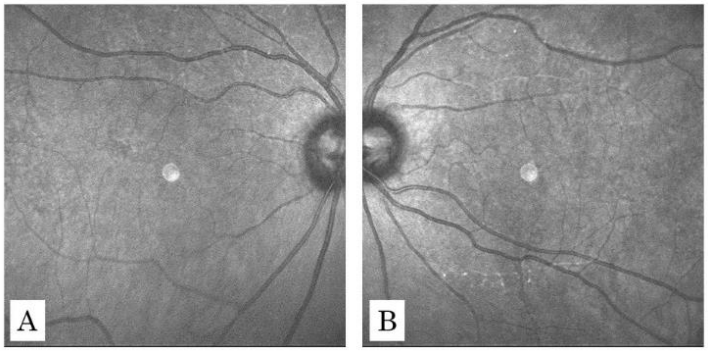

Fig-3: Infrared fundus photographs of the right (A) and left (B) eyes

The hyperreflective point at the center of the image is an optical artifact.

The following medical examination demonstrated a low plasma, total cholesterol (76mg/dL), HDL cholesterol (6mg/dL), and cholesterol ester (8mg/dL). The enzyme LCAT activity decreased to 45.4 U (normal range; 53.3～95.5). The genotype analysis detected a point mutation (Gly_{179}→Arg) in exon 5 of the *LCAT* gene. Some erythrocytes developed a target-cell configuration.

At the age of 70, her BCVA was 0.7 in the right eye and 1.0 in the left eye. Bilateral diffuse corneal opacity was not progressed (Figure 4). Nuclear opacity was gradually progressed in both lenses. Although fundoscopy was not possible, fundus examination showed no remarkable changes using infrared fundus photograph (Figure 5).

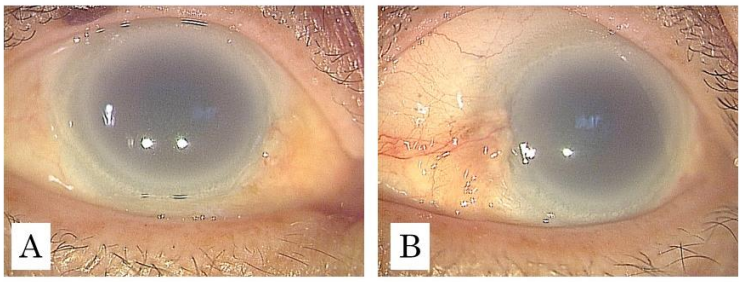

Fig-4: Gross appearance of the right (A) and left (B) eyes

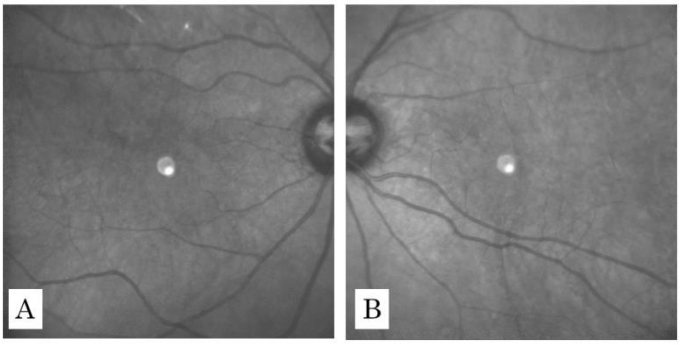

Fig-5: Infrared fundus photographs of the right (A) and left (B) eyes

Anterior segment optical coherence tomography (OCT) revealed a thinned corneal epithelium and increased homogeneous hyperreflectivity of the entire stroma (Figure 6). The central corneal thickness was 568µm in the right eye and 569µm in the left eye.

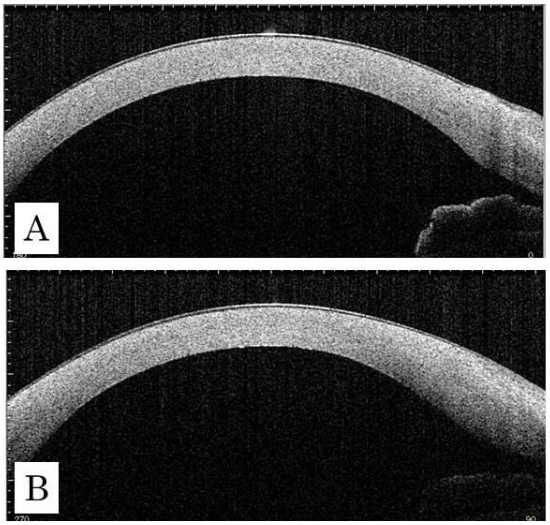

Fig-6: Anterior segment optical coherence tomography images of the right (A) and left (B) eyes

Note a homogeneous hyperreflectivity throughout the entire corneal stroma.

The initial ophthalmological manifestations of this case were previously described in Japanese [6].

DISCUSSION

Classic clinical findings of LCAT deficiency include markedly reduced plasma HDL, proteinuria, anemia, clouded plasma, triglyceridemia, and a typical bilateral corneal opacification [1-6].

Familial LCAT deficiency is a rare autosomal recessive disease, with 80 cases having been reported worldwide [1]. The LCAT gene (16q22) mutations cause familial LCAT deficiency and fish-eye disease [1, 7-11]. Mutations in the gene coding for LCAT cause a rare condition known as LCAT deficiency [2]. This key enzyme is responsible for esterifying free plasma cholesterol and may facilitate its uptake from peripheral tissues into HDL particles, possibly playing a major role in reverse cholesterol transport [7-11]. To date, multiple mutations in the human LCAT gene have been reported [2, 5]. In this present case, the genotype analysis detected a point mutation (Gly_{179}→Arg) in exon 5 of the *LCAT* gene.

Patients with LCAT deficiency display a loss of LCAT activity, HDL deficiency and systemic complications such as hemolytic anemia, proteinuria and renal dysfunction [7, 8]. The ocular abnormalities including optic disc hemorrhage, papilledema, retinal hemorrhage, and angioid streaks were reported [9]. In this present case, fundoscopy was not possible because of the hazy view. However, normal fundus appearance was clearly defined as examined by infrared fundus photography. These technologies can be complementary and may help in objectively monitoring disease progression and planning treatment.

CONCLUSIONS

Although our findings were based on single case, we consider that it is important to note ophthalmic images using infrared fundus photography is useful

method for detecting retinal complications in LCAT deficiency with corneal opacity. Accumulation of reports may be needed.

REFERENCES

1. Tateno H, Taomoto M, Jo N, Takahashi K, Uemura Y. Clinical and histopathological features of a suspected case of fish-eye disease. Jpn J Ophthalmol. 2012; 56(5): 453-457.
2. Palmiero PM, Sbeity Z, Liebmann J, Ritch R. In vivo imaging of the cornea in a patient with lecithin-cholesterol acyltransferase deficiency. Cornea. 2009; 28(9): 1061-1064.
3. Viestenz A, Schlötzer-Schrehardt U, Hofmann-Rummelt C, Seitz B, Küchle M. Histopathology of corneal changes in lecithin-cholesterol acyltransferase deficiency. Cornea. 2002; 21(8): 834-837.
4. Hesterberg RC Jr, Tredici TJ. Corneal opacification and lecithin-cholesterol acyltransferase (LCAT) deficiency: a case report. Ann Ophthalmol. 1984; 16(7): 616-621.
5. Tetsumoto K, Nakahori Y, Oku N, Kazusa R, Yaso S, Shinoda S, Maeda E. A pedigree of corneal opacity in LCAT deficiency presenting a point mutation (Arg_{99}→Cys) in exon 6 of the LCAT gene. Atarashii Ganka 1999; 16(8): 1127-1133.
6. Ishizaki K, Takezawa M, Makino S, Obata H, Ibaraki N. Corneal opacity in a case of LCAT（lecithin-cholesterol acyltransferase）deficiency. Rinshouganka 2011; 65(1): 12-15.
7. Ossoli A, Pavanello C, Calabresi L. High-density lipoprotein, lecithin: cholesterol acyltransferase, and atherosclerosis. Endocrinol Metab (Seoul). 2016; 31(2): 223-229.
8. Ossoli A, Simonelli S, Vitali C, Franceschini G, Calabresi L. Role of LCAT in atherosclerosis. J Atheroscler Thromb. 2016; 23(2): 119-127.
9. Barchiesi BJ, Eckel RH, Ellis PP. The cornea and disorders of lipid metabolism. Surv Ophthalmol. 1991; 36(1): 1-22.
10. Glukhova A, Hinkovska-Galcheva V, Kelly R, Abe A, Shayman JA, Tesmer JJ. Structure and function of lysosomal phospholipase A2 and lecithin:cholesterol acyltransferase. Nat Commun. 2015; 6: 6250.
11. Saeedi R, Li M, Frohlich J. A review on lecithin:cholesterol acyltransferase deficiency. Clin Biochem. 2015; 48(7-8): 472-475.