

Case report

A 43 year old male

BMI kg/m², 28.1

Lipid profile	Total cholesterol mg/dL	254
	Triglycerides mg/dL	394
	LDL-c mg/dL	169
	HDL-c mg/dL	6
	Apo A-I mg/dL	38
	Apo A-II mg/dL	12
	Apo B mg/dL	58

Renal function

eGFR 16.5

Proteinuria, albumin creatinine ratio 113.7 mg/mmol

Case report

Bilateral corneal clouding



Haemoglobin 8.4 g/dL

Carotid IMT

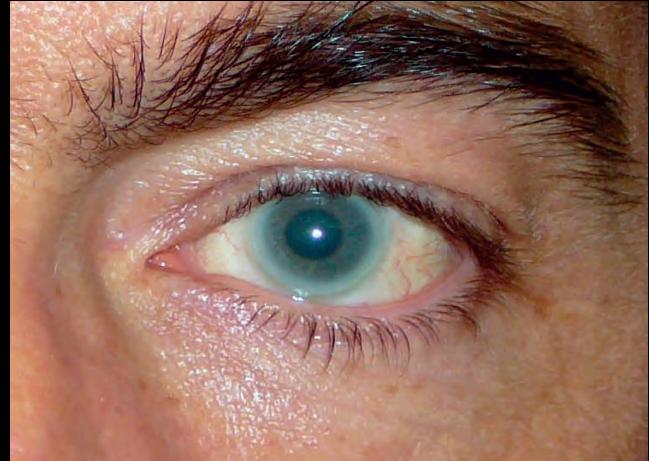
CC IMT, 0.52mm

Average IMT, 0.53 mm

Max IMT, 0.63mm

Case report

Bilateral corneal clouding



Haemoglobin 8.4 g/dL (12-17)

Therapies

furosemide

amlodipine

folic acid

vitamin D

erythropoietin

Case report

Lipid/lipoprotein profile

Total cholestreol mg/dL	254
Unesterified cholesterol mg/dL	254
Unesterified/total cholesterol (%)	100
Triglycerides mg/dL	394
Phospholipids mg/dL	442
LDL-c mg/dL	169
HDL-c mg/dL	6

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Lipid/lipoprotein profile

Total cholestreol mg/dL	254
Unesterified cholesterol mg/dL	254
Unesterified/total cholesterol (%)	100
Triglycerides mg/dL	394
Phospholipids mg/dL	442
LDL-c mg/dL	169
HDL-c mg/dL	6
CER nmol/mL/h	0
LCAT activity nmol/mL/h	0
LCAT mass µg/mL	3

LCAT gene sequencing

Two heterozygous mutations of the *LCAT* gene were identified:

c.321C>A (p.Tyr107* exon 3)

c.1034C>T (p.Thr345Met exon 6)

Genetic LCAT deficiency – treatments under development

CER-001	Synthetic HDL	Improvement in renal function	Phase 2
rhLCAT	Recombinant protein	ERT	Phase 2