A PATIENT WITH LCAT DEFICIENCY –REPORT OF A CASE

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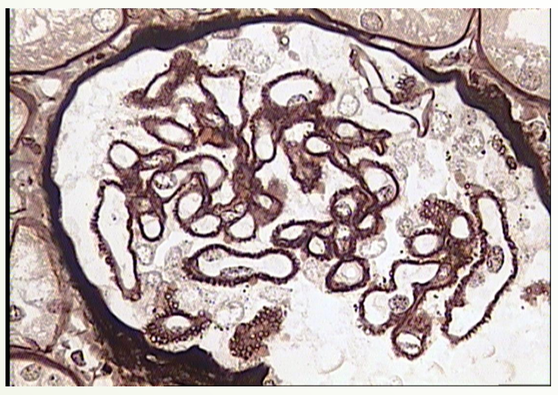
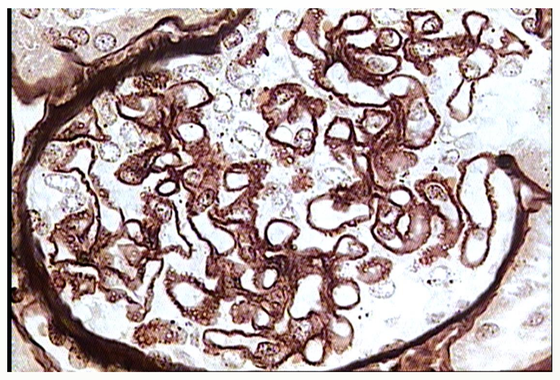
Background: Familial lecithin-cholesterol acyltransferase deficiency is a rare autosomal recessive disorder. It is characterized by defect esterification of plasma cholesterol. The disease was first described by Gjone and Norum in Norway.

Case report: A 30-year old man with nephrotic syndrome, corneal opacities, hepatosplenomegaly, anemia, low HDL levels and arterial hypertension was admitted to our department.
At admission he had elevated creatinine serum level (233 umol/l), 24h proteinuria was 12 g/dU, urine sediment was unremarkable, hemoglobin level was 90 g/l. Cholesterol and triglyceride levels were elevated (8.3 mmol/l, 16.58 mmol/l respectively) with low HDL-C levels (0.42 mmol/l). He had high blood pressure (200/100 mmHg). Abdominal ultrasound showed enlarged liver and spleen, kidneys appeared normal. Renovasular hypertension was ruled out, we found no endocrine causes of hypertension.
The patient was previously diagnosed with membranous nephropathy, he was treated unsuccessfully with immunosuppressive treatment (steroids, cyclosporine).
Renal function gradually decreased to stage IV CKD in 10-year period.
Re-evaluation of histopathological findings of kidney biopsy revealed massive deposition of lipid material in the glomerular basal membrane and in the mesangial region. The changes in glomeruli were similar to changes seen in liver cirrhosis.





Images 1 and 2. Patient's kidney histology – Jones silver stain. Courtesy of Prof. Coric, Department of Pathology, UHC Zagreb

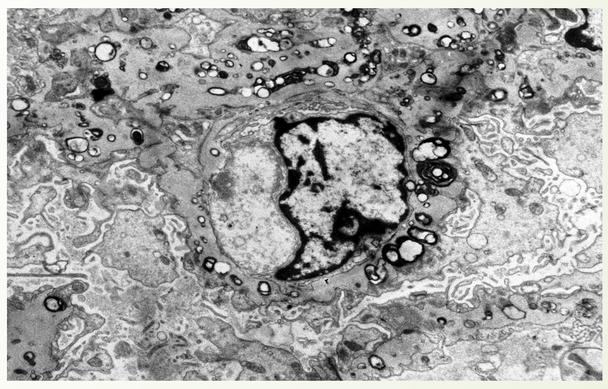


Image 3. Patient's kidney histology – electron microscopy.
Courtesy of Prof. Coric, Department of Pathology, UHC Zagreb

The disease was confirmed by plasma enzyme analyses; no functional LCAT activity was detected. Furthermore, cholesterol esterification rate was nearly zero. Genetic testing results show two novel mutations in the LCAT gene (c.496G>A.p (Ala166Thr) in exon 4 and c.1138T>C p.(Cys380Arg) in exson 6. The LCAT gene is located on the long arm of chromosome 16.

It is the first case report of LCAT deficiency in Croatia.

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