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Familial Lecithin: Cholesterol Acyltransferase (LCAT) Deficiency as a Cause of Chronic Kidney Disease ? A Case report

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Abstract: Background and Aims Genetic causes of chronic kidney disease are becoming more recognized . Familial lecithin: cholesterol acyltransferase (LCAT) deficiency (FLD) is a rare genetic disorder caused by loss of function mutations in LCAT gene. Patients present with abnormal lipid profile characterized with markedly reduced HDL-C, corneal opacification, anemia, and renal disease, which eventually progresses to kidney failure. Several studies reported genetic variants in LCAT gene that are associated with FLD , and others in certain apolipoprotein genes that act as risk factors for the disease. Incomplete form of FLD, caused by certain mutation, leads to fish-eye disease characterized by progressive corneal opacification. FLD was described in studies from Europe, Latin and North America, Australia and Japan. In Jordan, genetic and clinical studies in FLD patients are absent. Method We present a 36-year-old female who presented with nephrotic-range proteinuria, high serum creatinine, hypertension and corneal opacity. Further examination showed a severely reduced HDL level , an increased triglycerides level, anemia and mild thrombocytopenia. Patient's laboratory values at presentation are shown in Table 1. Her blood film showed normocytic and normochromic red blood cells, anisopoikilocytosis and target cells. Light and electron microscopy examination of the kidney biopsy revealed intramembranous, subendothelial and mesangial lipid deposition and vacuolization. In addition, the patient was found to have a family history of corneal opacity and chronic kidney disease. Therefore, FLD was suspected. Whole exome sequencing was employed to identify FLD variants. Results The patient was found to be homozygous at 154+5delG in LCAT gene, heterozygous at 388T>C in apolipoprotein E gene (APOE) and homozygous at 1114G>C in WW domain-containing oxidoreductase gene (WWOX). The patient's chronic kidney disease was managed supportively with low salt diet, moderate protein intake, statins, ang