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Can medullary nephrocalcinosis be a diagnostic clue to hereditary nephropathy with COQ8B mutation?

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Objectives: Nephropathy related to primary coenzyme Q 10 (CoQ_{10}) deficiency is a rare inherited disease. *COQ8B* (also known as *ADCK4*)-associated nephropathy has been reported in a Korean cohort with six patients accompanied by medullary nephrocalcinosis.

Methods: In here, we report a novel pediatric case with *COQ8B* mutation with medullary nephrocalcinosis and present the results from a systematic review on *COQ8B*-associated nephropathy. Electronic databases were searched using related terms (~Feb 2nd, 2020).

Results: A 4-year-old girl presented with isolated proteinuria, normal serum lab, and medullary nephrocalcinosis. We performed a direct Sanger sequencing for *COQ8B,* which found her with compound heterozygous mutations (c.737G>A, exon 9; c.1507C>T, exon 15). A kidney biopsy showed focal segmental glomerulosclerosis and abnormally increased mitochondrial accumulation was found in the podocytes cytoplasm. After 8 weeks of CoQ_{10} supplement and 4 weeks of cyclosporine treatment, her urine protein to creatinine ratio decreased to 0.5 mg/mg.
From 127 articles searched, there were 11 eligible studies with 48 patients with *COQ8B*-nephropathy. Male to female ratio was 1:2 and median age at diagnosis was 14.6 years. Major histology was FSGS (32/48, 67%) and mitochondrial aggregation in the podocytes were found in 7 (14%) patients. Patients had chronic kidney disease in 44% and progressed to end-stage renal disease in 25%. There were six transplantation cases with none recurred. Medullary nephrocalcinosis was reported only in 7 Korean patients. Of the 14 patients supplemented with CoQ_{10}, 7 reported improved outcomes. Calcineurin inhibitor was tried in 7 patients which induced partial remission in 4 cases.

Conclusions: *COQ8B*-associated FSGS is a rare hereditary nephropathy which can benefit from early diagnosis and CoQ_{10} supplement. So far, patients with *COQ8B* mutation reported in South Korea had medullary nephrocalcinosis. Although the disease is not immune-mediated, calcineurin inhibitor appears efficacious. Early suspicion based on this phenotype may improve the prognosis.