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**Nephrocalcinosis, tubular proteinuria and progressive kidney failure in patient with KIF11 mutation**

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**Case Study :** A one-day-old girl was transferred to our neonatal intensive care unit (NICU) with small for gestational age (SGA), microcephaly, and swelling of both ankles and feet. She had no abnormal findings other than oligohydramnios during regular antenatal care and delivery. In the diagnostic work-up for screening of multiple anomalies performed at our hospital, lymphatic malformation of the ankle and atrial septal defect (ASD) were confirmed, and renal glycosuria and medullary spongy kidney (MSK) were suspected. There were no abnormal findings in the chromosome study and brain MRI. During regular follow-up in an outpatient clinic, a significant amount of low molecular weight (LMW) proteinuria was confirmed along with renal glycosuria starting around 3-4 months of age. Developmental delay and facial dysmorphism were also suspected as the patient grew older, so a genetic study was conducted on these clinical manifestations in 2011 when she was 4 years old, but no genetic disorder was confirmed at that time. Afterwards, a severe fatty liver, multiple hepatic nodules and severe obesity were observed. Kidney failure began to progress at the age of nine, accompanied by hypercalciuria, and findings that were thought to be neonatal MSK were determined to be medullary nephrocalcinosis. Test related to inherited metabolic disease, including cystinosis, were also performed, but there were no abnormal findings. Additionally, type 2 diabetes was diagnosed at the age of nine, and retinal degeneration and atrophy were confirmed. Therefore, in 2020, when she was 13 years old, a genetic study was conducted again and the KIF 11 gene mutation was confirmed. At age 14, kidney biopsy revealed an increase of mesangial matrix, wall thickening of the arteriole, loss of focal foot process in epithelial cell, vacuolization in the tubules and increased mitochondria with focal structural abnormalities in the podocytes and proximal tubules.