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A PKD1 and SLC36A2 mutation in pediatric patient with polycystic kidney and nephrocalcinosis

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Case Study: Backgrounds: *PKD1* mutation is well known causative gene in autosomal dominant polycystic kidney disease (ADPKD). Solute carrier 36 member 2 (*SLC36A2*) encode pH-dependent proton coupled amino acid transporter. *SLC36A2* mutation shows autosomal and digenic recessive inheritance. Clinically *SLC36A2* mutation is associated with iminoglycinuria and hyperglycinuria which can be presented asymptomatically or with other symptoms: hypertension, glycosuria, nephrolithiasis, mental retardation and etc.
Case: A 3-year-old girl came to the hospital due to incidentally found multiple renal cyst with multiple bright echogenic foci suggesting medullary calcinosis in ultrasonography (US). Her mother was diagnosed as polycystic kidney disease (PKD) at her twenties, renal insufficiency aggravated after delivery of patient and intermittent hemodialysis was initiated at her early thirties. No other family member had PKD. The patient did not showed hematuria nor proteinuria and there was no renal insufficiency. The laboratory results were as follow: WBC 7010/uL, hemoglobin 13.4g/dL, platelet 275,000/uL, albumin 4.7g/dL, BUN 12.4mg/dL, creatinine 0.42mg/dL, uric acid 4.3mg/dL, calcium 7.9mg/dL, phosphorus 5.2 mg/dL and spot urine protein/creatinine ratio 0.22. As the patient and her mother were detected to have multiple renal cysts at a young age, in combination of medullary calcinosis in patient, we suspected possibility of unusual mutation or co-existence of second mutation. For this reason, we did whole genome sequencing and the patient was identified to have the heterozygous mutation c.G4609T in exon 15 of *PKD1* (p.E1537X) and heterozygous mutation c.91_94A on *SLC36A2* at chromosome 5, nonframeshift substitution. As result, she was diagnosed as having ADPKD with possibility of having iminoglycinuria or hyperglycinuria. She was tested for aminoaciduria and prolinuria was detected. Therefore, we are planning to test amino acid and organic acid level in serum and urine sample.
Conclusions: We report a first case of pediatric patient with polycystic kidney disease caused by *PKD1* with co- exist *SLC36A2* mutation.