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# A Rare Cause of Fanconi Syndrome With Recurrent AKI: Renal Tubular Dysgenesis-A Case Report

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**Case Study :** Considered for many years incompatible with survival, renal tubular dysgenesis has been lately described in a few clinical case reports in patients surviving past neonatal period. Defined as a faulty proximal tubulogenesis due to an abnormal expression of the renin-angiotensin-aldosterone system, renal tubular dysgenesis is usually clinically manifested in the neonatal period with severe oliguria and Potter sequence, frequently with patient's exitus. We hereby present the case of a 10-months old male patient, born from a non-consaguineous family, with no priors of nephropathy. From the patient's medical history we find out that he was born prematurely at 34 weeks of gestation due to severe oligohydramnios. He presented 3 episodes of AKI over the next 9 months. At 10 months of age, on arrival in our clinic, the patient had microcephaly (cranial perimeter His creatinine value was 1.29 mg/dl (eGFR of 22 ml/min/1.73 $m^2$), also presenting with severe anemia, metabolic acidosis and hyperkalemia, along with urinary features of proximal tubular dysfunction (tubular proteinuria, hypercalciuria, glycosuria). The renal ultrasound showed normal size kidneys, without corticomedullary differentiation, with some small hyperechoic lesions throughout both renal parenchyma. A whole-exome sequence (WES) was performed, which revealed a heterozygous compound mutation in ACE gene (c.3521del, p.(Gly1174Alafs*12) and c.1487G>A, p.(Arg496Gln). The patient was started on fludrocortisone (0.1 mg/day) and darbepoetin alfa (10 mcg/2 weks), with a smaller dose of alkali supplementation and high fluid intake. His eGFR increased and maintains at around 42 ml/min/1.73 $m^{2.}$ We conclude that renal tubular dysgenesis should be considered in patients with proximal tubular dysfunction even in the absence of the Potter's sequence. An NGS gene-panel or even a WES approach can establish the diagnosis in atypical cases.