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A clinical approach to Tubulopathies in children and young adults

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Tubulopathies can present with a variety of non-specific clinical features which can be diagnostically challenging. In this review, we build from this common anatomical and physiological understanding to present a tangible appreciation of tubulopathies as they are likely to be clinically encountered among affected children and young adults. The clinical presentation of tubular dysfunction in children and young adults is as equally varied as it is non-specific. Prominent features include polyuria, polydipsia, irritability, growth failure, nephrocalcinosis and blood pressure anomalies. It is important to elicit a history of polyuria and or polydipsia as these reflect a concentrating defect. This can be compounded by an osmotic diuresis secondary to increased solute delivery to the distal tubule. The body compensates with the release of ADH and aldosterone (if these mechanisms remain intact) and increased thirst. The loss of water and solutes, however, is constant, and despite polydipsia, children are often chronically or intermittently dehydrated resulting in irritability. Urine should be analysed with urine dipstick (for glucosuria), protein:creatinine ratio and calcium:creatinine ratio. Additional investigations for suspected proximal tubulopathy include beta-2 microglobulin (or equivalent tubular protein) and urine amino acid profile or metabolic screen. If initial testing is suggestive of a tubulopathy, paired urine and serum samples should be obtained to enable the calculation of fractional excretion of sodium (FeNA) and magnesium (FeMg) in addition to the trans tubular potassium gradient (TTKG) and tubular maximum phosphate reabsorption per glomerular filtration rate (TmP/GFR) . These fractional excretions can provide an insight into the electrolyte handling of the kidneys and evidence of the underlying pathology.