

Amelogenesis imperfecta and nephrocalcinosis syndrome

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An 18-year-old boy presented with pain in left flank of 2 days duration. He had no history of oliguria, dysuria, pyuria, hematuria, graveluria or swelling of feet or face. Examination revealed yellow colored teeth. The labial surfaces of lower teeth showed irregular horizontal enamel defects [Figure 1]. Rest of the general and systemic examination was unremarkable. Ultrasound abdomen revealed bilateral nephrocalcinosis. It was confirmed on a computed tomography [Figure 2]. The other investigations showed serum creatinine to be 0.9 mg/dl, blood urea 24 mg/dl, sodium 138 meEq/l, potassium 4.5 mEq/l, calcium 9.2 mg/dl, inorganic phosphate 3.2 mg/dl, alkaline phosphatase 180 IU/l, parathormone 69 pg/ml, vitamin D 25 ng/ml, bicarbonate 24 mmol/l and urine pH: 5.5. His parents' marriage was a consanguineous one. His elder brother and father also had yellow colored teeth. He was diagnosed amelogenesis imperfecta (AI) of hypoplastic type with nephrocalcinosis syndrome.

AI represents a group of developmental conditions, genomic in origin, which affect the structure and clinical appearance of enamel of all or nearly all the teeth in a more or less equal manner. The prevalence varies from 1:700 to 1:14,000. It may show autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns.^[1] The association of AI and nephrocalcinosis was reported in 10 patients till now.^[2] The autosomal recessive disorder, in which there are FAM20A gene



Figure 1: Yellow teeth with horizontal enamel defects

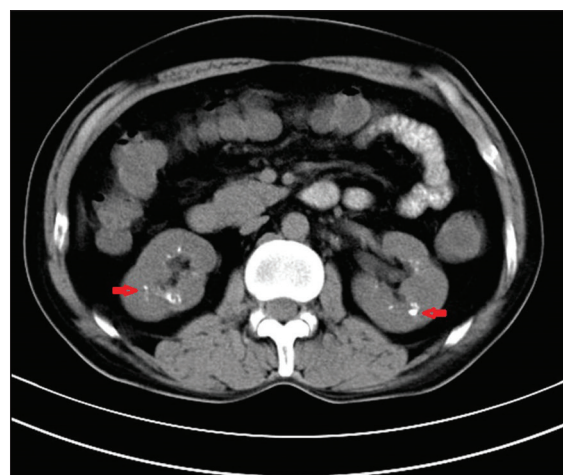


Figure 2: Computed tomography scan abdomen: Nephrocalcinosis

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mutations, causes nephrocalcinosis and AI (enamel renal syndrome).^[3] AI is also reported to be associated with Bartter's syndrome^[4] and distal renal tubular acidosis.^[5]

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