DISCUSSION

AI is a serious of problems resulting in poor oral health-related quality of life. AI affects the structure and clinical appearance of the enamel of all or nearly all the teeth in a more or less equal manner [8]. The morbidity and severity of AI with nephrocalcinosis is more and most of the cases were detected at the age of adulthood [9]. In our case AI was detected and then ultrasound of kidney was done. Mac-Gibbon et al reported in these cases renal function was stable from birth till adulthood but the progressive renal failure was observed [10]. Lubinsky et al and Hall et al described AI and nephrocalcinosis in siblings with normal serum calcium and phosphate level. The AI and nephrocalcinosis syndrome has also reported in consanguineous and non-consanguineous families. The common characteristics are the presence of thin or absent enamel, presence of intrapulpal calcifications, delayed tooth eruption bilateral, nephrocalcinosis and normal plasma calcium [11,12]. To avoid morbidity with nephrocalcinosis associated with AI, kidney ultrasound should be performed in all AI patients. Witkop et al & Peter et al reported in these cases AI patients' histological appearance of teeth with hamartomatous pericoronal proliferation [13,14]. Most of the research showed the molecular basis of AI. X-linked genetic defects were identified in autosomal forms. Only mutations in the amelogenin (AMEL X) gene have associated with the various X-linked AI forms [15]. Hotton et al reported that tissue non-specific phosphatase alkaline (TNSALP), the calcium sensing receptor (CaSR) and calbindin 28 kDa are proteins involved in the calcium and phosphate metabolism. These TNSALP, CaSR, calbindin 28kDa are present both in the kidney and teeth, defective mutations in above gene and protein result in the hypophosphatasia characterised by defective bone and teeth mineralization [16].

In summary, further research is necessary to clarify genetic defect in this syndrome, with two uncommon conditions factors, AI and nephrocalcinosis. The early diagnosis provided by the oral symptoms leads to a better renal prognosis.

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