Genotypic and Phenotypic Characterization of Turkish Patients with Vitamin D Dependent Rickets Type IA

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Background
- Vitamin D Dependent Rickets Type IA (VDDR-IA)
- Most common type of VDDR
- Caused by mutations in CYP27B1
- Aim: Analyze the genotypic and phenotypic features of our VDDR-IA patients.

Methods
- The patients with a clinical diagnosis of VDDR-IA were enrolled and analyzed for CYP27B1 gene mutations.

Results
- 12 (5 males) patients / 9 unrelated families
- Mean age of diagnosis: 3.48±4.00 (median: 1.1; min-max: 0.75-11.6) years
- 1 patient had toxic level of 25[OH]D (250ng/ml)
- Six patients had a history of high dose vitamin D intake (300000-1500000 IU)
- One had toxic level of 25(OH)D (250ng/ml)
- All patients with p.F443Pfs*24 mutation which leads to a truncated protein

Table 1: Clinical, laboratory, and genetic findings of patients at admission

Table 2: Comparison of clinical and laboratory findings of two common mutation

Conclusions
- We emphasized the importance of correct diagnosis in VDDR-IA for the proper management, and avoiding poor clinical outcomes.

*Patient had been diagnosed previously with hypophosphatemic rickets at the age of 17 months at another clinic therefore were already on calcitriol and phosphorus replacement therapy. No consanguinity, parents originates from close villages.