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Clinical and Genetic Features of Our Patients with Hypophosphatemic Rickets

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While *PHEX* gene mutation is the most common form of inherited rickets, limited data exist regarding genetic etiology of hypophosphatemic rickets in Turkey. The aim of this study was to investigate the type of genetic defect in 16 index children and their families (12 unrelated, 1 related).

Following clinical and laboratory assessment, *PHEX* analysis was made initially unless a mutation in another gene was suspected. If negative, *FGF23*, *SLC34A3*, *SLC34A1*, *CYP27B1*, *VDR*, *DMP1*, and *ENPP1* genes were analyzed sequentially.

Following the investigation of index cases and their families, we identified 21 patients (16 children, 5 adults) diagnosed with hypophosphatemic rickets from 13 families. Nineteen of them (91%) had findings related with rickets and 12 (56%) had short stature. Calcium levels were normal, phosphorus low, ALP markedly elevated, and parathormone normal (n=8, 38.1%) or mildly elevated (n=13, 61.9%) in all patients. We found 10 different *PHEX* mutations in 17 (80.9%) patients, one novel *SLC34A3* mutation in two siblings (9.5%), and no mutation in 2 patients (9.5%). Five *PHEX* mutations were *de novo*. Four novel *PHEX* mutations were: c.978_982dupCTACC (frameshift), c.1586+2T>G (splice site), c.436+1G>T (splice site), and c.1217G>T (p.C406F). Affected parents were all symptomatic but none were diagnosed previously.

The present study revealed that *PHEX* mutation seems to be the most prevalent mutation in Turkey as well. More attention should be paid to hypophosphatemia by the clinicians since some cases remain undiagnosed both during childhood and adulthood.

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The Relationship Between Gestational Diabetes Mellitus and Selenoprotein-P Plasma 1 (*SEPP1*) Gene Polymorphisms

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In this study, we aimed to investigate the relationship between *SEPP1* gene polymorphism and gestational diabetes mellitus (GDM).

The study included 40 patients with gestational diabetes mellitus (GDM group) and 40 healthy pregnant women (control group). Target single nucleotide polymorphisms (SNPs) were studied by using *rs4987017*, *rs13154178*, *rs146125471*, *rs28919926*, *rs16872762*, and TaqMan probes via ABI Prism StepOnePlus Real Time System (Applied Biosystems, Foster City, CA).

rs4987017 gene polymorphism was found to be AA homozygous in all subjects in GDM and control groups. There was no significant difference in *rs146125471*, *rs28919926*, and *rs16872762* polymorphisms between GDM and control groups (p=0.656, p=0.30, and p=0.627, respectively). However, a significant difference was detected in *rs13154178* polymorphism between the two groups (p<0.01). In the control group, 61.5% of subjects were AA homozygous, while there was no AA homozygous patient in the GDM group. When mutants and AA homozygous patients were compared, fasting blood glucose and blood glucose level on hour one of 50 g OGTT were found to be significantly higher in patients with polymorphism than those without (p<0.001 and p=0.01, respectively). When effects of *rs13154178* gene polymorphism on lipid levels were considered, it was found that LDL cholesterol, triglyceride, and total cholesterol levels were significantly lower in AA homozygous patients than in those carrying mutant gene (p=0.036, p=0.009, and p=0.006, respectively).

To the best of our knowledge, this is the first study investigating *SEPP1* gene polymorphism in GDM. Our study suggests that *rs13154178* gene polymorphism lead to predisposition to GDM in pregnant Turkish women.