5th International Conference on
HUMAN GENETICS AND GENETIC DISEASES
&
11th International Conference on
GENOMICS AND PHARMACOGENOMICS
September 21-22, 2018 | Philadelphia, USA

## Mutational screening in patients with familial hypophosphatemic rickets

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**Introduction:** Hypophosphatemic rickets, characterized by hypophosphatemia and defective bone mineralization, is predominantly inherited as an X-linked dominant condition caused by inactivating *PHEX* mutations. Autosomal dominant and recessive forms due to mutations in FGF23, DMP1, ENPPI, and SLC34A3 respectively are also documented.

**Aim:** To identify mutations in cases with familial hypophosphatemic rickets (FHR) and their family members.

**Patients and Methods:** The three FHR patients (probands) were born out of non-consanguineous marriages. In the family I, the patient and her mother were affected, in family II, the patient, her brother, and mother were affected while family III had two sisters, their father and paternal uncle affected. Clinical and family history was documented and 5ml blood drawn for DNA extraction. *PHEX* exons were screened by direct sequencing and result analyzed using in silico tools. Whole Exome Sequencing (WES) was done to find involvement of genes responsible for intra-familial variability seen in the affected members, and validation of mutations was done by Sanger sequencing.

**Results:** The patients had short stature, limb deformities, and low serum phosphate. *PHEX* screening revealed three mutations in three patients and their affected family members, of which one was novel missense (c.2048T>A) and two reported nonsense and missense (c.871C>T; c.1601C>T) mutations respectively. Two families presented with intra-familial variability in disease phenotype. The paternal uncle of family III had dental anomalies and mother of the family I had short stature. WES was done for the family I, and family III probands, father and paternal uncle. Results revealed a novel *PHEX* mutation G>C in exon 21 in the family I proband, which was absent in her mother but did not reveal any additional mutations, other than c.1601C>T in family III.

**Conclusion:** This study reports two novel *PHEX* mutations and also suggests that *PHEX* may be mainly responsible for FHR in India.

### Biography

Binata Marik is currently doing PhD in Human Genetics at All India Institute of Medical Sciences (AIIMS), New Delhi, India. She has done Masters in Biotechnology from AIIMS, New Delhi, India. She has an expertise in Pediatric Genetics and Cytogenetics. She is working on "Genetics of refractory rickets". She knows the laboratory techniques used for human and molecular genetics research such as DNA isolation, Polymerase Chain Reaction (PCR), Agarose gel electrophoresis, Polyacrylamide Gel electrophoresis (PAGE), Sanger sequencing and Karyotyping. She has knowledge in bioinformatics and knows how to analyze the massive data of exome sequencing and to predict the pathogenicity of the genetic variations.

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