FGF23-Mediated Hypophosphatemic Rickets: Phenotype, Genotype, and Comparison to Non-FGF23-Mediated Forms

Original Article

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ABSTRACT

OBJECTIVE

To compare the phenotypic, biochemical, and genotypic characteristics of hereditary FGF23-mediated hypophosphatemic rickets (FGF23-M-HR) and non-FGF23-mediated hypophosphatemic rickets (non-FGF23-M-HR).

METHODS

Clinical, biochemical, and radiological data of genetically proven FGF23-M-HR and non-FGF23-M-HR cases from a single center in western India were compared.

RESULTS

Thirteen probands (6 familial; 11 females) with FGF23-M-HR with median (IQR) age of symptom onset 2.0 (1.25, 26) years and age at diagnosis 10 (3, 30) years, were included. All, but one, presented with rickets and short stature. There were 12 (7 novel) unique PHEX mutations and one homozygous novel DMP1 mutation. FGF23-M-HR had significantly higher parathormone levels (81.9 vs. 25.1 pg/mL), lower 1,25 (OH) 2 D (54.9 vs. 103 ng/mL), and lower urinary calcium/ creatinine ratio (0.006 vs. 0.38). Parathormone > 65.3 pg/mL has 100% specificity for diagnosing FGF23-M-HR.

CONCLUSIONS

Parathormone, urinary calcium/creatinine ratio, and 1,25 (OH) 2 D levels can differentiate FGF23-M-HR from non-FGF23-M-HR.

Keywords: DMP-1 · Hypophosphatemic rickets · PHEX

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