

Disease

X-linked dominant hypophosphatemic rickets^{1, 2}

Gene

PHEX

Inheritance	XL
Current Treatments	burosumab, phosphate supplements, calcitriol
Evidence for Treatment	guideline ^{3, 4}
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum phosphate concentration, tubular resorption of phosphate corrected for glomerular filtration rate ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC009
Disease Frequency (per 100,000)	5 ⁵
Genes Included in Disease Frequency	PHEX
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Hypophosphatemic+Rickets&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK83985/>
- ² <https://www.ncbi.nlm.nih.gov/m/pubmed/29791829/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/31068690>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/35484227/>
- ⁵ <https://ghr.nlm.nih.gov/condition/hereditary-hypophosphatemic-rickets#diagnosis>