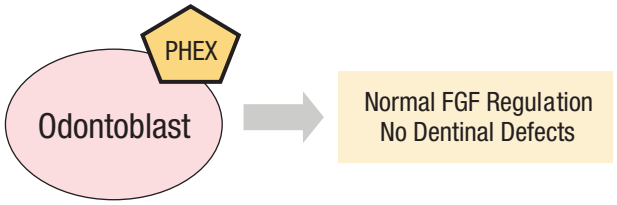
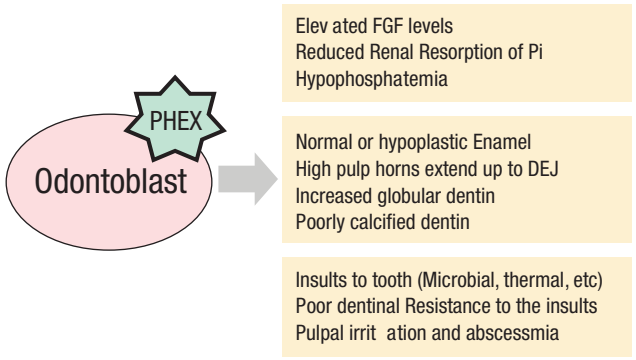
What causes Dental Manifestations?

Individual without XLH



Individual with XLH



Recommendations for Dental Management

- Early Diagnosis and Management
- Team Approach
- Communication between Health Care Providers
- Frequent Recall Visits
- Sealants on the Primary and Permanent Molars
- Fluoride Therapy
- Pulpectomy versus Extraction based on age and clinical diagnosis
- Space Maintainers
- Dental Implants
- Root Canal Therapy and Crowns should be considered

XLH NETWORK

The XLH Network, Inc.
911 Central Avenue, #161
Albany, NY 12206

www.xlhnetwork.org

Dental Manifestations of X-Linked Hypophosphatemia

Implications for Prevention & Treatment of XLH

www.xlhnetwork.org

What is XLH?

- X-linked hypophosphatemia (XLH) is an X-linked dominant disorder resulting in dental and skeletal abnormalities.
- XLH is the most frequent form of inherited rickets and osteomalacia.
- Loss-of-function mutations in the PHEX gene (expressed in odontoblasts, osteocytes and osteoblasts) result in elevated circulating levels of FGF-23 , a protein that acts on the kidneys and reduces tubular phosphate reabsorption.

Inheritance:

- X-linked dominant form

Prevalence:

- Approximately 1 in 20,000

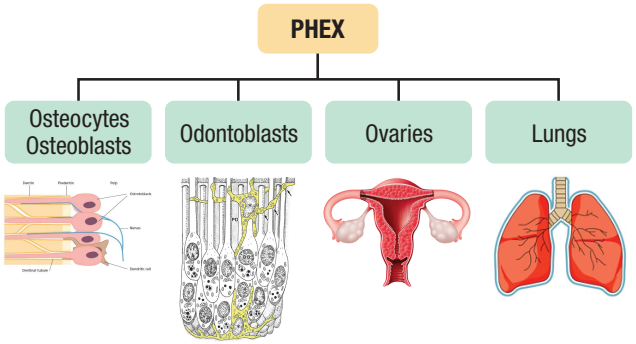
A similar phenotype can be seen in less common disorders such as:

- Autosomal Dominant Hypophosphatemia Rickets (FGF23 Mutations)
- Autosomal Recessive Hypophosphatemia Rickets (DMP1 Mutations)
- Sporadic cases with similar phenotype

Etiology and Pathogenesis

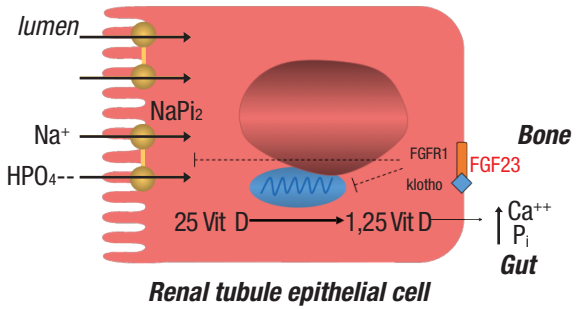
Mutations

PHEX gene located on Chromosome X and expressed in:



XLH is characterized by diminished proximal renal tubular phosphate transport due to elevated FGF23 leading to:

- Hypophosphatemia
- Phosphate wasting
- Impaired 1,25(OH)$_2$VitD synthesis
- Diminished Pi gut absorption



Diagnosis

XLH

Clinical	Radio graphic	Biochemical	
• Rickets-b ow or knock-knee deformity • Craniosynostosis • Short St ature • Dental Findings	• Frayed and widened gr owth pl ates • Bo wing of l ower extremities	Serum Calcium	Normal
		Serum Phosphorus	Low
		25-OHD	Normal
		1,25(OH)D	Low/Normal
		FGF23	High/Normal
		PTH	Normal/Slightly High (in children)
		Serum alkaline phosph atase	High/Normal

XLH - Dental Findings

Clinical	Radiographic	Histologic	
• Spontaneous abscess in absence of dental caries • Delayed eruption	• Reduced density of trabeculations • Loss of Lamina dura • Periapical radiolucency in absence of dental caries	Enamel	Normal or Hypoplastic
		Dentin	Large tubular clefts extend to pulp
			Wide predentin layer
			Increased globular dentin
			Partially mineralized dentin
		Pulp	Large pulp chamber
			High pulp horns extend up to DEJ