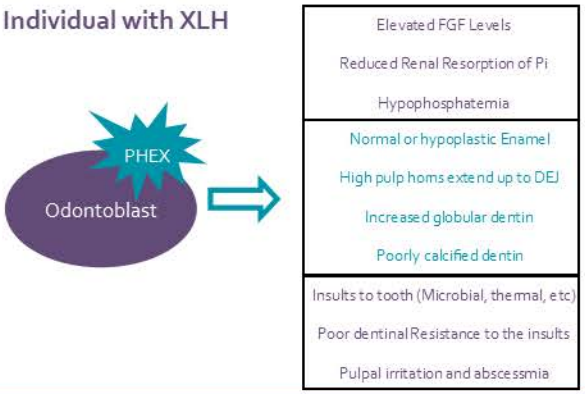
What causes Dental Manifestations?

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 vs. Extraction based on Age and Clinical Diagnosis
- Space Maintainers
- Dental Implants
- Root Canal Therapy and Crowns should be Considered

XLH NETWORK

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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 FGF23, 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:

- Osteocytes
- Osteoblasts
- Odontoblasts
- Ovaries
- Lungs

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

- Hypophosphatemia
- Phosphate wasting
- Impaired 1,25(OH)2VitD synthesis
- Diminished Pi gut absorption

Diagnosis

XLH

Clinical	Radiographic	Biochemical	
• Rickets-bow or knock-knee deformity • Craniosynostosis • Short Stature • Dental Findings	• Frayed and widened growth plates • Bowing of lower 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 phosphate	High/Normal

XLH – Dental Findings

Clinical	Radiographic	Histologic	
• Spontaneous abscess in absence of dental caries • Delayed eruption	• Reduced density of trabeculations • Loss of Laminadura • Periapical radiolucency in absence of dental carries	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