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Research Article

# UNRAVELLING THE DENTAL DEFECTS IN X-LINKED HYPOPHOSPHATEMIC RICKETS

Muhaseena[1*], Muhamood Moothedath[2], Moidin Shakil[3], Maji Jose[4]

[1]Assistant Professor, Department of Oral & Maxillofacial Pathology, Century International Institute of Dental Sciences and Research Centre, Kasaragod, Kerala, India
[2]Associate Professor & Head, Department of Public Health Dentistry, Century International Institute of Dental Sciences and Research Centre, Kasaragod, Kerala, India
[3]Lecturer,Department of Oral and Maxillofacial Surgery and Diagnostic Sciences, College of Dentistry, Alrass Qassim University, Kingdom of Saudi Arabia
[4]Professor and Head, Department of Oral & Maxillofacial Pathology, Yenepoya University, Mangalore, Karnataka, India

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*Corresponding Author: **Dr. Muhaseena,**
Assistant Professor, Department of Oral & Maxillofacial Pathology, Century International Institute of Dental Sciences and Research Centre, Kasaragod, Kerala, India, Mobile: +917204644232.

## ABSTRACT

**Introduction:** X linked hypophosphatemic rickets (XLHR) is a systemic disorder resulting from the mutation of PHEX gene and is associated with dental findings.
**Aim:** The present study aims at documenting the gross surface features, radiographic, and microscopic structural alterations of the exfoliated deciduous tooth obtained from XLHR patient.
**Materials & Methods:** Exfoliated deciduous teeth were obtained from XLHR affected patient. Gross surface features were observed using stereo-microscope and radiograph were taken. Ground sections and decalcified sections were prepared for microscopic examination.
**Results:** The teeth examined revealed numerous enamel depressions, reduced radio-density of enamel & dentin, enlarged pulp chamber and disturbed enamel rod morphology along with defective mineralization of dentin.
**Conclusion:** Dental practitioners should be aware of the manifestations of this disorder so that early intervention can be undertaken to prevent subsequent invasive dental procedures.
**Keywords**: Rickets, Hypophosphatemia, X Linked Hypophosphatemic Rickets (XLHR), Dental findings.

## INTRODUCTION

X-linked hypophosphatemia (XLH) or vitamin D-resistant hypophosphatemic rickets comprises of heritable disorders of renal phosphate regulation. Albright in 1939 first described it to be an X-linked disorder[1]and recently mutations in the phosphate regulating gene homologous to endopeptidases (PHEX) gene present on the X-chromosome which is located on Xp22.1was identified[2].
XLH manifests marked hypophosphatemia resulting from impaired reabsorption in the renal proximal tubule and low or normal serum calcium levels and subsequently characterized by growth retardation and osteomalacic bone disease[3]. Prominent clinical manifestations include growth failure, short stature, skeletal abnormalities, genu valgum, rachitic rosary, open fontanels, pathologic fractures, muscle weakness, and convulsions. Radiologic findings usually include fractures, generalized osteopenia and bow legs[4].
As this disease is characterized by the metabolic disturbance of calcium and phosphate, defective calcification of mineralized structures such as bones and teeth are common[5]. In general, both primary and permanent teeth will be affected in patients with X-linked hypophosphatemia. Various dental finding reported in earlier studies are: thin, hypo-calcified or hypoplastic enamel, grossly defective dentine, enlarged coronal pulp spaces[6]. In addition, other defects such as taurodontism, poorly defined lamina dura and a hypoplastic alveolar ridge also have been reported. Spontaneous dental abscess formation is often observed without dental caries or traumatic injury; attributed to large pulp chambers having high pulp horns, in association with enamel cracks and dentinal micro-cleavages allowing the ingress of micro-organism[7].

## MATERIALS AND METHODS

Exfoliated deciduous mandibular teeth were obtained from a 7 year old female patient affected with X-linked hypophosphatemic rickets. Gross surface defects were studied using stereo-microscope, radiographs were taken to study the morphology of pulp chamber, and to compare the radio-density of enamel and dentin. Ground sections and decalcified

sections were prepared for histologic examination to detect microscopic structural alterations. The findings were compared with normal teeth.

## RESULTS

On gross examination under stereo-microscope, a glassy appearance was noted indicating altered translucency of enamel compared to the normal tooth, along with some areas of enamel hypoplasia. Numerous cleavages and surface irregularities were also appreciated on the surface of the tooth affected with XLH rickets (Fig: 1).

Radiographic examination revealed reduced radio density of enamel & dentin and enlarged coronal pulp chambers with indistinct boundary compared to the normal tooth. Pulpal calcification was noted in the coronal chamber of one of the rickets affected tooth (Fig: 2).

Microscopic examination of both ground sections and decalcified sections showed various structural defects affecting enamel and dentin. Enamel was relatively thin with long cracks, disturbed enamel rod morphology and prominent Striae of Retzius when compared to the normal deciduous tooth. Dentin showed extensive interglobular dentin that could have resulted from the defective mineralization process with lack of fusion of the calcospherites (Fig: 3). A wide zone of pre-dentin and tubular defects was also observed in the XLH rickets affected tooth (Fig: 4).

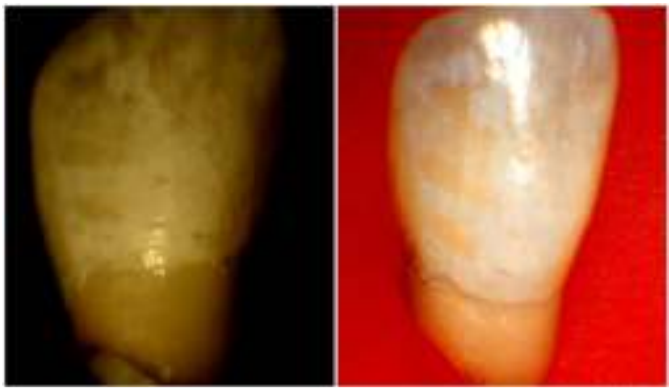

**Figure 1: Stereo-microscopic image of XLHR affected tooth showing dental translucency of incisal region giving a glassy appearance**

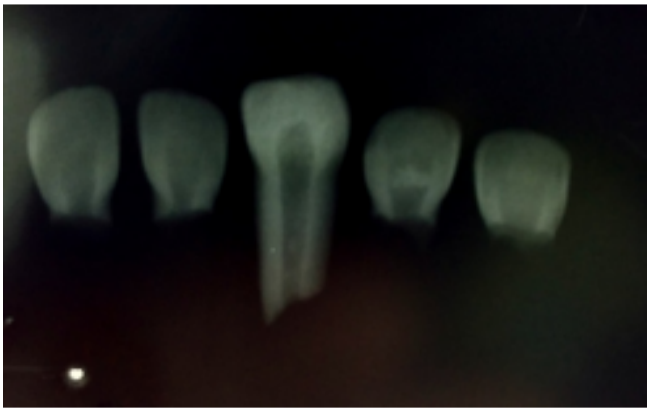

**Figure 2: Radiographic image showing reduced radio-density of enamel & dentin of rickets affected teeth compared to the normal unaffected tooth (middle tooth)**

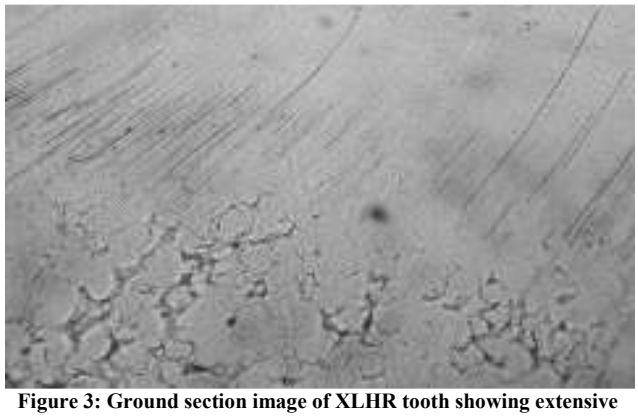

**Figure 3: Ground section image of XLHR tooth showing extensive interglobular dentin**

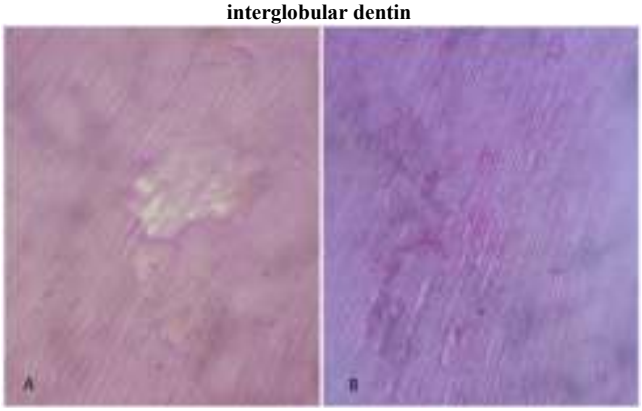

**Figure 4: Histopathologic picture of decalcified XLHR tooth showing wide interglobular dentin (A) and defective dentinal tubules (B)**

## DISCUSSION

X-linked hypophosphatemic rickets (XLHR) is considered to be due to a congenital impairment of phosphate transport and hypophosphatemia, resulting from reduced phosphate reabsorption in the brush border membrane on the luminal side of the proximal renal tubule and impaired phosphate absorption in the intestine[3]. Recently XLH rickets has been associated with mutations in the PHEX gene, which is predominantly expressed in osteoblast and odontoblasts[8].

Characteristic dental findings along with skeletal features that are common in patients affected by XLHR attributed to the metabolic disturbance of calcium and phosphate[5]. Most commonly reported dental abnormalities include thin enamel, presence of extensive interglobular dentin and large pulp chambers with pulp horns extending up to the DEJ (dentino-enamel junction). Odontoblastic function is reported to be normal in XLHR patients, but inadequate dental mineralization due to hypophosphatemia leads to a dysplastic and poorly mineralized dentin with areas of interglobular dentin[9]. Thus impaired dentin mineralization is a major abnormality of hypophosphatemic teeth.

In the present study, when the extracted tooth obtained from XLHR patient revealed various enamel defects as noted by various researchers; Godina et al, Pereira et al and Goodman JR et al[5,9,10]. Emin CT et al also reported that in these patients the dentition is highly susceptible to dental caries or attrition[3]. In addition on stereo-microscopic examination, we observed marked translucency of the incisal region which was also reported earlier by Pereira et al[9].

Similar to the reports of Pereira et al we also noted extensive interglobular dentin[9]. This structural defect has significant clinical implications as incompletely mineralized dentin may cause the entrapment of various micro-organisms which in turn may hinder the mechanical endodontic cleaning. The entrapment explains the existence of bacteria and failure of endodontic therapy.

Radiological examination showed enlarged coronal pulp chambers consistent with earlier reports[11]. Hillmann and Geurtsen stated that, the enlarged pulp chamber with extended horns, favor early exposure of pulp tissue resulting from any loss of the enamel and dentin layer and subsequent pulpitis[12]. In addition, we also noted small pulpal calcification in the coronal pulpal chamber of one of the XLHR affected tooth.

Pulpitis and spontaneous dental abscess formation is another dental complication often observed in patients with XLH, even without dental caries or traumatic injury. Enamel cracks and dentinal micro-cleavages in these teeth allow the ingress of micro-organism which easily reaches the pulp as pulp chambers is large having high pulp horns[7,10].

Generally this condition is diagnosed by the age of 8-10 months [13], by which the formation of all deciduous dentition would be completed and therefore the defects in the primary dentition usually cannot be prevented. However, once XLHR is diagnosed, supplementation of deficient minerals may be of benefit to reduce the dental defects of permanent dentition. Once the defects are formed, the only possible strategy in managing the dental defect is prevention of further complication. Therefore a good understanding of the dental manifestations of this condition would enable the clinicians to plan the treatment more effectively.

## CONCLUSION

Present study demonstrates various dental structural defects that may occur in XLHR affected patients through the stereomicroscopic, radiographic and microscopic examination. Due to these structural changes, these patients are more prone for the development of dental caries, pulpitis and spontaneous abscess formation. Clear understanding about these defects helps the dental professionals to plan and execute early preventive measures to prevent dental complications.

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