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Osteogenesis Imperfecta: A Radiological Case Study Of A 24-Year-Old Female

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Abstract
Background: *Osteogenesis Imperfecta (OI) is a heterogeneous group of inherited connective tissue disorders characterized by bone fragility, low bone mass, and skeletal deformities. The majority of cases are attributed to dominant mutations in the genes encoding type I procollagen, COL1A1 and COL1A2. Currently, nearly 19 subtypes have been described with variable phenotypic expression.*
Objective: *To present the radiological spectrum of OI in a 24-year-old adult female with atypical systemic manifestations including dental abnormalities and cardiac conduction defects.*
Methods: *Diagnostic imaging was performed using a Philips Brilliance 64-slice CT scanner and Carestream direct view Computed Radiography X-ray system. A comprehensive radiological evaluation of the axial and appendicular skeleton was conducted.*
Results: *Imaging revealed skeletal features consistent with OI, including osteopenia, gracile long bones, and characteristic craniofacial and dental anomalies. Additional findings included hypocalcemia-associated QTc prolongation on cardiac evaluation.*
Conclusion: *This case underscores the role of multimodality imaging in confirming OI in adulthood and highlights extra-skeletal associations that warrant multidisciplinary management.*
Keyword: *Osteogenesis imperfecta, Imaging, Radiography, COL1A1, COL1A2, Type I Collagen, CT, Dentinogenesis Imperfecta, QTc Prolongation*

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I. Introduction

Osteogenesis Imperfecta is a monogenic disorder of connective tissue primarily affecting type I collagen synthesis and structure. It manifests with a wide clinical spectrum ranging from perinatal lethality to mild forms presenting in adulthood. The classic triad includes bone fragility, blue sclera, and hearing loss. However, dental manifestations such as dentinogenesis imperfecta and systemic complications like cardiovascular involvement are increasingly recognized. The genetic basis in most cases involves mutations in COL1A1 and COL1A2, disrupting collagen triple helix formation and subsequent bone matrix mineralization.

II. Case Presentation And Materials And Methods

A 24-year-old female presented with chief complaints of short stature, recurrent dental caries with dental deformity, and fatigue. Clinical evaluation revealed features suggestive of a connective tissue disorder. Laboratory assessment noted hypocalcemia with electrocardiographic findings of QTc prolongation, primarily due to prolongation of the ST segment.

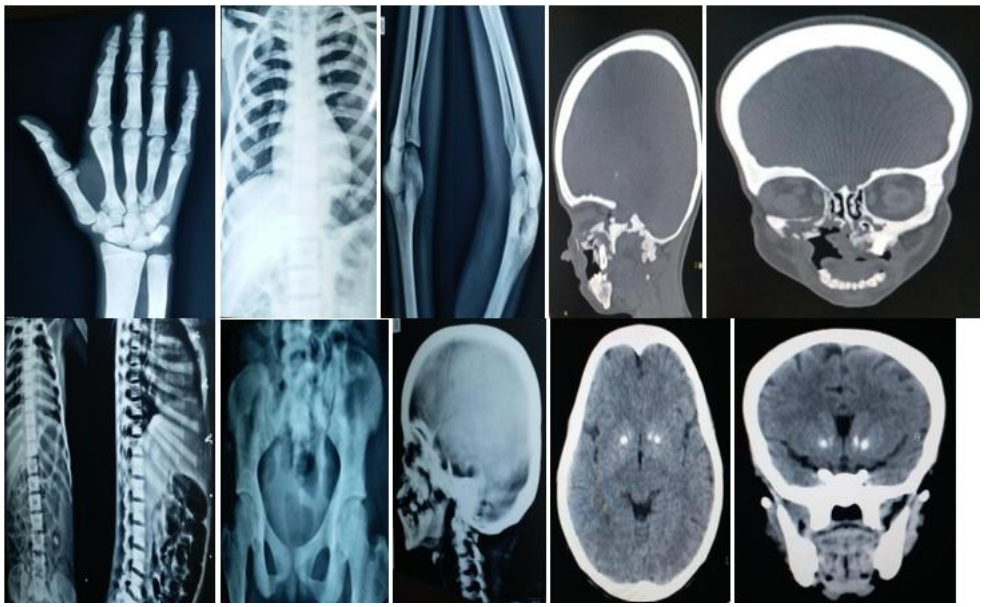
Radiological workup was performed to assess skeletal involvement. Imaging modalities included:
1. Computed Tomography: Philips Brilliance 64-slice scanner for high-resolution assessment of craniofacial and axial skeleton.
2. Digital Radiography: Carestream direct view CR system for evaluation of appendicular skeleton, including bilateral hands, ribs, spine, and pelvis.

III. Results

The radiological pictorial assay demonstrated findings consistent with Osteogenesis Imperfecta:
- Cranium: Thinned calvarium with Wormian bones suggestive of defective membranous ossification.
- Axial Skeleton: Osteopenic vertebral bodies with biconcave deformities and gracile ribs.
- Appendicular Skeleton: Long bones appeared slender with cortical thinning. Metaphyseal regions showed modeling defects.
- Dental: Radiographs revealed dental caries and structural dental deformity consistent with dentinogenesis imperfecta, a known extra-skeletal manifestation.

Cardiac evaluation correlated hypocalcemia with QTc prolongation. While cardiac involvement is not a primary feature of OI, electrolyte disturbances secondary to skeletal pathology and nutritional factors may contribute.

Below mentioned are the images describing the above features.



IV. Discussion

OI represents a diagnostic challenge in adults due to phenotypic variability and overlap with other metabolic bone diseases. Radiological assessment remains central to diagnosis and classification. The presence of osteopenia, bone deformities, and Wormian bones in this patient aligns with moderate to severe OI subtypes.

The association with dental caries and deformity reflects abnormal dentin formation, reported in types I, III, and IV. The observed QTc prolongation secondary to hypocalcemia emphasizes the need for systemic evaluation beyond skeletal features. Multidisciplinary follow-up including endocrinology, cardiology, and dentistry is recommended for comprehensive care.

Early radiological diagnosis aids in fracture prevention, surgical planning, and genetic counseling. Advances in CT and CR imaging allow detailed characterization of bone architecture, which is crucial for monitoring disease progression.

V. Conclusion

This case illustrates that Osteogenesis Imperfecta can present in adulthood with both skeletal and extra-skeletal manifestations. Comprehensive radiological evaluation using CT and digital radiography is essential for diagnosis and management. Recognition of systemic associations such as dental anomalies and cardiac conduction abnormalities can guide targeted therapeutic interventions.

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