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(CASE REPORT)

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Diagnostic challenges in distinguishing osteogenesis imperfecta from nutritional rickets in a 5-week-old preterm neonate with multiple fractures and rachitic rosary: A case report from Northern Tanzania

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Abstract

Background: Differentiating osteogenesis imperfecta from nutritional rickets in neonates is challenging in resource-limited settings because both conditions may present with bone fragility, fractures, and costochondral beading.

Case presentation: A 5-week-old male preterm neonate born at 33 weeks' gestation was admitted for prematurity-related care. On day 5 of life, he developed swelling and reduced movement of the limbs without a history of trauma. Examination revealed rachitic rosary and bilateral blue-grey sclera. Skeletal survey demonstrated fractures of both humeri and the right femur, with generalized osteopenia and thin cortices. Haemoglobin was 11.7 g/dL and serum calcium was borderline low at 2.13 mmol/L. Serum phosphate, alkaline phosphatase, and 25-hydroxyvitamin D could not be measured because reagents were unavailable. Although nutritional rickets was initially considered, the blue sclera, fracture pattern, and radiological osteopenia favoured osteogenesis imperfecta with possible concurrent rickets. There was no clinical or historical evidence of accidental or non-accidental trauma.

Conclusion: Multiple fractures in a neonate with rachitic rosary and normal or borderline-low calcium should prompt consideration of osteogenesis imperfecta, particularly when blue sclera and generalized osteopenia are present. In settings where advanced biochemical and genetic tests are unavailable, careful clinical examination, serum calcium measurement, and skeletal survey can support early provisional diagnosis.

Keywords: Osteogenesis Imperfecta; Nutritional Rickets; Preterm Neonate; Multiple Fractures; Blue Sclera; Rachitic Rosary; Resource-Limited Setting; Tanzania

1. Introduction

Osteogenesis imperfecta is a heterogeneous group of inherited connective tissue disorders mainly caused by pathogenic variants in *COL1A1* or *COL1A2*, which encode type I collagen [1]. Its estimated incidence is approximately 1 in 10,000–20,000 live births. Severe forms may present during the neonatal period with multiple fractures, bone deformities, osteopenia, and high mortality [2]. Nutritional rickets remains an important differential diagnosis in preterm infants and in settings where maternal vitamin D deficiency, limited supplementation, and restricted sunlight exposure are common [3]. Both conditions may present with chest wall beading and bone fragility, creating a diagnostic dilemma. While rickets is treated primarily with vitamin D and calcium supplementation, osteogenesis imperfecta requires multidisciplinary care, fracture prevention, orthopaedic management, and, where available, bisphosphonate therapy [2,4]. In resource-limited settings such as Northern Tanzania, the lack of biochemical assays and genetic testing can further complicate diagnosis [5,6]. We report a 5-week-old preterm neonate at Mawenzi Regional Referral Hospital who

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presented with multiple fractures and rachitic rosary, highlighting practical clinical approaches for differentiating osteogenesis imperfecta from nutritional rickets when diagnostic resources are limited.

2. Case Presentation

2.1. Patient information

A 5-week-old male neonate was born at 33 weeks' gestation by spontaneous vertex delivery with a birth weight of 1.3 kg. He cried immediately after delivery, with Apgar scores of 6 at 1 minute and 8 at 5 minutes. He was admitted to the Neonatal Intensive Care Unit at Mawenzi Regional Referral Hospital for prematurity, respiratory distress, and feeding difficulties. He improved with oxygen support and nasogastric feeding. There was no family history of bone disease, recurrent fractures, hearing loss, or dental abnormalities. The parents were non-consanguineous and denied any history of trauma to the infant.

2.2. Clinical findings

On day 5 of life, the mother noticed swelling, warmth, and reduced movement of both upper limbs and the right lower limb. There was no history of a fall, birth trauma, or rough handling. On examination, the head circumference was 30.5 cm, temperature was 36.6°C, respiratory rate was 57 breaths per minute, oxygen saturation was 95% on room air, and random blood glucose was 3.6 mmol/L. The infant was irritable on gentle handling. Chest examination revealed prominent beading at the costochondral junctions, consistent with rachitic rosary (Figure 1). There was no Harrison's sulcus. Ophthalmic examination showed bilateral blue-grey sclera. No facial dysmorphism, proptosis, or limb deformity was noted. There were no bruises, burns, or other cutaneous injuries suggestive of non-accidental injury.

2.3. Diagnostic assessment

Laboratory investigations showed haemoglobin of 11.7 g/dL and serum calcium of 2.13 mmol/L (reference range: 2.2–2.7 mmol/L). Serum phosphate, alkaline phosphatase, and 25-hydroxyvitamin D were not measured because reagents were unavailable at the Mawenzi Regional Referral Hospital laboratory at the time of admission.

Radiological assessment by skeletal survey demonstrated acute fractures of the mid-shaft right humerus, mid-shaft left humerus, and proximal third of the right femur (Figures 2 and 3). Generalized osteopenia and thinning of the cortical bone were also noted. Skull X-ray was not performed because the infant remained clinically stable without neurological signs. No evidence of neglect, abuse, or inconsistent history was found, and the parents' account was consistent with minimal handling.

2.4. Management and outcome

The fractures were managed conservatively with half casts and gentle handling for four weeks (Figure 4). Intravenous calcium was not administered because the serum calcium level was only borderline low and there were no clinical features of tetany. During admission to the neonatal intensive care unit, the infant received ampicillin 130 mg twice daily and gentamicin 4 mg once daily for five days because of the risk of infection. He was also started on folic acid 2.5 mg once daily, ferrous fumarate providing 8 mg of elemental iron once daily, and vitamin D 800 IU once daily. At five weeks of life, he developed progressive difficulty in breathing and was transferred back to the neonatal intensive care unit. Chest X-ray showed cardiomegaly. Before echocardiography could be performed, the infant developed cardiac arrest, and resuscitation was unsuccessful.

3. Discussion

3.1. Diagnostic dilemma: OI vs Rickets

Rachitic rosary and borderline hypocalcaemia in a preterm neonate may suggest nutritional rickets, which is an important consideration in Tanzania [3]. However, isolated rickets rarely causes multiple long-bone fractures involving both humeri and the femur in a 5-week-old non-ambulatory infant without marked biochemical derangement [6]. Bilateral blue sclera is a classic clinical feature of osteogenesis imperfecta and is not typical of nutritional rickets [1]. In addition, the fracture pattern, generalized osteopenia, and thin cortices on X-ray were more consistent with osteogenesis imperfecta than isolated rickets [2]. The borderline-low serum calcium level of 2.13 mmol/L may reflect concurrent nutritional deficiency related to prematurity. This overlap complicates clinical diagnosis when phosphate, alkaline phosphatase, and vitamin D testing are unavailable.

3.2. Challenges in resource-limited settings

In Northern Tanzania, diagnostic facilities such as dual-energy X-ray absorptiometry and genetic testing for *COL1A1*/*COL1A2* variants are generally unavailable at regional referral hospitals [7]. The inability to measure phosphate, alkaline phosphatase, and vitamin D in this case reflects a common diagnostic gap in many low- and middle-income settings [6]. Clinicians must therefore rely on clinical features, serum calcium, and skeletal survey to reach a provisional diagnosis. In this context, blue sclera, multiple long-bone fractures, and generalized osteopenia are useful discriminators in favour of osteogenesis imperfecta.

3.3. Clinical implications

This case highlights three key lessons for paediatricians working in resource-limited settings:

- Rachitic rosary does not exclude osteogenesis imperfecta. Rib beading may reflect healing rib fractures or chest wall changes rather than isolated metaphyseal changes of rickets.
- Early serum calcium measurement is useful because it is relatively affordable and widely available. Normal or borderline-low calcium in the presence of multiple fractures and blue sclera should prompt consideration of osteogenesis imperfecta.
- Preterm neonates may be at risk for both nutritional rickets and osteogenesis imperfecta. Vitamin D supplementation remains appropriate when deficiency is possible, but the fracture pattern and radiological features should guide further evaluation.

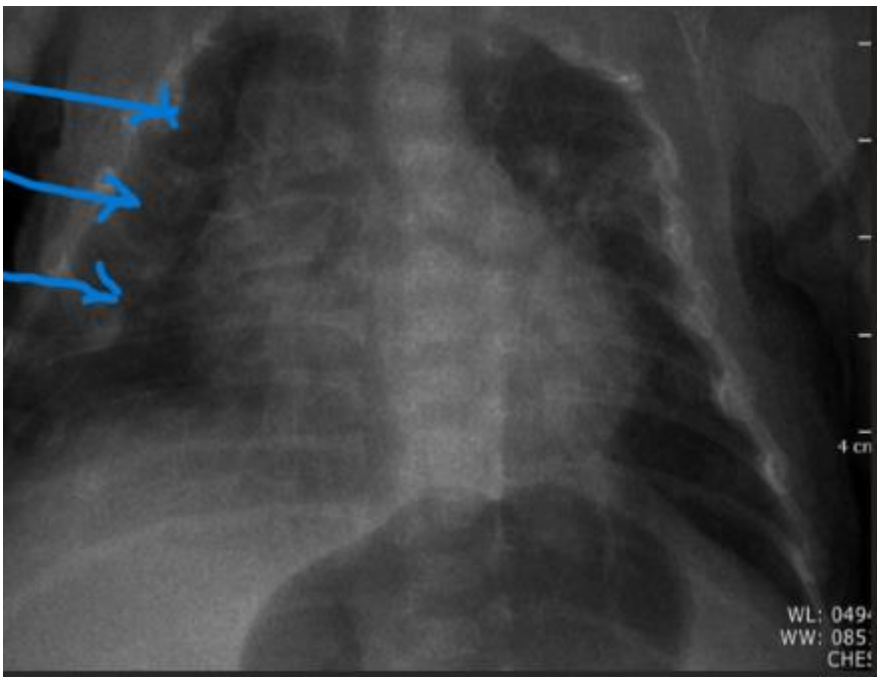

Figure 1 Clinical photograph showing rachitic rosary at the costochondral junctions.

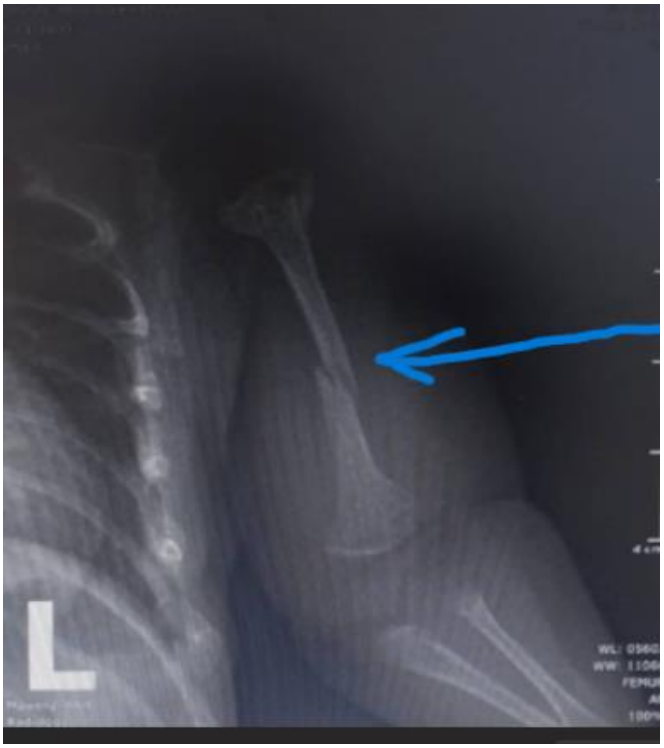

Figure 2 Anteroposterior X-ray of both humeri showing mid-shaft fractures and osteopenia.

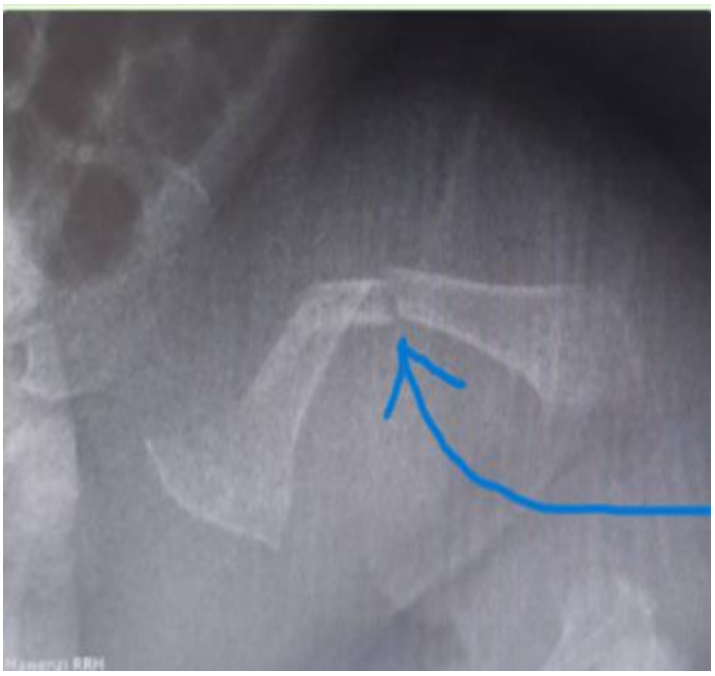

Figure 3 Anteroposterior X-ray of the right femur showing a proximal third fracture.

Figure 4 Clinical photograph showing limb immobilization with plaster of Paris after conservative fracture management.

4. Conclusion

We report a case of suspected osteogenesis imperfecta with possible concurrent nutritional rickets in a 5-week-old preterm neonate who presented with multiple fractures and rachitic rosary. Bilateral blue sclera, multiple long-bone fractures, generalized osteopenia, and borderline-low serum calcium were key findings that supported osteogenesis imperfecta rather than isolated nutritional rickets. Increased awareness and use of basic diagnostic tools can improve early recognition of osteogenesis imperfecta in resource-limited settings.

Limitations

Definitive diagnosis of osteogenesis imperfecta requires genetic testing for *COL1A1/COL1A2* variants, which was unavailable. Serum phosphate, alkaline phosphatase, and 25-hydroxyvitamin D levels could not be assessed because reagents were unavailable. Skull X-ray was not performed because the infant had no neurological signs before the terminal respiratory deterioration. The diagnosis was therefore based on clinical features, serum calcium, and skeletal survey findings.

Compliance with ethical standards

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Disclosure of conflict of interest

The authors declare no competing interests.

Statement of informed consent

Written informed consent was obtained from the mother for publication of this case report and accompanying images. Ethical approval was obtained from the Medical Officer in Charge. Patient identifiers were removed to ensure confidentiality.

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Authors' contributions

- JPK contributed to concept development, clinical care, first draft preparation, and manuscript review.
- EHD contributed to clinical care and drafting of the case report.
- FPK contributed to manuscript review and scientific editing. All authors read and approved the final manuscript.

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