Osteogenesis Imperfecta Congenita in a nigerian baby

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Introduction

Osteogenesis imperfecta, also called ‘Brittle Bone Syndrome’ is a rare disease of qualitative bone formation characterised by abnormal or deficient collagen, which has been demonstrated in bone, skin, sclerae, and dentine [4,10]. The mild form of osteogenesis imperfecta (tarda levis), Type I, is the most common and is dominantly inherited while the severe forms of the disease, osteogenesis imperfecta congenita Types II – IV, are sporadic, severe and present with fractures at birth [10]. Osteogenesis imperfecta congenita has an incidence of 1 in 50,000 live births [10]. The importance of osteogenesis imperfecta lies in the fact that it causes perinatal deaths or disastrous lifelong crippling [3]. We present a case of osteogenesis imperfecta congenita in a baby born with multiple united and fresh fractures at birth. This is the first reported case in our State, in Nigeria to the best of our knowledge.

Case Report

O.O. was a full term female new born who was referred to our centre on 27th February 2003 with multiple long bone fractures secondary to difficult breech extraction. She was a booked patient at the referral centre and received full immunisation during pregnancy. The pregnancy was reportedly normal. Ultrasound scan done in pregnancy prior to fall was said to be normal.

The labour was traumatic due to breech presentation. The baby was extracted and she cried spontaneously after birth. She was referred for further management on account of bony crepitus and abnormal positioning of the limbs which were noticed at birth.

O.O. was the 3rd child and the other two siblings were alive and well. There was no history of tendency to fracture in her nuclear and extended family.

On examination she was an average sized neonate with normal facies, uniformly pink in room air, anicteric, acyanosed but she had bluish sclerae. Her extremities were warm. Her weight
on admission was 2.75kg. She was tachypnoeic with a respiratory rate of 72/min. There was good air entry in both lung fields with vesicular breath sounds. The heart rate at 140/min was regular with normal heart sounds being heard. No cardiac murmurs were heard. The abdomen and spine were grossly normal. All the limb bones were clinically fractured i.e. both radius and ulna, both femora, both tibia and fibula as well as both clavicles. The skull was soft and had a crackling sound. The fontanelles were large and continuous. Examination of the central nervous system revealed an irritable baby with absent Moro reflex. She had poor grasp though sucking was good. No other congenital abnormality was detected.

X-rays were taken on admission, at about 5hrs post-delivery and showed a poorly ossified skull vault with Wormian bones, (Figure 1). There were fresh and also healed fractures of the ribs, which appeared beaded (Figure 2).

**Figure 1:**

*Skull x-ray demonstrating defective ossification of the vault and wormian bones in the coronal sutures*
The lungs were fully expanded and the cardiac shadow appeared normal. The bones of the limbs were all in varying stages of fracture union.

She was admitted to the neonatal ward with a diagnosis of osteogenesis imperfecta congenita presenting with multiple fractures. The serum electrolytes and urea, and the full blood count were normal. She had plaster of Paris (POP) casts applied above elbow bilaterally and above knee bilaterally. She was comfortable in casts. On the 5th day she became febrile and tachypnoeic, respiratory rate was 100/min and heart rate was 172/min. The lung fields were clear clinically. An impression of congenital malaria fever was entertained, which was confirmed by the presence of ring forms of malaria parasites in the blood specimen. Syrup chloroquine 25mg/kg base over 3 days was administered and fever lysed. The POP casts
were removed on the 14th day. The femoral, ulnar and radial fractures had united clinically but the bilateral tibial and fibular fractures were not clinically united. Thus bilateral above knee POP casts were reapplied. She was discharged home on the 16th day of life to be followed up at the out-patient clinic.

On her first outpatient clinic visit at age 33days, she presented with a fresh fracture of the right humerus just distal to the previous one sustained at birth. An above elbow POP cast was applied to the right upper limb while the lower limb casts were removed. The tibial and fibular fractures were clinically united bilaterally. At age 47 days, the above elbow POP cast was removed and the humeral fracture was united. At the last clinic visit at age 54 days, she was doing well. She has however defaulted from further follow up visits.

**Discussion**

There is no specific laboratory test for osteogenesis imperfecta [10]. The diagnosis of osteogenesis imperfecta congenita in the index patient was based on the clinical findings of blue sclerae, craniotabes and multiple fractures at birth. This was buttressed by the finding of defective ossification of the skull and the presence of wormian bones on the skull x-ray. A major finding in the congenital type of osteogenesis imperfecta is multiple wormian bones in the skull particularly in the parietal, temporal and occipital regions [1]. Wormian bones may also occur in cranial dysostosis, pyknodysoysis and rarely in cretinism and hydrocephalus [2,8]. The patient did not have appearances in keeping with any of these other conditions. The delivery was traumatic for the patient because she presented breech and had to be extracted. This might have accounted for the multiple number of fractures which she sustained at birth. It is interesting to note that the baby did not present with hypovolaemic shock or anemia despite having sustained such multiple fractures. She was however tachypnoiec at presentation and this was attributed to pain from the multiple fractures.
Plaster of Paris casts were applied for the management of limb fractures which all healed uneventfully. This is not unusual in osteogenesis imperfecta as healing of fractures and osteotomies usually is quite satisfactory even though the healed bone may be no stronger than the original [4]. Bone remodelling is also rapid [12] and non-union of fractures is rare in osteogenesis imperfecta [6]. The major problems that could arise, as the child grows older, include stunting of growth and limb deformity as a result of multiple fractures and the disorderly manner of bone healing. The multiple fractures may preclude ambulation [3,9]. Various treatment options aimed at enhancing the quality of life for patients with osteogenesis imperfecta have been described. Gerber et al [5] described a comprehensive rehabilitation programme with long leg bracing that results in a high level of functional activity with an acceptable level of risk of fracture in children with osteogenesis imperfecta. Sofield [11] also described a successful surgical method of multiple osteotomy, realignment, and medullary nail fixation in treating the deformities of this condition, which is still practised today. Papagelopoulos and Morrey [7] reported favourable results with total hip and total knee arthroplasties for severe joint mal-alignment in these patients. Our patient was scheduled for frequent follow-up in the out-patient clinic with the aim of proper management of fractures as well as prevention and early detection of deformities that may result. She however defaulted from clinic after all fractures had united.

References


