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Osteogenesis Imperfecta a Case Report in Owerri, Nigeria

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Abstract: A week old male infant presented with severe tenderness all over the body with associated continuous crying. There was bowing of both limbs. A total body radiograph revealed multiple healed and healing fractures in the long bones and ribs. The long bones were short, broad and bowing with thinning of the cortex. The spine shows compressed, flattened and irregular vertebral bodies. Some infants' deaths has been recorded in the family. Osteogenesis imperfecta is a rare disorder of connective tissue which is characterized by skeletal deformity, bone fragility, fractures, ligament laxity, hearing loss, blue sclera, dental abnormality and thin skin. All those features will not manifest in one case; they are therefore group into four types and subtypes. This case is more of Type 2 and Type 2A Osteogenesis imperfecta.

Keywords: Osteogenesis imperfecta; Multiple fractures; Short and broad long bones.

1. Introduction

Osteogenesis imperfecta is a group of connective tissue disorder of Type 1 collagen characterized by skeletal deformity, bone fragility, fractures, ligament laxity, hearing loss, blue sclera, dental abnormality and thin skin [1-4]. A deficiency of osteoblasts is the basic bone defect which results in congenital osteoporosis [3].

A patient cannot present with all these abnormalities. One may present with only three, four or five out of these depending on the 'Type' or severity [1, 2]. There are about four types with subtypes. Types 1 and 4 are milder and are inherited as autosomal dominant inheritance while Type 2 and 3 are probably inherited as an autosomal recessive inheritance [3]. Type 1 is the most common and about 70% of cases; Type 2 is 10 %, Type 3 is 15% while Type 4 is 5% of osteogenesis imperfecta cases, [2].

Depending on the type/severity, some cases die in the womb, some die immediately after birth, some few days or weeks, some live up to few years before they die while some may live up to adulthood.

In the second trimester, ultrasound antenatal diagnosis may often be made in more severely affected fetuses on the bases of multiple fractures with shortened long bones [2]. Thorax may be short and in assessing the fetal skeleton, three-dimensional ultrasound scan will be more helpful [2].

2. Objectives

This is to create awareness of osteogenesis imperfecta in our environment and to investigate families with increased infant/child mortality. This will help to pin point the cause of deaths and reduce some dispute/suspicious accusations among families and friends.

3. Case Report

A week old male infant, the first of a twin was brought by the parents to Paediatric Unit with severe tenderness all over the body and associated with continuous crying. The patient was very pale -almost paper white. There was bowing of the limbs and patient vomited after every meal. The second twin was relatively normal.

4. Family History

Patient is the fourth child. The first and second siblings were normal. The third sibling died immediately after birth and had the same features as the patient. He was also a male child. The patient's great grandfather had nine children and seven died in their childhood. One of the survivals is the grandfather of the patient.

An impression of osteogenesis imperfecta was made and the patient was sent for total body xray. An infantogram (total body x-ray) revealed multiple healed and healing fractures in the long bones and ribs (figure 1). The long bones were short, broad and bowing with cortical thinning. The spine showed compressed, flattened and

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irregular vertebral bodies (figure 2). The skull showed wormian bones with thinning of the cranial vaults (figure 3). A diagnosis of the type 2A osteogenesis imperfect was made.

5. Discussion

Though Osteogenesis Imperfecta is rare, some relatively normal people are carriers. Investigating families with increased neonatal/childhood mortality will help in the diagnosis of Osteogenesis imperfecta and other similar congenital abnormalities. A poor woman that had six still births should be investigated and family history taken. Ultrasound in the second trimester may also help in the diagnosis.

The knowledge of the four types and subtypes of osteogenesis imperfect is very necessary.

According to Sillence classification, the four types are as follows: Type 1 is the most common and largest, about 70% of the cases [2, 3]. It is autosomal dominant inheritance with blue sclera, mild bone fragility and mildly reduced stature. Deafness occurs in adult life due to ankylosis of ossicles and osteosclerosis [2, 3].

Subtype 1A has mild skeletal changes and normal teeth.

The subtype 1B has sever skeletal changes. The long bones are thin and gracile [2, 5]. There is also cortical thinning, bowing and osteoporosis of the long bones. Fractures occur in 10% and usually in young children. There may be no fracture at all in 10% of cases. The skull shows wormian bone formation (characteristic fissuring) [2, 3, 5, 6].

This Type 1B has also has dentinogenesis imperfect (translucent gray to yellow teeth involving both primary and permanent dentition and the enamel fractures easily, [2, 3, 5].

In this Type 1, one has to check the sclera and teeth of the child and watched out for deafness in adult hood. Total body x-ray should be done to check for skeletal changes.

About 10% of the cases are seen in Type 2. In this type, there is intrauterine death or early infancy death, fractures during delivery, dark blue sclera, and multiple healed or healing rib fractures at birth, grossly demineralized long bones with cortical thinning [2-5].

In Subtype 2A, the ribs are broad with bending multiple rib fractures [2, 5, 6].

This Type 2 and Type 2A show the typical features of this case report i.e. broad rib with multiple fractures and bending. The long bones of the patient were short, broad and bowed with multiple fractures.

In Type 2B the ribs have few or no beading at all; but the long bones have the same fractures as type 2A, [2, 3].

Type 2C has multiple fractures and thinning of the long bones [2, 3]. The ribs are thin and broad with gross callus formation and areas of union and non-union. They have enlarged skull with reduced mineralization and wormian bones are also seen [2, 3, 5].

Type 3 are 15% of cases and this is a severe and progressively deforming type [2, 4]. The long bones are thin and osteoporotic with multiple fractures in child hood which results in bowing [2, 3]. The spine show compression and flatting of the vertebral bodies resulting in kyphosclosis. There is generalized osteoporosis. The skull shows poor ossifications with widened sutures (pseudowidening of the sutures secondary to underossification of the skull) [2, 6]. The wormian bones persist. There is also dentinogenesis imperfecta [2, 3]. The sclerae is blue at birth but usually cleared and normal when patient becomes an adolescent [2, 3].

About 5% of the cases are seen in Type 4. At birth there is fracture with mild bony fragility [2, 4]. The sclerae is usually normal [2, 4, 5]. The benign form may not manifest till adult life. Numerous fractures may occur over a few years. Excessive callus formation may occur during healing stimulating sarcoma. Due to ankylosis of the ossicles and osteosclerosis, deafness is found in adult [2, 3, 5]. Protrusion of acetabula is present [2, 3, 5] and if there is further compression, child birth may be hindered [2]. There is a downward pull of intercoastal muscles due to softening and thinning of the ribs [2]. The sclerae is usually normal [2, 4, 5].

In Type 4A there is no dental lesion while in Type 4B there is dentinogenesis imperfecta [2, 5]. Formally osteogenesis imperfecta was divided only into two most common and simpler classification. The first is called osteogenesis imperfect tarda. This develops later in life with less severe autosomal dominant gene, [5, 7]. About 90% with this condition are presented with blue sclera but with fewer fractures [5]. The other condition is called osteogenesis imperfecta congenital. This one is less common and usually autosomal recessive with thick bone type but occasionally autosomal dominant with thin bone type is present [5, 7]. Death usually occur in infancy with frequent still birth [5].

Due to new mutation and multiple presentations, osteogenesis imperfect is now classified into four types with subtypes as stated above.

Deformity, fractures and demineralization are the three radiological features [7]. This agrees with the findings of this case report.

Differential diagnosis are battered baby syndrome with multiple healed, healing or fresh fractures. Diaphyseal fractures are seen in osteogenesis imperfecta rather than metaphyseal and there is often elevation of urinary hydroxyproline [2]. The second differential is idiopathic juvenile osteoporosis. There is metaphyseal fracture usually in the long bones of the lower limb [2]. The biochemical findings are normal and the calcium is negative only in severe cases.

6. Conclusion

Though osteogenesis imperfect appears to be a rare disorder, some cases are over locked. It is necessary to get family history and investigate those with frequent still birth, infant mortality, blue sclera, deformity, fractures and later manifestation of deafness. Second trimester ultrasound scan during antenatal period of every pregnant woman

at risk and subsequent follow up will help to screen these case out. This will help to reduce false accusations on ones neighbour for being responsible for death in a family

Legends to Figures

Figure-1. Long bones are short, broad and bowing with cortical thinning, multiple healed and healing fractures.

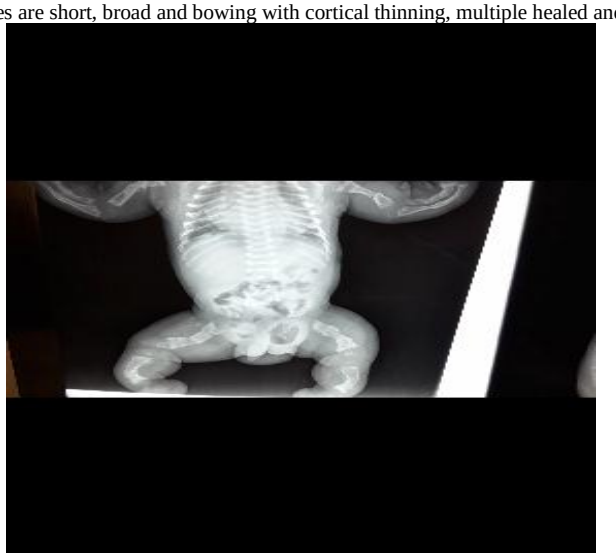


Figure-2. The X-ray of the spine showing compressed, flattened and irregular vertebral bodies

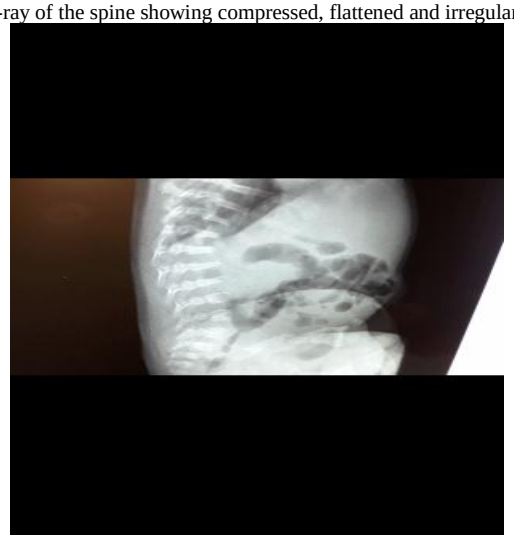
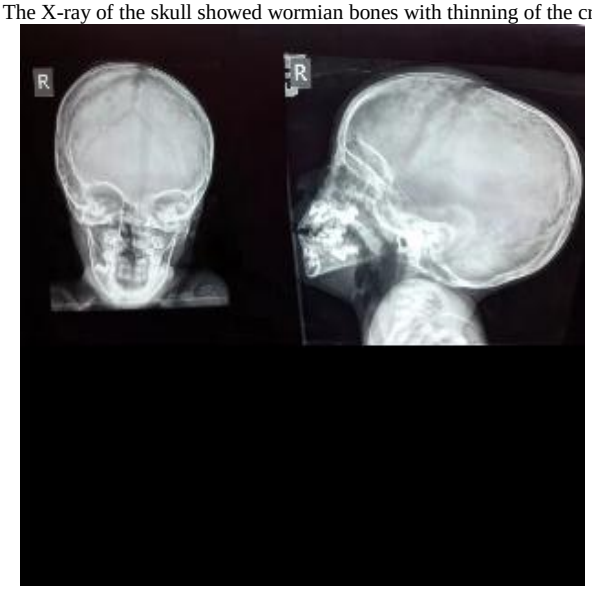


Figure-3. The X-ray of the skull showed wormian bones with thinning of the cranial vaults



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