International Journal of Applied and Natural Sciences (IJANS)
ISSN(P): 2319-4014; ISSN(E): 2319-4022
Vol. 4, Issue 4, Jun - Jul 2015, 5-8

International Academy of Science, Engineering and Technology
Connecting Researchers; Nurturing Innovations

OSTEOGENESIS IMPERFECTA: A CASE REPORT IN A SET OF TWINS IN LAUTECH TEACHING HOSPITAL, OGBOMOSO, NIGERIA

OLANREWAJU BAJOWA, TAIWO AKINBILE, OLUWASEYI OKE & OLUSEYI ATANDA

Department of Obstetrics and Gynaecology, Ladoke Akintola University of Technology (LAUTECH) Teaching Hospital, PMB 4000, Ogbomoso, Oyo State, Nigeria

Corresponding Author: OLUSEYI ATANDA

ABSTRACT

Osteogenesis imperfect is a rare form of congenital skeletal anomaly. The diagnosis of the condition in a set of twins is very rare, especially in this young tertiary institution, thus inspiring this write up. At birth, they were found to be small for gestational age. Their heads were large and their limbs were grossly deformed due to multiple fractures. They both had osteogenesis imperfecta (Type III) which is known to be a cause of perinatal deaths.

KEYWORDS: Bone, Collagen, Connective Tissue, Osteogenesis Imperfecta, Twins

INTRODUCTION

Osteogenesis imperfecta is a genetic connective tissue disorder affecting males and females equally, and is found in all races and ethnic groups. This condition also referred to as Brittle bone disease is characterized by increased bone fragility due to abnormal collagen formation. [1, 2, 3] There are different clinical types and the hallmark is increased bone fragility. [2] There are associated qualitative and quantitative defects in the synthesis of collagen Type 1. [4, 5] Incidence is about 1:20,000-50,000 live births. [5, 6] It could be either autosomal dominant (mild) or autosomal recessive (severe) .There are different types (I-VIII) with varying severity. Type I being the mildest and accounting for about 50% of cases.

Case Report

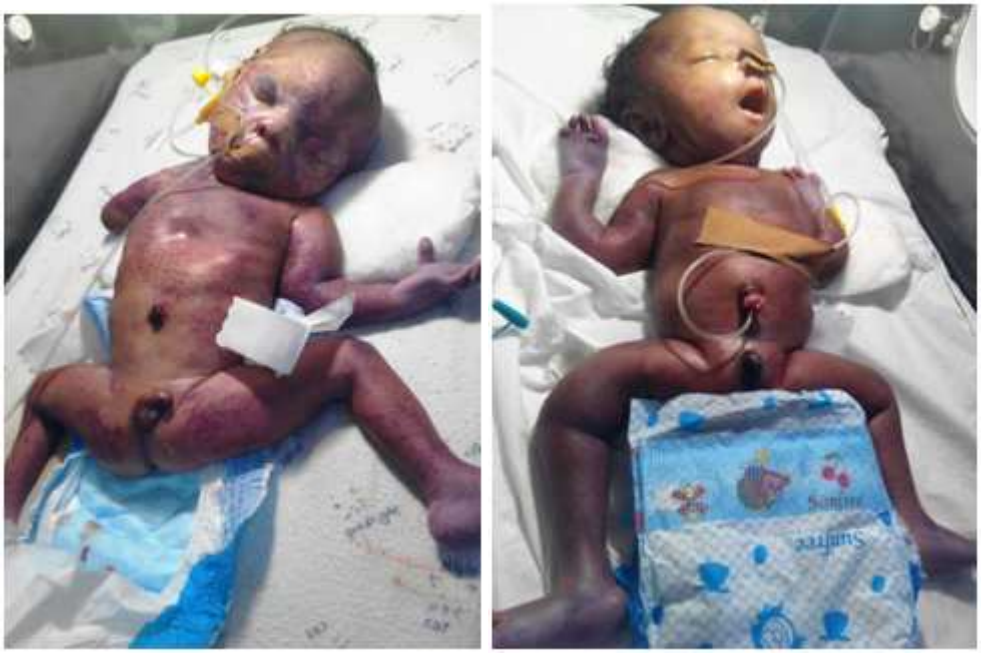
Babies AA and AB both male set of twins were born at 35 weeks gestation via emergency caesarean delivery on account of persistent occipito-posterior position of the leading twin in labor. The mother was an unbooked 28 year old primigravida who had no antenatal care. She denied family history of any skeletal anomaly and neither in the husband's family. The mother had no history of major illness during pregnancy. She routinely used daily over-the-counter medication throughout pregnancy. The babies had poor cry at birth with Apgar score of 3 and 6 at 1 and 5 minutes respectively for Baby AA and 4 and 6 at 1 and 5 minutes respectively for Baby AB. Their birth weights were 1.9 and 2.0kg respectively. Baby AA was 40cm in length and Baby AB was 41cm long. Their heads were disproportionately large with occipitofrontal diameters of 34cm and 35cm respectively. Extra digits were seen on the little fingers of both hands in Baby AA.

The central nervous system examination revealed babies with poor cry at birth and reduced neonatal reflexes. Both anterior and posterior fontanelles were palpable, soft and the sutures were widened and there was blue sclera in twin AB.

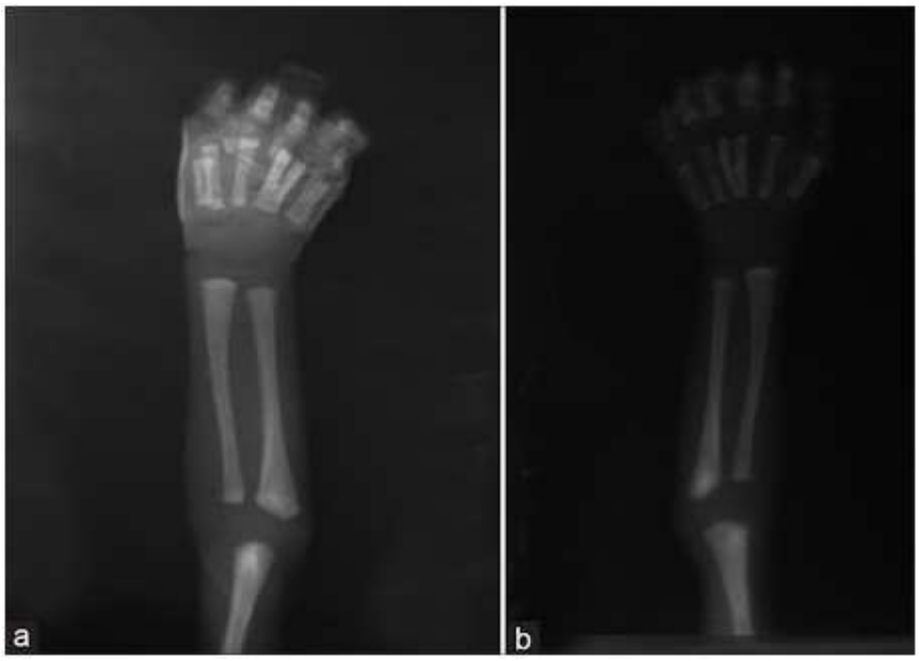
There were no relevant abnormal findings on examination of the gastrointestinal and cardiovascular systems. Clinical assessment of osteogenesis imperfecta was made. Other investigations done included a skeletal survey of both

twins, Full Blood Count and Differentials, Electrolytes, urea and creatinine. Of these, serum calcium was low in both twins while phosphate was low in Twin AB.

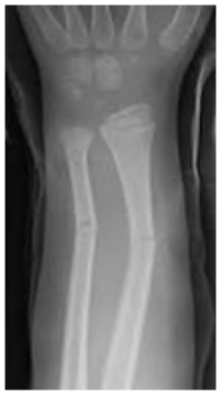
Radiological examination revealed multiple fractures at different stages of healing at right humerus, ulna, radius and left femur in Twin AA and both lower limbs in Twin AB with excessive callus formation and irregular outlines. The distal long bones were bowed in Twin AA. There was normal spinal column. The babies however died at 6 hours of life on account of perinatal asphyxia due to respiratory problems.



TWIN AA **TWIN AB**



Radiograph of TWIN AA (b) Showing Extra Digit Compared with TWIN AB



Radiograph of TWIN AA Showing Fractures of the Radius and Ulna

DISCUSSIONS

In the case presented, our findings are similar to those in previous reports. Literature has reported that multiple fractures can result even from minimal trauma. The cause of multiple fractures in the uterus is attributed to fetal movements in utero which the defective bone cannot withstand. [7] Fractures are therefore pathological and diagnostic of the disease entity as was seen in the set of twins presented. There was blue sclera in one of the twins which is typically seen in Type III. This was also found in the study by Bryan D.H (1996) who claimed that the sclera may be blue at birth and become less blue with age. [8] This is however not a reliable clinical sign in small babies as it can also be seen in normal newborns. The features of severe bone deformity, multiple fractures present at birth and severe respiratory problems along with the blue sclera in twin AB is highly suggestive of Type III Osteogenesis Imperfecta in this case report. Ultrasound antenatal diagnosis of osteogenesis imperfecta may be picked on routine scan, [7] however; this was an unbooked patient who had little or no care.

CONCLUSIONS

Osteogenesis imperfecta is a rare condition but occurs in our environment and is a known cause of perinatal deaths. [9, 10, 11] More attention should be given to antenatal care and presence of abnormal lie, presentation and twin gestations should warrant anomaly ultrasound scan to rule out such deformities and other congenital anomalies. Antenatal diagnosis would have lessened the enthusiasm, anxiety and burden of this condition on the patient, family and managing team.

ACKNOWLEDGEMENTS

The authors express their sincere gratitude to colleagues in the department, the midwives in the labour ward, the paediatric team and labour ward theatre team who contributed to the management of the case reported.

Consent of the couple was also obtained to report this rare skeletal anomaly in twin pregnancies.

There is no external source of funding nor conflict of interests declared.

REFERENCES

1. Akiode O, Ogunfowora OB, Shonubi A, Bajomo AA, Musa AA, Sotimehin SA. Osteogenesis Imperfecta Congenita in a Nigerian baby. SICOT Online Report 2003 E049

2. Mundlos S, Chan D, Weng YM, Sillence DO, Cole WG et al. Multiexon Deletions in the Type 1 Collagen COLIA2 Gene in Osteogenesis imperfecta Type 1B. JBC Online 1996, 271(15): 21068-21074

3. Jandl JH. Hereditary disorders of connective tissue: Osteogenesis imperfecta In: Blood Textbook of Haematology 2nd ed. Little & Brown Company U.S.A 1996, p1285

4. Rees MM, Rodgers GN. Hereditary disorders of connective tissue: Osteogenesis imperfecta. In: Wintrabe's Clinical Haematology Vol.2 10th ed. Edited by Lippincott Williams and Wilkins. Walter Kluwer Company. Philadelphia U.S.A 1998. p1637

5. Duthie RB, Bently G: Developmental diseases of the skeleton. In Duthie RB (ed): Mercer's Orthopedic Surgery. 9th ed., Bath Press, Avon, UK, 1996, pp. 262-270.

6. Fajolu IB, Ezeaka VC, Elumelu OJ, Onabajo OA, Ananti CH, Iroha EO, et al. Osteogenesis Imperfecta in a set of Nigerian twins-A case report. Internet J Pediatr Neonatol. 2012:14. doi:10.5580/2c29.

7. Osteogenesis Imperfecta. Source:http://en.wikipedia.org/wiki/Osteogenesis_imperfecta (date accessed: 17/03/2015)

8. Bryan DH. Inherited Osteoporosis: Osteogenesis Imperfecta. In: Nelson Textbook of Paediatrics: 15th ed. Edited by Behrman, Kliegman, Arvin. WB Saunders Philadelphia, U.S.A 1996. p.1978-80.

9. Smith R, Francis MJO, Houghton GR. The brittle bone syndrome. Osteogenesis Imperfecta. London: Butterwort; 1983.

10. Beighton P, Spranger J, Versveld G. Skeletal complications in Osteogenesis imperfecta. A review of 153 South African patients. S Afr Med J 1983; 64: 565-8

11. Glorieux FH, Rauch F, Plotkin H, Ward L, Travers R, Roughley P, Lalic L, Glorieux DF, Fassier F, Bishop NJ. Type V osteogenesis imperfecta: a new form of brittle bone disease. J Bone Miner Res. 2000*;* 15*(*9*):*1650–8.