Clinical Quiz

Yasir S. Siddiqui, *MBBS, MS (Orth),* ***Mohammad A. Sherwani,*** *MS (Orth), MCh (Liverpool),*
Abdul Q. Khan, *MBBS, MS (Orth),* ***Abhishek Gupta,*** *MBBS, D (Orth).*

From the Department of Orthopedic Surgery, Jawaharlal Nehru Medical College, Aligarh Muslim University, Aligarh, Uttar Pradesh, India.
Address correspondence to: Dr. Yasir S. Siddiqui, Clinical Orthopedic Registrar, Department of Orthopedic Surgery, Jawaharlal Nehru Medical College, Aligarh Muslim University, PO Box 71, Aligarh, Uttar Pradesh, India. Tel. +91 9837343400. Fax. +915 (71) 2702758.
E-mail: yassu98@gmail.com

Notice: Authors are encouraged to submit quizzes for possible publication in the Journal. These may be in any specialty, and should approximately follow the format used here (maximum of 2 figures). Please address any submissions to: Editor, Saudi Medical Journal, Armed Forces Hospital, PO Box 7897, Riyadh 11159, Kingdom of Saudi Arabia. Tel. +966 (1) 4777714 Ext. 6570. Fax. +966 (1) 4761810 or 4777194.

Osteogenesis imperfecta

Clinical Presentation

A 2-month-old male baby was brought to the Orthopedic Outpatient Department with complaints of deformities of both lower limbs following birth. There was no history of trauma and similar complaints in the rest of the 2 older siblings. The baby was born at term by spontaneous vaginal delivery, conducted at home. Antenatal history was unremarkable. The baby cried immediately after birth. The parents denied any family history of skeletal abnormalities and consanguinity. At presentation, the baby was active, afebrile, with normal respiration, and his weight was 3.5 kg. The baby appeared abnormal with bilateral bowed (curved) lower limbs. His head was relatively larger as compared to extremities. The rest of the systemic examination was normal. Laboratory investigations including complete blood count, erythrocyte sedimentation rate, serum calcium, and serum phosphate were within normal limits. However, his serum alkaline phosphatase was slightly high. The x-ray of the lower limbs is shown in **Figure 1**.

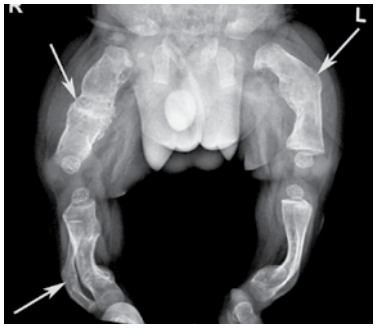

Figure 1 - An x-ray of both lower limbs - anteroposterior view showing multiple fractures in the long bones of the lower limbs at different stages of healing, with excessive callus formation (arrows). Radiograph also showed flaring and bowing of the ends of the long bones with bilateral genu vara.

Questions

1. What is the diagnosis?

2. What are the associated abnormalities?

3. What is the management?

Clinical Quiz

Answers

1. Clinico-radiological diagnosis of patient is osteogenesis imperfecta (OI). In the absence of any history of trauma, the presence of multiple fractures of the long bones, with excessive callus formation, flaring, and bowing of the ends of the long bones favors the diagnosis of OI. The differential diagnosis include battered baby syndrome, the fractures of which are usually metaphyseal.[1]

2. Along with fragility fractures, blue sclera, and dentinogenesis imperfecta are the associated abnormalities, which one can encounter in patients of OI.[1]

3. At present, there is no cure for OI. Treatment is directed at increasing global bone strength to check for fracture, and preserve mobility. Physiotherapy is recommended to reinforce muscles and to improve mobility, while curtailing the threat of fracture.[2] Bisphosphonates are being progressively administered to increase bone mass, and diminish the occurrence of fracture. It has a proven efficiency in dropping fracture rates in children,[3] however only a drift towards decreased fracture was seen in a small randomized study in adults.[4] While decreasing fracture rates, there is some concern that prolonged bisphosphonates therapy may delay the healing of fractures, although this has not been convincingly established. The ideal treatment of fractures and deformities of long bones is intramedullary nailing, and this allows the alignment of the fragments, a decreased number of fractures, and a better functional result.[5] Inserted nails in the long bones improve strength, and proved extremely useful in the rehabilitation and prevention of fractures.

Discussion

The OI is a group of hereditary disorders characterized by the aberration of amalgamation of type I collagen. Collagen is the chief structural protein in bone, ligaments, tendons, skin, sclera, and dentin.[2] The disease is expressed mostly through osseous fragility and deformities. They frequently present with multiple fractures in childhood, and it normally regresses by puberty.[5] The disease is also known as Lobstein syndrome, or brittle bone disease. The disease appears in childhood before the onset of puberty. The clinical presentation is constant with global osteoporosis, bluish sclera, reduced size, ligament laxity, deafness, gray and breakable teeth, and cardiopulmonary disorders.[5] The osteoporosis is responsible for the osseous fragility, which accounts for the frequent fractures even following trivial trauma. The most used classification is the modified Sillence classification (1979). The types vary in severity, age of presentation, and clinical features. No specific diagnostic test for OI is available. Early detection can improve morbidity. Treatment is aimed at increasing overall bone strength to prevent fracture and maintain mobility. Lifestyle modifications, such as the use of orthotics, and physiotherapy should be considered.[5] Medical management of OI, with the exception of bisphosphonates, has been largely unproductive. There is functional augmentation by intramedullary nailing of the long bones.

References

1. Sutton D. Cardiovascular system: Arteriography and interventional angiography. Textbook of Radiology and Imaging. 7th ed. London (UK): Churchill Livingstone; 2003. p. 72, 74, 411, 423, 476.
2. Strevel EL, Papaioannou A, Adachi JD, McNamara M. Case report: osteogenesis imperfecta elusive cause of fractures. ***Can Fam Physician*** 2005; 51: 1655-1657.
3. DiMeglio LA, Peacock M. Two-year clinical trial of oral alendronate versus intravenous pamidronate in children with osteogenesis imperfecta. ***J Bone Miner Res*** 2006: 21: 132-140.
4. Chevrel G, Schott AM, Fontanges E, Charrin JE, Lina-Granade G, Duboeuf F, et al. Effects of oral alendronate on BMD in adult patients with osteogenesis imperfecta: a 3-year randomized placebo-controlled trial. ***J Bone Miner Res*** 2006: 21: 300-306.
5. Elmrini A, Boujraf S, Marzouki A, Agoumi O, Daoudi A. Osteogenesis Imperfecta Tarda. A case report. ***Nigerian Journal of Orthopaedics and Trauma*** 2006: 5: 61-62.