A Retrospective Study of Osteogenesis Imperfecta in Dakhliya region. Sultanate of Oman

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Abstract :

Osteogenic imperfecta is one of the most common inherited bone diseases. The disease typically involves the bones, teeth, ligaments, eyes and skin and is characterized by bone fragility. Aim: To estimate the prevalence and the clinical characteristics of osteogenic imperfecta in the Dakhliya region of the Sultanate of Oman. Method: Retrospective study of all cases of OI diagnosed and followed in Nizwa hospital since 2000. Data was collected from the case files obtained from the medical record department.

Result: A total of 22 patients were identified in the study 10 (45.5%) were male and 12 (54.5%) were female. The mean age of diagnosis was 6.46 years. The parents of all patients had a first degree consanguineous marriage. A total of 66 fractures were sustained in these patients.

Conclusion: Osteogenic imperfecta is an important disease in Oman with high morbidity and mortality. There are new modalities of treatment like Bisphosphante which have been studied recently and showed remarkable improvements in the bone mineral densities of these patients. We suggest starting an OI registry in the country. This will enable the scientist and the medical professional to diagnose, treat and study these patients appropriately and promptly.

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Introduction

Osteogenesis imperfecta (OI) is one of the most common inherited bone disorders. The disease typically involves the bones, teeth, ligaments, eyes and skin, and is characterised by fragile bones that break easily. This bone disorder is usually present at birth as an inherited disease. Osteogenesis imperfecta (OI) is Classified into four major types (and further subtypes). All four types of OI are caused by defects in the amount or structure of Type 1 collagen, an important part of the bone matrix. The collagen problem usually results from a dominant genetic defect. In addition to a complete medical history and physical examination. Diagnostic procedures for OI may include a skin biopsy to evaluate the amount and structure of collagen. However, this test is complicated and not many facilities are available to perform the procedure. Additional diagnostic tests include: X-rays which may show thinning of bone and past or current fractures. An ultrasound may be used during pregnancy to detect limb abnormalities at 15-18 weeks gestation. However, these may not always be accurate.

The role of pharmacologic therapy has not been well defined, but some studies show a benefit of treatment with bisphosphanates and calcium supplements. Other medical interventions including bone marrow transplant, the use of growth hormone, and gene therapy are also under investigation. [1,2,3] Intramedullary fixation with use of a telescopic rod with a T-piece is one of the standard methods for long-bone stabilization in growing children with osteogenesis imperfecta. Prolonged immobilization after a fracture must be avoided. [4] Repeated respiratory infections, Basilar impression caused by large head and Cerebral hemorrhage caused by birth trauma are complications of OI. Patients with OI should be considered to be at high risk for complications of anesthesia. The life expectancy of subjects with nonlethal OI appears to be the same as that for the healthy population. Patients with lethal OI may die in the perinatal period, but individuals with extremely severe OI can survive until adulthood. The aim of our retrospective study was to estimate the prevalence of Osteogenesis Imperfecta in the Dakhliya region and analyse the patient's clinical presentation and management of fractures at Nizwa Hospital.

Methods

We included all the cases of osteogenesis imperfecta diagnosed and followed up in Nizwa hospital since 2000. Data was collected from the case files obtained from the Medical Records department.

The following information were collected, age at diagnosis, sex, history of consanguinity, any tribal relationship. The diagnoses based on either clinical or other [x-ray] investigations. Common fractures, the management sought for the fractures either conservative or surgical. The duration of treatment. Patient follow-up. The approximate time interval between fractures, fracture healing time, Cause of admission and cause of death.

Results

A total of 22 children were identified in our study 10[45.5%] were males and 12[54.5%] were females. Of these 10 were born in the Nizwa hospital. The mean age at diagnosis, which was the patients first presentation to the hospital is 6.46 years, 21 patients were diagnosed at the first visit, 1 patient diagnosed 23 days later at the

history. A total of 66 fractures were sustained in these patients. The decreasing order of bones involved – femur-23[34.8%], humerus-15[22.7%], forearm-6 [9%] and tibia-4[6%]. The management of the fractures was divided either into conservative comprising of Plaster of Paris slabs, Dynamic skin traction, Hip spica application, Cuff and collar slings or Surgery. Only 2 patients underwent surgical treatment, the procedures used were Tension Band Wiring of fracture olecranon for the first patient and multiple percutaneous K wiring for fractures of shafts of femur and humerus in the second patient. All the patients with limb fractures had regular follow up until fracture union and splint removal. The plasters or spica were kept until complete radiological union of the fracture which was approximately 6 weeks in lower limbs and approx 1 month in the upper limbs. The average interval between each fracture is 6.34 months. The average interval between 2 fractures of the same bone in the same limb is 21.1 months. The co-morbid conditions that were present in these patients were hydrocephalus – 1 patient, mental retardation- 2 patients, umbilical hernia- 2 patients, degenerative arthritis of the hip and shoulder in 1 patient. 5 patients died from the disease, 3 due to pneumonia with sepsis and 2 newborns with failure to thrive. The reasons for admission to the hospital other than fractures were pneumonia in 8 patients, and acute gastro-enteritis in 1 patient. The surgeries performed for other conditions were bilateral inguinal hernia repair in 3 patients. Hysterectomy in 1 patient and Extra Dural Hematoma evacuation in 1 patient.

The data obtained from patient records did not reveal if clinical features of Osteogenesis Imperfecta other multiple limb fractures were detected in these patients. We also did not have a complete record of all the patients treated since some the patients were also treated in other regional hospitals and the University.

Discussion

In O.I, Bone fractures that occur with little or no trauma are often the first indication that a child may have OI. Some milder cases are not diagnosed until the teen or adult years.

Emergency room physicians and others who see children with fractures need to consider O.I as a possible cause, particularly in cases involving multiple fractures or a family history of fractures." Not so long ago, parents were advised to "protect" their children by carrying them on pillows and avoiding recreational activities. However, this well-intentioned approach did not protect children from fractures and may have hindered development and achievement of independent functioning. Bone growth depends on muscle pull as well as loading (weight bearing) through standing, walking, and lifting. Swimming is an excellent conditioning exercise for many people with OI. In addition, physical rehabilitation and therapy can help. Immobilization may result in loss of muscle and skeletal mass. It can take as long as a year to restore this bone mass following a relatively short period of immobilization. Over the years, it has become clear that physical activity is an important part of managing OI in both children and adults because it promotes: general health optimal physical function to support independence in daily activities optimal psychological and social well-being by improving Self-confidence and the ability to interact socially with peers. Good nutrition and approved forms of exercise are key to building bone and muscle strength. Surgery to place metal rods in the bones can help strengthen them and prevent deformity. Braces and walking aids are helpful for some people. Bone marrow transplants have been successful in a few children. Transplants of mesenchymal stem cells or bone marrow stromal cells have resulted in better growth in a few children. Gene therapy to block the defective gene may prove useful in people with OI who have at least one normal gene. Growth hormone has been used with some success in children.

Our study shows that the number of osteogenesis imperfecta cases diagnosed in our region is fairly large. Almost all the cases that presented to the hospital were diagnosed at their first presentation. The initial presentation was mainly due to fractures. All the cases presented early in their life, the diagnosis were made clinically and later confirmed by x-rays. The most common fractures were that of the femur followed by the humerus. Most of fractures sustained by these patients were treated conservatively. Both patient and their parents had good compliance to treatment. They did not require aggressive management with complex surgical procedures, on the other hand minimally invasive procedures like k-wiring is sufficient to stabilise them since the fractures in these patients have a high potential for union. We do not have any experience with the use of medication.

In conclusion, OI is an important disease, which causes high morbidity and disability. In Oman, we do not have any antenatal screening programs to diagnose OI. Screening programs, Health Education to parents with family history of OI and Genetic counseling should be initiated. A regional OI community center should also be initiated to encourage, provide physical and train patients with OI to become self sufficient.

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