Research Paper

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Osteogenesis Imperfecta – A Case Report

Medical Science

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ABSTRACT

BACKGROUND: *Osteogenesisimperfecta is a congenital disease caused by defect in the gene that produces type I collagen fiber, an important building block of bone resulting in fracture of bones, reduced growth and bone deformity.*

CASE PRESENTATION: *A 16 years old female patient having the history of multiple bone fracture since 5 years. Her elder sister and her brother is normal. Routine clinical examination revealed that scoliosis of thoracic and lumbar vertebrae, diffused osteopenia and crowding of ribs on the left side noted. Digital X-ray of forearm shows fracture of distal end of radius and minimal displaced fracture of middle third of ulna noted.*

CONCLUSION: *Osteogenesisimperfecta is an autosomal dominant disease. Knowledge of osteogenesisimperfecta is important to provide better ability to plan prenatal diagnostic methods and to plan genetic counseling and preventive methods in affected families.*

INTRODUCTION:

Osteogenesis imperfecta is a genetic connective tissue disorder is characterized by fragile bones and growth disorders of varying severity. It affects both males and females equally, and is found in all races and ethnic groups. The incidence has an estimated 6 to 7 per 100,000 people worldwide. Type I and type IV are the most common forms of osteogenesis imperfecta, affecting 4 to 5 per 100,000 people. Individual with osteogenesis imperfecta have brittle bones most often as a result of mutations in the gene affecting type I collagen fiber, which is the most prevalent protein in bone, skin and other connective tissues. These mutations can lead to different levels of skeletal deformities and in some cases frequent multiple fractures (table 1).

The first four types of the disease arise from mutations in collagen type I genes, composed from COL1A1 and COL1A2 chains. A result of these mutations is the production of shortened or structurally defective protein. Individuals affected by OI forms V to IX have mutations in proteins encoded by following genes: CRTAP, LEPRE1, PPIB, FKBP10.

It has several clinical presentations, according to the severity of the involvement. Type I is the most common with mild involvement without major deformities, and normal stature. Type II is the most severe affecting neonates, and it is usually incompatible with life. In type III patients have short stature, triangular *fascies*, and bone deformities. Type V, OI with hypertrophic calluses and calcification of the interosseous membrane of the forearm was recently described in the literature.

80 to 90 percent of OI are caused by a dominant genetic defect. A person with a form of OI caused by a dominant mutation has a 50 percent chance of passing on the disorder to each of his or her children.

Some children who have the dominant form of OI inherit the disorder from a parent. Other children are born with the dominant form of OI even though there is no family history of the disorder. In these children, the genetic defect occurred as a spontaneous mutation.

Based on clinical signs the first OI classification from David Sillence (created in 1979) distinguished four types of the disease (I-IV). In the past, related to the development methods of analysis, such as molecular-genetic techniques and histological findings, new forms were identified in the IV group of OI - OI type V-IX [13].

TABLE 1:CLINICAL CHARACTERISTICS FEATURES :

Type	Mode of inheritance	Stature	Fracture	Deformity level	Sclera	Dentinogenesis imperfecta	Lethality
I	AD	Normal stature	Most of the fractures occur during the preschool years and are less common after puberty	Moderate	Blue sclera	Absent or present	Nonlethal
II	AD parental mosaicism	—	Exhibits extreme bone fragility and frequent fractures, which may occur during delivery	Severe	Blue sclera	Present	Many patients are still born and 90% of patients die before 4 weeks of age
III	AD	Short	Fractures may be present at birth	Progressively deforming	Normal or pale blue at birth, fades with age	Present	The majority of affected individuals die during childhood usually from cardiopulmonary complications caused by kyphoscoliosis
IV	AD	Short	Fractures present during childhood and decreases after puberty	Mild to moderate	Normal or pale blue at birth, fades with age	Present or absent	Nonlethal

AD: Autosomal dominant

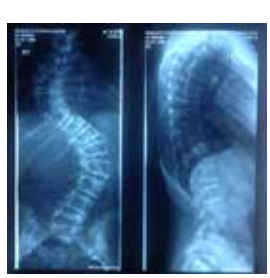
CASE PRESENTATION:

A 16 years old female individual came for the genetic counseling, having the history of multiple bone fracture since 5 years. The family history revealed the elder sister and her brother is normal.

Clinical examination:

Digital X-ray of dorsal spine shows scoliosis of thoracic and lumbar vertebrae with convexities to the right on thoracic vertebra and left on lumbar vertebra, rotation of lower dorsal and lumbar vertebrae, diffused osteopenia, crowding of ribs on the left side.

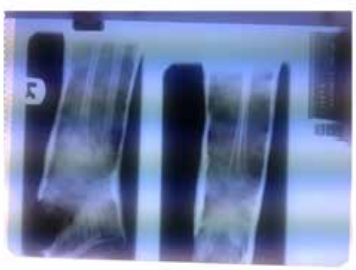
DIGITAL X—RAY OF DORSAL SPINE



Digital X-ray of forearm shows, fracture of distal end of radius, minimally displaced fracture of middle third of ulna noted with surrounding callus formation, POP cast in situ, joint space appear normal, soft tissue structures appear normal

DIGITAL X-RAY OF FOREARM

DIGITAL X-RAY OF FOREARM



Laboratory findings:
Vitamin – D is low

Calcium and phosphate is normal

Karyotyping is normal.

CONCLUSION:
Osteogenesis imperfecta is a hereditary pathology characterized by the osseous fragility which causes increasing severe deformities in patients. It affects children and it regresses by puberty. The disease is expressed by a reduction of the production either on the Type I collagen or by a production of abnormal collagen synthesis. The osteogenesis imperfecta also known as "LOBSTEIN" disease is a genetic disease with dominant hereditary transmission. Due to the decreased or abnormal synthesis of the type I collagen results in decreased mineralization that leads to osteoporosis of bone. This osteoporotic changes occuring in early age results in fracture of bones even minor injury.

By using "Sillence classification" we concluded that, this female individual is affected by type I A osteogenesis imperfecta with the help of clinical and the radiological findings.

In the future it is important to perform the molecular genetic analysis of complete sequences of both collagen type I genes and subsequently to compare the clinical manifestations of the disease in patients having the same form of OI and the same change in DNA.

REFERENCE

1.Sillence DO, Barlow KK, Garber AP, Hall JG, Rimoin DL. Osteogenesisimperfecta type II: delineation of the phenotype with reference to genetic heterogeneity. Am J Med Genet. 1984; | 17:407-423. | 2.Marini JC. Osteogenesisimperfecta. 2010. Available at:http://www.endotext.org/parathyroid/parathyroid17/parathyroid17.pdf. (Revised 1 March 2010). | | 3.Neves JFNP, Sant'Anna RS, Almeida JR et al. –Anestesiavenosa total empacienteportador de OsteogenesisImperfecta. Relato de caso. Rev Bras Anestesiol, 2004;54:668-671. | | 4.Sharma A, George L, Erskin K - OsteogenesisImperfecta in pregnancy: two case reports and review of literature. ObstetGynecolSurv, 2001;56:563-566. | 5.Assis MC, Plotkin H, Glorieux FH et al. - OsteogenesisImperfecta: novosconceitos. Rev Bras Ortop, 2002;37:323-327. | 6.Vogel TM, Ratner EF, Thomas Jr. RC et al. - Pregnancy complicated by severe OsteogenesisImperfecta: a report of two cases. AnesthAnalg, 2002;94:1315-1317. | 7.Cubert R, Cheng EY, Mack S et al. - OsteogenesisImperfecta: mode of delivery and neonatal outcome. ObstetGynecol, 2001;97:66-69. | 8.Chan B, Zacharin M - Maternal and infant outcome after pamidronatetreatment of polyostotic fibrous displasia and OsteogenesisImperfecta before conception: a report of four cases. J ClinEndocrinolMetab, 2006;91:2017-2020. | 9.Edge G, Okafor B, Fennelly ME et al. - An unusual manifestation of bleeding diathesis in a patient with OsteogenesisImperfecta. Eur J Anaesthesiol, 1997;14:215-219 | 10.Porsborg P, Astrup G, Bendixen D et al. - OsteogenesisImperfecta and malignant hyperthermia. Is there a relationship? Anaesthesia, 1996;51:863-865. | | 11.Esposito P, Plotkin H. Surgical treatment of osteogenesisimperfecta: current concepts. CurrOpinPediatr. 2008;20:52-7. | | 12.Zeitlin L, Fassier F, Glorieux FH. Modern approach to children with osteogenesisimperfecta. J PediatrOrthop B. 2003;12:77-87. | | 13.Luhmann SJ, Sheridan JJ, Capelli AM, Schoenecker PL. Management of lower-extremity deformities in | osteogenesisimperfecta with extensible intramedullary rod technique: a 20-year experience. J PediatrOrthop. 1998;18:88-94. | | 14.Khoshhal KI, Ellis RD. Effect of lower limb Sofield | procedure on ambulation in osteogenesisimperfecta. J PediatrOrthop. 2001;21:233-5. | | 15.Astrom E, Soderhall S. Beneficial effect of bisphosphonate during five years of treatment of severe osteogenesis imperfect. ActaPediatr. 1998,87:64-8. | | 16.Mulpuri K, Joseph B. Intramedullary rodding in | osteogenesisimperfecta. J PediatrOrthop. 2000;20:267-73. | | 17.Panigrahi I, Das RR, Sharda S, Marwaha RK, Khandelwal N. Response to zolendronic acid in children with type III osteogenesisimperfecta. J Bone Miner Metab. 2010;28:451-5.