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A CASE REPORT ON OSTEOGENESIS IMPERFECTA AND ITS COMPLICATIONS

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ABSTRACT

Osteogenesis Imperfecta(OI) is also known as brittle bone disease characterized by increased bone fragility resulting from the abnormal collagen formation. Blue sclera, dentinogeneis imperfecta, fractures, scoliois, pectus excavatum are the main clinical features of OI. Some OI patients have serious cardiac problems related to their disease, including mitral valve prolapse, aortic valve insufficiency and dilation of the aorta. The management of OI involves multidisciplinary care including general practioners, physiotherapist, rehabilitation and orthopedic surgery. Here we are reporting a rare case of osteogenesis imperfecta with mitral valve regurgitation and congestive cardiac failure.

KEYWORDS: Osteogenesis Imperfecta, Blue sclera, Scoliosis.

INTRODUCTION

Osteogenesis Imperfecta(OI) is also known as brittle bone disease characterized by increased bone fragility resulting from the abnormal collagen formation. OI is an autosomal dominant disorder of type I collagen with the incidence rate of upto 1/30000 and prevalence of 6 to 7/100000 births.[1,2] Extraskeletal clinical features like blue sclera, dentinogenesis imperfecta, adult onset hearing loss, fractures, presence of wormian bones are common in OI.[3,4] OI can be classified into 8 types out of which OI type 1 is the most common.[1] Diagnosis is commonly based on positive family history and blue sclera. The other findings like histomorphometry(bone biopsy), bone mineral density, osteocalcin, alkaline phosphatase and amino terminal telopeptide of type I collagen are useful for monitoring children with OI.[5]

The management of OI involves multidisciplinary care including general practioners, physiotherapist, rehabilitation expert and orthopedic surgery. Long term follow up is essential in patients with OI. Bisphosphonates like pamidronate reduce pain and fracture rates resulting in increase of bone mineral density. Cyclic IV pamidronate was considered to be the most widely reported treatment for children with OI.[6] Prophylactic supplementation of 500-1000mcg of calcium and 400-800IU of vitamin D also benefits in OI.[7] Some OI patients have serious cardiac problems related to their disease, including mitral valve prolapse, aortic valve insufficiency and dilation of the aorta.[8] Genetic analysis is an important aspect in diagnosis. The parents should need special instructions in positioning the child in the crib and handling the child with least possibility of causing fractures. Here we are reporting a rare case of OI with mitral valve regurgitation and congestive cardiac failure.

CASE REPORT

A 15 year old boy was admitted with the complaints of easy fatigability, generalized body pain and breathlessness for past 1 week. He has hearing impairment and a known case of congenital osteogenesis imperfecta. His mother received routine antenatal care and pregnancy was uneventful. She was taken iron and folic acid tablets during pregnancy time. The child was born at term by spontaneous vaginal delivery, baby cried immediately after birth. He had fracture in left femur at birth and birth weight was 2.5 kg. The treatment was initiated from third year onwards of age. Family history showed that there was no similar illness.

On examination the child was 92cm tall, 15kg weight and he had blue sclera. Head to foot examination shows dolicocephaly and webbed neck with high arched palate. Musculoskeletal system examination revealed pectus excavatum, short upper and lower limbs, pescavus, equino varus deformity and cryptorchidism. The CVS examination shows scoliosis, apex beat visible seen in left 5^{th} intercostals space in midclavicular line and visible epigastric suprasternal neck pulsation. The boy had palpable S_1 S_2 and systolic thrill present and characterstic pansystolic murmur (usually present in case of mitral regurgitation, tricuspid regurgitation, ventricular septal defect) was present. On laboratory assessment, elevated WBC (19.6 x 10^9 /L), blood urea (53mg/dl) was found except all were normal. ECG shows sinus tachycardia, biventricular hypertrophy and t- wave inversion. The USG abdomen confirmed partially thickened bladder. Finally it was diagnosed as OI with severe mitral valve regurgitation and congestive cardiac failure. Therapy with vitamin D was initiated,

cardiovascular problems were treated with Digoxin 0.25mg, Amiloride 2.5 mg and the patient was supported with intra nasal oxygen.

DISCUSSION

OI is a rare inherited disorder which causes bone fragility and fractures. It was also been called as osteopsathyrosis idiopathica, fragilitus osseum, glass bone disease, etc.[9] It is commonly present in children or young adults. Occurrence of mild OI may be present after infancy and proper care should be provided whenever children and adults have recurrent fractures. This patient experienced blue sclera, scoliosis, pectus excavatum and hearing impairment which shows the characteristics of OI. Scoliosis is the most common. The combination of chest deformities and scoliosis resulted in large number of deaths due to respiratory disorders and cardiovascular problems.[2] Stein D *et al* co-relates mitral valve abnormalities with the connective tissue disorder in two patients with OI.[10] Detection of osteogenesis imperfecta at early stage will help to initiate appropriate therapy as well as patient education. Genetic analysis and family screening is also an important aspect in OI.[11]

Mutations in any of COL I A 1 gene on chromosome 17 and COL I A 2 gene on chromosome 7 encoded the α –chain of collagen type I results in abnormal or decreased production of normal collagen, but some studies shows that molecular understanding of disease recognized newer autosomal recessive form of OI (type V, VI & VII) not associated with type I collagen gene defect.[12,13] Bisphosphonates notably pamidronate is the drug of choice for OI. Zeitin *et al* reported that there was an increased height and weight in OI children treated with pamidronate.[14] Another one study proposed that risedronate is the more suitable alternative to pamidronate because of its lower mineral affinity than the other nitrogen containing bisphosphonates such as alendronate and olpadronate.[15] In contrast to this Watts NB *et al* demonstrated that the long term use of bisphosphonates is associated with an increased risk of uncommon side effects such as osteonecrosis of the jaw and atypical fractures.[16] The findings from Chevrel G *et al* and Antoniazzi *et al* shows that the combination of bisphosphonates and growth hormone treatment results in terms of bone density, lumbar spine projected area and growth velocity than bisphosphonate alone.[5,17] The aim of all these treatment is to increase the bone strength and reduce the rate of fractures as well as to prevent deformities.

CONCLUSION

OI is a congenital and heterogenous group of disorders characterized by increased bone fragility resulting from the abnormal collagen formation. The primary aim in the treatment of OI is to promote general and physical well being, reduce the risk of fractures and increase the bone mineral density. Medical treatment improves the patient's quality of life rather than complete cure. This case study may pave the way for further research in future to improve the treatment strategies and to resolve the complications of OI.

REFERENCE

1. Marta Rebelo, jandira Lima, Jose Diniz Vieira, Jose Nascimento Costa. An unusual presentation of osteogenesis imperfecta type I. International Medical Care Reports Journal, 2011; 4: 25-29.
2. Akinola P, Discs E, Adewole O. Osteogenesis Imperfecta: A report of two cases: The internet journal of Pediatrics and Neonatology., 2007; 8(2).
3. Engelbert RH, Prujs HE, Beemer FA, Helders PJ. Osteogenesis imperfecta in childhood: treatment strategies. Arch. Phys. Med. Rehabil. 1998; 79(12): 1590-4.
4. Glorieux F.H, Rauch F, Plotkin H, Ward L, Travers R, Lalice L, et al. Type V Osteogenesis Imperfecta: A New Form Of Brittle Bone Disorder. J Bone Miner Res., 2000; 15: 1650-1658.
5. Chevrel G. Osteogenesis imperfect.Orphanet Encyclopedia, june 2004. http://www.orpha.net/data/patho/GB/uk-OI.pdf.
6. Plotkkin Gibis H, Rauch F, Bishop N, Montpetit K, Ruck Gibis J, Travers R , et al. Pamidronate Treatment Of Severe Osteogenesis Imperfecta In Children Under 3 Years Of Age. J clin Endocrinol Metab, 2000; 85: 1846-1850.
7. Glorieux FH, Bishop NJ, Plotkin H, Chahot G, Lanoue G, Travers R. Cyclic administration of Pamidronate in children with severe osteogenesis imperfect. N Engl J Med., 1998; 339: 947-52.
8. Arwin Lamanna, Trevor Fayers, Sophie Clarke, William Parsonage. Valvular and aotic diseases on osteogenesis imperfect. Heart, Lung and Circulation. June 2013; 22(10).
9. Brailsford JF. Osteogenesis imperfect.Br J Radiol., 1943; 16: 129-36.
10. Stein D, kloster FE. Valvular Heart Disease in Osteogenesis Imperfecta. Pubmed., Nov 1977; 94(5): 637-41.
11. Elizabeth L Strevel, Alexandra Papaioannou, Jonathan D, Marty McNamara. Case report Osteogenesis Imperfecta.

12. Mundlos S, Chan D, Weng Y.M, Sillence DO, Cole WG et al. Multiexon deletions in the type I collagen COL 1A2 gene in Osteogenesis imperfecta Type 1 B. JBL Online, 1996; 271(15): 21068-21074.
13. Bryan DH. Inherited osteogenesis imperfect In; Nelson Textbook of Pediatrics.15th edition. Edited by Behrman, Kliegman, Arvin WB Scanders Philadelphia, USA, 1996; 1978-1980.
14. Zeitlan L, Rauch F, Plotkin H, Glorieux FH. Height and weight development during four years of therapy with cyclic intravenous pamidronate in children and adolscents with osteogenesis imperfect types I, III and IV. Pediatrics., 2003; 111: 1030-6.
15. Bishop N, Harrison R, Ahmed F, Shaw N, Eastell R, Camphell M, et al .A randomized controlled dose ranging of Risedronate in children with moderate and severe osteogenesis imperfect. J Bone Minor Res., 2010; 25: 32-40.
16. Watts NB, Diab DL. Long term use of Bisphosphonates in osteoporosis. J Clin Endocrinol Metab., 2010; 95(4): 1555-1565.
17. Antoniazzi F, Monti E, Venturi G, Franceshi R, Doro F, Gatti D, et al.GH in combination with bisphosphonate treatment in osteogenesis imperfect. Eur J Endocrinol., 2010; 163: 479-87.