OPEN ACCESS Freely available online

Journal of Bone Research

Opinion Article

Osteogenesis Imperfecta: Classifications and Treatment Processes

Peter Thompson*

Department of Bone and Osteogenesis Imperfecta, Kennedy Krieger Institute, Baltimore, USA

DESCRIPTION

A change in the human DNA sequence sometimes causes diseases, these types of diseases are called genetic disorder. The genetic disease may causes to single body part or the whole body. Osteogenesis Imperfecta is also a genetic disorder, this is mainly effect on bones. Osteogenesis Imperfecta means imperfect bone formation. People who have this disease their bones break easily with no apparent cause or for simple stresses. In accidental cases severe multiple fractures are happen. Sometimes only few fractures are happen in a person's lifetime. But, the bad condition is bone breaks occur before birth. It is a rare clinical disease, worldwide occurring rate is between 1/10,000 and 1/25,000 [1]. It is a connective tissue disease with abnormal type I collagen which is caused by mutation. The symptoms in this disease are beading of the ribs, blue sclerae, multiple fractures, deformity of the skull, osteoporosis and advancing deafness.

The classification of osteogenesis Imperfecta is initially divided into four types from type I to type IV by Sillence in 1979 [2]. The type I is manifested by osteoporosis, blue sclerae and multiple fractures with mildest level. In this type fractures are quite common in childhood period, but rare in adult period.

Type II osteogenesis Imperfecta is severe type most cases dying before, or shortly after, birth. Severe osteoporosis, poor mineralization, beading of ribs, shortening of the long bones, and multiple fractures occur in this type.

Type III is milder than type II, but the most serious type in which affected children survive past infancy. Blue sclerae are rare in this type, but the incidence of fracture and clinical severity increase with time and only rare cases survive into adulthood.

Type IV patients do not have blue sclerae, but the rest of their clinical symptoms are similar to type I. However shortening of the long bones is more obvious after adult-hood has been reached. The obscure changes in gene caused to the type V and type VI. Type VII is related to Cartilage Associated Protein (CRTAP) gene and type VIII is related to Leucine Proline-enriched Proteoglycan (LEPRE1) gene. Type VII patients tend to have brittle bones and skeletal abnormalities, and type VIII patients have defects in mineralization and growth.

The general physical examination of patient includes that examining the eyes, teeth, spine, rib cage and measuring the head circumference, length of limbs. The detection methods involved in finding osteogenesis Imperfecta disease are ultrasound method, collagen analysis and genetic testing and radiography of the uterus method. And some other testing techniques are X-ray, bone density test and bone biopsy. The ultrasound method is mostly used for type II and fewer cases for Type III. The Multiplex Ligation-dependent Probe Amplification (MLPA) was the superior and more reliable method for finding genetic abnormalities. Radiography of the uterus method is used when ultrasound and genetic testing are failed. Radiography of the uterus method can cause radiation injury.

This disease causes some clinical challenges those are abnormal blood coagulation, airways obstruction, abnormal cardiovascular structure and delayed wound healing. In osteogenesis Imperfecta patients the strength of the chest muscles is weak. Abnormal platelet count, increased vascular fragility and reduced clotting factor will happen. The weakness of cardiovascular structure tends to lower tolerance for surgery.

The treatment processes for osteogenesis Imperfecta patients are bisphosphonates and estrogen, braces and surgery, Stem cell transplantation method. Bisphosphonate therapy is most widely using medical treatment for OI. All the studies on bisphosphonate therapy state that it increases the Bone Mineral Density (BMD) in individuals. Some studies states that estrogen and neridronate can be used to treat infant in osteogenesis Imperfecta. Braces and surgery are used when the correct fixation surgery is necessary. But, the braces surgery gives less impact in osteogenesis Imperfecta. The newly developed approach for the management of osteogenesis Imperfecta is stem cell transplantation. Some scientists are transplanted mouse bone marrow mesenchymal stem cells into the femoral cavities of osteogenesis Imperfecta mice for testing [3]. Mostly recommended suggestion from doctors is intake adequate vitamin D, but how much vitamin D intake needed is not well established. To strengthen the long bones metal rods are inserted inside the bones, but for small bones this not well recommended. Spinal fusion is can be performed in some cases, and avoided in some cases because of neurons presence.

Correspondence to: Peter Thompson, Department of Bone and Osteogenesis Imperfecta, Kennedy Krieger Institute, Baltimore, USA, E-mail: thompson.peter55@yahoo.com

Received: 04-Jan-2022, Manuscript No. BMRJ-22-15877; **Editor assigned:**06-Jan-2022, PreQC No. BMRJ-22-15877 (PQ);**Reviewed:**19-Jan-2022, QC No. BMRJ-22-15877;**Revised:** 25-Jan-2022, Manuscript No. BMRJ-22-15877 (R); **Published:** 31-Jan-2022, DOI: 10.35248/2572-4916-22.10.157.

Citation: Thompson P (2022) Osteogenesis Imperfecta: Classifications and Treatment Processes. J Bone Res. 10:157.

The prevention of this type of genetic disorder diseases is only possible when the prevention is done at the first place of birth. Parental screening and parental diagnosis is important to give healthy birth to a child. This parental testing can prevent this type of genetic disorders.

REFERENCES

1. Starr SR, Roberts TT, Fischer PR. Osteogenesis imperfecta: Primary care. Pediatr Rev. 2010;31(8):54-64.
2. Sillence D, Senn A, Danks DM. Genetic heterogeneity in osteogenesis imperfecta. J Med Genet. 1979;16(2):101-116.
3. Li F, Wang X, Niyibizi C. Bone marrow stromal cells contribute to bone formation following infusion into femoral cavities of a mouse model of osteogenesis imperfecta. Bone. 2010;47(3):546-555.