Universe International Journal of Interdisciplinary Research
(Peer Reviewed Refereed Journal)
© UIJIR | ISSN (O) – 2582-6417
SEP. 2020 | Vol. 1 Issue 4
www.uijir.com

OSTEOGENESIS IMPERFECTA: MANAGEMENT ROLE AND ITS PROGNOSIS

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Abstract

Osteogenesisim perfect is a like heterogeneous rare connective tissue disease which usually caused due to mutatic changes in collagen type 1 gene. It is a globalized disease which affecting the both adults and children. The typical signs are blue sclera observed in patient. The treatment is decided on the basis of type of condition. Various diagnostic findings are used to identify the disease and on the basis of this the treatment is planned which consists various process like gene therapy, physiotherapy, medical treatment and various surgical procedures were implemented for the corrections of bone defects. These underlying processes found effective to rule out the problems.

Keywords *: Osteogenesis,Prognosis*

INTRODUCTION

Osteogenesis Imperfecta is a condition which consists group of different disorders which specifically affecting the bones. The term osteogenesis imperfecta revealed the imperfect bone development. This condition is affecting the bony region in which bone break easily due to very mild traumas to bones or with no apparent reasons or cause. The Osteogenesis imperfecta first defined by Mckusick in year 1956. It is found that approximately 25000 cases are found suffering from the disease in USA.

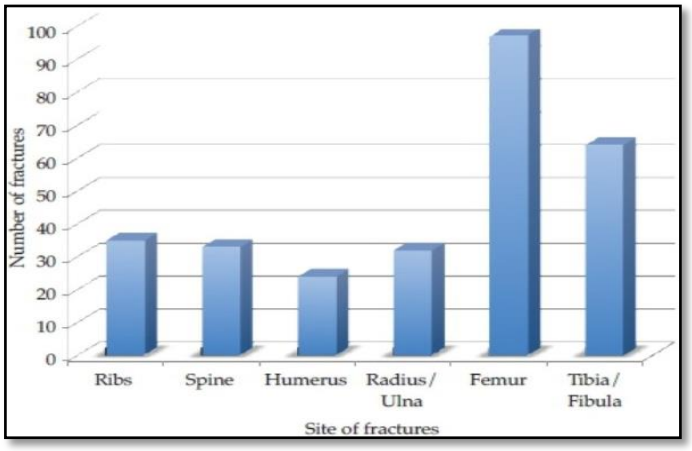

Fig 1: Presentation of fracture parts in various bones a bodydue toOsteogenesis Imperfecta

The symptoms of the disease may vary from mild to moderate or moderate to very severe condition. This disease also known as brittle bone disease. It is found that the basic cause is the mutation or changes in the genetic features. this disorder affecting the connective tissue with specific common characteristics of fragility of bony tissues. The basic cause leads to changes in collagen. The research revealed that there are above 150 mutations occurs in genes. The incidence ratio is 1:20,000 are found in all races.

2. CLASSIFICATION OF OSTEOGENESIS IMPERFECT a: This brittle bone disease or osteogenic imperfect a reflects various features which causes fragile skeletal structure. The variations or mutations in genetic structure COL1A1,COL1A2,CRTAP and P3h2 are leading to osteogenesis imperfect. The term Imperfect a itself expressed the imperfection of development of bone which leads to fracture, however severe condition might causes hearing loss, cardiac failures, and spinal bone problem or causes permanent damage.

Type	severity	Features	Inheritance
1.	Mild level of severity	Mild fragility features in bones, fractures, walking problems, deformities of bones, blue discoloration of sclera.	New changes or mutation, Autosomal dominant features.
2.	Lethal status of disease	Blue sclera features, severity in deformities, still birth, death of neonates, more number of fractures at intrauterine period.	New mutation features.
3	Severe deformity features	Normal status of sclera, dentinogenesis imperfect, frequently fractures episodes, neonates' death, and delivery after 20 weeks of gestation.	New changes in DNA sequence
4	Intermediate	Normal features of sclera, moderate status of deformities, short appearance of stature, possibility of deformities or dentinogenesis, imperfect condition	Variations in DNA sequence or new mutations.

3. CAUSES:

The basic causes of brittle Bone disease is due to a kind of defect occurs in gene which developing Type-1 collagen which is kind of protein consumed to create bony structure. The defected gene is usually inherited .it is found that one person in about 20,000 population were

found susceptible for brittle bone defect disease. The dominant mutatic changes is also basic cause of which happened on the gene structure COL1A2or either in COL1A2 gene. The recessive changes or mutations were found at LEPRE1 gene .the abnormal production of collagen leads to brittle features in bone structure. The collagen structure protein material is plays a crucial role for the production of cartilage, ligament structure, bones areas and skin.

4. SIGN AND SYMPTOMS

•The presence of muscle fatigue in body is found.

•The curvature of the spinal cord is changed or development of scoliosis is appeared in body.

•The appearance of the limbs become curved.

•The sclera of eyes looks blue or tint grey.

•The presentation of ears expressed the presentation of hearing loss later in adulthood age which is 20s and 30s age time period.

•The hearing loss is occurred due to inner ear deformities development and other three small bones also deformed which were present in the middle area of ear bone.

•The brittle bone feature and underdeveloped teeth.

•The bluish discoloration in osteogenesis imperfect patient appears easily.

•Constipation problems.

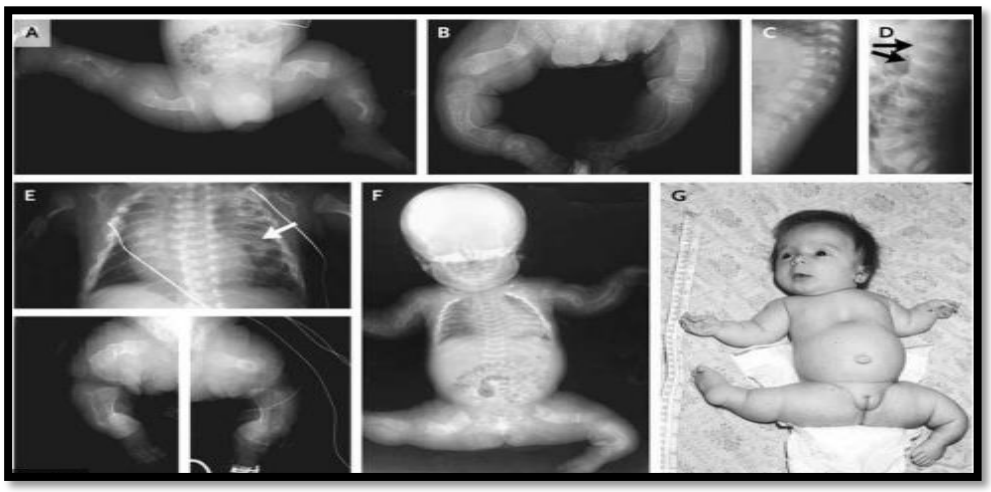

Fig2: The Clinical presentation of Osteogenesis Imperfect

•Heart defects

•Delayed motor development and activities

•The features of laxity in ligaments were observed.

•The liability of fracture is present.

•Poor development of teeth

•Flat feet presentation

•Poor development of teeth

•Presence of hypermobility due to features of loose joints

•Short stature and height appearance.

•Weakened bones because of the level of severity.

•Barrel shaped rib structure features appeared in type 3.

•Curve spine is found in type three and four.

•Triangular face appearance

5. DIAGNOSTIC EVALUATIONS

•X-Ray of the upper extremities areas like anteroposterior or lateral areas.

•Blood examination in which Vitamin-D, calcium level and phosphate level in blood stream

•DNA- Test:COL1A1,COL1A2 analysis must be done for gene defect.

•Collagen biochemical analysis.

•Complete blood analysis.

•CT SCAN AND MRI.

•Physical examination of the sclera part.

•Antenatal analysis of gene mutation in risk pregnancies of OI is done for the evaluation of uncultured chorionic villus cells structure.

•Positive family history must take.

•Antenatal Ultra sonography.

6. Management of osteogenesis imperfect:

Osteogenesis imperfect management process is designed for better prevention and control the disease situation which will be vary from person to person. The prior interventions are the good steps to rule out the disease and to implement the optimal standard of life and gain good prognosis of the disease. The management or the treatment process consists various process to overcome the disease.

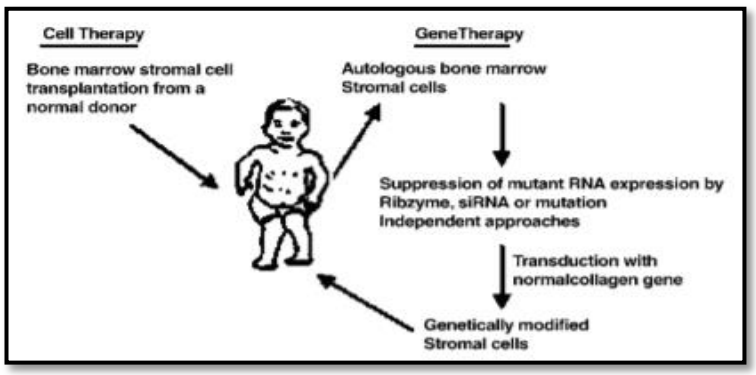

Fig3:Cell and Gene Therapy for Treatment

•**Fractured Bone Care:**The application of plaster cast,bracing the broken bones might found supporting. This process is very helpful to immobilize the bony broken areas to gain fusion. Becauseof this implementation sometimes bones becomes weak due to placing in one position for long duration. The health promotors were trying best to maintain the balance for both maintaining the healing of fractured area of bone and proper maintenance of bone strength.

•**Application of the Physical Therapy**:The implementation of the physical therapy treatment main objective is to maintain the functioning standards in all aspects of the life status as much as possible. In part of this treatment a fusion program which includes muscles strengthening along with aerobics conditioning. Numerous kids diagnosed for osteogenesis imperfecta are found with delayed mobility skills due to weakened muscles. Apackage of physical rehabilitation program consists deltoid muscles strength increment,biceps areas and much more the crucial part of lower muscles like gluteus Maximus,gluteus medius and also the area of trunk extensors. If these muscles found strong,kids can move their hands and legs independently in all directions against gravitational force.

•**Bracing Application:**For the patients of Osteogenesisimperfecta the application of braces or wearing in lower extremities found supportive for the weak muscles part, found effective for reducing pain and maintain the joints proper alignment. Braces application might be helpful or allowing patient to function activities more easily.

•**Surgical Interventions:**The surgical procedures are applied and are found the last step if

found necessary if other treatments are found not effective. In surgical procedure for the bony areas corrections the deformities like scoliosis and basilar invagination surgery is implemented. The Roding placement in which metal rods are imparted in long bone part of legs. These applications of rods may provide strength to legs and also prevent the fracture.

•**Medical Treatment**: The bisphosphates are the line of drugs utilized for treatment of osteoposrosis. These drugs might found effective particularly in pediatric patients. These drugs do not grow new cells but slow down the bone degeneration process, and also reduce vertebral compressions. But in part of motor skills no effective role is found of drug or to reducing pain in

•**Treatment for Related Problems:** Some other related conditions also found noticed in body of kid which specifically occurs due to osteogenesis imperfect:

•Implications of hearing Aids for hearing loss.

•Crown applications and dental devices are found useful for brittle dentures.

•Bone marrow transplantation is also evaluated as

•If any respiratory problems develop the administrations of oxygen is done.

7. Prognosis:

The outlook for the patients of OI are very for each type of diseases condition many children who were born with type-I OI were lived a normal healthy life as they proceed into adulthood. Very less severe symptoms may not affecting the live conditions the patients with type-III OI were significantly a reduce level of life patency as compare to general people. The notable access of death ratio for pediatric patients who were found less than 10 years of age due to pulmonary disease conditions. In part of cardiac condition the cause of death OI in patients diagnosed with type-III OI were facing the related problem of altered anatomical, thoracic structure and development of kyphoscoliosis. Large number of death were identified due to complications were occurs as occurrence of disease. Pediatric patients with OI had ratio of access mortality due to pulmonary condition and gastrointestinal disease and an increased risk of death due to trauma. Therefore, it is noticed that the modern pattern of treatment for pediatric patients diagnosed with OI were further result better outcomes and long life expectancy.

• The expectancy or the condition of patients depends on OI from which they are suffering:

• Type-I or may be mild OI is usually found the most common form of disease in which the patients live normal life condition.

• In part of Type-II is a severe condition which might often leading to death in the first year of the life.

• The Type-III picture showed a severe scenario of OI. In this form the patients might facing may fracture occurs in early stage of life and become more severe deformities of bones. Due to this situations patient is a need of use of wheelchair and short life status.

• The Type-IV is very likely same as Type-I features. The implications of braces or crutches were used for walking and have normal life status.

8. Conclusion: The overall out revealed that OI is a fatal disease which causes high ratio of morbidity and physical disability. In this hereditary disease the roll of occupational therapist might found effective but not much more. OI cannot be fully cured. The treatment helps to concur the problems as much as possible to improve the quality of life. The mutation in the collagen A1/A2were identified recent years. In various current researches and studies this disease may play a leading role or development of various bone diseasesor deformities. The collagen defects found very commonly which affecting the various internal parts of body and developing the secondary problems in the body. The picture of treatment is still vary from patient to patient. Still now also scientist are studying or investigating for the promising treatment for Osteogenesis imperfect.

References:

1)https://www.researchgate.net/publication/224920054_Hearing_Loss_in_Osteogenesis_Imperfecta_Characteristics_and_Treatment_Considerations

2)https://www.researchgate.net/publication/8918099_Gene_therapy_approaches_for_osteogenesis_imperfect

3)https://www.researchgate.net/figure/Expanded-Sillence-classification-of-OI_tbl1_51493605

4)https://www.slideshare.net/chaudharymahesh/osteogenesis-imperfecta-dr-mahesh

5)https://www.nature.com/articles/3302199

6) https://www.slideshare.net/kharr/osteogenesis-imperfecta-oi