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RESEARCH ARTICLE

OSTEOGENESIS IMPERFECTA (IO)

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ABSTRACT

Osteogenesis imperfecta (OI) is a hereditary osteoporotic syndrome, characterized by multiple fractures due to osteoporosis and excessive bone fragility. It is also known as brittle bone disease or Lobstein syndrome. Osteogenesis imperfetca congenita is a severe form or fatal type. It is caused by deficiency of type I collagen. Most cases are caused by mutations in the COL1A1 and COL1A2 genes. It is manifested with hearing loss Brittle teeth, Short stature, spinal curvature and sometimes barrel-shaped rib cage, Triangular, Loose joints (double jointed), Poor muscle tone in arms and legs. Medication, Immobilization of affected part and exercise are important to decrease the risk of future fractures.

INTRODUCTION

Osteogenesis imperfecta (OI) is a hereditary osteoporotic syndrome, characterized by multiple fractures due to osteoporosis and excessive bone fragility[1]. It is also known as brittle bone disease or Lobstein syndrome. Osteogenesis imperfetca congenita is a severe form or fatal type. People with ontogenesis imperfecta have a genetic defect that impairs the body's ability to make strong bones.

Definition

Osteogenesis imperfect (OI) is a hereditary osteoporotic syndrome, characterized by multiple fractures due to osteoporosis and excessive bone fragility.

Causes

It is caused by deficiency of type I collagen. Most cases are caused by mutations in the COL1A1 and COL1A2 genes[2]. In most cases of OI, children inherit the defective gene from one of their parents. In some children, neither parent has osteogenesis imperfecta. In these cases, the genetic defect is a spontaneous mutation (change) in the gene, and it stops functioning correctly.

Types

Type I Osteogenesis Imperfecta

It is a autosomal dominant disorder. It is a is the most common and mildest type of this disease. While the structure of the collagen is normal, there is less collagen than there should be. There is little or no bone deformity. It is manifested with:

- Bones fracture easily
- Slight spinal curvature
- Loose joints
- Poor muscle tone
- Discoloration of the sclera (whites of the eyes)

Type II Osteogenesis Imperfecta

It is the most severe form of the disease. The collagen does not form properly. Many infants with type II osteogenesis imperfecta do not survive. It is manifested with:

- Most cases die within the first year of life due to respiratory failure or intracerebral hemorrhage
- Severe respiratory problems due to underdeveloped lungs
- Severe bone deformity and small stature

Type III Osteogenesis Imperfecta

Type III osteogenesis imperfecta also has improperly formed collagen and often severe bone deformities, plus additional complications. It is manifested with:

- Short stature, spinal curvature and sometimes barrel-shaped rib cage
- Triangular face
- Loose joints (double jointed)
- The infant is often born with fractures.

- The whites of the eyes may be white, blue, purple, or gray[3].
- Spinal deformities, respiratory complications, and brittle teeth.
- Short stature, spinal curvature and sometimes barrel-shaped rib cage

Type IV Osteogenesis Imperfecta

It is autosomal dominant disorder. Collagen quantity is sufficient but is not of a high enough quality. It is manifested with,

- Less blue sclera
- Opalescent dentin
- Bones fracture easily, especially before puberty
- Short stature, spinal curvature and barrel-shaped rib cage
- Bone deformity is mild to moderate
- Early loss of hearing

Symptoms and Signs

- Hearing loss
- Brittle teeth
- Short stature, spinal curvature and sometimes barrel-shaped rib cage
- Triangular face
- Loose joints (double jointed)
- Poor muscle tone in arms and legs
- Discolouration of the sclera
- Loose joints
- Triangular-shaped face
- Breathing problems
- Bone deformities in both shinbones

Diagnosis

- Medical History and Physical Examination
- X-rays, blood or tissue samples for genetic testing.
- Ultrasound

Treatment

- Treatment is individualized and depends on the severity of the disease and the age of the patient. Medication. Medical bisphosphonates, these medications must be administered by properly trained doctors and require close monitoring.
- Immobilization. Casting, bracing, or splinting fractures is necessary to keep the bones still and in line[4].
- Exercise. After a fracture, movement and weight bearing are encouraged as soon as the bone has healed. Specific exercises will increase mobility and decrease the risk of future fractures.
- Nursing care on firm mattress and pillows, supervision of activities of daily living and prompt immobilization of fractures are essential measures
- Surgical Treatment
- Rodding
- Spinal fusion for scoliosis.

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