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Osteogenesis imperfecta (Brittle bone disease)

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Abstract
Osteogenesis imperfecta (OI) is an inherited (Genetic) bone disorder that is present at birth. It is also known as brittle bone disease. A child born with OI may have soft bones that break (Fracture) easily, bones that are not formed normally, and other problems. OI encompasses a group of mostly autosomal dominant hereditary alterations that are caused by several mutations in one of the two genes encoding the alpha chains, i.e., COL1A1 and COL1A2, of type 1 collagen. Bone fragility is conditioned by the quantity and quality of abnormal structural collagen protein. Therefore, bone deformities or fractures at minimal trauma are expected in patients with the disease. OI presentation varies from the lethal forms of intrauterine fractures to late cases in which fractures start in adolescence or even adulthood.

Keywords: Osteogenesis imperfect, autosomal dominant hereditary, collagen, collagen protein

Introduction
Osteogenesis imperfecta (OI) or brittle bone disease is a rare genetic disorder occurring in 1 in 15,000 to 20,000 births and is characterized by bone fragility and osteopenia [1]. Brittle bone disease (Osteogenesis imperfecta) is an inherited genetic condition that causes bone weakness [2].
This defect is caused by dominant or recessive mutations that lead to bone fragility and other skeletal manifestations, such as short stature and bone deformities. Extra skeletal tissues and organs can also be involved [3].
Osteogenesis imperfecta (OI) is a rare genetic condition characterized by increased bone fragility. Recurrent fractures, pain and fatigue have a considerable impact on many aspects of the life of a person affected with OI and their families [4].

Types [5]

Type I	Mildest and most common type. About 50% of all affected children have this type. There are few fractures and deformities
Type II	Most severe type. A baby has very short arms and legs, a small chest, and soft skull. He or she may be born with fractured bones. He or she may also have a low birth weight and lungs that are not well developed. A baby with type II OI usually dies within weeks of birth
Type III	Most severe type in babies who don't die as newborns. At birth, a baby may have slightly shorter arms and legs than normal and arm, leg, and rib fractures. A baby may also have a larger than normal head, a triangle-shaped face, a deformed chest and spine, and breathing and swallowing problems. These symptoms are different in each baby.
Type IV	Symptoms are between mild and severe. A baby with type IV may be diagnosed at birth. He or she may not have any fractures until crawling or walking. The bones of the arms and legs may not be straight. He or she may not grow normally.
Type V	Similar to type IV. Symptoms may be medium to severe. It is common to have enlarged thickened areas (hypertrophic calluses) in the areas where large bones are fractured
Type VI	Very rare. Symptoms are medium. Similar to type IV.
Type VII	May be like type IV or type II. It is common to have shorter than normal height. Also common to have shorter than normal upper arm and thighbones.
Type VIII	Similar to types II and III. Very soft bones and severe growth problems.

Etiology: It happens because of a mutation in the gene that makes the protein collagen. Collagen is an important building block of bones.

Children may inherit the mutation from a parent or it happens early in pregnancy when the baby is first forming. This makes their bones weaker and more brittle than normal bones. It also can lead to abnormally shaped bones [6].

Clinical Manifestation

Brittle bone disease has one or more of the following symptoms:

- Bone deformities
- Multiple broken bones
- Loose joints
- Weak teeth
- Blue sclera, or a bluish color in the white of the eye
- Bowed legs and arms
- Kyphosis, or an abnormal outward curve of the upper spine
- Scoliosis, or an abnormal lateral curve of the spine
- Early hearing loss
- Respiratory problems
- Heart defects [7]

Diagnostic Evaluation

Diagnosis is typically based on medical imaging, including plain X-rays, and symptoms.

An OI diagnosis can be confirmed through DNA or collagen protein analysis, but in many cases, the occurrence of bone fractures with little trauma and the presence of other clinical features such as blue sclerae are sufficient for a diagnosis.

A skin biopsy can be performed to determine the structure and quantity of type I collagen.

OI type II is often diagnosed by ultrasound during pregnancy, where already multiple fractures and other characteristic features may be visible.

OI can also be detected before birth by using an *in vitro* genetic testing technique such as amniocentesis [8].

Genetic testing

In order to determine whether osteogenesis imperfecta is present, genetic sequencing of the most common problematic genes, COL1A1, COL1A2, and IFITM5, may be done; if no mutation is found yet OI is still suspected [8].

Medical Management

Biphosphonates: Medical treatments with bisphosphonates are currently used as standard therapy in patients with a moderate or severe course in childhood and adolescence. Upon administration bisphosphonates bind to the hydroxyapatite crystals of the bone, which are resorbed by osteoclasts during bone remodeling and induce their apoptosis. These drugs effectively reduce bone resorption and thereby increase bone mass. Different bisphosphonates (Pamidronate, neridronate, zoledronate) have also been used and differ in treatment intervals. Oral bisphosphonates are less effective but can also be used in special indications [9, 10].

Surgical Management [11]

Intramedullary rod replacement	In patients with bowed long bones, intramedullary rod replacement may improve weight bearing and, thus, enable the child to walk at an earlier stage than he or she might otherwise.
Surgery for basilar impression	This procedure is reserved for cases with neurologic deficiencies, especially those caused by compression of brain stem
Correction of scoliosis	Correction of scoliosis may be difficult because of bone fragility, but spinal fusion injury may be beneficial in patients with severe disease.

Nursing Management [12]

Nursing Assessment	**History**: Assess the patient's medical history as osteogenesis imperfecta is a genetic disorder. **Physical assessment**: Fracture is a common occurrence in a patient with osteogenesis imperfecta and symptoms can be detected in a physical exam. **Laboratory values**: Laboratory results may reveal the occurrence of osteogenesis imperfecta.
Nursing Diagnosis	Risk for injury related to fragile bones. Impaired dentition related to genetic predisposition. Impaired physical mobility related to loss of integrity of bone structures.
Nursing Care Planning & Goals	Modify environment as indicated to enhance safety. Be free of injury. Display healthy teeth in good repair. Verbalize and demonstrate effective dental hygiene skills. Follow through on referrals for appropriate dental care. Increase strength and function of affected and/or compensatory body part.
Nursing Interventions	**Genetic counseling:** Offer genetic counseling to the parents of a child with osteogenesis imperfect. **Diet:** Encourage adequate calcium, vitamin D, and phosphorus intake, and ensure appropriate caloric management. **Activity:** Educate parents regarding positioning of the child in the crib and how to handle the child while avoiding fractures.
Evaluation	Modified environment as indicated to enhance safety. Free of injury. Displayed healthy teeth in good repair. Verbalized and demonstrated effective dental hygiene skills. Followed through on referrals for appropriate dental care. Increased strength and function of affected and/or compensatory body part.
Discharge and Home Care Guidelines	Physical therapy. Therapy should be directed toward improving joint mobility and developing muscle strength. Nutrition. Periodic nutritional evaluation and intervention should be implemented. Oral health. Patients with osteogenesis imperfecta require scrupulous oral hygiene and frequent follow up with a pediatric dentist who is familiar with the disorder.

Documentation Guidelines	Individual risk factors, noting current physical findings. Availability and use of resources. Individual factors influencing dentition problems. Description of oral cavity and structures. Level of function, ability to participate in specific or desired activities. Plan of care. Teaching plan. Individual responses to interventions, teaching, and actions performed. Attainment or progress toward desired outcomes. Modifications to plan of care.

Conclusion

Osteogenesis imperfecta also known as Brittle Bone Disease, is a genetic disorder caused by lack of collagen protein in the bone which cause bones to be more susceptible to fractures. For detecting this, amniocentesis is done during pregnancy stage and genetic study or x-ray should be done in childhood and adolescence age. It is treated by bisphosphonates. This disease is corrected by surgically rodding procedure.

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