Osteogenesis imperfecta bone

Osteogenesis Imperfecta or more commonly known as Brittle Bone Disease, is a

condition causing extremely fragile bones and is known to be a congenital disease. This means

you are born with it. It is caused by a defect in the gene that produces type I collagen which is an

important building block of bone. Most causes of Osteogenesis Imperfecta are inherited from a

parent caring the gene . Some cases are the result of new genetic mutations. People with

Osteogenesis Imperfecta are usually below average hight. Osteogenesis Imperfecta does not

seem to occur in any certain race, it effects all races male and or female. A person with

Osteogenesis Imperfecta has a 50 per cent chance of passing on the gene and the disease to there

offspring.

The symptoms of Osteogenesis Imperfecta are as follows:

Type I which accounts for 60 per cent of all cases

- Most common and mildest type of Osteogenesis Imperfecta

- Bones predisposed to fractures. Most occurring before puberty
- Normal to near-normal stature
- loose joints with low muscle tone
- Bone deformity absent or minimal
- Spinal curvature
- Collagen structure is normal, but the amount is less than normal
- Triangular shaped face
- Sclera (whites of the eyes) usually have a blue, grey or purple tint to them
- Brittle teeth
- Hearing loss
- The increase liability to bruising (thought to be due to the defective collagen)
- Hernias are more common in people with Osteogenesis Imperfecta
- Excessive sweating or intolerance of heat are common complaints, the cause is unknown

Type II

- Most sever form
- Death occurring shortly after birth, often due to respiratory problems
- Small stature with underdeveloped lungs
- Collagen is improperly formed

- Numerous fractures
- Severe bone deformities

Type III

- Bones fracture easily
- Fractures often present at birth
- Short stature
- Sclera (whites of eyes) to be a blue, grey, or purple tint
- Loose joints
- Poor muscle development in arms and legs
- Triangular shaped face
- Barrel-shaped rib cage
- Spinal curvature
- Bone deformity, often severe
- Brittle teeth
- Hearing loss
- Respiratory problems
- Collagen is improperly formed
- The increase liability to bruising (thought to be due to the defective collagen)
- Hernias are more common in people with Osteogenesis Imperfecta

- Excessive sweating or intolerance of heat are common complaints, the cause is unknown

Type IV

- Between Type I and Type III in severity
- Bones fracture easily, usually before puberty
- Shorter than average stature
- Sclera are white or near white, normal in color
- Mild to moderate bone deformity
- Barrel-shaped rib cage
- Triangular shaped face
- Spinal curvature
- Brittle teeth
- Hearing loss
- Collagen is improperly formed
- The increase liability to bruising (thought to be due to the defective collagen)
- Hernias are more common in people with Osteogenesis Imperfecta
- Excessive sweating or intolerance of heat are common complaints, the cause is unknown

The diagnoses of Osteogenesis Imperfecta.

Osteogenesis Imperfecta is diagnosed a few different ways. In most cases the diagnosis is

made from the pattern of fractures. In severely affected people X-rays may show characteristics

abnormalities. In the USA two specialized tests are sometimes used for the diagnosis of

Osteogenesis Imperfecta. One involves taking a small piece of the skin, culturing the cells and

chemically examining the collagen produced. The other uses a blood sample and searches for

mutations of the genes coding for the collagen of bone. Neither tests are more than 85 per cent

accurate in identifying cases of Osteogenesis Imperfecta. Chronic Villus Sampling maybe done

during pregnancy to determine if the fetus has the condition. However, because so many different

mutations can cause Osteogenesis Imperfecta, some forms can not be diagnosed with a genetic

test. Often the severe form Type II can be detected on an ultra sound when the fetus is as young

as sixteen weeks old.

The treatment of Osteogenesis Imperfecta.

There is no cure yet for this disease however curtain therapies can reduce pain and complications

due to this disease. Bisphosphonates are drugs that have been used to treat Osteoporosis. They

can increase the strength and the hardness of bone in people with Osteogenesis Imperfecta . They

have also been shown to highly reduce fracture rate. Swimming and low impact exercises help

maintain the strength of the bones. In more severe causes surgery may be used to place metal

rods into long bone in the leg to help reduce the risk of any further fractures.