What Is Osteogenesis Imperfecta?
Osteogenesis imperfecta (OI) is a genetic disorder characterized by bones that break easily, often from little or no apparent trauma. OI is highly variable, and signs and symptoms range from mild to severe. In addition to broken bones (fractures), people with OI sometimes have muscle weakness or joint laxity (loose joints). They often have skeletal deformities including short stature, scoliosis (curvature of the spine) and bowing of long bones. A classification system featuring different types of OI is commonly used to help describe how severely a person with OI is affected. OI occurs with equal frequency in males and females and among all ethnic and racial groups. OI is caused by genetic defects that affect the body’s production of type I collagen. Collagen is the major protein of the body’s connective tissue. It can be compared to the framework around which a building is constructed. In classical (dominant) OI, a person has too little type I collagen or poor quality type I collagen. In recessive OI, mutations in other genes interfere with collagen production. The result in all cases is weak bones that break easily.

What Is Osteoporosis?
Osteoporosis is a condition in which the bones become less dense and more likely to break. Fractures (broken bones) from osteoporosis can result in significant pain and disability. It is a major health threat for an estimated 44 million Americans, 68 percent of whom are women.

Risk factors for developing osteoporosis include:
- having a family history of the disease
- thinness or a small body frame
- for women, being postmenopausal, having early menopause or not having menstrual periods (amenorrhea)
- using certain medications, such as glucocorticoids
- not getting enough calcium
- not getting enough physical activity
- smoking
- drinking too much alcohol

Osteoporosis is a silent disease until a fracture occurs. It can often be prevented, however, if undetected, it can worsen over many years without symptoms. It has been called “a pediatric disease with geriatric consequences,” because building healthy bones in one’s youth is important to help prevent osteoporosis and broken bones later in life.

The OI – Osteoporosis Link
The term osteoporosis describes bone loss that is extensive enough to increase the risk of fracture. The term is a general one; not related to any specific cause for the bone loss. In fact, there are many causes of osteoporosis. Almost all people with OI have osteoporosis, because they do not develop appropriate bone mass at any age. Women and men with OI can experience additional bone loss, such as age-related bone loss, superimposed on OI. Symptoms of additional bone loss may appear at a younger age than commonly seen in people who do not have OI. When women and men with osteogenesis imperfecta become middle-aged, their risk of breaking bones more easily increases due to the combination of abnormal bone structure and factors that are not limited to OI. These factors include a diet low in calcium and vitamin D, smoking, decreased activity, and decreased levels of protective sex hormones (testosterone, estrogen, etc.). Lower bone density can lead to a return to the fracture cycles they experienced as children.
Osteoporosis Management Strategies
Strategies for prevention and treatment of osteoporosis in people with osteogenesis imperfecta are generally the same as those for people who do not have OI.

**Nutrition:** For healthy bones, it is important to have a diet with appropriate levels of calcium and vitamin D that are appropriate for the person’s size. Good sources of calcium include low-fat dairy products and calcium fortified foods and beverages. Supplements can help ensure that the daily calcium requirement is met when there is significant deficiency and urine calcium levels are not elevated. Vitamin D plays an important role in calcium absorption and bone health. It is made in the skin through exposure to sunlight. Vitamin D supplements may be needed if fortified foods are not part of the diet.

**Exercise:** Like muscle, bone is living tissue that responds to exercise by becoming stronger. Weight-bearing exercises that work against gravity are best for preventing bone loss. Some examples include walking, standing, and lifting. Swimming can also be a beneficial form of exercise. Because water activities do little to increase or maintain bone density, however, people with OI should also try to add walking or other weight-bearing exercise to their physical activity program if possible. Exercise can be challenging for people with OI who must cope with muscle weakness, bone fragility and deformity, joint instability and pain. However, regular exercise in moderation, such as walking, can help prevent bone loss and provide other health benefits. All adults, including those who spend most of their time in wheelchairs, need to consult with their doctor and a physical therapist about developing an appropriate exercise program. People with OI should only perform exercise that can be done safely and is well tolerated.

**Healthy lifestyle:** Smoking has a negative effect on all body systems including bones. Alcohol consumption in excess can also negatively affect bone health and predispose to falls and fracture.

**Bone density test:** Bone mineral density (BMD) tests measure bone density in various sites of the body. BMD measurements are often reported in terms of peak bone mass in young adults. However, people with OI, because of short stature, curvature of the spine, past vertebral fractures or the presence of metal rodding, may not be able to get an accurate reading. Almost all adults who have OI have low BMD. However, a base-line measurement in early middle age can be an important part of health management for men and women with OI, since that will give them a personal standard to compare to and determine whether their bone density is changing over time or as the result of a treatment.

**Medication:** There is no cure for osteoporosis. However, there are medications available to prevent bone loss, to increase bone mass and to treat the disease, which seem to be having success with women and men who have OI. Consult with your doctor to determine which medication is right for you. These medications usually require long-term use.