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, people with OI sometimes have muscle weakness or joint laxity (loose joints), and they often have skeletal malformations 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 and can be likened to the framework around which a building is constructed. In dominant (classical) OI, a person has too little type I collagen or a poor quality of type I collagen due to a mutation in one of the type I collagen genes. 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. In the United States, more than 40 million people either already have osteoporosis or are at high risk due to low bone mass.

Risk factors for developing osteoporosis include:

- Thinness or small frame.
- Family history of the disease.
- Being postmenopausal and particularly having had early menopause.
- Abnormal absence of menstrual periods (amenorrhea).
- Prolonged use of certain medications, such as those used to treat lupus, asthma, thyroid deficiencies, and seizures.
- Low calcium intake.
- Lack of physical activity.
• Smoking.
• Excessive alcohol intake.

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

The link between OI and osteoporosis

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. Nearly 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 a background of 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 OI reach middle-age, their risk of breaking bones more easily increases even further. Factors such as a diet low in calcium and vitamin D, smoking, decreased activity, and decreased levels of protective sex hormones (e.g., testosterone, estrogen) can compromise bone density and 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 levels of calcium and vitamin D that are appropriate for the person’s size. Good sources of calcium include reduced-fat dairy products and calcium-fortified foods and beverages. When a person has a significant calcium deficiency, supplements can help ensure that the daily calcium requirement is met, assuming that 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 foods fortified with vitamin D are not part of the diet.

Exercise. Like muscle, bone is living tissue that responds to exercise by becoming stronger. Weight-bearing and resistance exercises are best for preventing bone loss. Weight-bearing exercises force you to work against gravity. They include walking, standing, and dancing. Resistance exercises – such as lifting weights – can also strengthen bones. Swimming can also be a beneficial form of exercise. Because water activities do little to increase or maintain bone density, however, people with OI also should 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 malformations, 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.

Healthy lifestyle. Smoking has a negative effect on all body systems, including bones. Excess alcohol consumption also can have negative effects on bone health and predispose a person to falls and fractures.

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. Nearly all adults who have OI have low BMD.
Medication. There is no cure for osteoporosis. However, medications are available to prevent bone loss, increase bone mass, and treat the disease. Women and men who have OI seem to be using these medications successfully. Consult with your doctor to determine which medication is right for you. Many of these medications usually require long-term use.

Resources
For more information on osteogenesis imperfecta, contact the:
NIH Osteoporosis and Related Bone Diseases National Resource Center
Website: https://www.bones.nih.gov

Osteogenesis Imperfecta Foundation
Website: http://www.oif.org

If you need more information about available resources in your language or another language, please visit our website or contact the NIH Osteoporosis and Related Bone Diseases ~ National Resource Center.

For your information
This publication contains information about medications used to treat the health condition discussed here. When this publication was developed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

For updates and for any questions about any medications you are taking, please contact the U.S. Food and Drug Administration (FDA) toll free at 888-INFO-FDA (463-6332) or visit its website at https://www.fda.gov. For additional information on specific medications, visit Drugs@FDA at https://www.accessdata.fda.gov/scripts/cder/daf. Drugs@FDA is a searchable catalog of FDA-approved drug products.

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