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# OI Issues: Maintaining Health During the Adult Years

## Introduction

Osteogenesis imperfecta (OI) is a genetic disorder characterized by bones that break easily, often from little or no apparent trauma. It is caused by a genetic defect that affects the body's production of collagen, which is the major protein in connective tissue. Although most adults with OI experience a decrease in the rate of fractures (broken bones) after puberty, other medical problems, some of which are related to the basic genetic defect that causes OI, may require more attention.

For example, adults with OI should be concerned about weight gain, diabetes, cholesterol, and other cardiovascular problems such as high blood pressure (hypertension). Tendon, muscle, and joint problems may be aggravated with time, and hearing loss may become significant. Vigorous and consistent medical care for OI remains as important in adulthood as it is during childhood.

Adults with OI not only need to manage all of the same health issues as other adults, but also must cope with the musculoskeletal concerns associated with OI.

## The Health Care Team

An important part of managing OI and staying healthy is assembling a good health care team and having a solid working relationship with one's primary care doctor and medical specialists. The medical team may include an orthopaedist and an endocrinologist (doctors who specialize in diagnosing and treating conditions affecting bones and the endocrine system, respectively). The team also may

also include a nutritionist as well as rehabilitation professionals. Consultation with a pulmonologist (a doctor who specializes in lung diseases) and a neurologist (a doctor who specializes in caring for the nervous system) also may be needed.

Strategies for developing strong relationships with your health care team include keeping good records, keeping up to date on new information about OI and available treatments, and planning ahead for emergencies. Being a good health care consumer involves providing the doctor with accurate information about your health, listening carefully to the doctor's instructions, and asking questions until the information is fully understood.

People with OI who are small in stature may want to suggest that their doctors obtain a copy of a pediatric dose chart to keep in their medical file for reference. A pediatric blood pressure cuff and smaller instruments for gynecological exams also may be appropriate. Those with OI may want to provide any new doctor with a list of all rods or other surgical implants, because these may interfere with x rays or magnetic resonance imaging (MRI). In many cases, MRI exams can be performed in the presence of rods.

## **Health Concerns Related to OI**

**Bone density and osteoporosis.** Maintaining bone mass is a priority for adults with OI because fracture risk, in part, is related to bone density.

A bone mineral density (BMD) test is the best way to determine your bone health. BMD tests can identify osteoporosis, determine your risk for fractures, and measure your response to osteoporosis treatment. The most widely recognized BMD test is called a dual-energy x-ray absorptiometry (DXA) test. The test is painless—a bit like having an x ray, but with much less exposure to radiation—and can measure bone density in the hip and spine.

An initial DXA test may be obtained at any age and then yearly thereafter. If possible, the same DXA scanner should be used each year to avoid variations in test results caused by different equipment. Bone density can decline as a direct result of OI, from immobilization associated with casts or limited weight-bearing activity, and from age-related changes in bone and the endocrine (hormone) system.

Treatments may include calcium and vitamin D supplements (if the diet is inadequate), drug therapies including oral or intravenous bisphosphonates, diet, and exercise. Smoking, overuse of alcohol, and certain medications, including cortisone-like steroids, also can have a negative impact on bone health.

**Musculoskeletal and joint problems.** Adults often report pain in their lower back and hips. This can be the result of compression fractures of the spine, scoliosis (curvature of the spine), or joint deterioration. Other problems can include fractures that have failed to heal (nonunion fractures) and low muscle strength. Knee pain and ankle instability also are frequent complaints in people with OI. Exercise, orthotic devices, or braces to improve hip, knee, and ankle alignment and back or joint surgery may provide relief.

Many individuals have joint laxity (loose joints) or excessive joint flexibility. This is particularly a problem for the knees and ankle joints, which are subject to pressure over the years. Also, leg lengths may differ because of a history of previous fractures. Rolling in of the ankle joint is another common problem. Heel lifts and firm ankle supports are important in limiting wear and tear and improving gait.

Orthotic devices may help provide stability for lax joints of the knees, feet, and ankles. Joint replacement surgery may be a treatment option for some, but not all, adults with OI who have joint problems.

**Chronic pain.** Adults may experience pain from old fractures or compression fractures of the spine related to either OI or osteoporosis. Unstable joints may increase degenerative changes, which are the source of pain in many individuals. Pain management may include lifestyle adjustments to protect the spine, medications, and alternative treatments such as acupuncture. Adults should be wary of increasing the strength of pain medication to the point where it represses breathing or reduces consciousness. Reduced consciousness can increase the risk of falls and fractures.

**Pulmonary function.** Breathing problems are the main concern of many adults with OI, particularly those with Type III and Type IV OI and those with significant scoliosis (curvature of the spine). Decreased chest volume, chronic bronchitis, and asthma can lead to restrictive pulmonary disorder (a reduction of lung capacity).

Rib fractures and muscle weakness also may contribute to the problem. Sleep apnea, a related problem for some adults with OI, can be determined with an overnight sleep test. During the sleep test, blood gases also can be measured for use in guiding future treatment.

Exercise to promote deep breathing, regular testing of pulmonary function, and use of supplemental oxygen can help manage pulmonary function. A bilevel positive airway pressure (BiPAP) apparatus may help with sleep apnea or related pulmonary insufficiency.

Doctors recommend aggressive treatment of all upper respiratory infections in adults with OI. Chronic bronchitis and asthma may contribute to impaired pulmonary function and should be treated with bronchodilators, inhaled corticosteroids, and antibiotics when appropriate.

**Cardiac function.** A small number of adults with OI seem to have heart valve problems. The most common is called mitral valve prolapse. Dilation of the aorta also may occur, but is not common. High blood pressure is as common among adults with OI as in the rest of the population. High cholesterol and related lipid disorders that may occur in families can contribute to heart problems as well.

Medical management of these disorders includes appropriate diet, drug therapies, and regular monitoring by your primary care doctor. Along with diet, drugs such as statins can be helpful in controlling lipid problems. Coronary artery surgery has been successfully performed on people with OI, although precautions are necessary because of tissue fragility.

**Hearing.** Approximately 50 percent of all adults with OI will experience some degree of hearing loss during their lifetime. Hearing tests and MRI exams of the hearing canals can help your doctor understand the involvement of the bones in the ear. Treatment for hearing loss usually begins with hearing aids. Some adults are candidates for either stapedectomy or cochlear implant surgery.

**Vision.** The connective tissue problem in OI can extend to the eyes. Eye exams are recommended every 2 to 3 years. OI can affect the shape of the lens and the strength of the sclera (the whites of the eyes). For this reason, adults with OI should consult with an ophthalmologist before using contact lenses. Also, laser lens surgery is not recommended for people with OI.

**Gastric function.** Gastric problems are not uncommon in OI. These include gastric acid reflux, which is aggravated by a decreased length of the chest cavity, and chronic constipation. Small stature and frequent use of various pain medications can contribute to the problem.

**Kidney stones.** There appears to be a risk of kidney stones in about 20 percent of people who have OI. These may be caused by the increased calcium intake that results from changes in medications or diet. To see if calcium levels are too high, the doctor may recommend that a change in medications or diet be followed by a 24-hour urine calcium excretion evaluation.

**Basilar impression (BI).** Also known as basilar invagination, BI is a special problem for adults with Type III and IV OI. It involves pressure from the spinal column on the base of the skull. Symptoms of BI can include headache, muscle weakness, and tingling or numbness of hands and feet. Evaluation by a

neurologist, including an MRI exam of the cervical spine and base of the skull, is necessary. Not all individuals with BI have symptoms that get progressively worse.

## **Health Concerns in Common With Other Adults**

**Weight.** Maintaining a healthy weight should be a priority. Being overweight not only increases the risk for many health problems, such as diabetes and cardiovascular conditions, but also puts additional stress on the skeleton, which is particularly unhealthy for people with OI. Diet recommendations for people with OI should be individualized. Consultation with a nutritionist may be helpful to design a balanced diet and address cholesterol and blood pressure problems. Diet modifications also may be needed to help people with chronic constipation or gastric reflux. In general, a good daily multivitamin will be sufficient for adults with OI, and exotic vitamin supplements are not required.

**Diet.** Adults of small stature may require less calcium and vitamin D supplementation than usually prescribed. Total calcium intake of 800 to 1,000 mg (milligrams) per day is usually sufficient. Supplemental vitamin D intake should not exceed 800 IU (International Units) per day.

**Physical activity.** Physical activity to maintain or restore function is a goal of good health management. A low-impact exercise program that may incorporate swimming, a stationary bicycle, or a ski machine is highly recommended. It is important to exercise safely within the range of one's abilities. People with OI may need to consult with a physical therapist or rehabilitation professional to develop an individualized and appropriate program.

**Fatigue and weakness.** People with OI often report fatigue and weakness. Although muscle weakness may be involved, your primary care doctor should conduct a medical evaluation. Problems such as sleep apnea, anemia, or impaired pulmonary function may contribute to a sense of fatigue. In addition, a gynecologist should evaluate postmenopausal women experiencing fatigue and weakness. In some cases, hormone therapy may be appropriate despite concerns about negative side effects, because of the positive effects of this medication on bone strength.

**Alcohol use.** There are no statistics about OI and alcohol abuse. However, primary care doctors who are familiar with OI urge moderation in their patients who choose to drink alcoholic beverages. Individuals taking medications should ask their doctor or pharmacist whether alcoholic beverages are permitted. Impaired coordination resulting from too much alcohol or inappropriate mixing of alcohol and medications could lead to serious injury for adults with OI.

**Stress and mental health.** Health care professionals report that stress as well as the pressures of dealing with chronic health issues can put people at risk for mental health problems. Adults with OI, like other adults, should seek assistance if they experience excessive anxiety or depression.

**Annual physical exam.** After the age of 40, adults with OI should have a complete physical exam each year. The incidence of cancer and diabetes among adults with OI appears to be similar to rates seen in the general population.

### **Tips for Staying Healthy as an Adult With OI**

- Maintain a healthy weight.
- Eat a balanced diet with appropriate levels of calcium and vitamin D.
- Do not smoke.
- If you drink alcohol, do so only in moderation.
- Consult with a physical therapist to design a safe exercise program for maintaining and developing muscle strength and aerobic fitness. Consider a home exercise program or one that can be done at a local gym. Walking and swimming are beneficial activities.
- Have regular medical checkups as recommended for any adult, including, for women, gynecological exams and mammograms.
- Monitor kidney function to prevent development of kidney stones.
- Test for bone density every 1 to 2 years.
- Seek treatment for decreasing bone density, endocrine (hormone) problems, and osteoarthritis symptoms.
- Test hearing every 2 to 3 years.
- Test vision every 2 to 3 years.
- Manage blood pressure and cholesterol levels through diet, exercise, and medication.
- Have a baseline cardiac evaluation, possibly including an echocardiogram.
- Test pulmonary function every 1 to 2 years.
- Consider taking part in a sleep study if symptoms of sleep apnea occur.
- Aggressively treat all upper respiratory infections including colds.
- If a symptom is persistent or troubling, ask the doctor if it is being treated in the same manner as it would for a patient who does not have OI.

## Resources

For more information about osteogenesis imperfecta and osteoporosis, hearing loss, constipation, exercise, pregnancy, preparing for surgery, and other topics, contact:

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## **For Your Information**

This publication contains information about medications used to treat the health condition discussed here. When this fact sheet was printed, 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 Food and Drug Administration at 888-INFO-FDA (888-463-6332, a toll-free call) or visit its Web site at [www.fda.gov](http://www.fda.gov).

For updates and questions about statistics, please contact the Centers for Disease Control and Prevention's National Center for Health Statistics toll free at 800-232-4636 or visit its Web site at [www.cdc.gov/nchs](http://www.cdc.gov/nchs).

*Recognizing the National Bone and Joint Decade: 2002–2011*