



OI Issues: Maintaining Health During the Adult Years

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 type I collagen, which is the major protein in connective tissue. This fact sheet provides a brief overview of health issues of concern to adults with osteogenesis imperfecta (OI). Because OI is a highly variable disorder, it results in different physical features and clinical symptoms from one person to another. Therefore, people with OI should discuss these issues with their primary-care doctor to determine their individual risk and best course of treatment. Adults with OI need to cope with the musculoskeletal concerns associated with OI and also manage all of the same health issues as other adults.

Most adults with OI experience a decrease in the rate of broken bones (fractures) after puberty. Other medical problems, some of which are related to the basic genetic defect that causes OI, may require more attention. Tendon, muscle, and joint problems may be aggravated with time, and hearing loss may become significant. Vigorous and consistent medical care remains as important as it was during childhood.

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 orthopedist, endocrinologist (a doctor who specializes in hormones), a nutritionist, and rehabilitation professionals. Consultation with a pulmonologist (a doctor who specializes in lung diseases) and a neurologist (a doctor who specializes in the nervous system) also may be needed.

Strategies for developing strong relationships 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 one's health, listening carefully to the doctor's instructions, and asking questions until the information is fully understood.

People with OI who are short 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 may also be appropriate. Adults with OI should provide any new doctor with a list of all rods or other surgical implants, since these may interfere with x-rays or an MRI. In many cases, MRI examinations 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 is, in part, related to bone density. The most widely recognized bone mineral density (BMD) test is called a dual energy x-ray absorptiometry (DXA). It is painless: a bit like having an x-ray, but with much less exposure to radiation. It can measure bone density at the hip and spine.

An initial DXA test may be obtained at any age and then yearly thereafter. If possible, DXA scans should be done using the same machine 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 can 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, can also negatively impact 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, ankle instability, and rolling in of the ankle are frequent complaints of people with OI. These problems can be a result of joint laxity or excessive joint flexibility. This subjects knees and ankle joints to pressure over the years. Leg lengths may differ due to a history of previous fractures. Exercise, orthotic devices or braces to improve hip, knee and ankle alignment and back or joint surgery may provide relief. Orthotic devices may also assist in providing stability for lax joints of the knees, feet and ankles. Heel lifts and firm ankle supports can limit wear and tear on the joints and improve walking comfort. 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 respiration or consciousness. Reduced consciousness can increase the risk of falls and fractures.

Pulmonary function: Breathing problems are the main concern of many OI adults, particularly those with Type III and Type IV OI and those individuals with significant 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 is a related problem for some adults with OI and 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. BiPAP positive pressure breathing may help with sleep apnea or related pulmonary insufficiency.

Doctors recommend aggressive treatment of all upper respiratory infections in adults with OI. Asthma and chronic bronchitis 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 (hypertension) 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 and drug therapies and regular monitoring by a primary-care doctor. Drugs such as statins can be very helpful along with diet 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 examination of the hearing canals can help the doctor understand the involvement of the bones of 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 examinations are recommended every two to three years. OI can affect the shape of the lens and the strength of the coat of the eye, called the sclera. 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: 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. Short 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 changes in medications or in diet that increase the amount of calcium circulating in the body.

To see if calcium levels are too high, the doctor may recommend that a change in medication or diet be followed by a 24-hour urine calcium excretion evaluation.

Basilar Impression (BI): Also known as basilar invagination, this is a special problem for adults with Type III and IV OI. BI involves pressure from the spinal column on the base of the skull. Symptoms can include headache, muscle weakness, and tingling or numbness of hands and feet. Evaluation by a neurologist, including MRI examination of the cervical spine and base of the skull, is necessary. A neurologist should monitor BI symptoms. It has been reported that some people have BI, but their symptoms do not get progressively worse.

Health Concerns in Common With Other Adults

Weight: Maintaining a healthy weight should be a priority. Being overweight not only increases risk for many health problems, such as diabetes and cardiovascular problems, but 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 deal with cholesterol and blood pressure problems. Diet modifications may also be needed to help people with chronic constipation and gastric reflux. In general, a good daily multivitamin pill will be sufficient for adults with OI and exotic vitamin supplements are not required.

Diet: OI adults with short stature may require less calcium and vitamin D supplements than usually prescribed. Total calcium intake of 800 to 1000 mg (milligrams) per day is usually sufficient. Supplemental vitamin D intake should not exceed 800 IU/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 ski machine is highly recommended. It is important to exercise safely within the range of one's abilities. Adults with OI may need to consult with a physical therapist or rehabilitation professional to develop an individualized and appropriate program.

Fatigue and weakness: Adults with OI often report fatigue and weakness. While OI related muscle weakness may be one cause, a medical evaluation should be conducted by the primary-care doctor. Problems such as sleep apnea, anemia or impaired pulmonary function may contribute to a sense of fatigue. In addition, postmenopausal women experiencing fatigue and weakness should be evaluated by a gynecologist. In some cases use of hormone therapy may be appropriate in spite of concerns regarding negative side effects because of the positive effects of this medication on bone strength.

Alcohol use: There are no statistics regarding OI and alcohol abuse, but primary-care doctors who are familiar with OI urge moderation in people who choose to drink alcoholic beverages. People who are taking medications should check whether alcoholic beverages are permitted. Impaired coordination resulting from too much alcohol or inappropriate mixing of alcohol and medications could lead to a serious injury for an adult with OI.

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

Annual physical: After the age of 40, adults with OI should have a complete physical each year. It has been reported that the incidence of cancer and diabetes among adults with OI is 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.
- Use alcohol only in moderation.
- Consult with a physical therapist to design a safe exercise program to maintain and develop 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 check-ups 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 1-2 years.
- Seek treatment for decreasing bone density, endocrine (hormone) problems, and osteoarthritis symptoms.
- Test hearing every 2-3 years.
- Test vision every 2-3 years.
- Manage blood pressure and cholesterol levels through diet, exercise and medication.
- Have a base-line cardiac evaluation possibly including an echocardiogram.
- Test pulmonary function every 1-2 years.
- Consider a sleep study if symptoms of sleep apnea occurs.
- 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.

Many of the topics mentioned in this fact sheet as well as others of interest to adults with OI are covered in more detail in other OI Foundation publications and on the OI Foundation web site www.oif.org.

The National Institutes of Health
Osteoporosis and Related Bone Diseases ~ National Resource Center
assisted in the preparation of this publication
Revised November 2005 - Reviewed February 2007