

OSTEOGENESIS IMPERFECTA

ADULT HEALTH TOOLKIT



INFORMATION FOR ADULTS LIVING WITH OI, THEIR
FAMILIES, AND MEDICAL PROFESSIONALS

Thank You and Dedications

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The OI Foundation hopes this resource is helpful to you and your family. Please know that we are here to support the OI community. Questions may be directed to the OI Foundation at bonelink@oif.org.

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Please find an electronic version of the *Adult Health Toolkit* here: www.oif.org/adulttoolkit

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Greetings from the OI Foundation!

The Osteogenesis Imperfecta Foundation is pleased to offer the *Adult Health Toolkit: Information for Adults Living with OI, Their Families, and Medical Professionals* to help you navigate the many aspects of managing your health care as an adult living with OI.

Navigating the healthcare environment as an adult with OI is a community effort. It is important for adults living with OI to become their own advocate, as many providers may have a limited knowledge on the complexities of OI, and OI experts may be difficult to locate in your area. We hope you find this toolkit a helpful resource as you work to develop your own care team. This resource seeks to provide adult OI community members with tools to use in healthcare environments and everyday life.

We, at the OI Foundation, are committed to the development of resources that help serve the needs of the OI community. Through this toolkit and online resources, the OI Foundation provides the most up-to-date medically verified information possible.

We encourage you to connect with others living with OI for support. In addition, we can provide information on providers in your area. You can learn the latest about OI through our social media pages, website, or in-person at OIF regional and national conferences.

Do not hesitate to contact us. We are so happy to be here for you and your family!

Best,

A handwritten signature in black ink, appearing to read "Tracy Hart". The signature is written in a cursive, flowing style.

Tracy Hart
Chief Executive Officer







INTRODUCTION

Navigating Adult OI Care

The OIF Information Center receives approximately 12,000 inquiries per year. Many of these inquiries are from adults with OI, young adults with OI, teenagers with OI, or their parents and caregivers. We also get questions from members of the medical professional community that care for adult OI community members. *The Adult Health Toolkit: Information for Adults Living with OI, Their Families, and Medical Professionals* seeks to answer many of these questions. This toolkit includes medically reviewed information, some of which is interactive, to aid the adult OI community member in advocating for their health care.

Navigating the healthcare environment as an adult with OI is a community effort. This toolkit includes information to serve four different audiences:

-  Adults living with OI
-  Teens and parents navigating care transition to adult healthcare
-  Partners/family members of adults with OI/parents/caregivers
-  The medical professional community

All four audiences will find value in each section of the toolkit, though some information will be more pertinent to a particular subset of the audience.

Because health care needs change with age and are based on individual circumstances, the information that is required to address health care will also change or evolve. While this toolkit is designed to be a resource to guide you; it does not provide all the answers you may be seeking. If you need further information about particular topics, we invite you to visit the OIF website, specifically the Informational Factsheets, which provide a more in depth review of many of the topics included in this toolkit. If you still find yourself with questions, contact us directly using the information that can be found in the [Resources](#) section at the back of this guide.

Communication is key to managing your health care. You know your body best. To partner with the medical field and the members of your care team, it is essential to be able to communicate what you know.

- Keep detailed medical records that can be referenced quickly and easily during phone conversations. This toolkit includes several resources to help you organize your health history.
- Keep a summary of key points to be shared with new practitioners or doctors when traveling.
- Plan for emergencies: have ready information about how to contact the doctor, which hospital to go to, what to do on a weekend or holiday, and transportation. Discuss this plan with your care team, including what to do if your preferred doctor isn't available.
- When communicating with members of your care team and members of their staff, aim to recount events as best you can and answer questions directly. Remember, you know your body best; so, share all information, even details you may not feel are important.
- Be an attentive listener and take copious notes that can be referenced at later times.

OI Definition and Clinical Types

Osteogenesis imperfecta (OI) literally means “imperfectly formed bones”, and is a rare and complex genetic disorder that is often characterized by bones that break easily. While the vast majority of OI is caused by mutations (or variations) in genes that affect the structure of type I collagen or genes whose proteins interact directly with type I collagen. Also, mutations in many other genes that are important for normal bone formation and function have also been found to cause OI. These mutations not only affect how bones are made, how they are shaped, and how strong they are, they also affect the structure of other tissues. Therefore, while bones that break easily are a major feature of OI, many other body systems are also affected. Individuals with OI may also experience dental issues, hearing loss, muscle weakness, joint hypermobility, cardiac, respiratory, gastrointestinal and other health related issues.

The range of clinical features and their severity in OI are highly variable. The severity can range from the “mild” forms where affected individuals have few fractures, to “moderate forms” where individuals can have many fractures and bone deformities, to “severe forms” that can lead to death in the newborn period.

Even within families, clinical features such as fracture frequency, muscle weakness, hearing loss, dental malocclusion, chronic pain, or extra skeletal problems may vary.

Below is a table describing some medical characteristics of the most common types of OI. A more detailed Nosology Chart (a genetic classification system that includes a description of severity) can be found in the [Resources](#) section of this toolkit. More information about the genetics of OI and OI types can be found in the Informational Factsheets section of the OI Foundation website.

Type I (Mild)	<ul style="list-style-type: none"> • Most common and most mild type of OI • Height may be average or slightly shorter than average when compared with unaffected family members, but within normal range for age
Type II (Most Severe)	<ul style="list-style-type: none"> • Numerous fractures and severe bone deformities are evident at birth • Small stature with underdeveloped lungs and low birth weight • Infants may die within weeks from respiratory or other complications • Sometimes referred to as “lethal OI”
Type III (Severe)	<ul style="list-style-type: none"> • Fractures present at birth and X-rays may reveal healed fractures that occurred before birth • Progressive bone deformity is often seen • Short stature • Barrel-shaped rib cage • Spinal curvature and compression fractures of vertebrae
Type IV (Moderate)	<ul style="list-style-type: none"> • Between Type I and Type III in severity and height • Mild to moderate bone deformity • Spinal curvature and compression fracture of the vertebrae • Barrel-shaped rib cage
Type V (Moderate)	<ul style="list-style-type: none"> • Similar to Type IV in appearance and symptoms • Large hypertrophic calluses form at fracture or surgical procedure sites • Calcification restricts forearm rotation

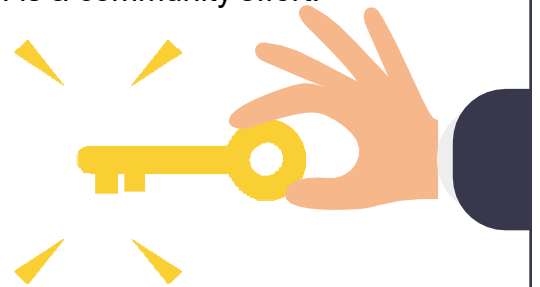
Transitioning from Pediatric to Adult Care

Teen and young-adult years are a critical time for major life changes. An important adjustment for youth who have OI is moving from pediatric care into the adult care system. While children's hospitals tend to have a multidisciplinary team who understand OI, the adult system is much less organized. The young adult will need to find a new team of doctors, take responsibility for communicating with them, and prepare for emergencies. Some children's hospitals offer ongoing multidisciplinary care for individuals with rare diseases like OI in their third and fourth decades of life. So it is worthwhile determining if this service is available at children's hospitals in your area. This transition typically occurs between the ages of 18 and 21; but, planning for it should begin much earlier. Many pediatricians and children's hospitals offer formal Transition Programs that instruct individuals in developing the skills mentioned above; some may have workshops on independent living skills, education/vocational options, dating and relationships, and understanding health insurance options. The following page contains some of the skills, knowledge, and habits that young people need to acquire as they transition to adult care.

Another useful resource is [Got Transition](#)[®], a nationally web-based resource center on health care transition. The website includes tools for youth, young adults, parents, and caregivers.

KEY POINTS

- 👤 Navigating the healthcare environment as an adult with OI is a community effort.
- 👤 OI exhibits wide variation in appearance and severity.
- 👤 Healthcare needs change with age and are based on situations. What is required to address healthcare or personal needs thus may change or evolve.
- 👤 Planning for the transition from pediatric to adult healthcare is multifaceted and should begin prior to the 18th birthday.



Skills for Navigating Adult Care



COMMUNICATE

- Be confident
 - Make and keep appointments in a timely manner
 - Be prepared to ask and answer questions clearly
 - Take responsibility for managing prescriptions
-



KNOW

- OI and your personal history
 - Trackers located in this toolkit help you prepare
 - Medical History Tracker
 - Surgery Tracker
 - Medication Tracker
 - Fracture/Ligament Tracker
-



FIND

- Adult primary care provider and orthopedist before one is needed
 - Pediatricians and pediatric specialists may have recommendations
 - The OIF can help locate providers with OI experience near you
-



DEVELOP

- Healthy habits that promote general health and minimize OI complications which include:
 - Exercise
 - Healthy diet
 - Avoid tobacco and alcohol products
-



PREPARE

- Prepare for OI-related injuries and emergencies before they happen
 - Carry:
 - a state photo ID
 - insurance card(s)
 - medication list
 - allergy list
 - health problem list. (Consider medical alert options and cell phone apps.)
 - Understand your health insurance and how to make a claim.
-



AWAY FROM HOME

- Find a local doctor, learn about the local hospitals, have an emergency plan.
- College students need to understand the Healthcare Services provided by their school and how to access them.
- People need to provide permission in writing for parents or another trusted person to have access to their medical information.



**BUILDING YOUR
MULTIDISCIPLINARY CARE TEAM**

Establishing a Multidisciplinary Team

An important part of managing OI and staying healthy is assembling a multidisciplinary team (MDT) and having a solid working relationship with one's primary care doctor and medical specialists. Simply put, your MDT is a group of healthcare professionals from different fields that collectively strives to provide comprehensive care based on one's situation and medical necessities.¹ There will be situations where it will be necessary to advocate for adequate care and services and this team will work together to make sure nothing is overlooked.



Because there are several professionals on an MDT, it is essential to identify a team leader to coordinate your care/treatment plan. Increasingly, that person is you. When this is the case, select a professional to coach you in your self-advocacy. A person whom you trust, who will guide and help you navigate what needs to be done and ensure nothing is overlooked. While this person can be anyone in the healthcare field, the primary care physician, geneticist, endocrinologist, or orthopedist is often identified as the team lead, or the person who will coach you as you manage your health care. There are a few things to consider when selecting your team leader, such as whether the person has a good understanding of OI and has the experience of treating people with OI. You should feel comfortable with and respected by your team leader, as they will be your advocate and voice with other members of your team. You should be able to have

frank and open conversations about your care using language you understand, where your ideas, thoughts, and concerns are considered and respected. The team leader should be responsive, either by phone or email, in a timely manner and they must be collaborative, as they will be coordinating care across several disciplines.

The following page provides space to record contact information for the members of your professional team. While this is not an exhaustive list, it may include more professionals than are currently on your team, and that's okay. You may not need all of these specialties. Fill in the spaces for those you have right now. As providers may change over time, it is recommended that you complete this section in pencil so it may be updated as needed. The pages following the care team chart will give a brief overview of the role of each professional listed as well as how everyone can function on an OI care team. When you finish recording your information, place a star next to the name of the leader of your care team or the person you select to help you manage.

Your Multidisciplinary Care Team

PROVIDER	NAME	CONTACT and PATIENT PORTAL INFORMATION (Phone Number, Web Address, Log-in info)
Primary Care Physician		
Orthopedic Specialists		
Spine		
Upper Extremity		
Lower Extremity		
Audiologist		
Cardiologist		
Dietician or Nutritionist		
Dentist		
Endocrinologist		
Geneticist		
Gynecologist		
Hand Specialist		
Mental Health Professional		
Neurologist		
Occupational Therapist		
Ophthalmologist		
Podiatrist		
Pulmonologist		
Physical Therapist/ Rehabilitation Professional		
Rheumatologist		

Multidisciplinary Team Member Roles

An important part of managing OI and staying healthy is assembling a multidisciplinary team (MDT). It is essential to identify a team leader or coach to help coordinate your care/treatment plan. The members of your MDT may change over time depending on the level of your needs. Academic medical centers are more likely to have medical practitioners familiar with rare conditions. Your primary physician or geneticist or endocrinologist can give guidance for which specialists/tests are appropriate for you and how often you should consult them, as well as guiding you in the interpretation of results.

Audiologist: Audiologists diagnose and manage conditions such as hearing loss and tinnitus (ringing in the ear) using a variety of devices to identify the extent and underlying cause of hearing loss.² Significant hearing loss has been reported in approximately 50% of people with OI beginning any time from childhood into middle age. While not everyone who has OI develops hearing loss, the incidence is much higher than in the general population. Regular hearing tests are encouraged for all people with OI.

Cardiologist: A cardiologist is a physician who specializes in the care of your heart and blood vessels. They can also specialize in specific areas, such as abnormal heart rhythms, heart failure, or heart valve problems.³ The implications of OI and cardiovascular health are discussed in the Cardiac Care section of this toolkit.

Dentist: A dentist, sometimes called a general dentist or family dentist, is a healthcare provider who diagnoses and treats oral health conditions. Dentists help keep your teeth and gums healthy with regular dental check-ups and cleanings. They can also perform a variety of oral health treatments, including dental fillings, crowns, and bridges.⁴ About half of the people who have OI have teeth that appear normal, and their major concerns are routine care, including cavity prevention and treating teeth fractures. However, the other half has a defect in the teeth called dentinogenesis imperfecta (DI), sometimes referred to as opalescent teeth or brittle teeth. Some individuals with OI may also need to see specialized dentists to treat malocclusions, or the misalignment of the upper and lower jaws and teeth. Your general or family dentist may refer you to a periodontist (specializes in gums), an orthodontist (specializes in alignment) or a dental surgeon for implants.



Dieticians and Nutritionists: Dietitians and nutritionists counsel patients on nutrition issues and healthy eating habits. Dietitians and nutritionists are experts in the use of food and nutrition to promote health and manage disease.⁵ Having a balanced diet and good intake of nutrients are important for bone health and reducing the risk of fracture, as is maintaining a healthy weight.

Endocrinologist: Endocrinologists are medical doctors who specialize in diagnosing and treating health conditions related to problems with the body's hormones and related tissues.⁶ Endocrinologists can be an essential part of a multidisciplinary care team and are most often experts in monitoring and managing bone health. They can determine whether a variety of treatments, including antiresorptive and anabolic therapies, may be beneficial to address increased fracture risk. For many adults with OI, this type of provider is crucial for monitoring and treating their OI.

Genetic Counselor: Genetic counseling is a medical specialty which deals with the occurrence or risk of recurrence of a genetic disorder in an individual or family. A genetic counselor is a healthcare professional trained in human genetics and psychosocial counseling that can provide information

about how a genetic condition like OI could affect an individual or family and support them in making the best possible adjustment to the diagnosis. Genetic counselors also help order and interpret genetic tests (DNA analysis) that are designed to help estimate the risk of a disease. A genetic counselor can help determine whether a person's OI was inherited or was the result of a spontaneous variant (mutation) and can help them make an informed decision about their medical situation and family planning.⁷ DNA analysis is useful for assisting in the diagnosis of OI and in family planning decisions.

Gynecologist: A gynecologist is a physician who specializes in diagnosing and treating diseases of the female reproductive system. Gynecologists are involved in a woman's reproductive health from puberty through menopause. They deal with all aspects of sexual health like preventive care, cancer screenings and physical exams. Some of the services and tests provided include pelvic exams, external genital exams, pap tests and cancer screenings.

Medical Geneticist: A geneticist is a medical doctor who is an expert in evaluating and managing patients with genetic disorders. OI is a genetic disorder caused by mutations in a number of different genes, most commonly type I collagen. Working with a geneticist can help you better understand your type of OI, its severity, symptoms, and know about potential treatment options that are more appropriate for the kind of OI you have. Geneticists can be an essential part of a multidisciplinary care team and can monitor and manage bone health, and determine whether a variety of treatments, including antiresorptive and anabolic therapies, may be beneficial to address low bone density. For many adults with OI, this type of provider is crucial for monitoring and treating their OI.

Mental Health Professional: A mental health professional is a healthcare practitioner or social and human services provider who offers services for the purpose of improving an individual's mental health or to treat mental disorders.⁸ People with OI can lead happy and fulfilling lives, but the physical stresses of the disorder, and society's often harmful view of chronic disorders, can put a psychological burden on the individual. Mental health professionals, including therapists, social workers, and psychologists, can help people with OI develop skills to manage stress and uncertainty that can arise from living with OI.

Neurologist: A neurologist is a medical doctor who specializes in diagnosing and treating diseases of the brain, spinal cord, and nerves.

Neurological diseases and conditions can affect nearly every part of your body and affect both adults and children.⁹ People with OI may include a neurologist in their care team to monitor things like back pain, nerve pain or basilar invagination, a condition that happens when the top of the spine presses into the base of the skull.

Occupational Therapist: Occupational therapists treat injured, ill, or disabled patients through the therapeutic use of everyday activities. They help these patients develop, recover, improve, as well as maintain the skills needed for daily living and working.¹⁰ Occupational therapy can help with fine motor skills and selection of adaptive equipment for daily living. Adults with OI benefit from safe and regular exercise to maintain bone and muscle mass. Swimming and water therapy are particularly well-suited for people with OI of all ages, as they allow independent movement with little fracture risk. Walking is also excellent exercise for those who are able (with or without mobility aids).



Ophthalmologist: An ophthalmologist is an eye care specialist. Unlike optometrists and opticians, ophthalmologists are medical doctors with specific training and experience in diagnosing and treating eye and vision conditions. 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.

Orthopedist: Orthopedic (also spelled “orthopaedic”) doctors and surgeons are doctors who specialize in the musculoskeletal system - the bones, joints, ligaments, tendons, and muscles that are essential to movement and everyday life.¹¹ While adults with OI often use orthopedic specialists or surgeons to treat fractures, they can also treat dislocated joints, back pain, and other types of chronic pain associated with OI.

Physical Therapist/Rehabilitation Specialist: Physical therapists are movement experts who improve quality of life through prescribed exercise, hands-on care, and patient education. Physical therapists examine each person and then develop a treatment plan to improve their ability to move, reduce or manage pain, restore function, and prevent disability.¹² A typical program includes muscle strengthening and aerobic conditioning. Physical therapy can be used to treat a specific injury, or it can be used as a preventative measure to increase strength and balance.

Physical Medicine and Rehabilitation (PM&R): Doctor, also known as a physiatrist, is a medical specialist that involves restoring function for a person who has been disabled as a result of a disease, disorder, or injury. PM&R doctors provide integrated, multidisciplinary care aimed at recovery of the whole person by addressing the individual’s physical, emotional, medical, vocational, and social needs. PM&R doctors are also trained to treat and control acute or chronic pain with medications and holistic approaches.

Primary Care Physician: A primary care physician (PCP), or primary care provider, is a healthcare professional who practices family medicine or internal medicine. Typically, PCPs are our first stop for general medical care and many of them can help coordinate complex care required by patients.

Some individuals may not have a PCP. In this case they can receive primary care from other healthcare providers such as nurse practitioners and physician assistants.

Pulmonologist: A pulmonologist is a doctor who diagnoses and treats diseases of the respiratory system. In addition to your lungs, your respiratory system includes your nose, throat, trachea, airways, muscles, and blood vessels.¹³ Pulmonary disease is the major cause of death for adults with OI, and therefore monitoring and maintaining pulmonary health should be a priority, regardless of type or severity of OI. Pulmonologists can monitor your respiratory health through regular pulmonary function tests, and then prescribe a variety of treatments. Pulmonary function tests should be conducted every 1-2 years (please see Routine Wellness and Lifestyle sections of this toolkit.)

Rheumatologist: A rheumatologist is a doctor with specialized training in inflammatory (rheumatic) diseases and conditions that affect the body’s bones, muscles, tendons, ligaments, and joints. They diagnose, treat, and manage a broad range of conditions including inflammatory disorders, connective tissue diseases, and autoimmune diseases. Because of their extensive knowledge in the body’s bones, muscles, tendons, ligaments and joints, rheumatologists can be an excellent resource for managing OI.



TAKE CHARGE OF YOUR HEALTH

Cardiac Care

There are compelling data that suggest some cardiovascular problems are more prominent in individuals with OI, but the information is scattered and to-date there have been no complete evaluations of the literature concerning pathophysiology, epidemiology, and clinical characteristics.



Studies have shown that a small number of adults with OI have heart valve problems. The most common is called mitral valve prolapse, but other valves may also be affected. Dilation of the aorta also may occur. 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 possible tissue fragility. There is current research underway to help better understand the implications of OI in cardiovascular disease.

Dental Care

In addition to bone fragility, OI may affect the growth of the jaws and teeth. About half of the people who have OI have teeth that appear normal, and their major concerns are routine care.¹⁴ However, the other half has a defect in the teeth called dentinogenesis imperfecta (DI), sometimes referred to as opalescent teeth or brittle teeth. These teeth may be misshapen, may chip or break easily, and will require special care. People with OI and DI need the same basic care as people without, but they also need to be monitored for cracking, chipping, and abrasion of the teeth. All teeth may not be affected by DI, and primary teeth usually are affected to a greater extent than the permanent teeth. Restorative treatment may be needed at some point.

Dental problems related to OI may include the following:

- A skeletal Class III malocclusion. This is when the teeth do not correctly match up making biting difficult. This is caused by the size and/or position of the upper jaw or the lower jaw.
- An open bite. There is a vertical gap between some of the upper and lower teeth.
- Impacted teeth. The first or second permanent molars do not erupt, or they erupt out of the usual location (ectopic).
- Dental development. Tooth development may be may be delayed or advanced in some individuals affected by OI
- OI does not affect the presence or absence of gum disease (periodontitis)

Adults who have OI with or without DI need routine care to prevent tooth loss. Treatments may include cast metal or ceramic crowns, implants, or veneers. Dentures are possible, but bone loss in the jaw requires monitoring. A conversation with a dentist about crowns should be started as soon as an individual notices wear on teeth to determine the next steps. It is important to have this conversation early on, so a dentist can determine when it is time to place a crown. The earlier the treatment is performed, the more likely the tooth is to survive. A literature review of available studies found people with Type I OI have similar success rates as the general population, greater than 90 percent. Reports of people with Type III and IV vary with a range from less than 50 percent to greater than 90 percent success.

People with OI taking bisphosphonates should be closely monitored by a doctor and a dentist. When possible, required dental surgery should be scheduled before starting bisphosphonate treatment or after treatment is finished. Bisphosphonate treatment should not be resumed until after the surgical area is healed. Elective jaw surgery, including dental implants, should be avoided during intravenous bisphosphonate therapy.

OI is a rare condition, which means that many dentists have never treated someone with OI or DI. When looking for a dentist, it is important to find someone trained in solving complex dental problems, even if that dentist has never treated an OI patient. For adults, periodontists, specialists in complex adult restorative dentistry, can be very helpful in coordinating care and connecting their patients with specialists who handle various dental issues. Periodontists can also help to determine the steps that need to be taken to maintain or improve a patient's teeth and determine the correct sequencing of procedures to enhance the best possible outcome.



Emergency Room/Department

Emergency rooms may be particularly stressful for people living with OI. While medical care should be approached as for any other adult of similar age, certain precautions should be considered as detailed in the “Emergency Department Management” Factsheet on the OIF website. Adults with OI vary in height and mobility. This should be considered in exams, procedures and treatments. Emergency room staff members should contact the individual's PCP or physician, who is familiar with OI as soon as possible. Additionally, adults with OI are often the best advocates for themselves and an excellent source of information about safe handling and whether a bone is likely to be broken. People with OI come to the emergency room most often for one of 4 reasons: fractures, pulmonary problems, cardiovascular problems, or accidental trauma. For more information regarding common tests run in the ER, please see the Informational Factsheets entitled “OI and Common Medical Tests and Procedures”, “Emergency Department Management,” and “Emergency Room Care Pocket Guide” on the OI Foundation website.

Family Planning

Genetics

Different types of OI have dominant, recessive or X-linked inheritance. It is important to have genetic testing to know the genetic defect causing OI for family planning and participation in OI research studies.

The most prevalent types of OI have dominant inheritance, which means that a mutation in one copy of a collagen or IFITM5 gene will cause the condition. This is the genetic pattern that will apply to most families with OI. People can inherit dominant OI from a parent, or they can have a new mutation causing OI. Either way, the person with dominant OI can pass this OI mutation to their children (50-50 chance in each pregnancy). The affected child of an OI parent has the same mutation as the parent and generally the same degree of severity, but severity may also be milder or more severe than in the parent.

Most of the rare (non-collagen) types of OI have recessive inheritance. In recessive inheritance, the affected individual has mutations in both copies of a particular OI-causative gene. They have inherited mutations from both of their parents, but the parents are generally not aware they are carriers because they have one normal copy of the gene in question. When a couple has a child with recessive OI, the risk of recessive OI to their other children is 25% per pregnancy. The children of affected persons with recessive OI will all be OI-carriers, because they will inherit a one of the two mutant gene copies from their affected parent, but would not be expected to be affected themselves.

One quite rare type of OI is X-linked, which means that the causative gene is on the X-chromosome. Since males have only one X-chromosome, it is expected that only boys would be clinically affected and that they would have inherited the mutant MBTPS2 gene from their carrier mother. Most carrier mothers will have no symptoms, but some can have mild symptoms. The daughters of affected males will themselves be carriers, but the sons of affected males would not be at risk for OI greater than the general population. A medical geneticist or a genetic counselor can provide information on OI genetics and reproductive decision making.

Pregnancy

Because OI is a variable disorder, a woman with OI who becomes pregnant may experience an uneventful pregnancy, or one with difficulties. Similarly, a baby with OI may be born with very few complications or with numerous fractures and other problems.



The results of a study on pregnancy in women with OI observed when compared with the general population, women with osteogenesis imperfecta had higher rates of diabetes in pregnancy, Cesarean (C-section) delivery, need for blood transfusion, and antepartum and postpartum fractures.¹⁵

They conclude that pregnancies for women with osteogenesis imperfecta are at an increased risk of complications, including hemorrhage (bleeding), fractures, diabetes mellitus, and increased neonatal morbidity.

Labor and Delivery

In general, decisions about the best mode of delivery (vaginal v. Cesarean) should be made on an individual basis. There are no definitive research data showing that Cesarean delivery is safer than vaginal delivery in women with OI who have normal pelvic dimensions and no other significant complications. In fact, it has been shown that delivery by cesarean section is not associated with decreased fracture rate. These results suggest that cesarean delivery should be performed only for other maternal or fetal indications, but not for the sole purpose of fracture prevention in OI.¹⁶

Gastrointestinal (GI) Care

Constipation

One digestive issue some individuals with osteogenesis imperfecta face is constipation. Constipation is defined as a decrease in frequency of stools or bowel movements with hardening of the stool. People with constipation may have gas and feel bloated but find it difficult to pass stools; as such, passing it may be uncomfortable and lead to further unwillingness to try. OI may contribute to constipation, especially in people who are short statured or have a pelvic deformity. The colon and bowel may be prevented from functioning normally if the hips and pelvis are narrow or deformed. Diminished mobility and low levels of physical activity and dehydration also contribute to constipation. Other causes include diet or medications. Some medications, especially pain medications, can be constipating because they decrease bowel wall motion. Additionally, the elasticity of the intestines and bowel due to OI collagen also contributes to decreased motility and it stretches to accommodate more waste instead of moving it through.

Managing constipation usually involves a combination of approaches. It is essential to determine the extent of the problem and develop a plan with a gastroenterologist. Keep a record of bowel movements, diet, and fluid intake. Be consistent with diet, exercise and activity, and fluid intake. Strive for a diet that keeps the stool soft. Too much fiber has the secondary effect of creating too much bulk for someone with a connective tissue disorder. It may be necessary to consult a nutritionist or dietician to achieve and maintain the right balance. Additionally, adding exercise and activity to one's daily routine can help prevent and relieve constipation.

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.

Reflux

Reflux or gastric acid reflux, happens when your stomach contents come back up into your esophagus. Gastric problems are not uncommon in OI and are 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. Oral bisphosphonates are not suitable when there is significant reflux. They are associated with an increased risk of inflammation of the esophagus (esophagitis), damage to the esophageal tissue lining (esophageal erosions), and opens sores in the lining of the esophagus (esophageal ulcers).^{17,18,19}

For more information on GI care, please see the "OI and Common Medical Tests and Procedures" Informational Factsheet on the OI Foundation website.

Hearing Loss

Approximately 50 percent of all adults with OI will experience some degree of hearing loss during their lifetime. A comprehensive hearing test can help the doctor understand what type of hearing loss you have and indicate any involvement of the bones of the ear; sometimes a CT scan or MRI imaging is also recommended. Treatment for hearing loss is dependent on the type of hearing loss and usually begins with hearing aids. Some people with OI are candidates for either stapedectomy or cochlear implant surgery.

Adults with OI should have a baseline evaluation in early adulthood (18-21 years old). If their hearing is normal, they are still considered an at-risk population, and hearing should be tested every three to five years, with three years being the goal. Those who had hearing deficits detected in childhood should not assume those deficits would be static.

There are three main types of hearing loss: sensorineural, conductive, and mixed. All three types of hearing loss can occur with all types of OI and can occur at any age. Sensorineural hearing loss is the most common type of permanent hearing loss and occurs when the inner ear is not transmitting the nerve signals normally to the brain. Conductive hearing loss usually results from a physical problem in the external or middle ear and can be temporary or permanent. It may occur as a result of an ear infection, blockage of the middle ear, or from fixation or fracture of the stapes. Mixed hearing loss is a combination of sensorineural and conductive hearing loss and means that the underlying issue may be in the outer or middle ear and the inner ear, and a combination treatment approaches may be used.

Since OI is associated with an increased risk for hearing loss, it is important to protect and preserve hearing to defer or minimize any loss. Use of hearing protection such as ear plugs or earmuffs in noisy environments is important. Controlling the volume on devices that send sound directly into the ear to a level no louder than 85 decibels for 8 hours is recommended; different technology has different outputs but setting your headphones or devices to no louder than approximately 60% output is one way to keep your devices at a safer listening level.

Tinnitus is ringing, buzzing, or other sounds you hear in your ears or head despite there being no external sound. Tinnitus after noise exposure is a sign that your exposure was too high. Tinnitus that occurs consistently or without noise exposure may be an early indicator of hearing loss.

Lifestyle

The keys to a healthy lifestyle with or without OI are weight management, healthy diet, appropriate exercise, avoiding illness, and especially for individuals with OI, maintaining bone mass. Taking care of yourself makes a difference! Please see the “Routine Wellness” chart located in the Share with Your Care Team section of this toolkit for information about annual wellness considerations for adults living with OI.

Tips for Staying Healthy as an Adult with OI:

- Have regular medical check-ups as recommended for any adult, including for women, gynecological exams and mammograms.
- Exercise to the extent you are able to in a safe and regular manner.
- Maintain a healthy weight, as extra weight is a strain on your skeleton, heart, and lungs. Walking, wheeling, and swimming are beneficial activities.
- Do not smoke and avoid secondhand smoke; smoking harms bones, heart, vessels, and lungs.

- Follow a healthy diet with adequate amounts of calcium and vitamin D. 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.
- Use alcohol only in moderation.
- Be vigilant about protecting your hearing.
- Use sunscreen when outdoors, as OI makes skin more fragile and a little thinner.
- Develop ways of coping with stress and anxiety. This may include exercise, mindfulness/meditation, consulting a mental health professional, etc.
- Identify and practice ways to manage pain.
- Monitor kidney function.
- Test for bone density every 1-2 years.
- Seek treatment for decreasing bone density, endocrine (hormone) problems, and osteoarthritis symptoms.
- Test hearing every 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, including an echocardiogram if recommended.
- Test pulmonary function every 1-2 years.
- Consider a sleep study if symptoms of sleep apnea (i.e. breathing stopping and starting during sleep, gasping for air during sleep) 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.



Mental Health

Living with a chronic health condition such as OI can affect mental health in a number of ways. People with OI may experience pain, low self-esteem, anxiety, depression and PTSD due to medical trauma. A lack of accessible transportation and other access barriers may make it difficult for many people with OI to get out into their communities. They may feel isolated, have difficulty developing friendships and intimate relationships, finding satisfying work, achieving and maintaining independence through changes in health, and living with confidence. They may have a fear of early death or feel depressed. Serious problems such as these often require help from medical and/or mental health professionals. People with OI who experience any of these feelings should discuss their symptoms with their primary care doctor or other trusted mental health professional. It can be difficult to find a mental health professional who is familiar with the challenges of OI. In that event, you may find that someone with experience working with people with disabilities or chronic illness will have the expertise to help.

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 the result of joint laxity or excessive joint flexibility. This subjects knee 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 adults with OI who have joint problems.

Manual wheelchair users should prioritize their shoulder health, as their shoulders are crucial for gross mobility. Improperly fitted mobility aids during childhood can lead to early onset shoulder breakdown, thus it is essential for parents and caregivers to ensure proper fitting. Additionally, maintaining a healthy weight is crucial to minimize stress on the shoulders during transfers.

Adaptive Mobility Aid Planning for Individuals with Osteogenesis Imperfecta

The process of prescribing and obtaining mobility aids is unique for every individual and may evolve with age. Physical and occupational therapists can recommend and provide appropriate aids like customized wheelchairs, orthotic devices, or mobility assistance tools. As OI individuals grow, their physical needs change and potential modifications to their mobility aids may be necessary to ensure continued safety and independence.

For more information on adaptive equipment, please see the “Adaptive Equipment” Informational Factsheet on the OIF website.

Orthopedic Concerns

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, and a neurologist should monitor BI symptoms. Symptoms may worsen over time in some people with BI, but in others symptoms can be quite stable.



Osteoporosis

Since lower bone mineral density (BMD) increases the chance of fracture, maintaining or even increasing bone density is important. Almost all people with OI have reduced BMD and BMD measurements commonly fall in the “osteoporotic” range in adults with OI. Osteoporosis is a condition in which the bones are less dense and more likely to break. The collagen abnormalities in OI cause a specific form of osteoporosis. In OI, fractures are most common during childhood, but the risk of fracture remains high throughout life. In addition, women and men with OI can experience additional bone loss superimposed on a background of OI, so it’s important to monitor bone density and to address any issues that may be causing bone loss.

Fractures and bone mineral density

Understanding Bone Density

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

Testing Bone Density

The most widely recognized bone mineral density (BMD) test is called dual energy x-ray absorptiometry (DXA or DEXA). 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, spine, and other areas. **Even young adults with OI should have a DXA scan, and then every 1-2 years thereafter, depending on the individual's clinical situation.** If possible, DXA scans should be done using the same machine each year to avoid variations in test results caused by different equipment.

Which Medical Professionals to Consult

Endocrinologists are medical doctors who specialize in diagnosing and treating health conditions related to problems with the body's hormones and related tissues. They are experts in managing bone health. Other specialists may also be experts in bone disorders (for instance, rheumatology).

Risk Factors for Declining Bone Density

Declines in bone density can result from a variety of factors. Immobilization associated with casts or limited weight bearing activity can cause bone loss, and aging is associated with a gradual decline in density. In women, the decline in estrogen during menopause commonly causes a loss in bone density. Smoking, overuse of alcohol and certain medications, including cortisone-like glucocorticoids, can also negatively impact bone health. Risk factors for bone loss include:

- having a family history of fracture or osteoporosis
- 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
- Some medical conditions, such as rheumatoid arthritis or thyroid overactivity
- calcium or vitamin D deficiency
- not getting enough physical activity
- smoking
- drinking too much alcohol



Treatments to reduce the risk of fracture

OI is a rare disease and it has been difficult to perform the large, definitive studies needed to demonstrate the effectiveness of anti-fracture therapies. Nevertheless, some approaches may be helpful. Treatments may include calcium and vitamin D supplements (if the diet is inadequate), nutritious diet, exercise, and drug therapies including oral or intravenous bisphosphonates. Teriparatide might also be useful in improving bone mineral density, especially in those with type I OI.²⁰ Newer anabolic medications should also be considered.

Fractures: Care and Management

Most adults experience a decrease in fracture rate after puberty as a result of hormonal and other metabolic changes affecting bone mass and structure. This can last into the 30's or 40's. Other medical problems, some of which are related to the basic collagen defect, may become more important as people age. These include tendon, muscle, and joint problems as well as respiratory compromise.

- The majority of fractures seen in people who have OI are nondisplaced and can be managed with immobilization.
- Care must be taken when manipulating OI bone because of the risk of causing additional fractures.
- OI bone is fragile and can easily fracture proximal to a cast of "normal" weight. Immobilizing a fracture should use the lightest materials possible.
- Adults may have intramedullary rods of different ages and types in different long bones. Their placement and condition should be evaluated if a fracture occurs in a rodded bone.
- Use of plates and screws to repair a fracture is rarely recommended for either children or adults who have OI, as poor bone quality leads to screw and plate instability. Further, plate rigidity can cause bone loss underneath the plate and fractures above and/or below the plate, and screw holes may add to bone fragility and predispose the bone to a new fracture.
- It is possible for some fractures not to be obvious on an x-ray. In this case, remember that you are your best advocate and you can ask for a second opinion if needed.

Other Orthopedic Concerns

- **Scoliosis:** Curves may progress and need to be monitored. Surgery is sometimes necessary.
- **Other spinal deformities:** Lordosis (when the lower back curves inward too much) and Kyphosis (when the upper spine curves out too far outward) may require treatment.
- **Non-Union Fractures:** In general OI bone tends to heal at the same rate as seen in the general population; however, non-union and slow healing fractures are not unusual. Reports show that the frequency of non-unions in adults with OI is higher than that seen in other adults. Researchers are looking into the use of bone morphogenic proteins (BMP) or "bone glue," and adult stem cells as possible solutions.
- **Rods:** Sometimes, rods migrate and become extremely painful. These may require surgery to repair, replace, or remove. Rods that have not moved and are not painful usually do not need to be removed or replaced.
- **Bowing of Long Bones:** The more bowed a bone becomes, the more vulnerable it is to slow healing or repeat fractures. Sometimes, this leads to a recommendation for surgery (usually intramedullary rodding) even if a fracture itself is minimally displaced.











Pain Management

People with OI can experience both acute pain (sudden, sharp; short duration) and chronic pain (persistent; long duration). A Brittle Bone Disease Consortium (BBDC) study on chronic pain found that 42% of the participants reported having chronic pain (30% of those being children and 61% of those being adults).²¹ Pain management requires assessment from a doctor and a personalized plan that provides a variety of strategies for coping with and managing pain. The goal for treatment is effective therapy that will not only reduce or remove the pain but will also achieve mental well-being and an improvement in physiological function.

The following are examples of different types of pain management strategies. More information can be found in the OI Foundation's Pain Management Informational Factsheet on our website.

Chronic pain treatment may include one or more of the following:

-  Medication for pain management (muscle relaxers or NSAID)
-  Physical therapy/physical strengthening
-  Cognitive behavior strategies for pain
-  Heat and ice
-  Exercise or physical therapy
-  Acupuncture
-  Relaxation training, mindfulness, meditation, and visual imagery
-  Biofeedback



Medications are sometimes needed for acute fracture pain and/or chronic bone pain. The majority of OI pain can be alleviated with over-the-counter medication, such as acetaminophen and ibuprofen; however, prescription medications are needed and can be helpful in cases of acute fractures and injuries. Most acute pain within five days should be able to be controlled with OTC medications and other modalities. Pain that persists longer than five days should be discussed with your physician. It is important to note that serious complications can be caused by either over-the-counter or prescription medications. Because of their small stature, many people who have OI cannot safely take the standard dose for either children or adults. Make sure that you and your doctor take this into account when starting a new medication. The 2023 IMPACT survey conducted by the OIF and OIFE (OI Federation of Europe) showed that pain is the main challenge for adults with OI. According to the survey, more than 80% of people with OI have chronic pain, and the percentage and complexity increases with age.

For more information about pain management, please see the "Pain Management" Informational Factsheet on the OI Foundation website.

Pulmonary Care

Respiratory problems can seriously diminish a person's quality of life and are a leading cause of OI-related death. The primary respiratory problem affecting people with OI is loss of lung capacity. Other problems include ineffective cough, poor secretion clearance, airway diseases such as asthma, sleep apnea, and low oxygen.

There are two main causes of lung problems specifically related to OI:

1. Abnormalities of lung tissue
2. Abnormal chest wall architecture (the size and shape of the chest cavity)

In addition, limited mobility and the effects of gastrointestinal problems such as constipation and reflux all contribute to poor pulmonary function.

Viral infections (colds and flu), bacterial infections (bronchitis and pneumonia), and allergies are common respiratory problems. Even when lung problems are not directly caused by OI, they can be more severe in people who have OI. Respiratory conditions are the leading cause of death for people with OI. For that reason, people with OI must protect their pulmonary systems from all types of viruses and be attentive to air quality alerts.

What You Can Do to Minimize Your Risk of Lung Problems:

Preventative Measures

- Do not smoke. Avoid secondhand smoke. Children with OI should never be exposed to secondhand smoke.
- Prevent infection by talking with your doctor about which vaccines may give you the most protection.
- Washing your hands and avoiding contact with people who obviously have a cold or “the flu” are simple but effective ways to stay healthy. When there is poor lung function, a “cold” can progress quickly to bacterial bronchitis or pneumonia. Be aware of infectious respiratory diseases of concern in your community (e.g., COVID-19, RSV at the time of this writing) and how to prevent and treat them.
- Avoid obesity.
- Attentively manage your other underlying lung problems such as asthma.
- Promptly seek care for and aggressively treat all respiratory infections.



Monitor Lung Function

- Monitor your lung function and the amount of oxygen in your blood (oxygenation). If additional workup is indicated, a blood gas test would be a second line recommendation.
- These tests should be part of your regular physical exams. Oxygen levels are most commonly tested by doing an “oximetry” by placing a special sensor on the finger. When lung disease is severe, it may be more appropriate to draw blood from an artery in the wrist and do an arterial blood gas (ABG) test. Sometimes supplemental oxygen or a positive pressure breathing device such as CPAP or BiPAP will be prescribed to help manage breathing problems.
- Evaluating lung disease by radiographs, pulmonary function tests, or sleep studies is helpful in monitoring the progress of respiratory issues in OI. Testing can be difficult due to anatomical differences associated with OI.

Talk to Your Doctor

- Keep the upper body strong. Effective breathing requires strong muscles. Talk to your doctor and physical therapist about an exercise program to help you improve your lung capacity.
- Seek prompt treatment for any breathing difficulties.

- Ask your doctor about whether you should have a sleep study. People with OI seem to have a very high frequency of sleep-related breathing problems.
- Promote good secretion clearance by staying well hydrated and speaking to your doctor or respiratory therapist about special coughing techniques and equipment.
- Get an annual flu shot and talk to your doctor about whether you're a candidate for the COVID and pneumonia vaccines. For adults 60 or older an RSV vaccine should be considered.

Treatments

- Treatments may include bronchodilators, airway clearance devices as appropriate for OI, supplemental oxygen, bi-level positive airway pressure, and pulmonary rehabilitation.
- For patients with OI who have asthma, it is recommended to avoid or minimize the chronic use of oral and inhaled steroids, if at all possible, to reduce the impact that daily steroid use can have on bone health.
- Patients with OI that require daily medication for severe asthma should, when at all possible, seek care at a center that specializes in the treatment of severe asthma to determine specific biologic therapies that can be used to manage their asthma.

Sexual Health

People with OI can have safe, healthy, and fulfilling intimate and sexual relationships with proper communication and precautions. In addition to following general safe sex practices, people with OI should also consider how certain positions could impact their physical health and its risks for causing pain, injury, and broken bones. When possible, have frank conversations with your potential partners before a sexual encounter, expressing your concerns. Share your concerns, discuss necessary modifications, and establish a plan if an injury does occur. Clearly communicating your needs as a person with OI is essential to thriving in all parts of adult life, especially in romantic and sexual relationships. Some professionals who can support this journey are clinical sexologists, sex therapists, pelvic floor therapists, and urologists.

Surgery

Effective communication with doctors before surgery is crucial for adults with OI, since OI may complicate some procedures. It is recommended to follow the following guidelines:

- Engage in pre-surgery consultations with a pulmonologist and anesthesiologist to assess breathing and anesthesia risks.
- Provide essential information about OI type, surgical history, physical differences, allergies, and bleeding tendencies.
- Share medication and supplement lists and inquire about personalized anesthesia for small stature.
- Plan pain management for all stages of surgery and recovery.
- Inform hospital staff about specific OI needs, such as positioning and gastrointestinal issues.
- Plan recovery by arranging home adaptations and consult with therapists as needed.
- Have a trusted advocate accompany you during hospitalization.
- Display the OIF flier, "Handle Adults with Care," for staff awareness.

For more information about considerations before surgery, please see the "Take Charge Before Surgery" Informational Factsheet on the OI Foundation website.

Treatments and Medications ^{22, 23}

Establishing care with a primary care physician, a physician who monitors bone density such as a medical geneticist or an endocrinologist, and an orthopedic physician is important across adulthood. Bone mass in OI is impacted by the balance between bone resorption and bone formation. When bone resorption exceeds bone formation, bone density and bone strength decline. This imbalance can be counteracted through either antiresorptive or anabolic therapies. Antiresorptive therapies target bone resorbing cells also called osteoclasts to decrease the rate of bone resorption. Anabolic therapies aim to increase the rate of new bone formation by targeting osteoblasts, the bone forming cells. Below is a list of antiresorptive and anabolic medications.

Class	Bisphosphonates	RANKL inhibitor	Parathyroid hormone analogues	Anti-Sclerostin antibody
Examples	Pamidronate, Zoledronate, Alendronate, and others	Denosumab	Forteo, Tymlos	Evenity
Mode of administration	Oral or intravenous (IV). Frequency of administration depends on the drug.	Subcutaneous (under skin) injection (SI) in upper arm, upper thigh, or abdomen. Every 6 months.	Subcutaneous injection. Daily.	Subcutaneous injection. Monthly.
Mechanism	Antiresorptive	Antiresorptive	Anabolic	Anabolic
Benefits	Longest experience in OI May help increase bone density	May help increase bone density	May help increase bone density	May help increase bone density
Limitations	Optimal duration of use in adults and fracture reduction effects are not well known. Studies have not supported a reduction in fractures ²⁴	Limited experience in OI Concern for rapid bone loss when treatment is stopped, with a rebound increase in the risk of vertebral fractures	Treatment duration usually 18-24 months, needs to be followed with an antiresorptive Unclear benefit in mild and severe OI	FDA approved for treatment of osteoporosis in 2019, but limited experience in OI Treatment duration 12 months, needs to be followed by antiresorptive therapy Possible cardiovascular risks in some individuals

The potential benefits and possible adverse effects of any drug therapy should be carefully discussed with the prescribing clinician.

Bisphosphonates

Both oral and intravenous (IV) options are available and can be prescribed at varying intervals, based on your own laboratory and x-ray data. It has been noted in the adult OI population that there may be a diminished response to bisphosphonates and a conversation with your provider would be necessary to help determine appropriateness of the medication. A provider can help to determine which agent and dosing would be best suited for the individual needs of a patient. Potential benefits of bisphosphonates may include a decrease in bone loss, increased bone mineral density, and in some cases an increase in quality of life.

Common Indications (reasons to be prescribed) for bisphosphonates include:

- Low and decreasing bone mineral density
- Vertebral compressions
- Fractures, but note studies have not supported a reduction in fractures²⁴

For more information about bisphosphonates, please see the “Bisphosphonates: Q&A” Factsheet on the OIF website.

Women’s Health

Girls with OI types I and IV can expect to begin menstruating at the same age as, or just slightly later than, girls who do not have OI; however, girls with OI Type III may experience a delay of several years before beginning to menstruate (Reed). This type of delay has been associated with an increased risk for osteoporosis in the general population. Once menstruation starts in girls with OI, their cycles are generally regular, although heavy bleeding may occur in girls and women who have a history of easy bruising or bleeding tendencies. There is no evidence to suggest that fertility is influenced by OI; however, miscarriage rates may be higher among women who have OI. Women should discuss routine wellness exams with their providers, but most recommendations include annual pap smears every three years starting at age 21, and mammograms yearly after age 40.

People with OI vary in height and mobility. This should be considered in exams, procedures, and treatments. Pelvic deformity and curvature of the spine may complicate positioning during routine exams; and depending on your size, pediatric equipment may be needed. Discuss this with your doctor prior to the examination.



Many women with OI are concerned about menopause and the possibility of more frequent fractures. The experience of postmenopausal women with OI varies greatly; some experience an increase in fractures, while others do not. Strategies including exercise, diet, medication, and more can help maximize their chances of staying active and healthy as they age. Women should start talking with their doctor about their bone health five years










before turning 51 (average age of menopause) because women begin to lose bone mass five years before they stop menstruating. Bone density measurements and medications or hormone replacement therapy (HRT) to prevent bone loss should be discussed with your physician.

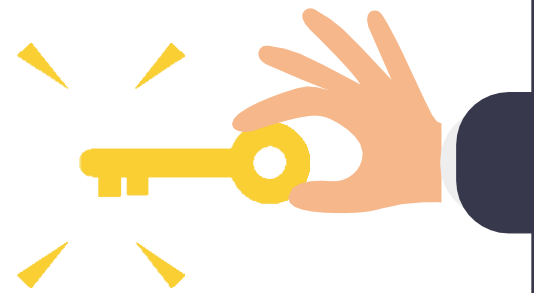
HRT is not a bone-building agent but is a bone-stabilizing agent. Before starting HRT, it is important to discuss the pros and cons of HRT with your provider, as well as to provide a detailed family history to determine whether the pros of HRT outweigh the cons. If even one member of a woman's family has had breast cancer, it is recommended to have genetic

testing to determine if there are any predisposing genetics that could increase the risk of developing breast cancer from HRT. If a woman taking HRT is still experiencing a decrease in bone density or is unable to take HRT, then a conversation about other available bone-building agents should occur.



KEY POINTS

-  The keys to a healthy lifestyle with and without OI are weight management, healthy ~~de~~ appropriate exercise, avoiding illness and (esp. for individuals with OI) maintaining bone mass.
-  Living with a chronic health condition such as OI can affect mental health in a number of ways and if you experience a decline in mental health it is best to seek help from medical and/or mental health professionals.
-  People with OI vary in height and mobility. This should ~~be~~ considered in exams, procedures, and treatments.
-  People with OI can have safe, healthy and fulfilling intimate and sexual relationships ~~with~~ proper communication and precautions.
-  Even if there is no other history of OI in someone's family, a person with OI can pass ~~the~~ OI mutation to their children because it is a genetic disorder.
-  The primary respiratory problem affecting people with OI is loss of lung capacity ~~and~~ restrictive lung disease.
-  In addition to bone fragility, OI may affect the growth of the jaws and may or may not ~~affect~~ the teeth.
-  Constipation, hearing loss and acute and chronic pain are often experienced by individuals with OI.
-  Most adults experience a decrease in fracture rate after puberty as a result of hormonal ~~and~~ other metabolic changes affecting bone mass and structure. This can last into the 30's or 40's.





SHARE WITH YOUR CARE TEAM

Compiling Your Health Care History

Maintaining your own health records is important as an adult with OI. There are many ways to organize your medical information: keeping a binder, storing hard copies of files, and having access to electronic files/records. Regardless of your preferred method, it is essential to keep detailed records, preferably in reverse chronological order, so that nothing is overlooked in your health care journey. In this section of the toolkit, you will find several interactive health trackers that may be useful in helping you organize your health care history. In addition to the information covered in the health trackers, other considerations may include:

Legal Documents

- Health Care Proxy: A document that designates another person to make healthcare decisions on your behalf if you become incapacitated.²⁵
- Durable Power of Attorney: A document that designates another person to handle private, financial and business matters on your behalf if you become incapacitated.²⁶
- Living Will: A legal document that tells doctors how you want to be treated if you cannot make your own decisions about emergency treatment.²⁷

Family Health Information

- For family planning and genetic counseling, it is most helpful to know your own genetic mutation. It can also be helpful to keep a list of relatives who have OI.
- It is a good idea to learn about your family health history. OI does not rule out being at risk for the other health issues that affect adults or that can run in families such as heart disease or cancer.

Orthotics and Equipment

- List the model number, manufacturer and dealer of any equipment (i.e. wheelchairs) you currently use.
- Note where items can be repaired.



Basic Health History, Medication Tracker, and Surgery Tracker

Your personal health profile will continue to evolve over time. Setting up an easy-to-use system to keep track of test results, hospitalizations and treatments is very beneficial. Your primary care doctor or insurance company may have suggestions for appropriate apps or websites. Some people keep pictures of health forms on their cell phones while others elect to use cell phone apps, such as the Health App on iPhones (set up a “Medical ID”) or the Safety and Emergency app on Androids (complete the “Medical Information”) to give first responders vital information in case of an emergency. Information from either app can be accessed on the phone’s lock screen. If you opt to store health information directly on your cell phone, whether via photos or apps, be sure to keep the information current by making regular updates.

Though it is necessary to maintain your detailed health history, a summary sheet is often the most helpful thing to share with new healthcare providers, whether they be a member of your care team or a physician in an emergent care facility. For that reason, please find on the following pages



four resources; the first details your health history, the second details your fracture and ligament history, the third details the various medications you’ve taken along with any adverse reactions to them and the fourth details your surgical history. Keep these with you, either electronically or a hard copy, and share them with your providers as necessary.

When discussing medications with your doctor, **make sure you discuss adjusting the dose of medication to your weight and height, rather than age;** monitor the use of NSAIDS due to the link to delayed bone healing after fracture; minimize the use of drugs that contain steroids due to negative effects on bone; and discuss your risk of spontaneous tendon rupture seen in people with a connective tissue disorder when using antibiotics known as fluoroquinolones (Cipro, Levaquin).

BASIC HEALTH HISTORY TRACKER

Complete the sections that apply to you and share with your provider(s).

Type/Mutation (if known):					
Date: _____		Determined by: <input type="checkbox"/> Clinical Exam <input type="checkbox"/> Skin/Collagen Biopsy <input type="checkbox"/> DNA Testing		Notes:	
OI RELATED TESTING/ASSESSMENT/SCREENINGS					
TYPE OF TEST	DATE	RESULTS	TYPE OF SCREENING	DATE	DIAGNOSIS
Bone Density			Breathing		<input type="checkbox"/> Asthma Sleep Apnea
Echocardiogram			Dental Care		<input type="checkbox"/> Dentinogenesis Imperfecta (DI) <input type="checkbox"/> TMJ <input type="checkbox"/> Osteonecrosis of the jaw
Pulmonary Function					I have the following: <input type="checkbox"/> Veneers <input type="checkbox"/> A partial <input type="checkbox"/> Caps <input type="checkbox"/> Dentures
Heart: Hypertension Valve Issues Family history of heart problems		<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Yes <input type="checkbox"/> No			
Hearing: Hearing Loss Hearing Aids Stapedectomy Cochlear Implant	Age: Type:	<input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Yes <input type="checkbox"/> No	Vision		
			Other		
Basilar Impression (Provide diagnosis and treatment information if this is part of your history)					
Gastrointestinal Issues (GI) Describe any current problems with constipation and/or GERD:					
GENERAL HEALTH INFORMATION					
Immunizations: (Give date of last inoculation.)					
Pneumonia PCV13 _____		Influenza (FLU Shot) _____		COVID _____	
Pneumonia PPSV23 _____		Other: _____		Other: _____	
Women's Health: (Give dates where applicable.)					
First period _____		Last period _____		Number of pregnancies _____	
Allergies/Sensitivities: (Foods, insects, plants, etc.)					
Mental Health (List mental health diagnoses/concerns/issues.)					

ORTHOPEDIC HISTORY TRACKER

Complete the sections that apply to you and share with your provider(s).

FRACTURE HISTORY: Check all major fractures and site of osteotomies. If a bone was broken more than once, list the most recent. Include rod information for any rods that are still in use.				
LOCATION	DATE	ROD TYPE	PLATE/SCREW	NON-UNION
Femur <input type="checkbox"/> Right <input type="checkbox"/> Left				
Tibia <input type="checkbox"/> Right <input type="checkbox"/> Left				
Pelvis <input type="checkbox"/> Right <input type="checkbox"/> Left				
Arm (Humerus) <input type="checkbox"/> Right <input type="checkbox"/> Left				
Spine Fracture(s)				
Tendon and/or Ligament Injuries: list any recurring injuries or those requiring medical attention below.				
INJURY	DATE	TREATMENT		
JOINT INJURY: Check all that apply.				
LOCATION	DATE	TREATMENT		
Knee <input type="checkbox"/> Right <input type="checkbox"/> Left				
Ankle <input type="checkbox"/> Right <input type="checkbox"/> Left				
Wrist <input type="checkbox"/> Right <input type="checkbox"/> Left				
Shoulder <input type="checkbox"/> Right <input type="checkbox"/> Left				

Routine Wellness

Audiology*	Baseline evaluation at 18-21 years old. If hearing is normal, continue with a hearing test every 3 years.
Cardiovascular*	Echocardiogram as a baseline to assess aortic or mitral valve disease and pulmonary artery pressure; annually if necessary.
Dental*	Dental cleaning and evaluation every six months.
Dermatology	Evaluate for signs of skin cancer annually.
Gastrointestinal*	Chronic constipation, assess for GERD, and history of kidney stones. Annual colonoscopy after age 45. Discuss with your doctor possible alternatives to colonoscopies, such as Cologuard testing.
General Health	Complete metabolic panel with fasting blood sugar, complete blood count, lipid panel, and HbA1c. Testing for Hepatitis C for those who had surgery before 1992. Assess calcium and vitamin D status as needed.
Gynecological	Pap smears every three years starting at age 21, and mammograms yearly after age 40.
Mental Health	Evaluate for depression, anxiety and substance abuse.
Neurology	Assess for symptoms related to basilar impression, limb length differences, changes in muscle strength; unequal gait, balance, risk for falling and evaluation of back pain.
Ophthalmology*	General vision testing; glaucoma; retinal detachment. Most insurances will cover an annual exam.
Orthopedic*	Bone density scan (DXA or DEXA) every 1-2 years. Assess scoliosis and other chest or back deformity; limb length discrepancy; osteoarthritis in joints.
Pain Management	Assess changes in general pain level or new pain.
Pulmonary	Pulmonary Function Test every 1-2 years and monitor the amount of oxygen in your blood (oxygenation). Consider a sleep study if symptoms of sleep apnea occur.
Vaccines	Stay current with annual vaccines including flu, pneumonia and COVID.

Ask your physician about which routine screenings to consider.

**These areas of health may be impacted more by having a diagnosis of OI and screening recommendations may differ from the general population.*

Additional Medical Considerations

- Conditions that may be made worse due to OI: asthma, osteoporosis, COPD, sleep apnea, dental issues, hearing.
- Conditions that may be masked by OI: arthritis, vocal cord problems.
- Conditions that appear to occur often in adult OI: anxiety disorders, reversible and irreversible airway obstruction and other breathing problems, non-union of fractures, glaucoma, hearing loss, heart valve and aortic disease, hypertension, pain syndromes, basilar impression, tendon and ligament injuries.
- Conditions that appear to occur more often in adults with more severe OI: shortness of breath and sleep apnea.
- Areas where bone, blood vessel and tissue fragility related to OI may complicate treatment or testing: cancer surgery, pregnancy, heart valve surgery, urinary tract infections, colonoscopy, ulcer diagnosis, other endoscopic procedures, and intubation for surgery.
- Areas where the effect of OI is not well understood: gastrointestinal function (includes GERD, constipation, celiac disease), heart problems and kidney stones.

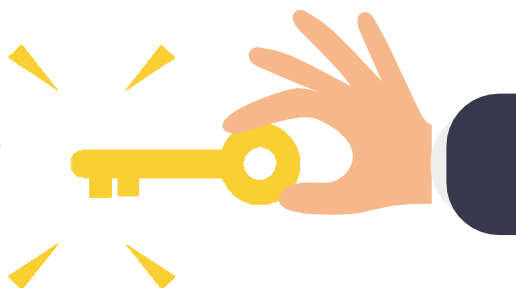
KEY POINTS



In addition to a detailed health history report, it is important to make a summary sheet with your health history and medications (incl. adverse reactions) for both every day and emergency hospital visits.



For medical professionals: To find medically verified resources by the OIF's Medical Advisory Council to consult or share with your patients please consult the "Resources for Medical Professionals" page.





RESOURCES

OIF Resources for Medical Professionals

The Osteogenesis Imperfecta Foundation (OIF) is the largest national health organization dedicated to helping improve the quality of life for people living with osteogenesis imperfecta. In addition to funding and directing research, the OIF provides the following information and resources to individuals living with osteogenesis imperfecta, their loved ones, and medical professionals. All the information is provided free of cost and much of it is available online at www.oif.org. The information provided is medically verified by the OIF's Medical Advisory Council.

The OIF has programs and services designed specifically for medical professionals treating patients living with osteogenesis imperfecta. To view a regularly updated list of public meetings for medical professionals, go to www.oif.org/medprocalendar.

Medical Professional Emails



Sign up to receive OIF medical professional emails to learn about upcoming events, programs, and resources to better understand and treat patients with osteogenesis imperfecta. Contact bonelink@oif.org to sign up.

These meetings provide an opportunity for medical professionals who are treating patients with OI, and those who are interested in bone health, to come together to learn about OIF resources and connect and collaborate with each other. The meetings are held twice per year, once virtually and once in-person. Attendance ranges from 50-100+ per meeting.

TeleECHO Clinic Series



The OIF hosts both a Rare Bone Disease TeleECHO series and an OI-specific TeleECHO series, each holding sessions one time per month. In each monthly virtual session, speakers present a brief didactic presentation, followed by participant-led case presentations and group discussion of the presented case. Both programs offer free CME credits through Project ECHO. A full schedule of upcoming Rare Bone Disease ECHO sessions can be found at www.rbdalliance.org/echo.

Educational Videos and Recorded Professional Meetings



The OIF has recordings of past professional meetings available on the OIF YouTube channel. This includes Rare Bone Disease and OI TeleECHO presentations, Rare Bone Disease Alliance Scientific Symposia, OI Research Updates, and more. Also, we have a wide variety of patient-facing recordings, including sessions of past OIF National Conferences in English, Spanish, and French. Visit the OIF YouTube channel at www.youtube.com/@oifoundation.

Research

Funding and helping direct OI research is a central part of the OIF's mission. In addition to the programs listed below, the OIF is a collaborator in many research initiatives and is a principal investigator in the National Institutes of Health Brittle Bone Disorders Consortium, and the recipient of several previous and ongoing awards from the Patient-Centered Outcomes Research Institute. The OIF is also a partner in the International OI Pain Project and the International OI Adult Health Project and helps administer working groups like the international OI surgeon's roundtable.

Michael Geisman Fellowship Grants

The OIF awards funding to post-doctoral trainees who are currently working on projects with clear relevance to OI, or who have projects that will enable them to develop expertise in OI research.

Annual OIF Scientific Meeting

Each year the OIF invites leading OI researchers and clinicians to attend this two-day meeting focused on sharing current and emerging research in OI.

Rare Bone Disease Alliance (RBDA)



The RBDA is a coalition of patient advocacy organizations focused on professional education for providers, expanding research, and assisting patients and communities affected by rare bone diseases. The RBDA is a program of the OI Foundation.

Consultations

The OIF can connect local providers with OI experts across the country, in many disciplines, to consult on treatment plans.

OI Registry



Individuals with OI (18 and older) and parents of children with OI are encouraged to join the OI Registry. The OI Registry is a database of individuals with OI who are interested in participating in OI research. Once you are enrolled, you will begin to receive information on upcoming studies and online surveys about OI. Learn more and join the OI Registry at www.oif.org/OIregistry.

OIF Resources for Community Members

The OIF Information Center responds to more than 12,000 inquiries from patients and medical professionals each year. Constituents can contact the OIF at bonelink@oif.org or call 844-899-7579 with questions about OI or OI related resources. In addition to answering individual inquiries, the OIF maintains the following resources:

Clinic and Provider Directories



The most frequent type of inquiries received by the OIF are community members in search of medical professionals with experience treating OI in their area. The OIF maintains a clinic directory of more than 60 OI and bone health clinics in the US, and a community-generated provider directory of physicians with experience treating OI.

Informational Factsheets



The OIF has published many Informational Factsheets on topics related to understanding and living with OI. Topics include genetics, surgery, family planning, dental care, and respiratory issues. For a full listing of Informational Factsheets, visit www.oif.org/factsheets.

Virtual Learning



The OIF website houses a library of video recordings on topics related to managing OI. The videos are a collection of lectures from virtual conferences, community calls and podcasts. Many of the videos are also available in French and Spanish. Visit the OIF YouTube channel at www.youtube.com/@oifoundation.

Newsletters

Sign up to receive the OIF's printed newsletter, Breakthrough, and monthly electronic E-News to learn about upcoming events, programs, and resources for OI community members.

Regional and National Conferences

The OIF hosts a biennial national conference that brings together 500-800 OI families for three days to attend educational sessions and social events. The regional conferences are a similar format but are one-day meetings and held several times per year in cities around the US. Financial assistance is available for families to attend both programs. Learn more at www.oif.org.conference.

Mutual Support

The OI Support Group Program aims to provide a space for members of the OI community to share, educate, and socialize with one another. The OI Support Group program is divided into five Regional Groups (Northeast, Southeast, Southwest, Midwest, and West) and OI Affinity Groups. Regional Support Groups aim to foster a sense of community and provide regional-based information to OI community members in specific regions. Affinity Support Groups are national-based groups of individuals with a shared interest or common identifying characteristic.

Opportunities to Participate in Research

The OI Registry gives members information about new OI research and the option to participate in IRB approved studies. More information on the OI Registry can be found at www.oif.org/oiregistry.

Financial Assistance

The Jeanie Coleman Impact Grant Program provides equipment and services that improve the quality of life for families living with OI. Items that have been awarded in the past include (but are not limited to) wheelchairs and ramps, hearing aids, technological devices (laptops, tablets), home adaptation, and accessible vans. Funding ranges between \$500-\$25,000.

The Edward D. Cranston College Scholarship is for students who are applying to or have been admitted to a college or university. Preference for the scholarship is based on a combination of merit and financial need. Scholarships range between \$250 to \$10,000 and can cover any costs associated with attending the school including tuition, books, and lodging.

OI Registry

Individuals with OI (18 and older) and parents of children with OI are encouraged to join the OI Registry. The OI Registry is a database of individuals with OI who are interested in participating in OI research. Once you are enrolled, you will begin to receive information on upcoming studies and online surveys about OI. Learn more and join the OI Registry at www.oif.org/OIregistry.

This flyer may be helpful to display in your room during hospital stays, or on your wheelchair, to educate those around you about precautions to consider when caring for a person with OI.

Patient's Name _____

CAUTION!! I HAVE OSTEOGENESIS IMPERFECTA (OI)

Handle me gently at all times.

I have osteogenesis imperfecta (OI), this means that I may have:

- Fragile bones
- Fragile skin
- Loose joints
- Fragile Teeth
- Lying flat may be difficult
- Respiratory problems

SUPPORT my entire body when lifting or turning me.

- **DO NOT** pull on my arms or legs to help me sit up or turn over.
- **DO NOT** force my head to turn.

My family and I can provide additional directions.

GENTLY remove any surgical tape or dressings; my skin is very fragile.

Check that the dose of all medicines has been adjusted to my small size.

Monitor blood pressure manually, using a pediatric cuff if necessary.

- **DO NOT** try to take a BP on a broken or curved arm.

Use Pediatric size equipment if/when necessary.

CONTACT my primary care doctor if you have questions about how to handle me.

Doctor's Name _____ Phone # _____

I have hearing loss and may not hear you if my aids are not in.

Nosology (Classifications) of Osteogenesis Imperfecta

OI TYPE	INHERITANCE	PHENOTYPE	GENE MUTATION
DEFECTS IN COLLAGEN SYNTHESIS, STRUCTURE, OR PROCESSING			
I	AD	Mild	Null COL1A1 Allele
II	AD	Lethal	COL1A1 or COL1A2
III	AD	Progressive Deforming	COL1A1 or COL1A2
IV	AD	Moderate	COL1A1 or COL1A2
XIII	AR	Mild/Severe	BMP1
DEFECTS IN BONE MINERALIZATION			
V	AD	Variable, Distinctive Histology	IFITM5
VI	AR	Moderate/Severe	SERPINF1
DEFECTS IN COLLAGEN MODIFICATION			
VII	AR	Severe (Hypomorphic) Severe/Lethal (Null)	CRTAP
VIII	AR	Severe/Lethal	LEPRE1
IX	AR	Moderate/Lethal	PPIB
XIV	AR	Severe	TMEM38B
DEFECTS IN COLLAGEN FOLDING AND CROSS-LINKING			
X	AR	Severe/Lethal	SERPINH1
XI/BRKS1	AR	Mild/Severe	FKBP10
BRKS2	AR	Moderate/Severe	PLOD2
DEFECTS IN OSTEOBLAST DEVELOPMENT WITH COLLAGEN INSUFFICIENCY			
XII	AR	Severe	SP7
XV	AR	Severe	WNT1
XVI	AR	Severe	CREB3L1
XVII	AR	Progressive Severe	SPARC
XVIII	XR	Moderate/Severe	MBTPS2
XIX	AR	Severe	FAM46A/TENT5A
XX	AR	Progressive Severe/Lethal	MESD
XXI	AR	Severe + Neurodevelopmental	KDEL2
XXII	AR	Severe	CCDC134

Abbreviations:

AD = autosomal dominant; the mutation is inherited in a dominant manner

AR = autosomal recessive; the mutation is inherited in a recessive manner

Glossary

Basilar Impression (BI)	Basilar Impression is when the top of the spine pushes up into the base of the skull. It causes pinching and pressing on the brain stem.
BBDC	The Brittle Bone Disorders Consortium is a multi-center program that focuses on understanding and providing better treatment options for all types of osteogenesis imperfecta (OI).
Bisphosphonates	A class of drugs, usually used to treat osteoporosis, that prevent loss of bone density.
Bone Density Testing	A test that helps to estimate bone density and the likelihood of breaking a bone.
Bone Mineral Density (BMD)	The amount of bone mineral (i.e. calcium) in bone tissue.
Brace	A medical device designed to address musculoskeletal issues; they are used to properly align, support, stabilize, and protect parts of the body as they heal from an injury.
Cochlear Implant	A cochlear implant is a small, complex electronic device that can help to provide a sense of sound to a person who is profoundly deaf or severely hard-of-hearing.
Collagen	The main structural protein in various connective tissues in the body.
DEXA or DXA Scan	A non-invasive test that measures bone mineral density. It is used to see if a person is at risk for a fracture.
Echocardiogram	A test of the action of the heart using ultrasound waves to produce a visual display; detects blood flow through heart valves and movement of heart walls.
Fiberglass Cast	A plaster cast made from fiberglass material, which is lighter than plaster of Paris.
Fracture	A crack or break in a hard object or material, typically a bone.
Handle Me With Care Flier	OI Foundation resource that can be printed and given to medical professionals that explains how to best work with people with OI.
HRT	Hormone Replacement Therapy (HRT) is a treatment used to relieve the symptoms of menopause. It replaces the female hormones that are at a lower level during menopause.
Kyphosis	An excessive outward curvature of the spine, which causes a rounding or hunching of the back.
Malocclusion	Malocclusion is when your upper and lower teeth do not align when you close your mouth.

Glossary

Mitral Valve Prolapse	Mitral valve prolapse, also called MVP, is a condition in which the two valve flaps of the mitral valve do not close smoothly or evenly, but bulge (prolapse) upward into the left atrium.
Multidisciplinary	Combining or involving several academic disciplines or professional specializations in an approach to a topic or problem.
Mosaicism	The state of being composed of cells of two genetically different cell populations, generally one normal and one carrying a mutation. Most often individuals who are mosaic for OI mutations may be quite mild or only come to medical attention when they have a severely affected child.
Nosology	The branch of medical science dealing with the classification of diseases.
NSAIDS	Non-steroidal anti-inflammatory drugs (NSAIDs) are medicines that are widely used to relieve pain, reduce inflammation, and bring down a high temperature.
Osteoporosis	Osteoporosis is a bone disease that develops when bone mineral density and bone mass decreases, or when the structure and strength of bone changes. This can lead to a decrease in bone strength that can increase the risk of fractures (broken bones).
PTSD	Post-traumatic stress disorder (PTSD) is a disorder that develops in some people who have experienced a shocking, scary, or dangerous event.
RANKL Inhibitor	RANKL is the abbreviation for receptor activator of nuclear factor kappa beta (NFkB ligand). RANKL inhibitors are used as a treatment in OI.
Reflux/Gastric Acid Reflux	When your stomach contents come back up into your esophagus.
Stapedectomy	Stapedectomy is a surgical procedure in which the stapes bone is removed from the middle ear and replaced with a prosthesis.
Osteogenesis Imperfecta (OI)	A group of genetic disorders that mainly affect the bones. Its hallmark characteristic is bones that break easily, but it affects many other systems in the body. Also known as brittle bone disease.
Osteogenesis Imperfecta Type I	The most common and most mild type of OI with few obvious symptoms. People with Type I OI may be of normal or near-normal height.
Osteogenesis Imperfecta Type II	The most severe type of OI. With Type II OI, there are numerous fractures and severe bone deformity at birth, and infants may die within weeks from respiratory or heart complications.
Osteogenesis Imperfecta Type III	A severe type of OI where progressive bone deformity is often seen. Fractures are present at birth, and X-rays may reveal healed fractures that occurred before birth. Spinal curvature and compression fractures of vertebrae are also common in OI Type III. People with Type III OI usually are short-statured, with a barrel-shaped rib cage.

Osteogenesis Imperfecta Type IV	A moderate type of OI that is between Type I and Type III in severity and height that has mild to moderate bone deformity, as well as spinal curvature and compression fracture of the vertebrae. People with Type IV OI usually have a barrel-shaped rib cage.
Osteogenesis Imperfecta Type V	A moderate type of OI that is similar to Type IV OI in skeletal severity. In Type V OI, large hypertrophic calluses form at fracture sites and calcification of the membrane between the radius and the ulna restricts forearm rotation, Type V OI also has dominant inheritance.
Osteogenesis Imperfecta Type VI	A progressive forming type of OI that is rare and is similar to Type III in appearance. Type VI OI is characterized by a mineralization defect in bone. The mutation for Type VI OI is not in the collagen pathway and is inherited in a recessive manner.
Osteogenesis Imperfecta Type VII	OI Type VII is a severe type of OI that has a recessive inheritance pattern and is commonly lethal. OI Type VII is caused by absence of the CRTAP protein, important for modification of collagen.
Osteogenesis Imperfecta Type VIII	OI Type VIII is a severe type of OI which can be lethal and has a recessive inheritance pattern. OI Type VIII is caused by the absence of P3HI enzyme, important for modification of collagen. In Type VIII OI, there is a severe growth deficiency and under mineralization of the skeleton.
Osteogenesis Imperfecta Foundation (OIF)	A voluntary health organization with the mission of improving the quality of life for those living with osteogenesis imperfecta through research, education, awareness, and mutual support.
Osteotomy	An osteotomy is a surgical procedure that involves cutting bone (and sometimes adding bone tissue) to reshape or realign your bones.
Procedure	A surgical operation.
Pulmonary Function Test	Non-invasive tests that show how well the lungs work.
Splinting	Any rigid material that is used to immobilize a fractured or dislocated bone, or to maintain any part of the body in a fixed position.
Surgery	The treatment of injuries or disorders of the body by incision or manipulation, especially with instruments.
Vital Signs	Clinical measurements, specifically pulse, temperature, respiration rate, and blood pressure, that indicate the state of a patient's essential body functions.

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Please find an electronic version of the *Adult Health Toolkit* here: www.oif.org/adulttoolkit