

OSTEOGENESIS IMPERFECTA

# NAVIGATING A NEW DIAGNOSIS



A TOOL KIT FOR PARENTS AND  
MEDICAL PROFESSIONALS

# Thank You and Dedications

This toolkit is dedicated to the memory of Michael David Johnston (06/20/1955-11/12/2018). When his daughter Emma was born with type IV OI, he and his wife Bonnie suddenly found themselves faced with a lot of new information and uncertainty. While the OIF helped them immeasurably along their journey with OI, this resource would have been invaluable. Mike served proudly on the OIF Board of Directors from 2001 to 2007. Mike was an activist, an artist, a cherished friend, and a wonderful father. He would have been excited about this toolkit and honored to have it dedicated to him.

*Sponsored in part by a memorial donation from Minnesota Neonatal Physicians*

In addition, the OI Foundation would like to thank Dr. Bonnie Landrum, Dr. Michael Bober, Dr. Ricki Carroll, Dr. Richard Kruse, Dr. Jeanne Franzone, Dr. Joan Marini, Dr. Reid Sutton, Dr. Deborah Krakow, Tina McGreal, Nikki Watson, Deborah Fowler, and Green Room Communications for their assistance in the development of *Navigating a New Diagnosis: A Tool Kit for Parents and Medical Professionals*. The OI Foundation hopes this resource is helpful to you and your families. Please know that we are here to support the OI community. Questions or concerns about this document can be directed to the OI Foundation at [bonelink@oif.org](mailto:bonelink@oif.org).

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Congratulations on your new baby and welcome to the OI family!

The Osteogenesis Imperfecta Foundation is pleased to offer *Navigating a New Diagnosis: A Tool Kit for Parents and Medical Professionals* to help you navigate the many questions you may have during this exciting, but sometimes stressful, time.

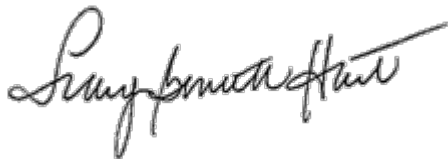
Our mission is to improve the quality of life for those living with osteogenesis imperfecta through research, education, awareness, and mutual support. We hope you find this tool kit a helpful resource.

Through this tool kit and our online resources, the OI Foundation provides the most up-to-date information possible, whether you are building your prenatal and pediatric care teams, navigating the neonatal intensive care unit or looking for information on what to expect when you take your baby home. We have also included a section that can be given to your physician if they are not familiar with osteogenesis imperfecta.

The OI Foundation is a resource for you and your child as he or she grows. We have built a website you can trust with medically verified information. We also encourage you to connect with others facing OI for support. You can learn the latest about OI through our social media presences or in-person at our regional and national conferences. Lastly, we can provide information on physicians in your area that have experience working with people with OI. All you need to do is contact us.

Again, congratulations! 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  
Osteogenesis Imperfecta Foundation

## About This Tool Kit

The information presented in *Navigating a New Diagnosis: A Tool Kit for Parents and Medical Professionals* has been reviewed by medical professionals with extensive experience in the osteogenesis imperfecta (OI) field. The booklet is full of useful information, but it is a lot to take in at one sitting, especially if you are new to OI. The following tips may be helpful as you read.



### Take pauses while reading

The lightbulb icon at the end of each section indicates a point to pause and consider the information.



### You aren't expected to know everything within the tool kit

The tool kit is meant to be an interactive resource. The paper icon indicates an opportunity for you to write in the tool kit; this is your copy. If you need an additional copy, the OI Foundation can have another emailed, faxed or mailed to you. A digital (PDF) version is also available via our website for your printing convenience.



### Check out the OI Foundation Podcast Series

The OIF Podcast series discusses innovative research studies and educational topics including spinal, dental, and cardiopulmonary issues in OI. Throughout the tool kit, we have included links to podcasts on different topics. The podcast series is available at [www.oif.org/podcast](http://www.oif.org/podcast)



### Contact the OI Foundation with questions

Through the OI Foundation, you can access up-to-date information on OI and reach members of our Medical Advisory Committee or other medical professionals. If needed, we can connect your baby's current doctor with OI experienced medical professionals or direct you to a nearby physician.



### Contact the OI Foundation

**Website:** [www.oif.org](http://www.oif.org)

**Phone:** (301) 947-0083 or toll free (844) 889-7579

**Email:** [bonelink@oif.org](mailto:bonelink@oif.org)

# Myths and Facts About OI

Osteogenesis imperfecta (OI), or “brittle bones disorder,” is a rare genetic disorder characterized by bones that break easily, often from little or no apparent cause. Osteogenesis imperfecta literally means “imperfect formation of bone.” OI is caused by a mutation (change) in a gene that codes for type I collagen, the major protein present in bone. These mutations affect bone composition, formation, and strength, as well as the structure of other tissues. The major feature of OI is a fragile skeleton, but many other body systems are also affected.

## Common Myths and Facts About OI

MYTH	FACT
People with OI are diagnosed at birth	<ul style="list-style-type: none"> <li>• OI can be diagnosed at many different ages from birth into adulthood</li> <li>• The most severe forms are usually diagnosed at birth or shortly after</li> <li>• Other forms may not be diagnosed until the child has a series of broken bones or an adult has unusually low bone density</li> <li>• OI Type I, mildest form of OI, is relatively common. It is rarely diagnosed at birth</li> </ul>
OI only affects the bones	<ul style="list-style-type: none"> <li>• Though fragile bones are the hallmark of OI, many parts of the body are affected by OI, including the lungs, skin, muscles, tendons, internal organs, and eyes</li> <li>• In addition, breathing problems, hearing loss, excessive perspiration, and dental problems affect some people with OI</li> </ul>
A baby with OI should always be carried on a pillow and discouraged from moving	<ul style="list-style-type: none"> <li>• Although there are handling techniques and precautions, it is in the child’s best interest to be held and touched and encouraged to explore independent movement to the greatest extent possible</li> <li>• Immobility increases the likelihood of bone loss and decreases muscle mass, leading to weakness, bone fragility, and more fractures</li> </ul>
OI is a childhood disorder; people grow out of it by their teens	<ul style="list-style-type: none"> <li>• OI is a genetic disorder that is present throughout a person’s lifetime</li> <li>• Many people with OI have fewer fractures after puberty when growth stops, but the genetic difference remains</li> <li>• Fractures and other complications occur throughout the lifespan and can increase again after menopause in women and after age 60 in men; non-skeletal problems can be serious</li> </ul>
Everyone who has OI is shorter than average, has blue sclera (whites of the eyes), and uses a wheelchair	<ul style="list-style-type: none"> <li>• The appearance of people with OI varies considerably</li> <li>• Although most people with OI are short-statured, people with milder forms may be in the normal height range and have no obvious symptoms of OI</li> <li>• About 50% of people with OI have tinted sclera, the white outer layer of the eyeball, that can range in color from nearly white to dark blue or gray</li> <li>• People with OI also have variable mobility, ranging from independent walking to full-time wheelchair use</li> </ul>
All children of a parent who has OI will have OI	<ul style="list-style-type: none"> <li>• When one parent has a dominantly inherited type of OI, there is a 50% chance with each pregnancy that the child will have OI</li> </ul>



**What questions do you have about OI? Are there myths you have heard about OI? Write them down here and you can share them with your doctor or with the OI Foundation.**

**We'd love to answer any questions you have about OI.  
Please send them to the OI Foundation at [bonelink@oif.org](mailto:bonelink@oif.org)**

## OI Clinical Types

Osteogenesis imperfecta (OI), or “brittle bone disease,” is a complicated and variable disorder. Its major feature is a fragile skeleton, but many other body systems are also affected. OI is caused by a mutation (change) in a gene that codes for type I collagen, the major protein present in bone. These mutations affect bone composition, formation, and strength, as well as the structure of other tissues. It is a lifelong disorder that occurs equally among males and females and in all racial groups. With good medical management and supportive care, most people with OI will lead healthy, productive lives and can expect an average life span. OI exhibits wide variation in appearance and severity. Severity is described as mild, moderate, or severe. The most severe forms lead to early death. Clinical features such as fracture frequency, muscle strength, or extra skeletal problems vary widely not only between types but within types and even within the same family. Some medical characteristics include:

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

For a detailed list of OI Types including clinical signs, degree of severity, and genetic mutation, please see the OI Foundation website ([www.oif.org](http://www.oif.org)).



**Has your baby received a genetic diagnosis of OI? If so, feel free to write the diagnosis below:**

Date of Diagnosis: \_\_\_\_\_

Diagnosing Doctor: \_\_\_\_\_

Diagnosis: \_\_\_\_\_

# Pregnancy

Congratulations on your pregnancy! Whether you are early in your pregnancy journey or preparing to give birth in the coming days, your mind and emotions may be going in several different directions. Our goal is to provide you with medically verified information regardless of where you are in your journey.

There may be a few different scenarios that bring you to read this section of the tool kit. Feel free to circle which scenario best fits your current situation. Additionally, there is a section included to write in your own scenario if it is not included below.

## Scenario A

Expectant parent of a baby with OI (family history of OI)

## Scenario B

Expectant parent of a baby with OI (no family history of OI)

## Scenario C

Expectant parent with OI (expecting a baby without OI)

## Scenario D

Relative or friend of an individual that fits either scenario/  
looking to gain further insight

***View OIF Factsheets***

at [www.oif.org/factsheets](http://www.oif.org/factsheets)

The following table directs you to sections of the tool kit that apply to your specific scenario.

SCENARIO(S)	SECTION
A, B, C, D	Managing the Pregnancy
A, B, D	Expecting a Baby with OI
A, C, D	Expectant Moms with OI
B, D	Expectant Moms without an OI Diagnosis (No Family History)
B	Diagnosis of OI (Prenatal)
A, B, C, D	Delivery Options

## ***Managing the Pregnancy***

Current research indicates that the standard amount of calcium and vitamin D and other minerals is appropriate for a pregnancy where OI is suspected. At this time, there are no treatments or dietary supplements that can prevent the child from having OI or that will make the type of OI milder. All pregnant women are encouraged to talk with a physician about appropriate diet and exercise during pregnancy to ensure optimum health for both themselves and their babies.

## ***Expecting a Baby With OI***

OI is typically the result of a mutation in one of the two genes that carry instructions for making type I collagen—the major protein in bone and skin. The mutation may result in either a change in the structure of type I collagen molecules or in the number of collagen molecules. Either of these changes results in weak bones that fracture easily and other connective tissue symptoms.

## ***Expectant Moms With OI***

Women with OI who are pregnant or considering becoming pregnant should consult an obstetrician/gynecologist. A specialist in high-risk pregnancies may also be helpful for women who anticipate pregnancy complications, either due to severe OI-related problems, or other problems, such as a history of preterm labor, multiple miscarriages, or other significant health concerns. Planning to deliver at a hospital with special services for high-risk mothers and babies is another consideration.

### **Specific Considerations for Women with OI Who Become Pregnant**

- Although many women with mild OI experience few adverse effects from pregnancy, they may have loose joints, reduced mobility, increased bone pain, and dental problems during pregnancy
- Potential anesthesia concerns for women with OI include hyperthermia (raised body temperature), or an inability to receive epidural anesthesia due to spinal curvature or compression
- All pregnant women experience changes in their bone density during pregnancy, but there is concern that women with OI do not regain the lost bone density after pregnancy, or do not regain it as quickly as other women
- A study of back pain in pregnant women who have OI suggests that vertebral crush fractures are common, and that cesarian section does not prevent this problem<sup>1</sup>
- Short stature, spinal curvature, and rib cage deformities can lead to complications when already crowded internal organs must accommodate a growing fetus. Complications can range from breathlessness and discomfort to more serious problems that necessitate early hospitalization or premature delivery. Monitoring of respiratory function may be indicated
- After the baby is born, the mother with OI may also experience increased bone pain, susceptibility to fracture, or other connective tissue problems
- Obstetrical manipulation during delivery may result in fractures
- Pregnancy is not clearly associated with increased maternal fracture risk. However, carrying a child to term can place additional stress on weakened bones and loose joints. A woman might be more likely to fall when her growing abdomen disrupts her balance
- Women with OI have reported several other pregnancy complications. It is not known whether they occur more frequently or severely in women with OI than in other women. These complications include preeclampsia (characterized by high blood pressure, protein in the urine, and body swelling); premature delivery; placenta previa (when the placenta covers the cervical opening); premature rupture of membranes; recurrent urinary tract infections; anemia (low red blood cell count); and calcium deficiency
- A history of pelvic fractures and/or pelvic deformities may necessitate cesarean delivery
- Women with a history of easy bruising, recurrent nosebleeds or bleeding tendencies following previous surgeries may be more susceptible to excessive bleeding after delivery. Blood coagulation and platelet tests may be prescribed prior to the delivery date as a precautionary measure

## **Expectant Moms Without an OI Diagnosis (No Family History)**

Sometimes, women who do not have OI become concerned about the likelihood of their child having OI during pregnancy. This may be because:

**1** The woman's partner has OI

**2** Prenatal testing suggests the presence of OI symptoms in the fetus

### ***The Woman's Partner has OI***

A person with a dominant form of OI has a 50% chance of passing the disorder on to each child. The child will have the same OI-causing mutation as the parent, although the child's symptoms may be different, milder, or more severe than the parent. It is possible that the child of a person with OI will have a spontaneous genetic mutation resulting in a different type of OI, but the chances of this happening are no greater for a parent with OI than they are for the general population.

Some individuals with very mild OI have been known to have a child with more severe symptoms. In these cases, it is believed that the parent is a mildly affected mosaic for OI. *Mosaicism* means that the individual carries a mutation for OI in only some of his or her cells. This can cause very mild symptoms of OI, or none at all, in the carrier. Excluding OI, the risk of other congenital disorders in pregnancies in which one parent has OI is the same as that of the general population.

### ***Prenatal Testing Suggests the Presence of OI Symptoms in the Fetus***

If an ultrasound indicates OI may be suspected in a fetus of an unaffected mother, additional, higher-level ultrasounds tests may be ordered and/or a referral to a center for high-risk pregnancies. Higher level ultrasounds may also be ordered for pregnant women who had a previously affected pregnancy.

The findings of the ultrasound may present certain medical and ethical questions to be addressed by the couple and their medical team. Questions include accuracy of the diagnosis, severity of the disorder, and prognosis for survival and development. At this stage it can often be difficult to determine with certainty whether or not the fetus has a lethal or non-lethal form of OI.

### ***Diagnosis of OI (Prenatal)***

Ultrasound can be used to examine the fetal skeleton for bowing, fractures, shortening, or other bone abnormalities consistent with OI. Ultrasound is generally most helpful for prenatal diagnosis of the more severe forms of OI. The fetal skeleton shows signs of OI as early as the second trimester. Fetuses with mild OI seldom show evidence of fractures or deformity before birth.

#### **Ultrasound**

A noninvasive, low-risk procedure. There are different levels of ultrasound, some of which are more useful than others in detecting OI.

#### **Chorionic Villus Sampling (CVS) and Amniocentesis**

Analyze cells obtained from the fetus for collagen defects and/or a genetic mutation that causes OI. CVS looks at placental cells, while amniocentesis examines fetal cells (amniocytes) shed into the amniotic fluid. Both procedures carry a risk of miscarriage (about 1 in 200 for amniocentesis and about 1% for CVS).

These prenatal tests are most useful for prognostication if the parent who has OI already has the results of his or her own collagen or DNA tests.

It can be challenging to ascertain prognosis and survivability from ultrasound or genetic testing. At this stage it can often be difficult to determine with certainty whether or not the fetus has a lethal or non-lethal form of OI.

Interpretation of these results also requires a specific expertise. Questions around accuracy of diagnosis, severity of disease, and prognosis should be addressed by someone well-versed in OI so that a family can make well-informed decisions. Seeking advice from a geneticist or genetic counselor is also highly recommended in these scenarios.

Geneticists and genetic counselors are trained to diagnose and discuss OI as well as options.

Perinatal palliative care physicians are trained to help families make difficult medical decisions, based on their goals and value.



Contact the OI Foundation for assistance locating a physician near you who is familiar with OI, including geneticists, genetic counselors, and perinatal palliative care physicians.

Email: [bonelink@oif.org](mailto:bonelink@oif.org)

### ***Delivery Options***

Decisions about the best mode of delivery (vaginal vs. cesarean) should be made on an individual basis.

Multiple studies have shown that Cesarean delivery does not decrease fracture rates at birth in infants with nonlethal forms of OI, nor did it prolong survival for those with lethal forms.<sup>2,3</sup> These studies also identified that breech presentation was more common when a baby had OI.<sup>2,3</sup> Breech presentation may require Cesarean section, as opposed to normal vaginal delivery.



## Building Your Prenatal Care Team

Having a prenatal care team is important for all pregnancies but is especially critical for planning for a baby with OI. Outlined below are different types of doctors that can provide care for you during this time:

**Geneticist:** A geneticist can assist in providing a diagnosis of OI and answer questions about inheritance.

**Obstetrician:** An obstetrician should be aware of your genetic history before giving birth.

**Genetic Counselor:** A genetic counselor can provide information on OI genetics and prenatal diagnosis and answer questions about inheritance. It is recommended that couples at risk of having a child with OI seek genetic counseling before conception or as early in the pregnancy as possible.

**Palliative Care:** Palliative care is aimed at improving quality of life for children with life-threatening and life-limiting conditions. Often comprised of a team of providers, palliative care providers partner with families to ensure their medical team is honoring their wishes for their child. Some of the things they can do are help bridge communication gaps, help families make difficult decisions, and provide emotional and spiritual support. They can also assist with pain and symptom management for your baby when he/she arrives.



When planning for delivery, here are other critical members of your care team who should have the skills necessary to handle a tiny, fragile baby who has OI:

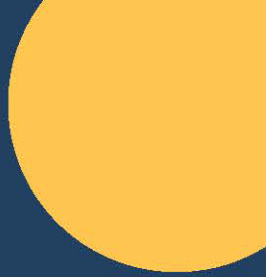
- Neonatologist
- Chief obstetrical nurse
- Nursery staff
- Doula/midwife
- Primary care provider
- Delivering physician



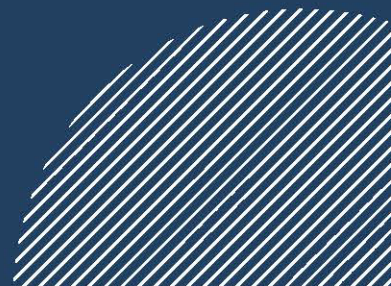
Below we have left some space for you to write down who is on your prenatal team:

NAME	SPECIALTY	PHONE





**THE BABY'S HERE!**



## Care Concerns for Babies With OI

Caring for children with OI needs to meet the needs of each child. No two children with OI are exactly alike. The development of a care plan helps to ensure your child's needs are being met. Elements of a care plan for a child with OI may include:

- Management of both skeletal and non-skeletal issues
- Rehabilitation to enhance function and encourage development of maximum bone mass
- Monitoring of growth and nutrition
- Surgical/hospital care that reflects knowledge of OI and respect for bone and tissue fragility
- Referrals to other specialists as needed, such as physical therapy
- Attention to normal childhood disease and immunizations

As your child grows, the following should also be monitored regularly:

- Screen routinely for vision, hearing, and dental care
- Check the spine for scoliosis and kyphosis, an excessive outward curving or hunching of the spine
- Test bone density to help monitor changes over time (a baseline test when a new course of treatment is started, and then another test approximately 6-12 months after a change in treatment)
- Conduct an echocardiogram in childhood, then the late teen years, and as a young adult
- Consult with an orthodontist to assess jaw development
- Perform a baseline pulmonary function test on all children with OI and again at maturity (age 20-25); repeat every two years if the test is normal

Your doctor will be able to work with you to determine when your child should have certain screenings done.

### ***Diet and Nutrition***



As with all children, it is important that children with OI have a balanced diet containing enough water, fiber, calcium, and vitamin D for their age and size. As your child grows, the following things are important to keep in mind:

- Slow weight gain in an infant may not be failure to thrive
- Nutrition counseling for the family may be beneficial
- A child with swallowing difficulties may need a referral to an occupational or speech therapist as well as a nutritionist who treats feeding disorders
- A small appetite may be seen in children with OI of all ages; this could be caused by slower growth, inactivity, pain, medications, and depression
- Constipation is seen in children of all ages and with all types of OI (and can be recurrent); short stature, inactivity, pelvic deformity, and difficulty with hydration are contributing factors
- Weight control is important; obesity places a strain on the fragile skeleton and can lead to loss of mobility



Listen to the podcast

***Multidisciplinary  
Care for the Pediatric  
Patient with OI***

[www.oif.org/podcast](http://www.oif.org/podcast)

## Development and Growth

It is important to track growth and development in all children. There are some specific considerations for children with OI when it comes to development:

- OI does not affect a child's ability to think and learn, but children with OI can demonstrate delays in meeting developmental milestones
- Delays can be the result of repeated immobilizations after fractures; physical and occupational therapy, braces, and use of adaptive equipment and mobility aides can assist in meeting developmental milestones
- Mild to significant short stature and a slow growth rate occurs in OI
- Hip and back pain should be evaluated by an orthopedist and/or a gait specialist

## Navigating the Hospital Setting

Navigating the hospital may be a new experience, but you don't have to go through this alone. Listed below are general guidelines and safety precautions for the emergency department. While the guidelines and precautions are specific to the emergency department environment, they may be helpful for other places, like the Neonatal Intensive Care Unit, or NICU.

### General Guidelines for Emergency Treatment

Don't be afraid to speak up about your opinions, wishes, advice, or instructions; you know your child best and your input can often help doctors

Fractures can be unpredictable in people with OI

Ask to stay with your child at all times; your presence not only provides comfort to your child, but you may be able to help with transferring or ensuring safety precautions are used to prevent further injury

### Safety Precautions

- Make sure that medical professionals are gentle and cautious during transfers; they should avoid sudden pulling of the limbs, neck, or spine. Limbs should never be twisted, bent, or straightened. As parents, you will be able to provide guidance on how to best move your child
- People with OI can bruise easily. IVs and blood draws should be done by the most experienced professional available
- Blood pressure cuffs or tourniquets can lead to bruising or fracture if placed too tightly. In addition, automatic blood pressure cuffs may put too much pressure on the arm bone. A baseline blood pressure should be taken by manual blood pressure measurements
- Medical professionals may not take the blood pressure of your child if it is not indicated, to avoid risk of fracture
- Ask for neonatal or pediatric nursing staff to help with medical procedures
- Doses of medicines should be determined by body weight, not the age of the child with OI
- Stretchers should be padded and without holes that a small person could slip through. Be careful when tightening straps on a stretcher to avoid causing a fracture



- Make sure that blankets and sheets are not too tight and be careful when removing them to avoid getting fingers, toes, etc., caught in the folds, which could cause a fracture
- Some people with OI develop hyperthermia under general anesthesia, and some are sensitive to inhaled anesthetics
- Make sure to communicate any allergies; some people with OI have an allergy to latex
- It may be helpful to bring a copy of the *Handle Me with Care* flyer from the OI Foundation



**Patient's Name** \_\_\_\_\_

## **CAUTION!! I HAVE OSTEOGENESIS IMPERFECTA (OI)**

**Handle me gently at all times.**

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

- Fragile bones
- Fragile skin
- Loose joints
- Fragile teeth
- Lying flat may be difficult
- I may have respiratory problems

**SUPPORT** my entire body when lifting or turning me.

- **DO NOT** pull on my arms 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** using a pediatric cuff if possible.

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

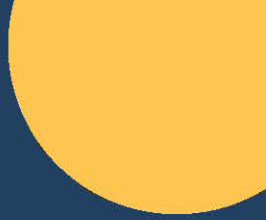
**USE** pediatric-sized equipment if necessary.

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

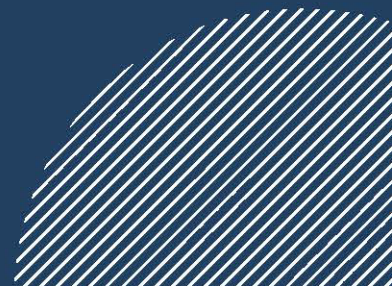
**Doctor's Name** \_\_\_\_\_ **Phone #** \_\_\_\_\_

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

Osteogenesis Imperfecta Foundation  
[www.oif.org](http://www.oif.org) • [bonelink@oif.org](mailto:bonelink@oif.org) • 844-889-7579 • 301-947-0083



# LIFE AT HOME



# Handling

The information below may be helpful when handling your baby. Consider printing this information and having it available throughout your home as reminder to family members and friends. Families should know that in severe OI, children can break bones, even when parents and caregivers are handling carefully.

- Remember that the bones are very fragile and can break with little or no pressure. Be especially careful of the long bones in the body: the arms, legs, and ribs
- You should NOT lift your baby under the armpits or pull on his or her arms or legs
- Babies with rib fractures should not be placed on their stomach
- When you change a diaper, lift the baby by the buttocks, not by the ankles as is customarily done. Spread your fingers apart as far as possible and put your hand under the buttocks with your forearm under the baby's legs to prevent them from dangling

- To lift the baby onto your shoulder or carry the baby, use the same technique, but with one hand placed behind the head and the other behind the buttocks, again with fingers spread as far as possible
- When lifting or moving your child, be careful that little fingers and toes do not get caught on the clothing you are wearing, such as shirts or blouses that button down the front
- When a child has a painful fracture, it is usually best to avoid lifting or moving him or her as much as possible. After some healing, the fracture will be less painful, and moving your child will be easier. Unfortunately, leaving your child in one position for a long time can cause skin rashes and sores



- Putting a child in different positions not only prevent skin rashes and sores, but also helps the child develop different sets of muscles, which is important for mobility
- Occasionally a gel pad is necessary to protect the back of the skull from flattening. Rolled blankets or soft foam wedges can be used to support side-lying
- Infants who spend an extended period of time in the nursery should be repositioned regularly. The unusually soft skull can be flattened from prolonged time in any one position
- Rib fractures, a deformed chest, etc., will preclude placing the baby in the prone position (on the stomach)
- When possible, have someone else help you with handling your baby. Four hands are always better than two

Remember to trust yourself. Common sense is the best guide when handling a child with OI. Do not be afraid to show affection to your child by cuddling, rocking, touching, and talking to him or her. Frequent stimulation is necessary for sound emotional and social development.

# Car Seats and Bedding

## Car Seats

You will need a car seat to take your baby home from the hospital. As with any child, it is important that the child with OI be safely confined in an approved car seat placed in the back seat.



- For maximum safety, never place a car seat in the front passenger seat, because airbags can be dangerous, especially for children with OI
- A car seat or in some cases a car bed will be necessary at the time of discharge from the hospital. This equipment is geared to the child's weight and ability to sit up
- Infant car seats are designed for children under 20 pounds in weight. Features to look for include a well-padded harness and a head hugger support pillow. This type of U-shaped pillow is commercially available and is used to position the baby's head at midline
- Small rolls or towels or other padding can be added to hold the child's hips in line
- All car seats should be approved for safety and anchored correctly in the vehicle
- It is not safe to place foam padding between the plastic shell and the padded cover. This will compress in the event of a car accident
- Removable cotton slipcovers are suggested because they are cooler and some children with OI perspire excessively
- It is always best to place a car seat in the back seat of the vehicle
- Many parents place a label on the top edge of the car seat stating the diagnosis, physician name and phone number, emergency contact phone number, and HANDLE WITH CARE (included in the Resources Section) instructions in case a car accident occurs

## Bedding

A standard crib mattress is most suitable for a baby with OI. Waterbeds and soft bedding should not be used.

# Feeding and Dressing

## Feeding

Some babies with OI display a weak sucking reflex and may require small, frequent feedings. The combination of small stature, feeding problems, and slow growth may be mistaken for failure to thrive.

**Breastfeeding:** Breast milk is an excellent source of calories for virtually all infants, including those with OI. Breastfeeding can create a special bond between the mother and child. Babies with all but the most severe forms of OI should be capable of being breastfed. However, those with the most severe forms of OI may have breathing difficulties that interfere with the ability to suck. Rapid respirations can predispose to aspiration. If the baby is not able to breastfeed, the mother may opt to pump breast milk and feed the child breast milk from a bottle, nasogastric tube, or G-tube.

**Handling During Feeding:** When feeding the infant, the mother should be especially careful to avoid having the baby positioned with an arm behind the back or a leg pressed against the mother's body in such a way as to put pressure on it at an abnormal angle.

**Burping:** Burping should be done very gently to reduce the chance of fractures, especially of the ribs. Soft taps, possibly with padding over the hand, are recommended.

- To pick up the infant for burping, lay the baby on his/her back while the caregiver bends over to pick up the infant. The caregiver's shoulder should very gently touch the baby at which point the infant is supported under the back and positioned on the shoulder as the caregiver moves up and backwards. Gently rubbing the baby's back while taking gentle bouncing steps may also be beneficial



## Dressing

There are certain precautions that you should make when dressing your baby, including:

- Loose, lightweight, cotton clothing seems to be the most comfortable for babies with OI because children with OI are frequently affected by warm temperatures and are often bothered by excessive sweating
- Look for clothes with buttons or snaps down the front and at the crotch
- When dressing the infant, bring garments over the limb; do not pull the limb through the sleeve or pants leg
- Using clothing sizes larger than what is needed may prevent pulling, twisting, or getting caught in clothing can cause fractures
- Parents should be advised to choose clothing with wide openings that allows them to slide the garment over the baby's arms or legs without pulling the limbs
- Clothing without ribbons, buttons, pockets, and ruffles is also safer for the baby

# At Home Fracture Care

Fractures may occur no matter how careful you are. Below is a guide to recognize fractures and begin treatment.

## Recognizing Fractures

- Sudden onset of pain or unexplained fussiness may indicate a fracture; consider other reasons for fussiness (ex. colic, constipation, other illness)
- Sensitivity or avoidance of movement of an arm or leg
  - Was an arm or a leg twisted or positioned unusually before the onset of pain? This may help identify the area that is possibly injured
- Sudden cry or outburst from your child; you might actually hear or feel the snap of the bone in some instances

## First Aid

- Avoid further motion of your child and allow time for your child to calm down
- Once your child is calm, very slowly and carefully investigate by gently touching or barely lifting each limb; you can usually pinpoint the location of the fracture
- When you lift or move the fractured bone even slightly, your child will wince or cry out; this reaction will help you determine where the fracture has occurred
- Once you identify an injured area, apply a splint, including the joint above and the joint below
- If the extremity is deformed or crooked, do not attempt to straighten it – splint it in the position in which it lies
- To ease the initial discomfort after a bone fractures, you can administer pain medicine as prescribed by your physician

## Splinting at Home

For babies and small children, positioning on a pillow or a soft protective surface and avoiding movement of the extremity may provide enough comfort. For some fractures, a soft cotton wrap will provide sufficient immobilization ([See photo](#)).

In some instances, your OI care team will give you a splinting kit and will instruct you on how to splint a fracture at home. There are many types of commercial splints available. It is recommended that you have splinting materials with which you are comfortable available at all times (at home and on the road).

If you don't have a kit, there are two other methods that parents have found to be effective:

**Thigh Bone:** A broken leg can often be protected (especially for sleeping) by simply placing a small folded hand towel between the child's legs and wrapping both legs together with an elastic bandage or gauze wrap. The towel will prevent chafing and will lend some rigidity to the legs. Or, cut an oval piece of cardboard that is 4 to 5 inches wide and as long as the child's thigh from hip to knee (or from hip to ankle). Bend it to curve around the leg, cupping the bone like a cast. Pad the cardboard with soft fabric or a blanket. Wrap an elastic bandage or gauze roll loosely around the cardboard brace.



A soft cotton wrap immobilizes the left arm

Wrapping is most easily accomplished by two people, one of whom wraps and one of whom holds the leg. When wrapping with an elastic bandage or gauze roll, roll the bandage on without stretching it to allow for swelling. Leave the toes exposed and frequently check for color changes that indicate a lack of circulation. Watch also for any swelling and color changes in the splinted limb. A deep pink or red color indicates that the splint is too tight; the first step would be to loosen the wrap.



Example of a home splint for the right leg



Right arm is supported with a soft cotton wrap and held to the body with Surgilast

**Upper Arm Bone:** Support the arm against the body, limiting motion as much possible (*See photo*). For an effective temporary sling, simply pin the sleeve of a long-sleeved shirt to the shirt body above and below the wrist and at the elbow.

**Lower Arm Bone:** Pad a magazine with a small towel, wrap it around the arm, and secure it with an elastic bandage or gauze wrap.

In a child old enough to describe pain, ice may be applied with a protective layer between the ice and the skin. Apply ice for up to 20 minutes, then off for 20 minutes. Do NOT apply ice directly to the skin.

Elevation is an important step to help improve swelling and discomfort:

- For a lower extremity, elevate the injured limb, if possible, by laying your child down and gently elevating the leg. The toes should be above the level of the hip
- For an upper extremity, place pillows or small soft blankets under the injured arm. The hand should be above the elbow

## ***Communicating with Your Healthcare Team***

Have a contact or phone number readily available to reach your OI team or your local provider who has experience with OI patients. It is also recommended that families have a letter from the primary OI doctor describing the condition, what to do, and who to contact in case of fracture or emergency.

- Discuss with your provider next steps regarding the role for X-ray or evaluation/follow-up
- Although almost all fractures can be cared for with splinting and scheduled follow-up, seek urgent care if:
  - There is extreme discoloration of the skin indicating potential circulation problems
  - If the limb is quite deformed (crooked)
  - If there is bleeding at the fracture site
  - If there is concern for a neck or spine injury with loss of extremity function or change in bowel or bladder control
  - If medical advice is not otherwise available

## ***Additional Information***

At first, you will want to take your baby or child to the orthopedic surgeon whenever a fracture occurs. As you become more familiar with your child's condition, you will have a better grasp of your child's needs and may begin to be comfortable managing the fractures at home.

Set up a system to access a care team comfortable with OI in order to minimize having to go to an Emergency Room.

After initial splinting, your provider may decide that additional immobilization by casting or splinting is necessary.

## Mental Health and Support

Living with a lifelong health condition like OI can be stressful at times. But many people with OI and their families cope well with both the physical and the social/emotional problems associated with OI to lead happy, interesting, and successful lives. Being aware of trouble areas can help people be better prepared and avoid or minimize some potential problems. By putting support systems in place, additional help will be available when you need it.

Some helpful strategies include:

- Being well informed about OI
- Developing an effective support network
- Resolving the social and emotional needs of the person with OI and their family members as they arise
- Including interesting and fun activities in your schedule

We also recognize that mental health needs are different for every member of your family. Your needs as a parent may be different than the needs of grandparents and even siblings.

### ***Issues that May Arise for Family Members***

**Parents:** When an unborn child, infant, or young child is diagnosed with OI, parents can feel a range of emotions. Sometimes a family waits months or even years for a diagnosis, as the condition is rare. The journey to diagnosis can be stressful with concerns about a child's health paramount. It may come as a relief when the diagnosis is finally made and if child abuse allegations were part of the experience, parents may feel vindicated. Once the diagnosis is made, parents deal with the uncertainty of raising a child with a lifelong medical condition and the challenge of finding physicians experienced with the condition.

Some other emotional issues and decisions that parents may have to deal with are the:

- Impact of the unexpected diagnosis on all members of the family in terms of their emotions, everyday activities, career choices, and finances
- Uncertainty of the condition, such as: when the next fracture might occur; when a child will reach a developmental milestone, such as rolling over; which medical treatment option is the best; what the effects of an experimental treatment might be; or whether vacation or holiday plans will be interrupted by an injury
- Realization that no matter how careful a caregiver tries to be, it is impossible to protect a fragile child from the pain of broken bones
- Struggle to find the right balance between protecting from harm and encouraging the child to try new things
- Meeting the needs of a child who has OI while caring for other children and adults in the family; caring for a child who has OI can be more time consuming than caring for other children
- Coping with fatigue, stress, and sadness that can affect parents' health and relationships and put a strain on marriages



The OI Foundation  
addresses mental health  
aspects of OI in the podcast

***Disability Identity  
and OI***

[www.oif.org/podcast](http://www.oif.org/podcast)

Strategies to help parents cope with the stress of OI include:

- Developing techniques for dealing with risk and uncertainty and a system for coping with broken bones and other medical emergencies
- Creating a support network that includes extended family, friends, and professional services
  - Grandparents often become important parts of the support system
  - Doctors, nurses, hospital social workers, and genetic counselors can provide information about local services
  - OI Foundation can provide information about the condition and available resources
- Cultivating the habit of open and clear communication between all family members, including children
- Paying attention to their own needs for rest, emotional support, and adult friendships

**Siblings:** It is normal for brothers and sisters to feel jealous of the time their parents must devote to caring for a child with OI. Siblings may feel left out, angry, sad, or afraid. They may even feel guilty over not having OI or causing a fracture. Many siblings worry when the child with OI must be hospitalized, or when their parents are away from home. It is common for children to feel resentment when they perceive that the child with OI is treated differently than the rest of the family.

The following strategies for parents can help siblings develop a lifelong affection and respect for each other:

- Communicate openly and calmly with children; information can help children feel more secure and reassured
- Spend special time with children who do not have OI
- Be sensitive to the sibling's point of view
- Expect age appropriate behavior from all children in the family
- Include the child with OI in family projects and daily chores
- Teach all children in the family what to do in an emergency

Siblings have a need for information about OI provided in age appropriate language. They also need rules for playing with their fragile brother or sister. At the appropriate age, they will need information about genetics and their future as parents.

**Grandparents and Others:** As part of the extended family, grandparents, aunts, uncles, cousins, friends, and neighbors will all be affected to some extent when a child has OI. They too have a need for information and support. The OI Foundation sponsors a biennial national conference, an interactive website, and publishes materials that can fill this need. The extended family can be an important part of each family's support system.

For more information on mental health, please check out [www.oif.org](http://www.oif.org).



**You are not alone! List the members of your support group below** (*i.e., family, friends, doctors*). The OI Foundation offers support groups on our website ([www.oif.org](http://www.oif.org)).

**Use the space below to list the support group closest to you and the phone number for the leader, if you ever want to expand your support network.**

# Child Abuse Accusations

Parents of children with OI may be mistakenly suspected of child abuse because of symptoms like fracture. This can be a scary thought, let alone a situation to face; but, there are some ways that you can be prepared.

- Be prepared to answer questions about your child's condition calmly and with understanding when you find it necessary to seek medical help away from your regular doctor
- Carry a letter from your doctor at all times stating that your child has a diagnosis of OI and explaining what that means; many parents also carry a copy of this letter in the glove compartment of the car
- Bring copies of your child's medical records with you when traveling; knowledge of OI is not as widespread as we all would like it to be

# Pain Management and Treatments

## *Pain Management*

People with OI can experience both acute and chronic pain. 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, and more information can be found on the OI Foundation [fact sheet](#) on pain management.

- Heat and ice
- Exercise or physical therapy
- Acupuncture
- Relaxation training, mindfulness, medication, and visual imagery
- Biofeedback
- Medications for pain management

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 sometimes helpful in certain circumstances. It's important to note that serious complications can be caused by either over the counter or prescription medications. Because of 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 medicine.

## *Treatments*

Currently, there is no cure for OI. However, there are a variety of treatment options that your doctor may recommend to you for your child. Treatments for OI are tailored to the needs of each individual child. More information on treatments can be found at [www.oif.org](http://www.oif.org).

**Medications:** Bisphosphonate drugs, which are currently approved by the U.S. Food and Drug Administration (FDA) to prevent and treat osteoporosis are sometimes used to increase bone density in children and adults with moderate and severe OI. Learn more about bisphosphonates at [www.oif.org/factsheets](http://www.oif.org/factsheets). Other drugs that were developed to treat osteoporosis and a drug based on the parathyroid hormone are also used to prevent age-related bone loss in adults who have OI. Treatments under study include growth hormone and gene therapies. The search continues for a drug treatment that is specific to OI.

**Physical Therapy:** Goals for physical therapy include expanding and maintaining function and promoting independence. A typical program includes muscle strengthening and aerobic conditioning. Physical therapy often begins in infancy to counteract the delay in motor skill development many children experience due to OI-related muscle weakness. Adaptive devices may be needed.



### **Occupational Therapy and Safe**

**Exercise:** OT and safe exercise helps with fine motor skills and the selection of adaptive equipment for daily living. As a child with OI grows older and gains more independence, he or she will benefit from continued physical activity. Adults with OI also 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).

**Surgery:** Surgery may be needed to repair a broken bone, correct bone deformities such as bowing, stabilize the spine, or repair tiny bones in the middle ear and improve hearing. Many children with OI undergo a surgical procedure known as rodding where metal rods are inserted into the long bones to control fractures and improve deformities that interfere with function. Both non-expandable and expandable rods are available.

**Healthy Lifestyle:** People with OI benefit from a healthy lifestyle that includes safe exercise and a nutritious diet. Adequate intake of nutrients, such as Vitamin D and calcium, are necessary to maintain bone health. However, extra-large doses of these nutrients are not recommended. Maintaining a healthy weight is important since extra weight adds stress to the skeleton, heart, and lungs and reduces the ability to move easily. In addition, people with OI should avoid smoking, secondhand smoke, excessive alcohol or caffeine consumption, and steroid medications, which all reduce bone density.

**Other Treatments:** Other treatments include hearing aids, crowns for brittle teeth, supplemental oxygen for people with breathing problems, and mobility aids.



The following podcasts dive deeper into treating children with OI

***Surgical Updates on Treating Children with OI***

***Physical/Occupational Therapeutic Strategies for Children with OI***

***Rodding Surgery and OI***


at [www.oif.org/podcast](http://www.oif.org/podcast)





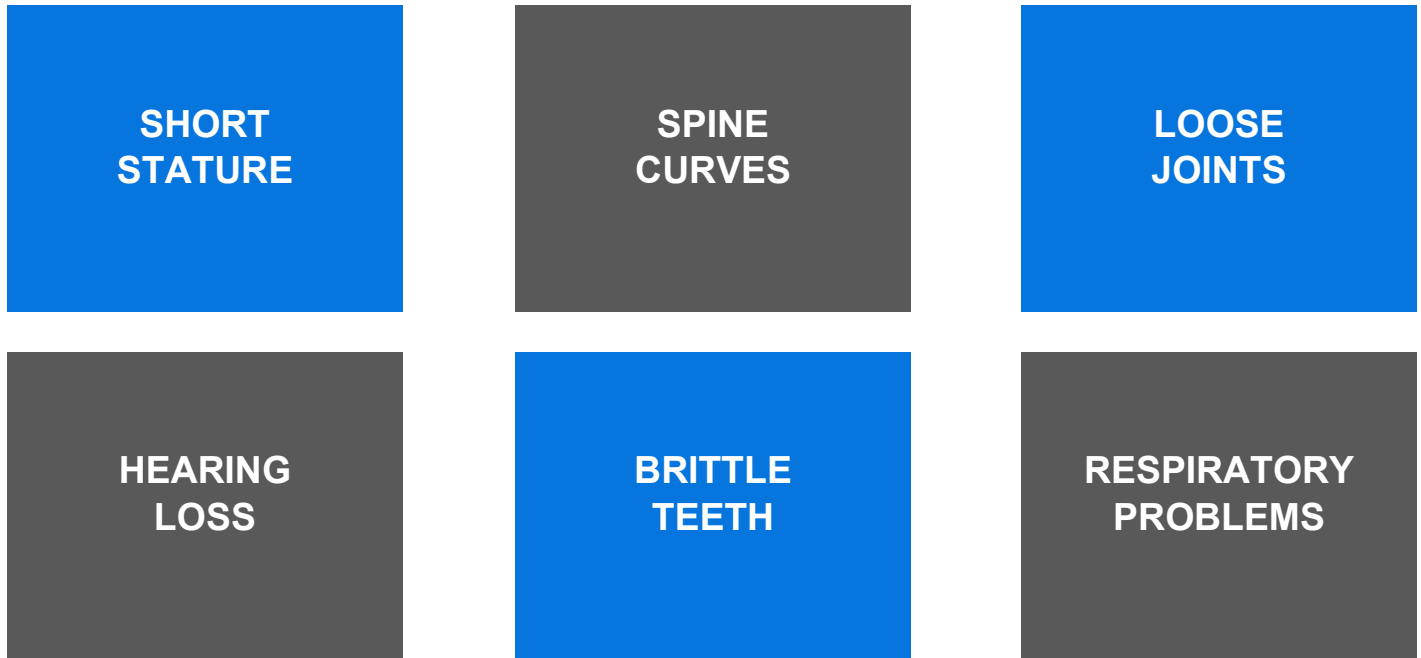
# SHARE WITH YOUR DOCTOR

Parents are encouraged to keep a log (electronic or paper) with important information like reports, fractures and questions to help you remember details between appointments with your provider(s).



## Introduction and Type Chart

There are more than 15 types of OI, ranging from mild, to moderate or severe. The clinical features of the condition vary by type, within types, by age, and even within the same family. Below are some of the distinguishing features of the major types of OI.



To read the full list of characteristics, or to view the OI specific Growth Chart, visit [www.oif.org/informationcenter](http://www.oif.org/informationcenter).

### *Types of OI*

Hundreds of mutations involving more than a dozen genes have been associated with OI. Currently, there are several systems for addressing the clinical and genetic variability of OI. Some focus on clinical severity as the overriding measure while others focus on specific gene causes to distinguish types. Others integrate both. Broadly speaking, based on severity of the clinical picture, OI can be grouped into four classes – mild, moderate, severe, and extremely severe or lethal. The great majority of OI cases (85-90%) are inherited in an autosomal dominant manner where gene mutations affect the quantity or quality of type I collagen. These cases are described in Types I-IV on this chart. Genotype/phenotype relationships have not been clearly established for either dominant or recessive OI and knowing the OI type is not predictive of future function. Knowledge of the mutation provides useful information for genetic counseling. Knowledge of the Type or degree of severity can help establish an initial treatment plan.

The following chart is a genetic classification system that includes a description of severity. Additional clinical features are included at [www.oif.org/informationcenter](http://www.oif.org/informationcenter).

## Nosology of Osteogenesis Imperfecta<sup>4</sup>

OI TYPE	INHERITANCE	PHENOTYPE	GENE MUTATION
<b>Defects in collagen synthesis, structure, or processing</b>			
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
<b>Defects in bone mineralization</b>			
V	AD	Variable, Distinctive Histology	IFITM5
VI	AR	Moderate/Severe	SERPINF1
<b>Defects in collagen modification</b>			
VII	AR	Severe (Hypomorphic)	CRTAP
VIII	AR	Severe/Lethal	LEPRE1
IX	AR	Moderate/Lethal	PIIB
XIV	AR	Severe	TMEM38B
<b>Defects in collagen folding and cross-linking</b>			
X	AR	Severe/Lethal	SERPINH1
XI/BRKS1	AR	Mild/Severe	FKBP10
BRKS2	AR	Moderate/Severe	PLOD2
<b>Defects in osteoblast development with collagen insufficiency</b>			
XII	AR	Severe	SP7
XV	AR	Severe	WNT1
XVI	AR	Severe	CREB3L1
XVII	AR	Progressive Severe	SPARC
XVIII	XR	Moderate/Severe	MBTPS2

**Abbreviations:**

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

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

# Collecting Vital Signs and Handling

## Collecting Vital Signs

Children with OI undergo numerous medical procedures, when collecting vital signs on a baby with OI:

- Keep the process as short and painless as possible
- If possible, have a parent hold the child
- Before drawing blood, ask the patient or caregiver to identify sites of previously successful venipunctures
- To accommodate small veins, use the highest gauge needle feasible to obtain samples
- Avoid direct contact with skin when tying a tourniquet by placing it over a patient's sleeve (do not use a tourniquet if the patient has an acute humeral fracture)
- For patients with severe OI, a manual blood pressure cuff is preferable over a tourniquet because it disperses pressure on the underlying bone (the cuff should not be inflated over 80 mm Hg)
- Measuring blood pressure in a patient with OI requires precautions such as weight-distributed support, protective padding, avoidance of impacts, twisting, and straightening of bent limbs. Blood pressures are obtained only when necessary
- Appropriately sized equipment should be used
- A manual blood pressure cuff is recommended
- If possible, avoid taking blood pressure on an arm that is bowed or has been repeatedly fractured. If both arms have malformations, blood pressure may be obtained on the thigh
- Measure length instead of height in patients who cannot stand- because leg lengths may differ, measure both sides of the body
- A platform scale may be needed to weigh non-ambulatory patients
- People with OI generally have higher baseline body temperature and greater sensitivity to heat than other patients

## Handling

The infant who has OI has some special characteristics. The infant may have an unusually soft skull, startle easily, and have bone deformity and fractures, often of the ribs or long bones, that are in various stages of healing. When handling a baby with OI:

- All movements should be slow, methodical, and gentle
- Never push, pull, twist, bend, apply pressure, or try to straighten arms or legs
- Infants with OI should not be picked up under the axillae or around the rib cage because this can cause rib fractures
- The head and trunk should be supported with one hand while the other hand supports the buttocks
- Keep fingers of the caregiver spread apart to provide a wider base of support and an even distribution of support pressure
- When lifting or turning the baby for feeding, dressing, or diapering, apply support to the broadest possible area. One safe and effective way is to slide one hand underneath the child's buttocks to the back with some support under the head. Place the other hand on the chest and abdomen "sandwiching" the baby between the two hands
- When diapering the baby, do not lift the baby by the ankles (as this could result in a fracture). Slide your hand under the buttocks to gently roll the baby onto one side to remove/replace the diaper
- Infants with fractures may be immobilized with a cast or splint to reduce motion and provide stabilization. Such infants must not be placed prone on their stomachs because suffocation can occur
- Care should be taken when changing dressings and bedding to protect the infant's arms, wrists, and fingers

- When dressing the infant, bring garments over the limb; do not pull the limb through the sleeve or pants leg. Pulling, twisting, or getting caught in clothing can cause fractures
- It is important that babies with OI receive affection and are held and touched by parents and other caregivers

## Feeding and Bedding

### *Feeding*

Infants with OI can be poor feeders. Some babies display a weak sucking reflex and may require small, frequent feedings. The combination of small stature, feeding problems, and slow growth may be mistaken for failure to thrive. Here is the information you need to know about feeding for a baby with OI.

**Breastfeeding:** Breast milk is an excellent source of calories for virtually all infants including those with OI. Breastfeeding can create a special bond between the mother and child. Babies with all but the most severe forms of OI should be capable of being breastfed. However, those with the most severe forms of OI may have breathing difficulties that interfere with the ability to suck. Rapid respirations can predispose to aspiration. If the baby is not able to breastfeed, the mother may opt to pump breast milk and feed the child breast milk from a bottle, nasogastric tube, or G-tube.

**Handling:** When feeding the infant, the mother should be especially careful to avoid having the baby positioned with an arm behind the back or a leg pressed against the mother's body in such a way as to put pressure on it at an abnormal angle.

**Burping:** Burping should be done very gently to reduce the chance of fractures, especially of the ribs. Soft taps, possibly with padding over the hand, are recommended. To pick up the infant for burping, lay the baby on his/her back while the caregiver bends over to pick up the infant. The caregiver's shoulder should very gently touch the baby at which point the infant is supported under the back and positioned on the shoulder as the caregiver moves up and backwards. Gently rubbing the baby's back while taking gentle bouncing steps may also be beneficial.

### *Bedding*

A standard crib mattress is the most suitable for the baby with OI. Waterbeds and soft bedding should never be used.

# Fracture Care, Pain Management and Treatment

OI is characterized by frequent bone fractures. As a result, people with this disorder may spend a considerable amount of time immobilized, often in a cast. Understanding the different methods that are used to immobilize a fracture as well as the proper techniques for taking care of a cast will enable them to play a more active role in their recovery.

## **Fracture Care**

Correct immobilization is critical in promoting proper healing of fractures, relieving pain, and allowing some freedom of movement during the healing process. The following is a brief listing of the different forms of immobilization that may be used. A more extensive list can be found on [www.oif.org](http://www.oif.org).

**Plaster Cast:** commonly used for the initial treatment of a fracture because it is pliable and economical. It is generally used in the acute stage of fracture healing because it is easily molded to the contour desired by the physician to hold the fracture in the correct position.

**Fiberglass Cast:** are lighter and stronger and the exterior is more water resistant than plaster. If the cast becomes wet, it will not change form, but the padding and lining beneath it will remain wet and irritate the skin. When a fiberglass cast is used in conjunction with a GORE-TEX cast liner, the person can participate in activities involving water with no special drying procedures.

**Brace:** used to maintain a fracture while allowing adjacent joints the freedom of movement.

**Splint:** used to immobilize and maintain a particular position of a bone or joint. The splints are secured to the limb with elastic bandages.

Immobilization should be kept to a minimum and weight bearing, or other functional use of the affected extremity, should be encouraged as quickly as possible to prevent bone loss. Casting and splinting are usually administered by an orthopedic technologist, working under a physician's directions, who has been trained in proper cast application and has an understanding of OI.

## **Pain Management**

People with OI experience both acute and chronic pain. Pain management requires adequate assessment and a personalized plan that provides a variety of strategies for coping with and managing pain. Pain management often requires a multidisciplinary approach. 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, and more information can be found on the OI Foundation [fact sheet](#) on pain management.

- Heat and ice
- Transcutaneous electrical nerve stimulation (TENS)
- Exercise or physical therapy
- Acupuncture
- Relaxation training, mindfulness, medication, and visual imagery
- Biofeedback
- Medications for pain management\*

\*Serious complications can be caused by either over the counter or prescription medications. Because of small stature, many people who have OI cannot safely take the standard dose for either children or adult medications.

## Treatments

**Physical Therapy:** Goals for physical therapy include expanding and maintaining functioning and promoting independence. A typical program includes muscle strengthening and aerobic conditioning. Physical therapy often begins in infancy to counteract the delay in motor skill development many children experience due to OI-related muscle weakness. Adaptive devices may be needed.

**Occupational Therapy (OT) and Safe Exercise:** OT and safe exercise helps with fine motor skills and selection of adaptive equipment for daily living. As a child with OI grows older and gains more independence, he or she will benefit from continued physical activity. Adults with OI also 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).

**Surgery:** Surgery may be needed to repair a broken bone, correct bone deformities such as bowing, stabilize the spine, or repair tiny bones in the middle ear and improve hearing. Many children with OI undergo a surgical procedure known as rodding where metal rods are inserted into the long bones to control fractures and improve deformities that interfere with function. Both non-expandable and expandable rods are available.

**Medications:** Bisphosphonate drugs, which are currently approved by the U.S. Food and Drug Administration (FDA) to prevent and treat osteoporosis are sometimes used to increase bone density in children and adults with moderate and severe OI. Other drugs that were developed to treat osteoporosis, as well as a drug based on the parathyroid hormone, are also used to prevent age-related bone loss in adults who have OI. Treatments under study include growth hormone and gene therapies. The search continues for a drug treatment that is specific to OI.

**Growth:** Except for the mildest forms of OI, the length and weight of children with OI often falls below standard pediatric curves by 1 year of age. Sometimes this must be distinguished from failure to thrive or cardiorespiratory problems. Standardized growth curves for girls and boys with types III and IV OI are available to see how a particular child compares to others with the same OI type in weight and length. They can be downloaded free from this medical article:

Barber, L.A.\*, Abbott, C.\*, Nakhate, V.\*, Dang Do, A., Blissett, A.R., and Marini, J.C. (2018)  
Longitudinal growth curves for children with classical osteogenesis imperfecta (types III and IV) caused by structural mutations in type I collagen. *Genet Med* 21:1233-39

**Healthy Lifestyle:** People with OI benefit from a healthy lifestyle that includes safe exercise and a nutritious diet. Adequate intake of nutrients, such as vitamin D and calcium, are necessary to maintain bone health. However, extra-large doses of these nutrients are not recommended. Maintaining a healthy weight is important since extra weight adds stress to the skeleton, heart, and lungs and reduces the ability to move easily. In addition, people with OI should avoid smoking, secondhand smoke, excessive alcohol or caffeine consumption, and steroid medications, which all reduce bone density.

**Other Treatments:** Other treatments include hearing aids, crowns for brittle teeth, supplemental oxygen for people with breathing problems, and mobility aids.

More information on treatments can be found at [www.oif.org](http://www.oif.org).

# Discharge Planning and Pediatric Care Team

## *Discharge Planning*

The following considerations should be made for when a patient is ready to be discharged from the hospital:

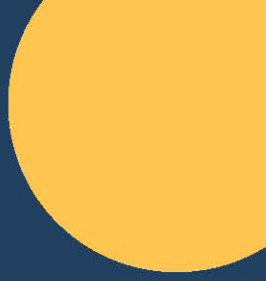
- Parent education prior to discharge should include explanations and demonstrations of the procedures for holding, lifting, diapering, and general infant care. The demonstrations will ensure that the parents are comfortable with each skill. If possible, demonstrate these skills to other family members and friends who will be assisting in caring for the child at home
- Because new fractures are to be expected, parents should also receive training through their pediatric orthopedist in how to recognize a fracture and how to protect the injured body part while traveling to the hospital or clinic
- A letter on hospital letterhead, stating that the child has been diagnosed with OI can facilitate obtaining appropriate care if the family returns to the Emergency Department or needs to seek treatment at a different facility.
- A car seat or, in some cases, a car bed will be necessary at the time of discharge. This equipment is geared to the child's weight and ability to sit up. Car seats are designed for children under 20 pounds in weight. Car seats should also include a well-padded harness and a head hugger support pillow. This type of U-shaped pillow is commercially available and is used to position the baby's head at midline. Small rolls or towels or other padding can be added to hold the child's hips in line. All car seats should be approved for safety and anchored correctly in the vehicle. It is not safe to place foam padding between the plastic shell and the padded cover
- It is always best to place a car seat in the back seat of the vehicle
- Parents should be advised to choose clothing with wide openings that allows them to slide the garment over the baby's arms or legs without pulling the limbs. Clothing without ribbons, buttons, pockets, and ruffles is also safer for the baby
- Discharge planning should include referral to local OI support groups or to another family with a child with OI (with the family's approval)

## *Pediatric Care Team*

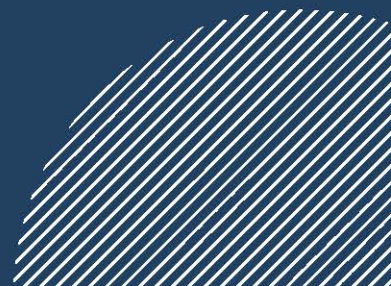
In addition to a referral to local support groups, there are many other clinical specialties to refer a family to so that they can have multidisciplinary care, including but not limited to: a pediatrician, a pediatric orthopedist, a physical therapist, an audiologist, an optometrist, a registered dietician, mental health professionals, and any other services that offer support for children with OI.

In many cases, the OI Foundation can assist with referrals. Parents and hospital personnel are encouraged to contact the OI Foundation for assistance.





# RESOURCES



# Osteogenesis Imperfecta Foundation Resources

The OI Foundation can support you and your family as your child grows and has different needs. We look forward to welcoming you into the community and hope that we can provide the information you and your family needs as your child grows. Here are some of the resources that we have available:

**OI Foundation Website:** Our website ([www.oif.org](http://www.oif.org)) has information about all types of topics related to OI. Fact sheets and other resources provide medically accurate information and can be printed out at any time.

**Clinic Directory and Physician Referral System:** The OI Foundation website lists clinics that provide multidisciplinary medical care by state. The organization can also help locate a doctor by specialty. Contact the OI Foundation Health Educator at (301) 947-0083, for more information.

**OI Foundation Events:** The OI Foundation hosts a national conference every two years on various OI topics. The conference provides an opportunity for the OI community to come together, get to know one another and socialize, while learning the latest on OI research. The organization also holds day-long, regional conferences on important OI topics. The location of the regional conferences change. All events are listed at: [www.oif.org/events](http://www.oif.org/events).

**Support Networks:** The OI Foundation also lists support groups by state on its website. These community-based get-togethers are first and foremost a social gathering. Speakers may be scheduled from time to time to provide information on topics of interest to the group. Visit [www.oif.org/supportnetworks](http://www.oif.org/supportnetworks) for more information.

**Social Media:** Follow the OI Foundation on Facebook, Instagram, and Twitter to learn more about progress we are making and the latest news and research.

**Contact Us:** The OI Foundation is here for you! Our email address is [bonelink@oif.org](mailto:bonelink@oif.org) and our phone number is (301) 947-0083 or toll free (844) 889-7579.



Patient's Name \_\_\_\_\_

## CAUTION!! I HAVE OSTEOGENESIS IMPERFECTA (OI)

**Handle me gently at times.**

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

- Fragile bones
- Fragile skin
- Loose joints
- Fragile teeth
- Lying flat may be difficult
- I may have respiratory problems

**SUPPORT** my entire body when lifting or turning me.

- **DO NOT** pull on my arms 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** using a pediatric cuff if possible.

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

**USE** pediatric-sized equipment if 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.**

Osteogenesis Imperfecta Foundation  
[www.oif.org](http://www.oif.org) • [bonelink@oif.org](mailto:bonelink@oif.org) • 844-889-7579 • 301-947-0083

# Glossary

<b>Audiologist</b>	A medical professional who diagnoses and treats hearing and balance problems.
<b>Bisphosphonates</b>	A class of drugs, usually used to treat osteoporosis, that prevent loss of bone density. Bisphosphonates are sometimes used in children with OI.
<b>Bone Density Testing</b>	A test that helps to estimate bone density and the likelihood of breaking a bone.
<b>Brace</b>	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.
<b>Collagen</b>	The main structural protein in various connective tissues in the body.
<b>Dietician</b>	A medical professional that is an expert in nutrition.
<b>DEXA Scan</b>	A non-invasive test that measures bone mineral density. It is used to see if a person is at risk for a fracture.
<b>Echocardiogram</b>	A test of the action of the heart using ultrasound waves to produce a visual display; used for the diagnosis or monitoring of heart disease.
<b>Failure to Thrive</b>	A child is said to have failure to thrive when they don't meet recognized standards of growth.
<b>Fiberglass Cast</b>	A plaster cast made from fiberglass material, which is lighter than plaster of Paris.
<b>Fracture</b>	A crack or break in a hard object or material, typically a bone.
<b>Geneticist</b>	A medical professional who is an expert in heredity and the variation of inherited characteristics.
<b>Genetic Counselor</b>	A medical professional that educates and provides support to individuals and families at risk for, or diagnosed with, genetic conditions.
<b><i>Handle Me With Care</i> Flyer</b>	OI Foundation resource that can be printed and given to medical professionals that explains how to best work with people with OI.
<b>Head Hugger Support Pillow</b>	A type of pillow used in car seats to provide support for the head and neck.
<b>Kyphosis</b>	An excessive outward curvature of the spine, which causes a rounding or hunching of the back.
<b>Mental Health Professionals</b>	Can include, but is not limited to, psychologists, psychiatrists, psychiatric nurses, and mental health counselors who provide support on social and emotional issues, including grief.

<b>Mosaicism</b>	The state of being composed of cells of two genetically different types.
<b>Neonatal</b>	Relating to newborn children.
<b>Neonatologist</b>	A medical professional who is trained to handle complex and high-risk situations with newborns.
<b>Neonatal Intensive Care Unit (NICU)</b>	An intensive care unit in the hospital that specializes in the care of ill or premature newborn infants.
<b>Nurse</b>	A person trained to care for the sick, especially in a hospital.
<b>Obstetrician</b>	A medical professional who specializes in pregnancy, childbirth, and the reproductive system.
<b>Occupational Therapy</b>	A form of therapy that encourages rehabilitation through the performance of activities required in daily life.
<b>Orthopedist</b>	A medical professional who works to correct deformities or impairments of the skeletal system.
<b>Osteogenesis Imperfecta (OI)</b>	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.
<b>Osteogenesis Imperfecta Type I</b>	The most common and most mild type of OI. People with Type I OI may be average or near-average height.
<b>Osteogenesis Imperfecta Type II</b>	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.
<b>Osteogenesis Imperfecta Type III</b>	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.
<b>Osteogenesis Imperfecta Type IV</b>	A moderate type of OI that is between Type I and Type III in severity and height that has mild to moderate bone deformity and spinal curvature and compression fracture of the vertebrae. People with Type IV OI usually have a barrel-shaped rib cage.
<b>Osteogenesis Imperfecta Type V</b>	A moderate type of OI that is similar to Type IV OI in appearance in symptoms. 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 a mutation that is not in the collagen pathway and has dominant inheritance.
<b>Osteogenesis Imperfecta Type VI</b>	A moderate type of OI that is extremely rare and is similar to type IV in appearance. Type VI OI is characterized by a mineralization defect in biopsied bone. The mutation for Type VI OI is not in the collagen pathway and is inherited in a recessive manner.
<b>Osteogenesis Imperfecta Type VII</b>	OI Type VII is a severe type of OI that has a recessive inheritance pattern.

<b>Osteogenesis Imperfecta Type VIII</b>	OI Type VIII is a very severe type of OI that is similar to Type II but has a recessive inheritance pattern. In Type VIII OI, there is a severe growth deficiency and under mineralization of the skeleton.
<b>Osteogenesis Imperfecta Foundation (OIF)</b>	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.
<b>Palliative Care</b>	Specialized medical care for people living with a serious illness. This type of care is focused on providing relief from the symptoms and stress of the illness to improve the quality of life for the individual and the family.
<b>Physical Therapy</b>	The treatment of disease, injury, or deformity by physical methods such as massage, heat treatment and exercise.
<b>Plaster Cast</b>	A bandage stiffened with plaster of Paris, molded to the shape of the limb that is broken, and is used to support and protect it.
<b>Preeclampsia</b>	A condition in pregnancy that is characterized by high blood pressure.
<b>Prenatal</b>	Before birth; during or relating to pregnancy.
<b>Procedure</b>	A surgical operation.
<b>Pulmonary Function Test</b>	Non-invasive tests that show how well the lungs work.
<b>Social Workers</b>	Professionals who are trained to help individuals and families to develop their skills and their ability to use their resources and community resources to resolve problems.
<b>Spica Cast</b>	Used to immobilize the hip or thigh. It is used to facilitate healing of hip joints or fractured femurs.
<b>Splinting</b>	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.
<b>Surgery</b>	The treatment of injuries or disorders of the body by incision or manipulation, especially with instruments.
<b>Ultrasound</b>	A type of test that uses sound or other vibrations that have an ultrasonic frequency that produces a medical image.
<b>Vital Signs</b>	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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