

## Osteogenesis Imperfecta

### Goals and Objectives

#### Course Description

"Osteogenesis Imperfecta" is an asynchronous online continuing education course for occupational therapists and occupational therapy assistants. This course presents updated information about Osteogenesis Imperfecta including sections on etiology, symptomology, diagnosis, assessment, therapeutic intervention, medical services, and social services.

#### Course Rationale

The purpose of this course is to present occupational therapists and occupational therapy assistants with current information about Osteogenesis Imperfecta. A greater understanding of Osteogenesis Imperfecta will enable occupational therapists and occupational therapy assistants to provide more effective and efficient rehabilitative care to individuals affected by this condition.

#### Course Goals and Objectives

Upon completion of this course, the participant will be able to:

1. Identify the symptomology of OI
2. Differentiate between the different types of OI and identify their causes.
3. Recognize and understand the tests utilized to diagnose OI.
4. Define safe OI neonatal and infant care.
5. Recognize the primary care needs of individuals with OI.
6. Recognize the precautions associated with providing medical care to individuals with OI.
7. Identify the special care requirements of OI individuals receiving ER services
8. Identify the special care requirements of OI individuals receiving surgical services.
9. Identify the therapeutic needs of individuals with OI.
10. Identify therapeutic strategies to address the needs of individuals with OI.
11. Recognize the social services needs of individuals with OI.
12. Recognize other OI associated health issues.
13. Identify social, emotional, and family issues often affecting individuals with OI.

**Course Provider** – Innovative Educational Services

**Provider Contact Information** – [information@cheapceus.com](mailto:information@cheapceus.com)

**Course Instructor** – Michael Niss, DPT

**Financial/Non-financial Disclosure** - Neither the Provider nor the instructor have any financial or non-financial conflict of interest related to the presentation of this CE program.

**Target Audience** – Occupational Therapists, Occupational Therapy Assistants

**OT Scope of Practice** – OT Service delivery (evaluation, intervention, outcomes); Foundational knowledge (Human body diagnoses and conditions)

**Course Educational Level** – This course is applicable for introductory learners.

**Course Prerequisites** – None

**Method of Instruction** – Distance Learning – Independent; Asynchronous online text-based home study

**Location** - Cheapceus.com

**Date** – Continuously available on-demand

**Course Completion Requirements / Criteria for Issuance of CE Credits** – Completion of instructional materials and a score of 70% or greater on the course post-test.

**Continuing Education Credits** – Four (4) contact hours / .4 AOTA CEUs / NBCOT 5 PDUs

**Course Fee** - \$39.95

**Registration Information** – No pre-registration required; available on-demand at Cheapceus.com

**Special Needs Requests** – Email: [information@cheapceus.com](mailto:information@cheapceus.com) or phone: 954-663-4101

**Cancellation by the Learner** – Learners may cancel their participation at any time and receive a full refund of all paid fees.

**Cancellation by the Provider** – Registrants for the cancelled activity will be contacted by an IES staff member via phone call or email within 12 hours of the cancellation decision. If the decision to cancel is made less than 14 hours prior to the event, each participant will be contacted no later than 2 hours prior to the scheduled start time of the activity. Registrants will receive a full refund if an activity is cancelled by the Provider. The refund will be processed back to the Registrant within 12 hours of the cancellation.

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**Refund Policy** - Unrestricted 100% refund upon request. The request for a refund by the learner shall be honored in full without penalty or other consideration of any kind. The request for a refund may be made by the learner at any time without limitations before, during, or after course participation.



Innovative Educational Services is an AOTA Approved Provider of professional development. PD activity approval ID# 5471. This Distance Learning – Independent PD activity is offered at .4 CEUs; Introductory; OT Service delivery, Foundational knowledge. The assignment of AOTA CEUs does not imply endorsement of specific course content, products, or clinical procedures by AOTA.

# Osteogenesis Imperfecta

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## Osteogenesis Imperfecta

### Overview

Osteogenesis imperfecta (OI), also known as brittle bone disease, is a genetic disorder of connective tissue characterized by bones that fracture easily, often from little or no apparent trauma. It is highly variable in severity from patient to patient, ranging from very mild to lethal. In addition to having fractures, people with OI often have muscle weakness, joint laxity, skeletal malformations, and other connective tissue problems.

The prevalence of OI is approximately 1 in 20,000, including patients diagnosed after birth. OI occurs with equal frequency among males and females and among all racial and ethnic groups. Patients with OI have the full range of intellectual capabilities as seen in the general population. There is nothing inherent in the disorder that affects cognitive abilities. Life expectancy varies according to the underlying severity of the disorder and ranges from very brief (Type II OI) to average. Medical treatment for OI is increasingly understood.

Patients with osteogenesis imperfecta usually have a faulty gene that instructs their bodies to make too little type I collagen or poor quality type I collagen. Type I collagen is the protein "scaffolding" of bone and other connective tissues. Inheritance, in nearly all cases, follows an autosomal dominant pattern, although sporadic cases are common. When there is no family history of OI, the disease is caused by new dominant mutations.

Patients are often knowledgeable about their health status and the problems associated with OI. Accordingly, the opinions, requests, and instructions of adult patients and parents of children with OI should be respected.

### Symptomology

Depending on the severity of OI, the following characteristics may be seen:

- skeletal malformation
- short stature – Growth impairment is severe in all those individuals with Type II and Type III OI, moderate in those with Type IV, and relatively less in those with Type I.
- muscle weakness
- ligamentous laxity
- smooth, thin skin
- triangular face

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- dental manifestations – Dentinogenesis imperfecta is present in about 50 percent of patients with OI. Deciduous teeth are usually more severely affected than permanent teeth.
- blue sclerae – Approximately 50 percent of people with OI have blue, purple, or gray-tinted sclerae.
- respiratory complications – Lung complications, such as pneumonia, represent a significant cause of death for those with Type II and III OI. Pneumonias are seen in children and adults, and cor pulmonale, a type of heart failure, is seen in adults.
- cardiac complications – Mitral valve prolapse (laxity) is seen but is not as common as in some other connective tissue disorders.
- hearing loss – In those with OI, hearing loss is frequent.
- thermal instability – Those with OI experience slightly higher than normal body temperature, sensitivity to heat and cold, excessive sweating, pseudomalignant hyperthermia after anesthesia.
- blood vessel fragility – Patients may exhibit easy bruising, frequent nosebleeds, and, in a small number of patients, profuse bleeding when injured.
- neurologic manifestations – Basilar invagination of the skull, hydrocephalus, and syringohydromyelia of the spinal cord may be seen in patients with the more severe forms of OI.

### Prognosis

The prognosis for an individual with OI varies greatly depending on the number and severity of symptoms. Respiratory failure is the most frequent cause of death for people with OI, followed by accidental trauma. Despite numerous fractures, restricted activity, and short stature, most adults and children with OI lead productive and successful lives. They attend school, develop friendships and other relationships, have careers, raise families, participate in sports and other recreational activities, and are active members of their communities.

### Types of OI

In 1979, Sillence and others devised a classification scheme that divides OI into four types based on clinical, radiographic, and genetic distinctions. Features of OI vary not only between types but within each type as well. Patients with OI may present with some but not all of the clinical features. Children and adults with milder OI may have few obvious signs. Some patients appear to have characteristics of several types. Patients may walk unassisted; require the assistance of walkers, crutches, or braces; or be wheelchair-dependent. All types of OI may include dentinogenesis imperfecta and varying degrees of blue sclera.

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The frequency of fractures may decrease after puberty. An increase in fractures may be seen in women following menopause and in men in later life.

While the Sillence classification is part of the commonly accepted language of OI, therapists are urged to look beyond type alone. The key to optimal care is to be aware of the patient's specific symptoms and capabilities and to treat each patient individually.

- **Type I** – Mildest form of the disorder. Manifests with relatively few fractures, minimal limb deformities, blue sclera, and high incidence of hearing loss. Stature may be average or slightly shorter than average for the unaffected family members. Hearing loss onset is primarily in young adulthood but may occur in early childhood. Some patients have few fractures or obvious signs of OI. Some patients experience multiple fractures of the long bones, compression fractures of the vertebrae, and have chronic pain. Dentinogenesis imperfecta may or may not be present. Life expectancy seems to be normal.
- **Type II** – Most severe form; features severe osteoporosis. Infants are frequently premature or stillborn and are small for gestational age. Multiple fractures in the womb lead to bowing and shortening of the long bones at birth. The head is large for body size, with severe undermineralization. The rib cage is small and narrow, and palpation of the rib cage reveals "beading" from calluses due to rib fractures *in utero*. The sclerae are almost uniformly dark blue/gray. In the newborn period, it can be difficult to distinguish between Type II and severe Type III OI. Infants with Type II usually die in the immediate postnatal period from respiratory and cardiac complications. Rare cases of infants surviving into childhood have been reported.
- **Type III** – Most severe type for those patients who survive the perinatal period. Multiple long bone fractures may be present at birth but without the severe thoracic malformation seen in Type II OI. Frequent fractures of the long bones, tension of muscle on soft bone, and disruption of the growth plates lead to bowing and progressive malformation with short stature. Marked short stature, kyphoscoliosis, compression fractures of the vertebrae, and pectus carinatum or pectus excavatum occur frequently. The head is large for body size. Sclera may be white or tinted blue, purple, or grey, and dentinogenesis imperfecta may be present or absent. Patients with Type III are generally diagnosed at birth due to multiple fractures. Many patients with Type III use wheelchairs or other mobility aids. Some are independent ambulators within the home. Use of assistive devices to independently perform activities of daily living is common. Surgery may be required to support and straighten bowed limbs. Life span may be somewhat reduced. While some individuals are living into their sixties and seventies, there appear to be clusters of mortality due to pulmonary complications in early childhood, teens, and thirties to forties.

- **Type IV** – Moderately affected, with the diagnosis possibly made at birth but more frequently later, because the child may not fracture until he or she is ambulatory. Bowing of the long bones is present to a lesser extent than in Type III. Patients have moderate-to-severe growth retardation, which is one factor that distinguishes them clinically from Type I OI. Scoliosis and ligamentous laxity may also be present. Dentinogenesis imperfecta may be present or absent. Although the Sillence classification indicates that patients have white sclera, blue sclera have also been seen. Type IV OI can range in severity from similar to Type I to resembling Type III. Life span is not affected.

Recently, researchers have reported additional types that do not involve a defect of type I collagen. Clinically, these patients are similar to Type IV OI. Additional radiographic or histologic data are required to diagnose Types V and VI.

- **Type V** – Moderate in severity and similar to Type IV but also characterized by large hypertrophic calluses that develop at sites of fractures or surgical procedures. Calcification of the membrane between the radius and ulna restricts forearm rotation.
- **Type VI** – Extremely rare, moderate in severity, and only identified through bone biopsy.

### Additional Forms of OI

The following conditions are rare, but they feature fragile bones plus other significant symptoms. More detailed information on them can be found in *Pediatric Bone: Biology and Diseases*, Glorieux et al, 2003.

- **Osteoporosis-Pseudoglioma Syndrome:** This syndrome is a severe form of OI that also causes blindness. It results from mutations in the low-density lipoprotein receptor-related protein 5 (LRP5) gene.
- **Cole-Carpenter Syndrome:** This syndrome is described as OI with craniosynostosis and ocular proptosis.
- **Bruck Syndrome:** This syndrome is described as OI with congenital joint contractures. It results from mutations in the procollagen-lysine, 2-oxoglutarate 5-dioxygenase 2 (PLOD2) gene encoding a bone-specific lysyl-hydroxylase. This affects collagen crosslinking.
- **OI/Ehlers-Danlos Syndrome:** This recently identified syndrome features fragile bones and extreme ligament laxity. Young children affected by this syndrome may experience rapidly worsening spine curves.

## Etiology

With rare exceptions, OI results from mutations in the type I collagen genes and is considered to be a dominantly inherited disorder. On the basis of a limited number of population surveys, the overall frequency of OI in the general population is about 1 in 20,000. Because some infants die at birth and would not be included in these surveys, the birth incidence is slightly higher, perhaps 1 in 15,000 -18,000 births. In families in which OI occurs in more than one generation with clearly dominant inheritance, the risk of recurrence of OI is 50 percent for each pregnancy.

When parents who have no symptoms of OI have a child with OI, they will inquire about how this occurred in their family and about the risk of recurrence. For the great majority of these families (about 90 percent), their child's OI was caused by a new mutation that took place in the egg or sperm near the time of conception. Their risk of recurrence in subsequent pregnancies is approximately equal to the risk of OI in the general population. In the remainder of these families (about 10 percent), the child's OI results from mosaicism for the mutation in one parent. A mosaic parent has the mutation in some of the cells of his or her body, including some of the egg or sperm cells. The mosaic parent usually appears to be unaffected or only mildly affected. Parental mosaicism can be determined by genetic testing. For these parents, the risk of subsequent affected children is between 10 and 50 percent per pregnancy.

There are very unusual forms of OI that seem to be inherited in a recessive fashion, which means that each parent is a carrier and contributes one altered gene each to their child, who is then affected with OI.

Another aspect of OI genetics is its prevalence in the general population, that is, its occurrence among all living individuals. Individuals with OI will have children of their own. In general, this occurs more frequently at the milder end of the OI spectrum. When prevalence is considered, children with OI who are born into families with OI will constitute a larger proportion of cases with OI. In a recent survey in Finland (Kuurila et al, 2000), about 65 percent of individuals with OI were in families where a prior generation was affected, and the remaining 35 percent were isolated cases without a family history.

Osteogenesis imperfecta is a disorder of connective tissue. Defects in the structure or quantity of type I collagen cause most cases of OI. Type I collagen is the primary structural protein of bone and skin. It is composed of three chains that are twisted together to form a triple helix. Two of the chains are identical. These are called the alpha 1, or  $\alpha 1$ , chains. The third chain is similar to the first two but not identical to it. This is called the alpha 2, or  $\alpha 2$ , chain.

Each of the three chains is made up of uninterrupted repetitions of the amino acid triplet Gly-X-Y, where Gly is glycine, X is often proline, and Y is frequently hydroxyproline. The presence of a glycine at every third position along the chains

is crucial for proper folding of the collagen helix inside the cells where it is produced. Glycine is the smallest amino acid and the only amino acid that can fit in the internal space of the triple helix. Bonds formed between glycine residues on one chain and X-position residues on an adjacent chain are important for the stability of the helix. After the collagen helix is formed in the cells, it is secreted into the matrix and processed into its mature form. The mature collagen helices then spontaneously assemble into bundles of collagen, called fibrils, in the extracellular matrix of bone and skin.

OI is usually caused by a mutation in one of the two genes, either COL1A1 or COL1A2, that code for the  $\alpha 1$  and  $\alpha 2$  chains of type I collagen, respectively. Patients with Types V and VI OI do not have mutations in type I collagen; the gene(s) involved in these conditions is not known. For mutations in type I collagen, there is a general correlation between the type of collagen mutation and the Sillence types.

Type I OI is usually caused by a quantitative defect of the  $\alpha 1$  chain. One copy of the gene does not produce collagen chains so that patients make only half the proper amount of type I collagen. All of the collagen made by these patients is of normal structure; it is simply the amount that is reduced.

Types II, III, and IV OI are caused by structural mutations in one of the collagen genes. About 85 percent of these mutations are changes that result in the substitution of a larger amino acid for one of the glycine residues that should occur in every third position along the chains. The term used by geneticists for these mutations is "missense mutations." Subsequently, folding of the collagen helix inside the cell is delayed at the substituting amino acid, allowing time for extra sugar groups to be added to the collagen alpha chains. The remaining 15 percent of collagen structural mutations are more unusual rearrangements.

Identifying the OI-causing mutation contributes to genetic counseling for the family and to OI research, but the mutation is not the only factor that determines the severity of OI in an individual patient. In some cases, patients with an identical collagen mutation may have phenotypes (i.e., clinical features) that differ enough that they are classified into different Sillence types. For optimal clinical care, the key is to treat each patient individually rather than by the "label" of his or her Sillence type or specific mutation.

## Diagnosis

The diagnostic process may include:

- physical exam
- medical history, including pregnancy and childbirth information
- family history

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- bone density testing
- x rays
- collagen (protein) testing using a skin biopsy
- molecular testing
- blood and urine tests to rule out conditions other than OI.

### **Physical Exam**

The physical exam includes assessment for abnormalities in:

- skull formation
- fontanel closure
- head circumference
- facial shape
- scleral hue
- dentition
- hearing
- chest shape
- shape of spine, presence/degree of scoliosis, and kyphosis
- shape of long bones
- segment measurements (upper and lower extremities)
- height/length (compared to unaffected children)
- body proportions
- bruising/scarring
- joint mobility
- development (physical and cognitive).

Some characteristics are age-dependent. Hearing loss may not be apparent in infancy or childhood. Bone malformation may not be present in an infant or young child with mild disorder. Pale blue sclerae are normal up to 18 months of age. Intense scleral hue and its presence past 2 years of age can suggest the need for further evaluation for OI. While tinted sclerae are a characteristic of OI, it is important to note that only some patients exhibit blue sclera.

### **Genetic Testing**

As with all genes in the body, DNA is the basis for inheritance. DNA contains sections that are expressed (exons) and sections that are not expressed

(introns). DNA is translated into RNA, which contains only those sections that are expressed. The RNA is then used to make proteins, which are the building blocks for the human body. The protein, RNA, and DNA can all be tested to diagnose OI.

- **Biochemical Testing of Collagen Protein** – Biochemical (protein) testing allows researchers to learn about the structure and amount of the collagen protein that the cells of the body are producing. About 85 percent of people with OI will have a positive collagen protein biochemical test abnormality. Fifteen percent of cases diagnosed clinically and radiographically as OI will have a negative collagen protein biochemical test. This test requires a skin biopsy to obtain living cells that produce collagen. The punch biopsy, about 1/16 inch diameter, is taken from the arm or leg under lidocaine anesthetic. Foreskin taken during circumcision can also be used. The skin cells are grown in a culture medium. The collagen produced by the cells is isolated from both the culture medium in which the cells are growing (secreted collagen) and the inside of the cell (intracellular collagen). Collagen chains are compared by gel electrophoresis to the collagen chains of control subjects without OI. Patients with OI who have structural defects of collagen often show two populations of chains, one with normal gel migration and one with delayed migration. This results in protein gel bands that appear wider in OI than in controls or that may even migrate as two distinctly separate bands on the gel. Diagnosis of the collagen quantitative defect of Type I OI is also apparent on gel electrophoresis.
- **Sequencing RNA or DNA to Identify the Specific Collagen Mutation** – RNA or DNA sequencing allows researchers to determine the specific change in a person's genes for type I collagen that causes OI. Either RNA or DNA testing can locate the mutation that causes OI if it results in changes in the amino acid sequence of the protein. For many mutations, both methods are equally sensitive, but the RNA-based testing will not identify a mutation if the mutation results in an unstable RNA (i.e., premature termination codons). Such mutations are only detectable in the DNA.
- **RNA (cDNA)-based Testing** – This approach uses the RNA (complementary DNA) made by the COL1A1 and COL1A2 genes. RNA-based testing usually requires a skin biopsy, although in some instances enough collagen RNA can be isolated from white cells. The RNA is then copied to make a DNA copy, which is then sequenced in its entirety.
- **DNA-based Testing** – DNA sequencing can be performed using a routine blood sample or skin biopsy. Because the collagen genes are moderately large, laboratories do not sequence entire genes. Instead, the genes are tested exon by exon (i.e., the expressed portions of genes). In this strategy, regions are amplified using the polymerase chain reaction and then either sequenced directly or tested by a technique such as conformation sensitive gel electrophoresis (CSGE) to detect differences

between the two copies of the gene. Only exons that have abnormal CSGE findings are sequenced. This method detects most structural defects of collagen protein (those that cause Types II, III, and IV OI) and also detects chain termination mutations that cause Type I OI. The DNA-based testing is more likely than the RNA-based testing to detect mutations at exon boundaries. However, some mutations are overlooked because not all mutations cause abnormal CSGE. No functional correlation between mutation and biochemistry is determined in this testing.

### Other Diagnostic Tests

- **Radiography** – X rays should be obtained on any child in whom OI is suspected. X rays can usually confirm the diagnosis and may reveal osteoporosis, bowing of long bones, and vertebral compressions, depending on the severity of the condition. In some children and adults with mild forms, these alterations may be difficult to identify. In addition, x rays can help identify subclinical or old healing fractures. Lateral x rays of the skull in infants may show wormian bones, which can be a component of OI. Wormian bones are not seen in all patients with OI and are not unique to OI.
- **Dual Energy X-ray Absorptiometry (DXA)** – DXA (bone density testing) can be a useful adjunct to clinical examination and diagnosis. However, the bone density must be compared to age- and sex-matched peers. Z-scores (*not* the T-scores routinely used for adult patients) are essential for analyzing bone densities in children. Generally, a Z-score of -1 to +1 standard deviation (SD) is considered within normal range, with 0 being the mean for healthy children of the same age and sex.
- **Urine and Blood Tests** – Urine and blood tests, other than DNA analysis, are not conclusive or diagnostic for OI. They may, however, be used to rule out other conditions, such as hypophosphatasia.

### Prenatal Testing

When OI is present in one of the parents or in a previous child in the family, prenatal testing is often requested. Ultrasound of the fetal skeleton or amniocentesis or chorionic villus sampling to obtain cells and DNA are the available tests.

- **Ultrasound** may detect bone malformation as early as 16 to 20 weeks gestation. It is extremely difficult to differentiate between Types II and III OI based on ultrasound.
- **Amniocentesis** is useful for molecular detection of previously known mutations but not for biochemical tests of collagen protein.

- **Chorionic villus sampling (CVS)** can be performed between 10 and 13 weeks of pregnancy. The cultured chorionic villi cells can be used for molecular detection of a previously known mutation or to detect the protein abnormalities previously identified.

### **Educating the Family**

Most parents of a child with osteogenesis imperfecta can report exactly where and when they first learned about their child's diagnosis. The quality and quantity of information and the manner in which it was given profoundly affect the parents, the child, and the whole family for years to come. A sensitive, accurate, and hopeful presentation of information can help foster a sense of partnership between the health care providers and the family and avoid misunderstandings. Ongoing communication with families will be necessary due to the large amount of information. Learning that one's child has a genetic disorder can be shocking and confusing for the parents and extended family.

Parents need to be reassured that nothing they did prior to or during pregnancy caused OI. They need to be reassured that OI does not affect a child's ability to think and learn. With some adaptations in the physical environment, children who have OI go to school, make friends, grow up, and have families and careers of their own.

If the person receiving an initial diagnosis is an older child, teenager, or young adult, counseling about lifestyle changes and precautions for avoiding fractures may be necessary. Teens and adults also will benefit from information about the genetics of OI or a referral to a genetic counselor.

In addition to the need for information, there is an ongoing need for emotional support for patients of every age and their families. Teens and the newly diagnosed young adult may be in particular need of this support.

Coping with and adjusting to having a child diagnosed with OI can be stressful for families. It is normal for families to go through the stages of grief (i.e. denial, anger or resentment, bargaining, depression, and finally acceptance). Families will continue to experience these feelings at different stages during their child's life. Therapists can encourage families to express their thoughts and assure them that their feelings of disappointment, anger, and sadness are normal. It is also important to help families find support for coping with their feelings. A referral to family social services may be helpful.

### **Treatment**

Because there is no cure for OI, its management or treatment currently focuses on minimizing fractures and maximizing mobility and independent function.

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Aggressive rehabilitation is an important part of treatment for most types of OI. Prolonged immobilization can further weaken bones and lead to muscle loss, weakness, and fracture cycles. Many orthopedists prefer to treat fractures with short-term immobilization in lightweight casts, splints, or braces to allow some movement as soon as possible after a fracture.

When managing OI in children and adults, rehabilitative treatment is increasingly emphasized. Goals include improving cardiovascular function; prevention or reduction of problems associated with misalignment of hips, knees, and ankles; and development of optimal peak bone mass. The need for exercise starts in infancy and continues throughout life. Various orthotics to support ankles, knees, and wrists are often necessary. Braces may also be part of a treatment program.

Swimming and water therapy have been shown to be not only enjoyable but of benefit to children and adults with OI.

Orthopedic surgical treatments may include inserting rods in the long bones of the arms and/or legs to control fractures and improve malformations that interfere with function. Spinal surgery is sometimes necessary to correct scoliosis or prevent it from worsening.

Treatment with fluoride, calcitonin, vitamin D, and high doses of calcium has generally been judged as ineffective for osteogenesis imperfecta.

### **Experimental Treatments**

#### **Bisphosphonates**

Experimental drugs for OI, including bisphosphonates, are being investigated at multiple centers in the United States and in other countries, either in clinic or clinical research settings. As significant numbers of patients are currently receiving bisphosphonates, rehabilitation professionals should be aware of the potential benefits and drawbacks.

Bisphosphonates are potent inhibitors of bone resorption (breakdown) and were originally developed as treatment for postmenopausal osteoporosis and hypercalcemia. They are currently approved by the U.S. Food and Drug Administration (FDA) for the prevention or treatment of osteoporosis and the treatment of Paget's disease of bone in women and men. They are frequently prescribed "off label" for adults who have OI with symptoms of osteoporosis. In adults, bisphosphonates can stabilize bone density. And in many cases of bone density loss due to osteoporosis, bisphosphonates can increase bone density. In the early 1990s, some investigators began treating children with OI with intravenous pamidronate, a second-generation bisphosphonate (Glorieux et al, 1998). Reports appeared that pamidronate decreased pain and fracture rate as well as improved mobility and bone density in affected children. Research continues with this drug and the newer IV and oral bisphosphonates. Questions that remain to be answered include long-term side effects as well as impact on

bone quality and bone health, the dosing schedule that maximizes benefits and minimizes detrimental effects, and the time to end or cut back therapy as well as the benefit to patients with milder OI. Potential drawbacks of bisphosphonate administration seem to be emerging from wider studies. Surgeons have reported delayed healing of osteotomies in some cases. There are anecdotal reports of increased bone brittleness and fracture rate after prolonged treatment with bisphosphonates.

### **Bone Marrow Transplant**

A highly experimental and unproven treatment for OI is bone marrow transplantation. Although articles published from this research suggest a positive effect on growth and fracture rate, there is little data to support these claims. At this time, bone marrow transplantation is not a recommended treatment for OI.

### **Gene Therapy**

While gene therapy holds promise in theory, the actual application of such therapies is not imminent.

### **Growth Hormone (rGH) Therapy**

Short stature is a significant feature of osteogenesis imperfecta. Studies of growth hormone administration reported that some children with Type I or Type IV OI responded to the treatment with increased linear growth. A few children with Type IV OI attained normal height curves. Responders to rGH also experienced positive changes in bone histology and small increases in bone density. Combining growth hormone therapy with bisphosphonates is being investigated.

## **Neonatal and Nursery Care**

There are certain medical concerns to consider when caring for an infant with OI. The infant may have an unusually soft skull, startle very easily, and have malformation and fractures in various stages of healing.

### **Handling Suggestions**

- It is important that babies with OI receive affection and are held and touched by parents and other caregivers.
- 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 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 lift the baby, then remove and 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 limbs; do not pull a limb through a sleeve or pants leg. Pulling, twisting, or getting caught in clothing can cause fractures.

### 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.

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.

To avoid fracture, the same care taken for other activities should be taken when holding and positioning the infant. 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 an arm or leg at an abnormal angle.

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. Gently rubbing the baby's back while taking gentle bouncing steps may also be beneficial. When picking up the infant for burping, it is important to provide both front and back support. The caregiver should lay the baby on his or her back and

bend over to pick up the baby. The caregiver's hands should be positioned under the infant, as described under Handling Suggestions, while the caregiver's shoulder should very gently touch the baby. At this point, the baby is supported under the back and positioned on the shoulder as the caregiver moves up and backward.

### **Bedding**

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

### **Positioning**

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. Occasionally a gel pad is necessary to protect the back of the skull. Rolled blankets or sheets or soft foam wedges can support sidelying. Rib fractures, a malformed chest, etc., will preclude placing the baby in the prone position (i.e., on the stomach).

### **Parent Education**

Parent education should include explanation and demonstration of the procedures for holding, lifting, diapering, and general infant care. The return demonstration will ensure that the parents are comfortable with each skill. If possible, these skills also should be demonstrated 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 in conjunction with the orthopedic surgeon on how to recognize a fracture and 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 osteogenesis imperfecta can facilitate obtaining appropriate care if the family arrives at the Emergency Department or needs to seek treatment at a different facility.

Parents will need to acquire a car seat that, in some cases, will also be used as a baby carrier. A contoured foam insert may be needed to provide a good fit.

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 a local OI support group or contact with another family of a child with OI (with their approval).

## Primary Care

### Children and Teens

In addition to care provided by orthopedic surgeons, pulmonologists, and endocrinologists who specialize in pediatric patients, children with OI must receive primary pediatric services to maintain health and prevent illness. The primary care provider is an important resource for healthy living and preventive care as well as for common acute illnesses not directly related to the child's OI, such as otitis media, strep throat, etc.

The primary care staff needs to work closely not only with the child and family but also with the orthopedic surgeon, physical and occupational therapists, and OI clinic or research facility, as appropriate. Multidisciplinary care is paramount for optimal outcomes. The primary care provider may be asked to provide referrals and recommendations to specialists and may also need to make recommendations for the patient's physical limitations in activities at school and in other settings. It should be recognized that parents and older children with OI become knowledgeable about both the disease and the special accommodations or limitations that may be needed.

Patients with OI should be seen by a primary care provider regularly, just like any other patient of the same age. Health care providers caring for infants and children in the primary care setting should be aware that osteogenesis imperfecta is a highly variable disorder, ranging from very mild to very severe. Some children with the mild form of the disorder may have few fractures, whereas those with a more severe form fracture frequently. Some children are not able to stand independently for measurements of stature and may need to be measured lying down. Pressure should not be applied to bowed legs in an attempt to straighten limbs for measurement. The child may lag behind his or her peers in physical development and may not crawl or walk independently without intervention from physical therapy. The patient may instinctively develop alternate methods of locomotion, such as commando crawling or scooting.

Hearing may be impaired. The patient may wear hearing aids and require more structured communication.

Referral to a pediatric dentist is especially important for patients with OI, who may also have manifestations of dentinogenesis imperfecta.

Despite the wide-ranging effects of OI on multiple body systems, parents and children still need information on normal child health and safety issues. Information should be tailored to the patient and family's specific situation. Always consider the patient's age and level of function, not size, when providing information or guidance. Occupational and physical therapists can be quite helpful with providing suggestions for adaptations and accommodations for patients with OI.

## **Transition to Adult Care**

Transition to adult medical care may be difficult for patients who have received frequent, highly specialized care from pediatric specialists and children's hospitals. Families often have developed strong bonds with nurses, assistants, physicians, therapists, and other health care providers. Patients may have concerns about insurability, future care with new health care providers, etc. However, transition to adult care can occur successfully for patients with OI if the process is carefully coordinated and initiated well before the patient outgrows a system or provider. It is helpful to:

- encourage the older child to be his or her own advocate, by speaking to health care providers directly and providing accurate information about his or her own medical history.
- provide the family with a referral list of physicians in the community who are familiar with OI.
- obtain signed "authorization to release information" forms so complete medical records can be sent to new health care providers.
- be aware of insurance requirements and work with the family to facilitate any transition between insurance carriers.

## **Adults**

Ongoing primary care is essential for the adult. Presence of a chronic disorder does not preclude the patient from experiencing unrelated health problems. Patients with OI should receive care that follows the same guidelines for routine assessments as other patients. The health care provider needs to address childbearing and sexuality issues – just as he or she would with any other adult patient – while also considering the diagnosis of OI. Women with OI should receive the full spectrum of well-woman care.

In adults after puberty, the fracture rate usually decreases as a consequence of hormonal and other metabolic changes affecting collagen and bone. Other medical problems related to the basic collagen defect, such as tendon, muscle, and joint problems, may assume more importance. Later in life, fractures may become more frequent, especially in women after menopause. Maintaining bone density is a primary concern for men and women. Recurrent dismissal of symptoms as simply an aspect of OI may lead to diagnostic errors.

All patients should be counseled to maintain a healthy lifestyle, which would include not smoking or quitting if they currently smoke, exercising safely within the spectrum of their abilities, and eating a healthy, well-balanced diet. Swimming and other low-impact recreational activities, performed with

appropriate cautions, are often ideal for the patient with OI. Physical therapy may be ordered to assist in developing a targeted exercise program.

### Medical Procedures

Previous experience can help dictate the best way to perform medical procedures for a patient. Parents, older children and adults with OI, and the primary care nurse are all resources for this information. The diversity of patients with OI makes it difficult to generalize and requires multiple options for procedures.

### Assessing the Patient

In addition to the information required in a medical history, the following information should be considered during medical assessment:

- **Age** – Many individuals with OI are markedly short in stature and can be mistaken for someone who is chronologically and intellectually much younger.
- **Head Size** – Head size tends to be appropriate for age in persons with short stature so that there is the appearance of macrocephaly.
- **Eyes** – Assess for evidence of tinted sclera.
- **Hearing** – Assess for impairment that compromises conversations, safety, etc.
- **Teeth** – Assess for evidence of dentinogenesis imperfecta.
- **Drugs and Supplements** – Inquire about prescription medications as well as any herbs or dietary supplements. Some individuals receive drugs as participants in a research protocol.
- **Rods** – Since rodding surgery is a common treatment for people with OI, it is important to document the presence and location of rods. In some cases, rods may interfere with MRI, DXA, or CT studies or other radiology procedures.
- **Bleeding** – Some individuals with OI have a history of excessive bleeding. This is related to capillary fragility and may manifest itself in frequent nosebleeds, bruising, and greater than normal bleeding during surgery. Drugs such as aspirin may contribute to prolonged bleeding time.
- **Respiratory Function** – Many individuals with the more severe forms of OI have a compromised respiratory system due to scoliosis, chest malformation, or short stature. The compromise can be serious enough to impact activities of daily living and lower resistance to infection. Nurses are advised to be alert to the early stages of respiratory distress or respiratory failure to prevent a crisis from developing.

### **Taking Blood Pressure**

- Guard against inflating the blood pressure cuff too tightly, because this could lead to bruising or fractures in patients with severe OI. An automatic BP cuff with the pressure preset cannot be controlled and should not be used.
- Avoid obtaining a blood pressure in an arm that has repeatedly fractured and/or has a bowing malformation, as the bone may be especially predisposed to fracture. The blood pressure may be obtained in the thigh for those patients with bilateral arm malformations.
- The blood pressure cuff size should be appropriate for the patient. Small-stature adults may need pediatric-sized equipment.
- Young patients may tolerate the procedure best if a parent holds the child on his or her lap. This will also assist in keeping the child still. To reduce the child's anxiety, demonstrate the procedure on a stuffed animal or doll and explain to the child that you are going to give the arm a gentle "hug."

### **CPR**

As a lifesaving measure, CPR can be performed on patients with OI, despite the possibility of higher risk of rib fractures. In one report, manual chest compressions (two-finger method) on an infant with Type II OI with multiple pre- and postnatal fractures did not result in new rib fractures as evidenced by serial chest radiographs (Sewell & Steinberg, 2000). The force used for the compressions can be adjusted to achieve the desired depth in each situation. Children with OI will likely require less force than healthy children to achieve the proper compression depth.

When performing CPR on an adult patient with OI, the patient's chronological age should be used as a reference for the proper ratio of compressions to breaths, keeping in mind the size of the patient. Less force may be required to achieve desired compression depth.

### **Emergency Department Care**

Patients with OI may have substantial experience with fracture management and orthopedic procedures due to ongoing medical and surgical needs. They are often very knowledgeable about their health status and problems associated with OI. Accordingly, the opinions, requests, and instructions of adult patients and parents of children with OI should be respected.

Children and adults with OI may also present to the Emergency Department with problems unrelated to OI that are seen in the general population and need to be evaluated as such.

### **General Guidelines**

A fall, head injury, accident, nontraumatic fracture, or respiratory compromise may bring the OI patient to the Emergency Department. In these situations, the degree of injury may be more severe than typical for the degree of trauma. When informed that a patient has been diagnosed with OI, medical personnel should record this information prominently in the hospital chart.

Patients should be asked about whether they have had rodding surgery. Extra care must be used when handling the patient's limbs or removing clothing from a limb that has been rodded. Some rods interfere with radiology studies, such as MRI, CT, and dual energy x-ray absorptiometry (DXA).

Caution should be taken when transferring a patient. Sudden pulling of limbs, neck, or spine should be avoided. Never twist, bend, apply pressure, or try to straighten a limb, since some limbs cannot be straightened due to an existing malformation of the bone or contractures. Many patients have scoliosis, which affects positioning. Parents, family members, and older patients can provide guidance about appropriate alignment for the patient.

Parents should be allowed to stay with their child at all times in the Emergency Department to comfort their child and help with transferring or positioning the child. This should include accompanying the child into the examining room and Radiology Department.

Side rails of stretchers should be padded to prevent the child or small adult from slipping through an opening and to protect against injury. Always make sure the side rails of the hospital bed are secure. Use caution when tightening straps on a stretcher, applying splints, or using restraints so as to avoid fracture. Blankets and sheets should be kept loose, and care should be taken when removing them to prevent catching the patient's fingers or toes in a fold of the blanket or sheet.

Medical equipment sized by age of the patient may not be appropriate for the patient with OI. Small adult or pediatric-sized equipment may be needed for some adult patients. Equipment for the head or face, such as an oxygen mask, is usually determined by age.

### **Acute Fracture Care**

Bones affected by OI fracture easily, and therefore, the severity of a fracture is not always related to the level of trauma. No external sign of injury may be apparent. Soft tissue injury around a fracture is less likely because the bone usually cracks before the ligaments and tendons tear. The patient may

experience a serious fall or accident with no fractures or may unexpectedly fracture performing normal daily activities.

Patients frequently experience microfractures that are not visible on x ray immediately following the injury. Due to low bone density, possible nondisplaced fracture, decreased soft tissue reaction, and bone malformation in some patients, the fracture may not be discernible. Followup x rays 1 to 2 weeks after the event may reveal the fracture due to the formation of callus. If a fracture is suspected due to pain, swelling, the patient's inability to use the limb (especially in a child), or the patient's or family's insistence of likely fracture, the limb should be treated as if it is fractured until followup x rays are obtained. If the bone is indeed fractured, the patient will be much more comfortable with the limb immobilized. If a fracture is not diagnosed, a few days of immobilization should not cause permanent damage.

If an orthopedist is not immediately available, appropriate splinting techniques should be used to immobilize the affected limb until there is definitive orthopedic treatment. If the patient uses an orthotic device, it can often serve as an effective splint. Bone malformations require added consideration when immobilizing a fracture.

### **Pain**

It is a myth that patients with osteogenesis imperfecta feel less pain than patients without OI. If possible, the staff should minimize handling until after pain relief is administered. Do not hesitate to use splints and wraps to reduce motion of a painful limb and to minimize spasm. Adequate pain relief is paramount prior to any procedure, assessment, or x ray.

### **Other Related Issues**

Staff in the Emergency Department may also see patients with OI for respiratory and cardiac problems. Any adult with OI, but particularly those with Type III OI or those with short stature, may have compromised cardiorespiratory functions due to malformation of the rib cage, scoliosis and kyphosis, rib fractures, pneumonia, etc. Patients may have heart valve problems, including aortic valve insufficiency, aortic aneurysm, mitral valve regurgitation, and mitral valve prolapse. Medical therapy for mitral valve prolapse may include antibiotics to prevent rhythm abnormalities and valve infections.

Patients should be instructed to seek prompt treatment for respiratory infections and difficulty breathing. Due to the decreased chest volume, restrictive pulmonary disorder is commonly seen in severe cases of OI. Pulmonary complications can occur due to rib fractures, muscle weakness of the chest wall, heart valve disorders, chronic bronchitis, or asthma. Respiratory complications including pneumonia present as significant morbidity threats for those with Type II and III OI.

In adults with basilar invagination of the skull base, complications may occur with central (brain stem) respiratory control. Headache upon coughing may be an important clinical symptom.

Medical bracing of the ribs will not improve pulmonary function in adults. However, controlled oxygen therapy, BiPAP, inhaled bronchodilators for asthma, inhaled glucocorticoids, and antibiotic use for chronic bronchitis may be indicated.

Some patients have hyperhydrosis and may need significant fluid replacement. Otherwise, metabolic chemistries should be unremarkable if there is no dehydration.

Medication dosages may need to be adjusted for patients with OI. Medicines should be titrated to body weight, not age, even in the case of the adult patient with OI.

Kidney stones are sometimes associated with OI and can cause hematuria and flank pain. OI is not related to a lack of calcium. High urine calcium levels are observed in some patients.

### **Child Abuse Allegations**

The child with OI may present to the Emergency Department with a fracture that is more severe than the reported trauma. Thorough examination may reveal multiple fractures in various stages of healing. These signs are commonly taken as evidence of non-accidental injury or child abuse. Health care providers unfamiliar with OI may not recognize an undiagnosed case of OI. Medical professionals need to be aware that no particular type of fracture is typical or indicative of OI (Marlowe et al, 2002). Clinical features vary widely. Children with mild OI may have few obvious clinical features.

It has been estimated that seven percent of children who have unexplained fractures have an underlying medical condition (Wardinsky et al, 1995). Other disorders that may include unexplained fractures are Ehlers-Danlos Syndrome, hypophosphatasia, and disorders of vitamin D metabolism. The physician and nurse should consider these uncommon conditions when a patient presents to the Emergency Department with fractures of uncertain cause.

Parents are advised to carry a letter from the child's primary care physician stating the OI diagnosis and provide hospital staff with this information. Such documentation should be prominently displayed on the patient's chart.

## **Surgical Services**

### **Anesthesia**

Some anesthesia risks are associated with patients with OI that distinguish them from the general population. Anesthesia personnel need to be aware of the susceptibility of fracture from movement, impact, or stress. Alternative procedures for intubation, such as fiber optics inserted nasally, may be necessary when treating some patients. Dosage may also be affected by the patient's smaller body size.

Chest and rib malformations and scoliosis may compromise breathing. Other risks include fragile dentition, joint stiffness, or heart valve disease.

The small stature of a person with moderate to severe OI often determines the choice of equipment. The endotracheal tube size used should be determined by the size of the head instead of the size of the body.

Patients with OI can exhibit increased body temperature during anesthesia and after surgery. The reason for this is unclear but may be related to an increased metabolic rate. The increased temperature is most often not associated with malignant hyperthermia. Precautions, such as avoiding the use of warming blankets or heavy drapes, are often effective and sufficient. Occasionally, ice packs or other cooling measures may be needed.

Some anesthetic agents such as atropine should be avoided because they may exacerbate increased body temperature. In general, anesthesia is safe and well tolerated in children and adults with OI.

### **Positioning**

Positioning the patient for surgery requires awareness of specific needs for the patient by the surgical nurse, anesthesiologist, and surgeon. Skeletal malformations necessitate extra planning and attention. All limbs should be supported and well padded. Soft padding may be needed to prop up bowed legs or to support the patient's curved spine. Keep in mind that the patient's skin is thinner and more prone to bruising than that of a patient without OI. Restraints should be applied after the patient is relaxed.

### **Intubation**

Intubation can be challenging due to fragile teeth, scoliosis, neck malformation, and joint laxity. Care should be taken to avoid hyperextending the neck when inserting an endotracheal tube. Alternative procedures for intubation, such as fiber optics inserted nasally, may be necessary when treating some OI patients. The endotracheal tube size used should be determined by the size of the head

instead of the size of the body. Spinal malformation, such as scoliosis or kyphosis, may impair pulmonary function. The occurrence of restrictive lung disease and cardiac problems is increased.

### **Rodding**

Rod insertion is a common surgical procedure performed on patients with OI. Rodding is most often used to treat children with moderate to severe OI. In teens and adults, rodding and other surgery is usually reserved for difficult fractures that are not healing well or for alignment problems. Rodding is performed to correct long bone malformation or decrease fracture recurrence at a particular site. It is performed more commonly in the lower limbs than upper limbs. The style of rod and the purpose of the surgery will determine the specific procedure. Osteotomies may be performed and wedges cut from the bone to correct malformation. An intramedullary rod is then inserted. Reaming of the medullary cavity may be required. The limb is immobilized in a cast until union of the bone is achieved. Depending on the style of rod chosen, the rod may need to be replaced as the child grows.

### **Physical and Occupational Therapy**

Children with all types of OI often have motor delay and can be assisted by developmental assessment through referral to rehabilitation professionals, such as physical therapists, occupational therapists, and physiatrists. The increased risk of gross motor delay – caused by skeletal malformation and decreased strength and endurance – warrants early referral. Children with mild OI may only require occasional services. Children with Types III and IV OI will require more extensive assistance. Every effort must be made to provide the child with a means of independent mobility. Equipment recommendations may consist of bracing, walkers, or crutches for ambulation and ADL adaptive equipment. In the case of non-ambulatory patients, consideration will be made for manual or power wheelchair mobility. As the child enters school, mobility is particularly important because it facilitates age-appropriate social interaction.

The need for strength and endurance training, postfracture rehabilitation, and continued rehabilitative treatment frequently persists later in life. Adults may need to work with a rehabilitation professional to develop an appropriate program that can be performed at home or at the local gym. Maintaining bone density, developing strength, expanding cardiorespiratory function, and remaining mobile are important goals for the therapy program for adults as well as children with OI.

The therapist must partner with parents and patients to set feasible and appropriate goals for formal physical / occupational therapy and/or for exercise and recreational activities at home. Programs based on the individual's interests

– which include activities that are enjoyable as well as beneficial – are more likely to be successful.

### **Role of Therapy**

The long-term goal for people with OI is independence in all life functions (e.g., self-care, locomotion, recreation, social interaction, education, and work), with adaptive devices as needed or, in the case of very severely affected people, the ability to direct their own care.

For many years, the parents of children with OI, particularly in severe cases, were told to carry their children around on a pillow and not to expect them to achieve independent function. This led to their being treated as infants even as they grew into school age and beyond. It is now clear, however, that most people with OI can achieve some level of independent mobility and function with the help of physical and occupational therapy, appropriate exercise, surgical intervention, medications, adaptive equipment, and environmental adaptations at home, school, or the workplace.

Maximizing a person's strength and function not only improves his or her overall health and well-being. It also improves bone health, because mechanical stresses and muscle tension on bone help to increase bone density. Encouraging people with OI to adopt different body positions during the day will help strengthen different muscle groups and possibly prevent or minimize malformations, such as a flattened skull, a highly scooped lower back, or tight hip flexor muscles.

Ongoing or intermittent physical and occupational therapy is appropriate in a number of circumstances:

- **Delays or weakness in motor skills.** Because of fractures, muscle weakness, and joint laxity, many children with OI (even those who are mildly affected) experience delays in motor skill development, which then interfere with function. As soon as it is evident that an infant has muscle weakness or motor skill delays compared with unaffected same-age peers, therapy should begin. It should continue until a child reaches appropriate therapy goals. In some cases, an infant or young child may have delays. But after gaining sufficient strength, he or she may become able to sit, stand, and walk. After fractures or surgeries, these children may require additional intensive rehabilitation or physical therapy until they are able to regain the previous level of motor function. In other cases, certain motor skills may be unattainable due to weakness or skeletal malformations. For example, walking is not possible for some people with OI. However, the therapist can help the person maximize function by developing other skills and using adaptive equipment, energy conservation, and joint protection concepts.

- **Recovery from a fracture or surgery.** Because fractures and surgery are frequent for many people with OI, it is particularly important for them to regain as much function as possible during recovery, both to maximize independence and to maintain bone and muscle strength. After recovery, a person may need to relearn skills that he or she had previously mastered as well as regain strength in the affected limb(s). The therapist can help a person with OI develop alternate strategies and use appropriate equipment to make self-care skills easier. Therapists will need to teach the use of gravity-eliminated activity (e.g., aquatics) postoperatively and the principles of leverage (e.g., supporting the whole limb when giving resistance) that are safer for the individual with OI.
- **Fear of movement and trying new skills.** In some cases, the biggest obstacle to independent function is fear. A child who has had fractures may become fearful of moving or trying new things. Parents may respond fearfully to a child's attempts at independent movement, finding it difficult to allow the child to test his or her physical limits because of the pain and difficulty that a fracture will bring to the whole family. In some families, these fears can lead to the child's complete dependence on a parent for all aspects of daily function and self-care. This becomes increasingly problematic as the child enters school – and later, the adult world – unable to do most things for him- or herself and unable to rely on anyone other than the parent for help. While therapists must acknowledge these fears as understandable, they can also suggest ways that the child can practice new skills in a safe environment. This is usually successful if skills are broken down into small steps, allowing the child to succeed at something relatively easy, moving step by step through more difficult skills rather than failing repeatedly when expectations are too high. Protective equipment (e.g., clamshell splints in the pool, hinged circumferential forearm splints when doing some weightbearing), positioning, protected movement, and water therapy can also be helpful. Encouraging the child to direct some of his or her own care, transfers, and handling will also build the child's confidence.
- **Learning a new skill, or a new way of performing a known skill.** Many key self-care skills, such as toileting, dressing, bathing, grooming, and preparing food, pose challenges to people with OI. Some may lack the strength to perform certain tasks or have trouble using standard household equipment because they are short-statured or use a wheelchair. Due to injury, aging, or progressive malformation, children and adults with OI often have to relearn how to do a task in an entirely different manner. Through a combination of strengthening activities, use of adaptive equipment, and creative problem-solving, many obstacles to independent self-care can be overcome. It is essential for therapists to listen to individuals with OI and their families. By respecting their problem-solving approaches, therapists may learn new ways to approach problems they may not have encountered before.

In working with individuals and families living with OI, therapists should keep two principles in mind:

- With the proper environment and equipment, most people with OI can function well in many or most areas of daily life.
- Individuals and families living with OI are truly the experts in how the disorder affects them. Listening to their concerns and ideas, building on their strengths and interests, and working with them as a team will help ensure success.

### **Safe Handling**

There are some basic principles of safe handling that are important to follow anytime a therapist is working with someone who has OI. Fractures can occur simply because a part of the body was slightly twisted, pushed, pulled, or compressed. People with OI and their caregivers have extensive knowledge of what handling practices are safe for their individual cases. They should be encouraged to tell others in new situations that they are experienced in safe handling to prevent injury.

- Ask the parents or the individual with OI to show you the safe handling techniques they have developed. Before handling the person or moving a limb, state what you are going to do and how you are going to do it. If they ask you to stop, stop! For young children, using a floppy doll to demonstrate a motion and to problem-solve with parents will make new transitions easier. When the child hears, "It's your turn," and the parent helps the child with OI mirror the doll's movement, even a child as young as 10 months of age can understand and start the activity with less anxiety.
- It is often preferable to have the parent or caregiver do all handling of a young infant or child at the beginning of a therapy relationship, then gradually involve the therapist. As the child becomes secure and comfortable in the environment, he or she will gradually accept hands-on help from other individuals. The ideal is to structure the environment so it is easy for the child to perform the task without an adult's hands-on help. This approach is safe, puts the child at minimal risk, and reinforces the child's awareness of what he or she can do independently.
- It is critical to remember that people with OI do not have impaired coordination or sensation and do not require the complex, neurologically based interventions used for children who do. The main constraint to movement in OI is weakness. Once a child gets stronger, he or she typically figures out how to accomplish a task and often does not need the guidance of hands-on therapy to do it. This is especially true if the therapists and parents focus on constructing the environment to help the

child achieve the task, rather than using external caregiver hands-on assistance.

- Never pull, push, or twist a limb. Avoid passive rotation of the arms, legs, head, or trunk.
- Lift an infant with OI with the widest base possible. Lift by placing one open hand under the buttocks and legs and the other under the shoulders, neck, and head. Do not lift the child from under the armpits, which puts pressure on fragile ribs and loose shoulders, and do not lift the buttocks by pulling on the ankles, especially during diapering.
- Be aware of where the person's arms and legs are at all times, to avoid awkward positions or getting a hand or foot caught in clothing or equipment.
- Provide adequate support when the child or adult is in a standing position so that the legs don't "crumple" under him or her. Examples of support are straddle-riding toys, gait trainers with sling seats, and clamshell braces or splints on the legs.
- Avoid positions and motions of great leverage that stress bones, such as hip flexion ("jackknife" position with the person leaning far forward while sitting) that stresses the femur and diagonal trunk rotation that stresses the vertebrae. The "bridging" exercise (lifting the buttocks with knees flexed while lying on the back) should also be avoided because it stresses tibial bones in the same plane in which they tend to bow.

### **Therapeutic Strategies**

- Before learning personal care skills, a child must first develop gross motor skills such as reaching and sitting, which may be delayed or difficult for those with moderate to severe OI.
- Protective handling, protective positioning, and protected movement contribute to safe development of motor skills.
- Equipment, ranging from simple pillows to specialized wheelchairs, can help children and adults achieve motor and personal care goals even if they have weakness or are recovering from a fracture.
- Water therapy provides the opportunity for children with OI to develop skills in a gravity-free environment before trying them on land and for adults with OI to relearn or maintain motor skills.
- Patience and task analysis are both necessary to develop a successful therapy program. Therapy may progress more slowly for individuals with OI than for other therapy patients. Developmental concepts and specific skills need to be analyzed closely, so that many small improvements can lead to achieving a particular therapy goal.

## **Protective Positioning**

A key method for helping a person with OI maximize strength and function is to encourage him or her to adopt various positions throughout the day or, in the case of an infant or young child, to encourage parents and other caregivers to place the child in different positions. Position changes not only strengthen different muscle groups but also help prevent contractures and malformations that can limit mobility and increase pain. It is important to keep the hips and spine as straight as possible, prevent flattening of the back of the head from lying supine, and promote head-turning in both directions.

In many cases, everyday objects can be used to make different positions easier and safer. For example, towel rolls and padding can be used to encourage upright posture and avoid "frog-leg" positioning in a wheelchair, car seat, or stroller. An infant or child can be encouraged to try prone positioning by lying on the parent's chest, a partially inflated beach ball, or a foam wedge.

## **Protected Movement**

New positions and skills should be introduced gradually to allow the person to feel safe and promote gradual strengthening of muscles and bones. Providing adequate support is important to overcome weakness and prevent injury. According to Marnie King, an occupational therapist with extensive experience with children who have OI,

"Protected movement...is a hard concept because caregivers tend to protect the child from fractures by limiting potentially harmful movement experiences. But not moving creates the problems of decreased strength, bowing from constant sitting, fear of movement and being moved, and dependence in all activity... [T]he child with OI will require slow, graded introduction to being moved in progressively less supported positions."

Therapists can begin by assessing the person's current functional abilities. Is he or she stuck at a particular level of progression? The goal in therapy will be to gain the next level or improve within the lying, sitting, and walking levels. Thus, the goal for a very severely affected person might be to lie in a supported, inclined position. For a severely affected person, learning to sit-scoot might enhance his or her self-care skills. More mildly affected people may gain walking skills with or without braces or other aids. Very mildly affected people may function at the same level as their unaffected peers with occasional modifications or limitations, such as no high-impact activities. Activity analysis will need to be done in very small increments to assess progress. Parents of children with OI need to know that many developmental milestones might not be met but will be compensated for by building skills around them. For example, a particular child might not be able to crawl or independently get into a sitting position, but he or she can improve sitting skills and may use a power reclining wheelchair later to compensate.

King suggests a specific developmental progression for children with OI, as follows:

### **Progressive Modifications**

#### **Supine positioning**

Use a curved concave skull pad or gel pad, or provide sufficient position changes to prevent flat skull. If child cannot get hands to midline, use trough shaped foam bed pad to guide shoulders forward. Provide support or splint forearms to prevent humerus and forearm from bowing toward body.

#### **Prone positioning**

Use a chest wedge, practice on steep incline first (e.g., parent's chest). However, a chest wedge may not work for some children; they become locked in position and can't move.

#### **Inclined sitting**

Use a concave skull pad or gel pad. Blanket rolls along torso for support can also serve as arm rests. Position to decrease wide hip abduction. Provide place for feet to rest flat. Use very wide straps or vest for trunk support.

#### **Sidelying**

Support under head and below axilla. May need support under upper arm and leg if position used for sleeping.

#### **Rolling**

May not be comfortable for child. Start using blanket like a hammock and slowly tilt child, or position child to reach for a ball or object. Then partial rolling in a blanket on a firmer surface. Once child is able to tolerate side motion, use slight wedge to roll downhill.

#### **Supported sitting**

May be done in positioning chair as with inclined sitting (above). Provide head and neck support at first. Slowly decrease support as head turning gets better.

#### **Unsupported sitting**

Avoid ring sit. Work toward positioning on chair or bench (with close spotting).

#### **Getting to a sit**

Start in pool, child sitting beside parent, side-leaning on parent's thigh and trying to get to a sit. On land, side-lie on a wedge or parent/therapist thigh and forearm (not extended wrist). For adults, work on abdomen strength to use a sit-up method.

#### **Sit-pivot, sit-scoot**

Start sitting on bench in pool/tub with water to chest height and shift side-to-side to get floating toy. Lower height of water until water is child's hip height. Then try

on land on slippery bench (spot child!). This position will be a transfer method from chair to bed to toilet during fractures and if legs are not strong enough to stand.

### **Crawling**

Start in kneeling position with chest supported by partially inflated beach ball and aim up hill on wedge. Start static reaching for toy above child. Progress to less chest/abdomen support. If legs abduct, use "mermaid suit" of stretchy 6- to 8-inch wide tubigrip, old panty hose top, or wide stockinette from child's waist to ankles.

### **Kneeling/pulling to a stand**

"Mermaid suit" (see above) and high kneel for trunk development. In the pool, progress from high kneel to half kneel. Also in pool (water depth to waist when standing), lower to sitting and stand up again. Then try crawling in water the height of child's knees.

Some people with OI will achieve all of these skills, although interruptions in progress and reverting to previous skills are common because of fractures and surgery. Others will achieve only some of these skills. However, supplemented by equipment and environmental adaptations, any level of proficiency with these skills will increase potential for independent function and self-care.

### **Water Therapy**

Water provides an ideal environment for people with OI to practice protected movement and learn new skills. Water not only cushions bones and joints and protects the person from falls, but it also provides gentle resistance along the entire length of bones. This resistance helps to strengthen bones and muscles, and prevent fractures that are caused when too much pressure is applied to an isolated area. Swimming and other water exercise often become favorite fitness activities for older children and adults with OI.

Practicing the above-listed developmental progressions in the water can reduce fears and help make transitions from one position to another easier. Once the skills are mastered in the water, they can be tried on land. (King 2001, p. 109 for a diagram of how progressions can be practiced in the water.)

Examples of water therapy that can promote new skills are:

- "Shimmy-sitting" and scooting by sitting on steps in the water and scooting from side to side or up and down the steps.
- Standing and walking, starting with water up to the chest. Provide support, such as lightweight splints on the legs, a foam "noodle" or kickboard for the person to hold onto, and/or a flotation vest to promote upright posture. Move into more shallow water (less buoyancy) as confidence and strength

increase. Use a shoe lift during this activity if the person has a leg length discrepancy.

## **Adaptive Equipment and Aids to Independence**

The equipment available to help a person with OI function independently is practically unlimited if one considers both traditional adaptive equipment as well as "homemade" solutions to everyday challenges. Important concepts to consider when choosing equipment are energy conservation, joint protection, mobility, and accessibility. In considering these concepts, the goal is for the person with OI to be as independent as possible in his or her daily life.

### **Energy Conservation**

To help a person function most efficiently, evaluate what tools and environmental adaptations might be needed so that he or she can accomplish common tasks without excessive strain or fatigue. Establishing workstations, such as a homework station, toothbrush station, or hair-drying station, with all needed materials in one place and within reach, will prevent unnecessary reaching or traveling around the room searching for things. Baskets or bags attached to a wheelchair, walker, or crutches allow the person to carry things from room to room. Clothing that is easy to put on, such as pullovers and pants with elastic waists, will minimize the effort needed to dress and undress.

### **Joint Protection**

To help avoid overstretching and injury, teach people with OI to use their strongest muscle groups (usually the largest) to accomplish tasks. Suggest tools that will minimize strain during day-to-day tasks, such as jar openers and electric can openers, and low shallow shelves to help access items. Reachers must be used carefully because the added length increases the weight of the object.

### **Mobility**

Many people with OI use a mobility aid at some point in their lives. Some may only need assistance when they are learning a new skill or recovering from a fracture or surgery, while others will use a walker, crutches, wheelchair, or other aid most of the time.

### **Accessibility**

Physical environments at home, school, or work can be modified to allow maximum independence. While extensive structural changes – such as ramps or lowered kitchen and bathroom surfaces – are sometimes called for, some accessibility problems can be addressed with creative use of assistive devices, rearrangement of furniture and other equipment, and thoughtful consideration of how the person with OI can best use his or her home, classroom, or office. Because the world is not modified for short-statured people, using a wheelchair with a seat elevator should be considered when needed.

An important byproduct of making homes as accessible as possible for children with OI is that the children can better participate not only in their own care but also in household responsibilities. It is vital, both for the child's well-being and the family's healthy functioning, that children with OI take responsibility for appropriate household tasks. To help children do their jobs safely, families may need to modify room arrangements and storage of household items.

The following lists many commonly used types of equipment that help to maximize function for people with OI and some factors to be considered when choosing appropriate equipment. It is vital to choose equipment that matches a particular individual's strengths, weaknesses, and interests as well as those of his or her family.

### **Types of Equipment and Considerations**

#### **Walkers**

Supported walking allows weight bearing in legs, which increases strength and bone density. Need sufficient upper-body strength to grasp/move walker. Posterior walkers may be useful for encouraging upright posture. Some people with OI report feeling more secure/steady with an anterior walker. Baskets attached to walker can help with independence.

#### **Crutches and canes**

Supported walking allows weight bearing in legs, which increases strength and bone density. Need sufficient upper-body strength to grasp/move crutches. Crutch/cane tips may need to be specially ordered if the standard tips are not sufficiently slip-resistant. They should be replaced often to maintain maximum slip-resistance.

#### **Wheelchairs**

Manual chairs can enhance upper-body strength in people whose arms are long and strong enough to push the wheels without pain or risk of fracture. Armrests should support the whole arm and flip up for pushing to avoid forearm bowing. Manual chairs are lighter and easier to transport than power chairs. Power chairs allow people with arm fractures, short arms, and/or arm deformities to move independently. Features such as a power reclining back and a power seat elevator are helpful for some people with OI.

#### **Other Mobility Aids**

Scooter boards, riding toys, tricycles, etc. Four wheels are best to prevent tipping/falls. Seat belts/safety harnesses are necessary. Seats with back rests provide more support and promote good posture. These aids are particularly helpful for young children who are not candidates for walkers or wheelchairs, but who will benefit from independent mobility.

### **Braces/splints**

Should be circumferential, perforated (because of excessive sweating), and lightweight. Leg braces can help with alignment and promote standing and walking. Their role in preventing fractures is limited. Lightweight forearm splints (bivalve or hinged for support to the entire surface—1/16" perforated Aquaplast® is perfect for children) can provide stability to people with forearm bowing, weakness, or pain, and help them with ADLs, weight bearing, reaching, lifting. After a short period of casting, fractured limbs are often immobilized in a lightweight splint or brace that can be removed for bathing and other activity. They may permit greater activity when worn during water therapy while a fracture is healing.

### **Positioning aids**

Pillows, bolsters, towel rolls, gel pads, etc. Promote 90/90/90 position in car seat, wheelchair, stroller, etc., rather than "frog leg" position. Encourage sidelying and prone positions for infants and young children with OI, who often spend a lot of time on their back.

### **Standers**

Promote vertical weight bearing posture, which benefits bone growth and density. Supine standers are preferred to prone standers because standing can be introduced and increased gradually. Tray attachments can allow a child to use stander while coloring, doing homework, games, working on computer, etc.

### **Infant/child car seats**

Use towel rolls, stuffed animals, or bolsters to promote good sitting posture and keep head in midline. Extra wide padding on straps can help prevent injury in a sudden stop or traffic accident. Car-bed style safety seats may be useful for very small infants and infants/children who are unable to sit up. Look for breathable fabrics for padding and seat covers, as children with OI tend to overheat easily. Many families affix a noticeable tag or sticker to the seat indicating the OI diagnosis, in case of a traffic accident.

### **Self-care aids**

Transfer benches, bath chairs, grab bars, reachers, etc. The goal for all people with OI is to be as independent and as safe as possible in self-care tasks. Self-care aids can help them overcome limitations due to weakness, short stature, use of a wheelchair, etc.

### **Self-Care Tasks**

Toileting, bathing/grooming, dressing, and food preparation are four key self-care tasks. These tasks are often challenging for people with OI, particularly if they are short-statured, use a wheelchair, and/or are recovering from a fracture or surgery. Helping people with OI become independent or semi-independent in these key tasks will do a great deal for their overall well-being and sense of self-

esteem. Therapy goals should be to assist the individual to the next higher level of independence for the greatest freedom. Levels of independence are:

1. **Independent** – able to do all aspects with no modifications.
2. **Independent with modifications** – e.g., in a one-level, wheelchair-accessible home and workplace.
3. **Independent with intermittent help** – requires assistance shopping, carrying in/out, and cleaning but can do personal care, toileting, light cooking without help.
4. **Independent with daily help** – requires assistance, as above, and also for bathing and meal setup.
5. **Dependent in most activities** – needs help for toileting, dressing, is able to do light tabletop activity, is safe alone for two to three hours.
6. **Dependent in all activities.**

### **Toileting**

Toilet teaching a toddler often involves a "one step forward, two steps back" pattern. With children who have OI, that pattern may be even more pronounced, as a child who is learning to use the toilet may go back to diapers when recovering from a fracture or surgery. But while toilet teaching may take longer than average in some cases, people with OI can achieve independent or semi-independent toileting with the help of modified or specialized equipment.

- If a standard potty chair or toilet poses problems for a child with OI (e.g., not enough sitting support or too high off the ground), a specialized potty chair can be made from a child-sized resin chair. Cut the legs so the chair sits low to the ground, cut a hole in the seat, place a bucket underneath, and add a safety harness and cushions as needed.
- Some toilet manufacturers offer models that are lower to the ground than average. Families may wish to consider installing one of these models for a short-statured person.
- A toilet-paper reacher can extend the reach of the hand if a person's arms are particularly short. Reachers can be purchased. They also can be made using plastic-covered solid core copper wire twisted around a broom handle and finished with electrician's tape or liquid plastic so that the reacher is washable.
- A sliding transfer bench and/or grab bars, located near the toilet, will allow for easier transferring from wheelchair to toilet. The "shimmy-sit" transfer will be used.

### **Bathing/Grooming**

Bath time often gives a severely affected infant some of his or her first experiences of independent movement, and does so with less chance of fracture.

Placing a folded towel, gel pad, or foam pad on the bottom of the tub provides a comfortable, slip-resistant surface for the infant to be bathed. Older children and adults may benefit from such adaptations as a sliding transfer bench, a shower seat, grab bars, and a hand-held shower head. Long-handled scrub brushes or sponges, washcloths, and "soap on a rope" can be hung on easy-to-reach hooks in the shower or tub enclosure.

For grooming tasks, such as brushing teeth or styling hair, it is helpful to use the "work station" concept discussed previously. For example, a hair dryer can be plugged in and mounted to the wall next to a low mirror, with combs, brushes, and styling aids in a drawer nearby.

### **Dressing**

Infants with OI should be dressed in clothing that minimizes stretching, pushing, and pulling of limbs, such as t-shirts that snap open up the front and play suits with snaps along both legs and the torso. For older children and adults with OI, simple, easy-on clothes, such as pants with elastic waists, are useful. They may find it easiest to dress while sitting on a bed or bench. Clothes often need to be modified for people who are short-statured or are wearing a cast. For example, a seam can be cut and hook-and-loop material sewn onto the seam so it can be opened and closed around a cast. Dressing tools, such as reachers and sock donners, may be useful for some people. Closet rods can be lowered by using ropes or chains to hang a broom handle horizontally from the existing closet rod. Other clothing should be stored on shelves or in drawers that the person can easily reach.

### **Food Preparation**

From a child who wants to fix an after-school snack, to an adult living in his or her own home, people with OI benefit from having an accessible kitchen where they can prepare food. While a custom-designed kitchen with lowered counter tops and appliances, long-handled faucets, and adjustable shelving is ideal, any kitchen can be made more accessible to a person with OI. Commonly used items should be stored in low drawers or cabinets near where they will be used (e.g., cups stored near the refrigerator, pots stored near the stove, etc.). Lazy Susan turntables and pull-out shelving make items easier to reach. A loop of rope or fabric can be attached to the refrigerator door to allow a person to hook the loop onto his or her wheelchair and pull the door open. Beverage containers can be stored on the lowest door shelves for easy access, and the contents of large beverage containers can be divided into smaller containers so they are not so heavy. A miniature "ramp" can be placed in front of the microwave, so the person can slide a plate or bowl out of the microwave onto the counter without having to lift a hot, heavy item. "Heat and eat" convenience foods can be useful for people with limited mobility and strength, as they can be prepared in the microwave in lightweight plastic containers.

## Transportation

Parents will often ask about the best way to transport their child in different situations. Equipment for infants and children should meet local requirements for safety, provide head support, be easy to use, and be appropriate for the child's size and weight. Commercially available equipment may need to be modified to meet the needs of a smaller than average infant or young child. Because of the child's slow growth, the patient will most likely need to remain in a car seat or booster seat much longer than his or her friends. Therefore, it is important to be sensitive to the child's feelings.

Some adults with OI also require special accommodations for travel. The use of booster seats, harness style seat belts, and professionally installed tie-downs for wheelchairs may be necessary.

### Car Seats

Car seats are geared to the child's weight and ability to sit up. 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. If a softer surface is required, it is better to use a folded, thin baby blanket. Removable cotton slipcovers are suggested because they are cooler, and children with OI often 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 instructions in the event of a car accident.

- **Infant Car Seats** are designed for children who weigh less than 20 pounds. Features to look for include a well-padded harness and a head hugger support pillow. This commercially available U-shaped pillow is used to position the baby's head at midline. Small rolls of towels, baby blankets or other padding can be added to hold the child's hips in line. Some infant car seats can be fitted with a contoured foam insert.
- **Full Size Car Seats** are designed for children who weigh more than 20 pounds. Additional features to those already listed include low sides, a 5-point harness, and no flip-down bar. Such seats are more easily adapted to children in different types of casts. Examples of infant seats and full-size car seats suitable for children with OI include the Snug Seat ([www.snugseat.com](http://www.snugseat.com)) and the Britax Roundabout ([www.britax.com](http://www.britax.com)).
- **Booster Seat** laws vary from State to State. Some States have adopted laws requiring booster seats for children age 8 to 12 who are shorter than 4 feet 9 inches or weigh less than 60 pounds. Some States are also advising adults of short stature to consider using a booster seat in private vehicles. Booster seats offer significant protection to the passenger, and

their use is recommended. There are two types of booster seats: high backs and backless. Both can be used in any vehicle equipped with lap-shoulder belts if the passenger weighs more than 40 pounds. If a backless seat places the head above the top of the car seat, a high-back booster is recommended to safely support the head.

### **Transporting a Child in a Cast**

Parents should discuss different cast options with their physician prior to surgery to allow adequate preparation to bring the child home from the hospital. Any time a child is in a spica cast or long leg cast, especially after a rodding surgery, special car seat arrangements may be needed. A standard car seat may be used if it can be set in a reclining position that still allows for easy entry and exit. If that is not possible, several manufacturers make child-size car seats that are especially adapted to accommodate spica casts or braces. In many communities, the Easter Seals organization or children's hospital has programs to rent or loan these seats to families. The seats can also be purchased or rented from a local durable medical equipment company, but they may need to be ordered in advance. A prescription from the physician is usually required for a spica car seat. Some insurance companies provide coverage of at least part of the purchase or rental cost. Older children who are too large for a car seat may require the use of a vest-type restraint that allows them to safely recline on the back seat. The E-Z-On Vest Company (800-323-6598 or [www.ezonpro.com](http://www.ezonpro.com)) sells commercially available vests that meet this need.

Parents should become familiar with the mechanics of the car seat before discharge day. A child in a cast is awkward and heavy to handle. By using good body mechanics, the parents will avoid injury to themselves or additional injury to their child.

Pain management options should be discussed with the physician during the discharge planning period. Administering a dose of pain medication prior to the trip home may allow maximal comfort for the child while riding in the car. Additional support and comfort can be added to the seat by using soft padding around the child.

### **Other Equipment**

A stroller is an important piece of equipment for any child. For the child with OI, the following features should be considered when purchasing a stroller: light weight; sturdy; easy to fold; adjustable seat positions allowing reclining, semireclining, and upright sitting; support for the spine and head; and easy entry and exit (i.e., no leg holes or fixed bar across the front). It can be very helpful if the stroller is wide enough to accommodate casts. Strollers that have a good suspension and swivel wheels provide a smoother ride than other models.

## **Travel by Airplane**

If it is necessary to travel by air to receive medical treatment in another city, families and adult patients should be informed about the various Mercy Medical Airlift programs that function in each state to provide air transportation. If the patient/family is using a commercial airline, they should inform the airline prior to the departure date about any special needs and become knowledgeable about rules related to traveling with oxygen, wheelchairs, or walkers.

## **Public Transportation**

Children and adults with OI who use public transportation, such as city buses, school buses, and taxi cabs, should be made aware of some safety concerns. The patient/parent should look for appropriate use of wheelchair tie-downs, avoid riding in the back of a bus, choose to ride in the back seat of a taxi or other passenger vehicle, and wear seat belts. Harness-style seat belts are available for installation in some school buses. The child with OI should be seated in a middle seat of the school bus to reduce "bouncing" when the bus travels on rough surfaces. These concerns should be discussed with the health care provider. Some communities require referral from the physician to gain access to paratransit programs.

## **Air Bags**

Air bags in automobiles may present a risk of injury to drivers and passengers who have OI. If the driver must sit closer than 10 inches from the steering wheel, an on-off switch can be installed that allows the air bag to be manually disabled. Any decision to disconnect an air bag needs to be discussed by the person with OI and his or her physician. It is advisable to disconnect the air bag only when the physician feels that injury from the air bag is a greater risk than injury from hitting the steering wheel, dashboard, or windshield in the event of an accident. Injury from hitting these areas can occur even with the use of seat belts.

## **Social Services**

The services that social workers provide can be very valuable for patients with OI and their families. There are many issues involved in caring for a person with OI, such as education, funding for specialized equipment and medical services, and coping with and adjusting to this life-long disorder. Knowledge of the resources available to deal with these issues will greatly benefit families and individuals coping with OI.

Social services can assist with identifying the following resources:

Innovative Educational Services

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## Osteogenesis Imperfecta

- Hospital and community-funded programs for counseling, support, and information
- Early intervention programs for infants and toddlers
- Public education services for eligible preschool children
- The Federal law, *Individuals with Disabilities Education Act* (IDEA)
- The National Dissemination Center for Children with Disabilities (NICHCY), at 1-800-695-0285, and each State's office of special education, which is within its department of education
- Each State's Children with Special Health Care Needs Program. This program provides health and support services to children with special needs (from birth to age 21) and their families. Eligibility and services vary from State to State. The *Directory of State Title V CSHCN Programs: Eligibility Criteria and Scope of Services* can be accessed at <http://cshcnleaders.ichp.edu/TitleVDirectory/directory.htm>.
- Each State's departments of social services and public health (which may administer some of the programs)
- Osteogenesis Imperfecta Foundation
- Easter Seals
- Family Voices
- Each State's vocational rehabilitation program
- Mental health services
- Independent living centers
- Nutrition counseling
- Medicare, Medicaid, and Social Security information
- Mercy Medical Airlift program

### **Nutrition**

Children and adults with OI should be advised to eat a balanced diet, which includes a variety of vitamins and minerals and is low in fat and added sugar. Excessive weight gain should be avoided to reduce stress on compromised bones, lungs, and heart. Extra calcium does not reverse the collagen defect that causes OI and could lead to an increased risk of kidney stones. However, adequate calcium and vitamin D are necessary to optimize bone mass and prevent bone loss. Patients taking bone-altering medications may need supplemental calcium if dietary intake is insufficient, but a calcium supplement should be used only under the advice of a clinician. Because RDA guidelines were developed for people of average height and weight, the required amounts of nutrients may vary for people with OI.

Innovative Educational Services

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Constipation is a problem for some patients. Eating a high-fiber diet, drinking plenty of water and other fluids (particularly for those patients prone to excessive perspiration), and being physically active may help reduce constipation. Medications may also be ordered to alleviate the symptom.

Some infants may show slow weight gain. Some toddlers and children with OI who are short in stature may eat very little at any one time, which could be mistaken for failure to thrive.

### **Adult Health Issues**

While most adults with OI experience a decrease in the rate of broken (fractured) bones after puberty, other medical problems, some of which are related to the basic genetic defect that causes OI, may require more attention.

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

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

### **Bone Density and Osteoporosis**

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

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

An initial DXA test may be obtained at any age and then yearly thereafter. If possible, DXA scans should be done using the same machine each year to avoid variations in test results caused by different equipment. Bone density can decline as a direct result of OI, from immobilization associated with casts or limited weight-bearing activity, and from age-related changes in bone and the endocrine (hormone) system. Treatments can include calcium and vitamin D supplements (if the diet is inadequate), drug therapies including oral or intravenous bisphosphonates, diet, and exercise. Smoking, overuse of alcohol, and certain

medications, including cortisone-like steroids, can also negatively impact bone health.

### **Musculoskeletal and Joint Problems**

Adults often report pain in their lower back and hips. This can be the result of compression fractures of the spine, scoliosis, or joint deterioration. Other problems can include fractures that have failed to heal (nonunion fractures) and low muscle strength. Knee pain and ankle instability also are frequent complaints in people with OI. Exercise, orthotic devices, or braces to improve hip, knee, and ankle alignment and back or joint surgery may provide relief. Many individuals have joint laxity or excessive joint flexibility. This is particularly a problem for the knees and ankle joints, which are subject to pressure over the years. Also, leg lengths may differ due to a history of previous fractures. Rolling in of the ankle joint is another common problem. Heel lifts and firm ankle supports are important in limiting wear and tear and improving gait. Orthotic devices may help provide stability for lax joints of the knees, feet, and ankles. Joint replacement surgery may be a treatment option for some, but not all, adults with OI who have joint problems.

### **Chronic Pain**

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

### **Pulmonary Function**

Breathing problems are the main concern of many OI adults, particularly those with Type III and Type IV OI and those with significant scoliosis. Decreased chest volume, chronic bronchitis, and asthma can lead to restrictive pulmonary disorder.

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

Exercise to promote deep breathing, regular testing of pulmonary function, and use of supplemental oxygen can help manage pulmonary function. BiPAP

positive pressure breathing may help with sleep apnea or related pulmonary insufficiency.

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

### **Cardiac Function**

A small number of OI adults seem to have heart valve problems. The most common is called mitral valve prolapse. Dilation of the aorta also may occur, but is not common. High blood pressure is as common among adults with OI as in the rest of the population. High cholesterol and related lipid disorders that may occur in families can contribute to heart problems as well. Medical management of these disorders includes appropriate diet, drug therapies, and regular monitoring by your primary care doctor. Drugs, such as statins, can be helpful along with diet in controlling lipid problems. Coronary artery surgery has been successfully performed on people with OI, although precautions are necessary because of tissue fragility.

### **Hearing**

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

### **Vision**

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

### **Gastric**

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

## **Kidney Stones**

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

## **Basilar Impression (BI)**

Also known as basilar invagination, 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. Not all individuals with BI have symptoms that get progressively worse.

## **Weight**

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

## **Diet**

OI adults with short stature may require less calcium and vitamin D supplementation than usually prescribed. Total calcium intake of 800 to 1,000 mg (milligrams) per day is usually sufficient. Supplemental vitamin D intake should not exceed 800 IU per day.

## **Constipation**

Constipation is a problem for some people with osteogenesis imperfecta (OI). Two studies indicate that adults with OI Type III and pelvic malformation are more likely to complain of constipation and abdominal pain. The colon and bowel may be prevented from functioning normally if the hips and pelvis are narrow or malformed. A specific pelvic malformation, known as acetabular protrusion, can affect some people with OI Type III and cause a predisposition to constipation. Diminished mobility and low levels of physical activity also contribute to constipation.

Dehydration is a well-known factor contributing to constipation. Individuals with OI who have increased perspiration should be closely monitored so they remain hydrated.

Some medications, especially pain medications, can be constipating because they decrease the normal motion and movement of the bowel. Adults with OI and parents of children with OI are encouraged to check with their doctor and/or pharmacist about side effects and possible drug interactions from their medicines and dietary supplements.

Fecal impaction, or clogging of the bowel with hardened stool, can be a serious complication. Treatment of constipation in people with OI is often challenging.

### **Social, Emotional, and Family Issues**

Many people with OI Type I do not appear disabled, so others may misunderstand or underestimate the disorder. Parents may provide information about preventing fractures to teachers, babysitters, or other caregivers, only to have the caregivers dismiss them as being "overprotective." Meeting with teachers and other school staff and providing written information and a letter from the child's doctor briefly explaining the OI diagnosis and the recommended precautions - can help reinforce the information provided by parents. It is important to set up a system to ensure that substitute teachers also are aware that a child with Type I OI is in the class.

In addition, it is important for a child's siblings and peers to receive age-appropriate information about OI. It is common for peers to wonder why their classmate does adapted activities during physical education, or can't participate in contact sports. Some children with mild OI are accused of being "clumsy" or "lazy," or of "faking it" when they have yet another injury. In most cases, such teasing comes out of ignorance, not malice. Many children with OI or their parents give a brief presentation to the class at the beginning of each school year to explain OI. Visual aids and props - such as the child's braces, or a cast or splint - are particularly meaningful to young children.

Some families with mildly affected children have been accused of child abuse after their child went to the emergency room with unexplained fractures. Once an OI diagnosis is made, families should ask for a letter on medical letterhead confirming the diagnosis and explaining what it means. Copies of the letter should be kept in the diaper bag, in the car, with the child's medical and school records, and anywhere else where they may be useful, particularly when the family is traveling or visiting the emergency room.

Adults with Type I OI usually know about their diagnosis when deciding whether to have children. There is a 50 percent chance that a person with Type I OI will

pass the disorder on to his or her child, and that chance remains the same for each child. Usually, a child will inherit the same type of OI as his or her parent. However, it is possible that the child's signs and symptoms will be different than the parent's - either milder or, in some cases, more severe. Adults with Type I OI who are considering having children may wish to consult a genetic counselor to obtain information about their options, including pre-implantation genetic diagnosis (PGD). It is worthwhile to obtain a skin biopsy or DNA analysis to confirm one's own OI diagnosis before conceiving a child. Having this information on file expedites testing a newborn for OI, if the parents desire it. Type I OI does not appear to affect fertility or predispose women to particular pregnancy complications.

Research suggests that pregnancy and breastfeeding may affect a woman's bone density, and may increase the risk of fracture. It is, therefore, particularly important that women with OI eat a calcium-rich diet and exercise appropriately while pregnant and breastfeeding.

People with Type I OI often have to deal with the conflict between their outwardly healthy appearance and their underlying fragility. Deciding who to tell about their condition in social or employment situations can be difficult. Concern about strength, stamina, and changes as a person ages can also affect decisions about family life, housing, and careers. Adults with Type I OI recommend developing an effective personal support network.

### Myths About OI

**Myth:** A baby with OI should always be carried on a pillow and discouraged from moving.

**Fact:** Although there are handling techniques and precautions, it is in the child's best interest to be held and touched and to explore independent movement to the greatest extent possible. Immobility increases bone loss and decreases muscle mass, leading to weakness, bone fragility, and more fractures.

**Myth:** Fractures caused by OI can be easily distinguished from those caused by child abuse

**Fact:** Children with OI can have all types of fractures, including spiral, rib, skull, incomplete, and displaced fractures. Distinguishing OI fractures from child abuse requires a thorough assessment by a medical professional who is familiar with the full range of OI characteristics.

**Myth:** OI only affects the bones.

**Fact:** Though fragile bones are the hallmark of OI, other medical problems, including loose joints, early hearing loss, brittle teeth, respiratory problems, and easy bruising, are also part of the disorder.

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**Myth:** OI is a childhood disorder; people grow out of it by their teens.

**Fact:** OI is a genetic disorder that is present throughout a person's lifetime. The frequency of fractures may decrease after puberty, when growth stops. Later, it may increase again in women with the onset of menopause and in men due to age-related changes in their endocrine system.

**Myth:** People with OI are usually diagnosed at birth.

**Fact:** OI Type I, the most common and mildest form of OI, is rarely diagnosed at birth unless a parent has OI. In fact, some very mild cases are only diagnosed when a person has a child with OI Type I, and a review of the person's medical history reveals a pattern of fractures and other features of OI. OI is primarily a clinical diagnosis. Collagen studies and/or DNA analysis can identify the mutation and confirm the clinical diagnosis. Negative results on these tests do not eliminate the diagnosis of OI.

**Myth:** People who have OI cannot have children.

**Fact:** OI does not affect fertility. Many men and women who have OI have children. Some women who have OI may experience pregnancy complications due to skeletal problems. It is important that all young people with OI receive information about their condition and reproductive health.

**Myth:** All children of a parent who has OI will have OI.

**Fact:** When one parent has a dominantly inherited type of OI, there is a 50 percent chance with each pregnancy that the child will have OI. There is a 50 percent chance that the child will not have OI. In the rare instances where OI is transmitted as a recessive trait, parents are healthy carriers and their children have a 25 percent chance to be affected and a 50 percent chance to be carriers.

## Supplemental Information

### [Osteogenesis Imperfecta](#)

Morello, R., & Esposito, P. W. (2012). Osteogenesis Imperfecta. In Y. Lin (Ed.), *Osteogenesis*: InTech. CC BY 3.0

### [A Systematic Overview of Osteogenesis Imperfecta](#)

Alharbi, S. A. (2016). A Systematic Overview of Osteogenesis Imperfecta. *Molecular Biology: Open Access, 2016*. CC BY 4.0

### [Recent developments in osteogenesis imperfecta](#)

Shaker, J. L., Albert, C., Fritz, J., & Harris, G. (2015). Recent developments in osteogenesis imperfecta. *F1000Research, 4*. CC BY 4.0

### [Initial report of the osteogenesis imperfecta adult natural history initiative](#)

Tosi, L. L., Oetgen, M. E., Floor, M. K., Huber, M. B., Kennelly, A. M., McCarter, R. J., ... & McKiernan, F. E. (2015). Initial report of the osteogenesis imperfecta adult natural history initiative. *Orphanet journal of rare diseases, 10*(1), 1. CC BY 4.0

### [Quality of life in children and adolescents with Osteogenesis Imperfecta: a qualitative interview based study](#)

Hill, C. L., Baird, W. O., & Walters, S. J. (2014). Quality of life in children and adolescents with Osteogenesis Imperfecta: a qualitative interview based study. *Health and quality of life outcomes, 12*(1), 1. CC BY 2.0

### [From pediatric to adult care: strategic evaluation of a transition program for patients with osteogenesis imperfecta](#)

Dogba, M. J., Rauch, F., Wong, T., Ruck, J., Glorieux, F. H., & Bedos, C. (2014). From pediatric to adult care: strategic evaluation of a transition program for patients with osteogenesis imperfecta. *BMC health services research, 14*(1), 489. CC BY 2.0

### [The impact of severe osteogenesis imperfecta on the lives of young patients and their parents—a qualitative analysis](#)

Dogba, M. J., Bedos, C., Durigova, M., Montpetit, K., Wong, T., Glorieux, F. H., & Rauch, F. (2013). The impact of severe osteogenesis imperfecta on the lives of young patients and their parents—a qualitative analysis. *BMC pediatrics, 13*(1), 153. CC BY 2.0

### [A Systematic Overview of Osteogenesis Imperfecta](#)

Alharbi, S. A. (2016). A Systematic Overview of Osteogenesis Imperfecta. *Molecular Biology: Open Access, 2016*. CC BY

### [Multiple Osteotomies and Corrective Surgery with Intramedullary Nailing For Multiple Fractures in Osteogenesis Imperfecta](#)

Demet Dogan, E. (2011). Multiple Osteotomies and Corrective Surgery with Intramedullary Nailing For Multiple Fractures in Osteogenesis Imperfecta. *Orthopedic & Muscular System*. CC BY

### [Osteogenesis imperfecta complicated with psychosis secondary to complex partial seizures](#)

Samadi, R., Manteghi, A. A., Haghighi, M. B., & Assari, S. (2015). Osteogenesis imperfecta complicated with psychosis secondary to complex partial seizures. *International Journal of Case Reports and Images (IJCRI), 6*(6), 332-337. CC BY

### [A Therapeutic Role for Hematopoietic Stem Cells in Osteogenesis Imperfecta](#)

Mehrotra, M., & LaRue, A. C. (2013). A Therapeutic Role for Hematopoietic Stem Cells in Osteogenesis Imperfecta. In M. Puiu (Ed.), *Genetic Disorders*: InTech. CC BY 3.0

### [Involving Families with Osteogenesis Imperfecta in Health Service Research: Joint Development of the OI/ECE Questionnaire](#)

Forlino, A., Cabral, W. A., Barnes, A. M., & Marini, J. C. (2011). New perspectives on osteogenesis imperfecta. *Nature Reviews Endocrinology, 7*(9), 540-557. CC BY PD

### [Radiographic Features of Osteogenesis Imperfecta](#)

Renaud, A., Aucourt, J., Weill, J., Bigot, J., Dieux, A., Devisme, L., ... & Boutry, N. (2013). Radiographic features of osteogenesis imperfecta. *Insights into imaging, 4*(4), 417-429. CC BY

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## Osteogenesis Imperfecta

### Post-Test

- Osteogenesis imperfecta is also known as \_\_\_\_\_. (p. 3)
  - Arthroprogenesis
  - Osteopenia
  - Juvenile arthritis
  - Brittle bone disease
- Osteogenesis imperfecta does not affect cognitive abilities. (p. 3) A. True B. False
- Which of the following is NOT a characteristic typically associated with OI? (p. 3-4)
  - Muscle weakness
  - Thermal instability
  - Gastrointestinal complications
  - Hearing loss
- Individuals with \_\_\_\_\_ OI typically do not survive beyond infancy. (p. 4-6)
  - Type I
  - Type II
  - Type III
  - Type IV
- Which type of OI does NOT involve a defect of type 1 collagen? (p. 5-6)
  - Type II
  - Type III
  - Type IV
  - Type V
- With rare exceptions, OI results from mutations in the type I collagen genes and is considered to be a dominantly inherited disorder. (p. 7) A. True B. False
- Protein, RNA, and DNA can all be tested to diagnose OI. (p. 10) A. True B. False
- A DXA z-score of -1 to +1 standard deviations is considered positive for diagnosing OI. (p. 11) A. True B. False
- Which prenatal test(s) may be utilized to assess the fetus for OI? (p. 11-12)
  - Ultrasound
  - Amniocentesis
  - Chorionic villus sampling (CVS)
  - All of the above
- Which of the following supplements has proven to be effective for treating OI? (p. 13)
  - Fluoride
  - Calcitonin
  - High doses of calcium
  - None of the above
- Touching and holding of the infant by parents and caregivers should be strictly limited. (p. 14) A. True B. False

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12. Most babies with OI are capable of being breastfed. (p. 15) A. True B. False
13. Waterbeds or very soft bedding is recommended to prevent pressure points and resultant fractures. (p. 16) A. True B. False
14. In adults after puberty, the fracture rate usually decreases as a consequence of hormonal and other metabolic changes affecting collagen and bone. (p. 18) A. True B. False
15. Individuals with OI typically feel less pain than most other people do. (p. 22) A. True B. False
16. Passive rotational ROM of the limbs is a benign way to initiate therapy on a child with OI. (p. 29) A. True B. False
17. Which developmental progression sequence is correct per the King recommendations? (p. 31)
  - A. supine, inclined sitting, prone, sidelying
  - B. prone, supine, inclined sitting, sidelying
  - C. supine, sidelying, prone, inclined sitting
  - D. supine, prone, inclined sitting, sidelying
18. Braces for individuals with OI should be \_\_\_\_\_. (p. 35)
  - A. Circumferential
  - B. Perforated
  - C. Lightweight
  - D. All of the above
19. \_\_\_\_\_ is an associated health issue frequently seen in adults with OI. (p. 42-46)
  - A. Gastric acid reflux
  - B. Aortic aneurysm
  - C. Glaucoma
  - D. Chronic diarrhea
20. Which one of the following statements is TRUE? (p. 47-48)
  - A. Children with OI should be held and touched.
  - B. Fractures caused by OI can be easily distinguished from those caused by child abuse.
  - C. OI only affects the bones.
  - D. People with OI are usually diagnosed at birth.

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