**Osteogenesis Imperfecta (Brittle Bone Disease):**

|  |
| --- |
| Osteogenesis imperfecta (OI) is a genetic disorder characterized by fragile bones that break easily. It is a heritable condition of connective tissue. OI can be caused by a dominant or a recessive mutation. Approximately 35% of children with OI are born into a family with no family history of OI. Most often this is due to a new mutation to a gene and not b anything the parents did before or during the pregnancy. Spontaneous new mutations are common, and can cause any type of OI, not just OI Type II or III. OI occurs with equal frequency among males and females and among all racial and ethnic groups. In addition to fractures, people with OI often have muscle weakness, hearing loss, fatigue, joint laxity, curved bones, scoliosis, blue sclera, dentinogenesis imperfecta, bleeding and easy bruising, and short stature. OI is caused by a mutation on a gene that affects the body’s production of the collagen found in bones and other tissues. Collagen is an important building block in bones. OI is not caused by a deficiency in calcium. *Clinical Features:* OI is a highly variable disorder. A person who has it might exhibit only a few of the common characteristics. Some characteristics are age-dependent and will not be seen in an infant or young child. Other characteristics are present in only certain types of OI. The clinical features of OI may include the following:1. The sclera (whites of the eyes) might appear darker than normal, with a blue or gray tint.
2. Dentinogenesis imperfect – characterized by transparent, discoloured and fragile teeth that may fracture easily. DI is evident in approximately 50% of people with OI, particularly those with the severe forms. Dental abnormalities are usually apparent when the first tooth erupts.
3. Bone malformations can include abnormal rib shape, pectus carinatum (round chest) or pectus excavatum (hollowed chest), curving of the long bones, vertebral compressions, spinal curves, scoliosis, mild kyphosis (hunchback)and an abnormal skull shape.
4. Osteopenia (low bone mineral density) may be apparent on x-ray or with bone density tests
5. Head circumference may be greater than average, or the head may appear large relative to the person’s small body.
6. Wormian bones, which are small bone islands in the skull where normally there is an intact sheet of bone, are seen in approximately 60% if people who have OI.
7. A triangular facial shape is characteristic in the more severe forms
8. Hearing loss may start in the young adult years.
9. Short stature and the body may be disproportional
10. Infants may have a low weight for their age. Older children are frequently overweight for their size
11. The skin may feel soft and bruise easily
12. The joints may be lax and unstable and the feet may be flat.
13. Gross motor development may be delayed due to fractures and/or hypotonia
14. The intellect is normal

https://encrypted-tbn3.gstatic.com/images?q=tbn:ANd9GcTVcGNs-zvi2noKSYrbfqqTKKMupHJaI1c4Fz2moPWzB2bIEiBnhttp://2.bp.blogspot.com/_gwtpUd4El9A/S3QZMdQuxCI/AAAAAAAAEHM/-xWuimpftCY/s400/6.JPGhttp://orthoinfo.aaos.org/figures/A00051F01.jpghttp://upload.wikimedia.org/wikipedia/commons/thumb/8/83/Pectus1.jpg/230px-Pectus1.jpghttp://img.tfd.com/mosby/thumbs/500063-fx7.jpghttps://encrypted-tbn2.gstatic.com/images?q=tbn:ANd9GcQ6yR6q5m0Gmru6RDPCDFXEhDScCmHMIShw6OBjXaO1julOoxWSShort stature + disproporionalWormonian BonesCurving of the long bonesBlue ScleraPectus excavatumDentinogenesis ImperfectaDecreased chest volume and deformities may lead to restrictive pulmonary disorder in severe cases of OI. Pulmonary complications can occur due to rib fractures, kyphoscoliosis, muscle weakness of the chest wall, heart valve disorders, and chronic bronchitis or asthma. Lung collagen abnormalities have a negative impact on lung function. Respiratory complications, including pneumonia, are a significant cause of death among the OI population, especially among those with OI Type III. Supplemental oxygen, bilevel positive airway pressure, or bronchodilators for asthma may be indicated. Even when not caused by OI, lung problems can be more severe in people who have OI of any type. People with OI of any type have higher rates of asthma and pneumonia than people who do not have OI.*Types of OI:* There are 8 types of Osteogenesis Imperfecta. These range in severity from a lethal form to a milder form with few visible symptoms. There can be significant variation within the types of OI in terms of severity of symptoms. http://www.nejm.org/na101/home/literatum/publisher/mms/journals/content/nejm/2006/nejm_2006.355.issue-26/nejmicm062996/production/images/large/nejmicm062996_f1.jpegPictureType 1 is the mildest, and accounts for 50% of all cases of OI. It is dominantly inherited. Type 1 is characterized with mild bone fragility, relatively few fractures and minimal limb deformities. The child might not fracture until he or she is learning to walk. Some children have few obvious signs of OI. Others might experience multiple fractures of the long bones, compression fractures of the vertebrae and chronic pain. After growth is completed, the incidence of fractures decreases considerably. Blue sclera are often present. Typically a child’s stature may be average or slightly shorter-than-average as compared with unaffected family members. There is a high incidence of hearing loss. Onset primarily occurs in young adulthood, but it may occur in early childhood. Dentinogenesis imperfecta is often absent. Significant care issues that arise with OI Type I included gross motor developmental delays, joing and ligament weakness and instability, muscle weakness, the need to prevent fracture cycles and the necessity of spine protection. The treatment plan should mazimize mobility and function, increase peak bone mass and develop muscle strength. Physical therapy, early intervention programs and as much exercise and physical activity as possible will improve outcomes. OI type II is the most severe form. At birth, infants with OI Type II have very short limbs, small chests and soft skulls. Their legs are often in a frog-leg position. The radiological features are characteristic and include absent or limited calvarial mineralization, flat verbebral bodies, very short, telescoped broad femurs, beaded and often broad short ribs and evidence of malformation of the long bones. Intrauterine fractures will be evident in the skull, long bones or vertebrae. The lungs are underdeveloped and respiratory and swallowing problems are common. Infants with OI Type II will normally die within a few weeks. http://www2.marshfieldclinic.org/wissp/wisspers/w13f6.gifOI Type III is the most severe type among children who survive the neonatal period. The degree of bone fragility and the fracture rate vary widely. At birth, infants generally have mildly shortened and bowed limbs, small chests and a soft calvarium. Respiratory and swallowing problems are common in newborns. There may be multiple long-bone fractures at birth, including many rib fractures. Frequent fractures of the long bones, the tension of muscle on soft bone and the disruption of the growth plates lead to bowing and progressive malformation. Children have a markedly short stature, adults are normally shorter than 102 cms. Spine curvatures, compression fractures of the vertebrae, scolosis and chest deformities occur frequently. The head is often large relative to body size and the face has a triangular shape due to overdevelopment of the head and underdevelopment of the face bones. Dentinogenesis imperfecta is common, but not universal. The majority of OI Type III cases result from dominant mutations, so genetic counseling is recommended regarding future pregnancies. Significant care issues that arise with OI Type III include the need to prevent fracture cycles, the appropriate timing of rodding surgery, scoliosis monitoring, respiratory function monitoring, the need to develop strategies to cope with short stature and fatigue, the families need for emotional support and medication. People with OI Type IV are moderately affected, but it can range in severity from relatively few fractures as in OI Type I, to a more severe form resembling OI Type III. People with OI Type IV have moderate to severe growth retardation, which is one factor that distinguishes them clinically from people with Type I. Bowing of the long bones is common, but to a lesser extent than in Type III. The sclera are often light blue in infancy, but may lighten to which later in childhood or early adulthood. The childs height may be less-than-average for his or her age. It is common for the humerous and femur to be short. Long bone fractures, vertebral compression, scoliosis and ligament laxity may also be present. Dentinogenesis imperfecta may be present or absent. OI Type IV has an autosomal dominant pattern of inheritance. Many cases are the result of a new mutation. Significant care issues that arise with OI Type IV include the need to prevent fracture cycles, the appropriate timing of rodding surgery, scolosis monitoring, the need to develop strategies for coping with short stature and fatigue. http://adc.bmj.com/content/vol92/issue4/images/large/ac96552.f3.jpegOI Type V is moderate in severity. It is similar to OI Type IV in terms of frequency of fractures and the degree of skeletal deformity. The most conspicuous feature of this type is large, hypertrophic calluses in the largest bones at fracture or surgical procedure sites. These calluses can also arise spontaneously.OI Type VI is extremely rare. It is moderate in severity and similar in appearance and symptoms to OI type IV. This type is distinguished by a characteristic mineralization defect seen in biopsied bone. OI Type VII and VIII are recessively inherited types of OI. Unlike the dominantly inherited types, the recessive types of OI do not involve mutations in the type 1 collagen genes. Recessively inherited OI has been discovered in people with lethal, severe and moderate OI. There is no evidence of a recessive form of mild OI. Recessive inheritance probably accounts for fewer than 10% of OI cases. Parents of a child who has a recessive type of OI have a 25% per pregnancy of having another child with OI. Unaffected siblings of a person with a recessive type have a 2 in 3 chance of being a carrier of the recessive gene. Some cases of OI Type VII resemble OI Type IV in many aspects of appearance and symptoms. Short stature and coxa vara are common. It is common for leg bones, humerus and femur to be short. Cases of OI Type VIII are similar to OI Types II or III in appearance and symptoms except for white sclera. OI Type VIII is characterized by severe growth deficiency and extreme under-mineralization of the skeleton. Other medical conditions that share some of the same clinical signs as OI include hypophophatasia, juvenile Paget’s disease, rickets, idiopathic juvenile osteoporosis, some inherited defects in vitamin D metabolism, Cushing’s disease and calcium deficiency. *Diagnosis:* Diagnosis is primarily based on clinical evidence. It may be difficult to make a clinical diagnosis of the milder forms of OI in infancy. Laboratory studies can rule out other conditions, provide information that is useful in medical management, and in most cases, confirm the diagnosis through mutation identification. There are many laboratory studies that can be done to test for OI, though blood tests are unreliable. Diagnosis is also made through a medical history, a family history and a physical examination. X-ray findings can include osteopenia (low bone density), fractures (new, subclinical, or old-healing), bowing of the long bones, vertebral compressions, and wormian bones in the skull sutures. *Treatment:* There is no cure for OI. Treatment focuses on minimizing fractures, maximizing mobility, mazimizing independent function and general health. Treatments include:1. Physical therapy and safe exercise including swimming
2. Casts, splints or wraps for broken bones
3. Braces to support legs, ankles, knees and wrists as needed
4. Orthopedic surgery, often including implanting rods to support the long bones in arms or legs
5. Medications to strengthen bones – bisphosphonates, growth hormones, increased vitamin D intake
6. Mobility aids such as canes, walkers or wheelchairs.
7. Behavioural and lifestyle modifications to prevent situations that might cause a fracture.
8. Scoliosis management

The prognosis for a person with OI varies greatly depending on the number and severity of symptoms. Life expectancy is not affected in people with mild or moderate symptoms. However, life expectancy may be shortened in those displaying more severe symptoms. The most severe form of OI results in death at birth or during infancy. Respiratory failure is the most frequent cause of death for people with OI, followed by accidental trauma. **Managing Osteogenesis Imperfecta:**The most important tenants for managing OI in children are safe handling, protective positioning and safe movements. These should be taught to parents as soon as possible. Children and youth need to learn which activities to avoid, and how to practice energy conservation. The number of fractures usually decreases in adulthood. Other important behavioural, lifestyle and medical considerations include: 1. Safe handling of the infant
2. Constipation
3. Dental Care
4. Exercise and physical activity
5. Pain management
6. Nutrition
7. Hearing Loss
8. Musculoskeletal and Orthopedic Issues
9. General Health
10. Infections and respiratory health

**Safe Handling of the Infant:** Children who have OI need protective handling, good positioning and protected movement. Caregivers should remember to encourage age appropriate cognitive development through language development and by encouraging small motor skills. Children with severe OI may appear to be much younger than their chronological age. In general, parents need to pad surfaces that the child may bump including bed rails, car seat, stroller and tub. When handling an infant with OI, all movements should be slow, methodical, and gentle. The infant who has osteogenesis imperfecta (OI) has some special characteristics. The infant may have an unusually soft skull, startle very easily, have bone deformity and have fractures, often of the ribs or long bones, in various stages of healing. Never push, pull, twist, bend, apply pressure to, or try to straighten an infant’s arms or legs. There are a couple of guiding principles that should be followed: 1. Never pull or push on a limb, or bend it into an awkward position.
2. Infants with OI should not be picked up under the axillae or around the rib cage. Such actions can cause rib fractures. Instead, you should support the infant’s head and trunk with one hand, while your other hand supports the buttocks.
3. When holding an infant with OI, keep your fingers spread apart to provide a wider base of support and an even distribution of support pressure.
4. When lifting the baby for feeding, dressing, or diapering, apply support to the broadest possible area along the buttocks, back, and head.
5. 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.
6. Care should be taken when changing the infant’s clothing, bedding, or bandages to protect his or her arms, wrists, fingers, legs, and ankles.
7. When dressing the infant, bring garments over the infant’s limbs; do not pull the limbs through sleeves or pants’ legs. Pulling, twisting, or getting a limb caught in clothing can cause fractures.
8. It is important that babies with OI receive affection and that they are held and touched by their parents and other caregivers.
9. Infants can be carried against the caregiver’s shoulder. The caregiver should bend down to pick up the infant from a flat surface rather than bring the infant all the way up to adult height before positioning him or her against the caregiver’s own trunk and shoulder.
10. Regularly alternate which shoulder you use to support the infant to encourage the infant to develop different neck muscles.
11. Once infant has achieved head and neck control, he or she can be carried facing forward, against the caregiver’s chest. Place one hand around the child’s chest and another under the buttocks.
12. Hydrocephalus occurs in a large percentage of children with Type III OI. Extra care is needed to support the head if it is oversize for the small body.
13. Be aware of where the baby’s arms and legs are at all times to avoid awkward positions or getting a hand or foot caught.
14. When a fracture is suspected, minimize handling of the affected limb.
15. Clothing: Simple, lightweight cotton clothing is recommended. Clothes with buttons or snaps or velcro down the front and at the crotch are easier to put on than more complicated garments. Roll up sleeves or pants legs and gently pull the garment over the arm or leg. Do not pull the arm or leg through the sleeve or pant leg.

*Preventing Head Flattening* http://radiographics.rsna.org/content/32/7/2101/F3.small.gifAll infants with OI have soft skulls. To prevent skull malformations, every effort should be made to reduce pressure on the back of the head. The following strategies are beneficial:1. Put gel pads under the infant’s head when he or she is on her back.
2. Position the infant in a propped, side-lying position.
3. Frequently change the infant’s position throughout the day.
4. Carry the infant on your shoulder or in an approved sling carrier.

**Constipation in Osteogenesis Imperfecta:**Constipation is a problem for some people with osteogenesis imperfecta. Constipation, in medical terms, is a decrease in frequency of stools or bowel movements with hardening of the stool. As a consequence, people with constipation may have feelings of being bloated and having gas but find it difficult to pass stools. Because the stool becomes quite hard, passing it may be uncomfortable and lead to further unwillingness to try, especially in infants and young children. 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.Causes of Constipation Constipation can be caused by inadequate dietary fiber, lack of exercise, and dehydration. Colon obstruction and illness are additional causes. Some medications, especially pain medications, can be constipating because they decrease bowel wall motion. OI may contribute to constipation especially in people with the short stature and pelvic deformity associated with OI Type III. The colon and bowel may be prevented from functioning normally if the hips and pelvis are narrow or deformed. A specific pelvic deformity known as acetabular protrusion is known to affect some people with OI Type III and causes a predisposition to constipation. Diminished mobility and low levels of physical activity also contribute to constipation. Dehydration is a well-known contributing factor to constipation. Children with OI who have increased perspiration should be closely monitored so they remain hydrated.Managing Constipation Managing constipation usually involves a combination of approaches. It is essential to determine the extent of the problem and develop a plan to correct it. 1. Keep a record of bowel movements, diet and fluid intake. Even when a child appears to be "regular," keeping records about diet, illnesses, exercise and bowel movements will help caregivers figure out if something in particular triggers the problem.
2. Be consistent about diet, exercise and activity and fluids.
3. Children and adults with OI often respond to diet changes, a change in activity level, and mild home remedies.

Treatment options include the following:1. Diet and fluids
2. Exercise and activity
3. Home remedies
4. Medications

*Diet and Fluids:*Dietary changes can include the following suggestions. 1. Add fiber to the diet including whole grain breads, whole grain cereals, bran cereals or muffins, and popcorn. Ground beans can be added to other foods.
2. Increase amount of fruits, fruit juice and vegetables.
3. Add beans and nuts to the diet.
4. Reduce amount of processed foods and refined sugars.
5. Avoid “junk” foods that are high in fat and low in fiber.
6. Include foods such as yogurt with active cultures that contain the bacteria lactobacillus acidophilus.
7. Limit soft drinks and drinks containing caffeine such as colas or tea.
8. Drink water throughout the day.

Strive for a diet that keeps the stool soft. Too much fiber has the secondary effect of creating too much bulk for someone with a connective tissue disorder. This can put pressure on the rectum. Pressure, plus the lax or elastic muscles seen in the pelvic floor of a person with OI, inactivity, too much prolonged sitting, and chronic constipation can lead to a more serious problem called rectal prolapsed. *Exercise and Activity* Adding exercise and physical activity can help prevent and relieve constipation. Exercise helps move digested food through the intestines. Infants, children and adults who sit, recline, or use a wheelchair require regular position changes. Infants benefit from water play that encourages them to kick. Children and adults can benefit from swimming, walking or bicycling.*Home Remedies* Home remedies can include: 1. Karo syrup
2. Prune juice mixed with a good quality apple juice or applesauce to make it more palatable.
3. Allowing enough time on the toilet. Some physicians suggests 10-15 minutes at least twice a day.

*Medications* For some children, regular medication or even a prescription laxative may be necessary. Use of medication, whether prescription or over-the-counter, must be discussed with your physician. Children on a medication need to be carefully monitored. Taking a medication too often can reduce its effectiveness or aggravate negative side effects. Medications can include: 1. Suppositories
2. Mineral oil
3. Stool softeners
4. Laxatives such as Senokote®
5. Prescription laxatives such as Miralax®

Persistent, painful constipation should not be ignored. Staying well hydrated, eating a healthy diet and increasing exercise can reduce the frequency of constipation in children and adults who have OI.**Dental Care in Osteogenesis Imperfecta:**Osteogenesis imperfecta (OI) is always associated with bone fragility. In addition, OI may affect the growth of the jaws and may or may not affect the teeth. About half of the people who have OI have teeth that appear normal, and their major concerns are routine care. However, the other half has a defect in the teeth called dentinogenesis imperfecta (DI), sometimes referred to as opalescent teeth or brittle teeth. These teeth may be misshapen, may chip or break easily, and will require special care. Oral cavity problems related to OI may include the following: 1. A skeletal Class III malocclusion. The teeth do not correctly match up making biting difficult. This is caused by the size and/or position of the upper jaw or the lower jaw.
2. An open bite. There is a vertical gap between some of the upper and lower teeth.
3. Impacted teeth. The first or second permanent molars do not erupt, or they erupt out of the usual location (ectopic).
4. Dental development. Tooth development may be delayed or advanced in some individuals affected by OI.

OI does not affect the presence or absence of gum disease (periodontitis). Dentinogenesis imperfecta can be part of osteogenesis imperfecta (DI type I) or it can be a separate inherited dominant trait without OI (DI type II). DI occurring with OI seems to run in families but can vary in severity from one member to another. DI has a variable affect on the color, shape, and wear of both primary and permanent teeth. If someone has OI and DI, all of their teeth may not be affected to the same degree. Teeth affected by DI have essentially normal enamel. Affected teeth will have an increased incidence of fracture, wear and decay. Dentinogenesis imperfecta may be diagnosed with the first baby tooth. If the tooth looks gray, bluish, or brown, DI should be suspected. Children should be taken to a dentist when the first teeth are erupting. This may happen as early as 6 months to 1 year of age. Radiographs, or X-rays, can be useful but may be difficult to obtain until the child is older. Sometimes there are changes visible on the X-rays that are not obvious just by looking at the teeth. Crowns appear bulbous and roots may be shorter and more slender than standard. Primary teeth are usually more affected than the permanent teeth. General Care for People With OI and Without DI A dentist should see a child with OI by 6 months after the eruption of the first baby tooth at the latest. Baby teeth require care. They are important for chewing, speaking, holding space for the permanent teeth to grow in, and growth of the jaws. There appears to be minimal risk of jaw fracture from routine dental care and dental extractions. No particular precautions are needed other than those that would be taken anyway, such as support of a very thin lower jaw when an extraction procedure is being done. Good care involves brushing and flossing the teeth of young children, then teaching them how to do it themselves and checking them as they grow older. Soft toothbrushes are good for everybody and easier on gums, since gums also need brushing. Use of fluoride toothpaste is recommended. Children should use a small dab of toothpaste, or a children’s toothpaste, and be taught to spit it out well after brushing so they do not swallow excessive amounts. Before going to bed, children should spit out the toothpaste after brushing, but not rinse their mouths. This will leave more fluoride in the mouth to work overnight. General Care for People With OI Plus DI Children with OI and dentinogenesis imperfecta need the same basic care as discussed above, but they also need to be monitored for cracking, chipping and abrasion of the teeth. Special care will be needed even with the baby teeth. All of the teeth may not be affected by DI, and primary teeth usually are affected to a greater extent than the permanent teeth. Restorative treatment may be needed at some point. Regular care is needed so the teeth will last as long as possible and to prevent abscesses and pain. Brushing and cleaning has not been shown to cause damage, but will not make teeth affected by DI white. Sealants should be effective on teeth affected with DI as long as the enamel is intact. Older children and especially adolescents with DI are often embarrassed by their discolored teeth. Different types of veneers can sometimes hide the problem. If the teeth are wearing excessively, caps (also called crowns), will probably need to be placed on at least some of the teeth. Caps serve to keep the teeth in place and encourage proper development of the jaw. More specialized treatment may be more appropriate for permanent teeth. **Exercise and Physical Activity in Osteogenesis Imperfecta:** Not so long ago, parents were advised to “protect” children who have OI by carrying them on pillows and avoiding recreational activities. This well-intentioned approach did not protect children from fractures (broken bones), and may have hindered development and achievement of independent functioning. Bone growth depends on muscle pull as well as loading or weight bearing through standing, walking and lifting. Immobilization may result in loss of muscle and skeletal mass. It may take as long as a year to restore bone mass following a relatively short period of immobilization. Over the years it has become clear that physical activity is an important part of managing OI in both children and adults. Research indicates that physical activity is important because it promotes: 1. general health – including cardiovascular fitness, mental alertness, weight control, improved sleep quality, improved ability to handle infection, reduced risk for some cancers
2. maximum bone density
3. optimal physical function to support independence in daily activities
4. optimal psychological and social well-being by improving self –confidence and the ability to interact socially with peers.

Children and adults with OI will benefit from a regular program of physical activity to promote optimal function through muscle strengthening, aerobic exercise and recreational pursuits. Specifics of the exercise program vary depending on the person’s age, level of function, severity of OI, and needs and desires. A well-designed program can combine activities to prevent problems as well as to restore function. Activity programs may include specific exercises recommended by rehabilitation professionals (physiatrists, physical therapists, occupational therapists, and recreation therapists), as well as sports and other recreational activities. Having fun and gaining a sense of accomplishment are legitimate goals for an exercise program. In addition, diet, weight control, and commitment to a healthy lifestyle are essential to longevity and an improved quality of life. The optimal long-term goal for children with OI is good health and independence in all areas of function (social, educational, self-care, locomotion, and recreation) using adaptive devices as needed. Goals for adults with OI include maintaining independence, preserving bone density, and supporting cardiovascular function. To achieve these goals, it is often necessary to improve muscle strength and body alignment. When to Begin: The first year of life includes many motor skill transitions and is a critical window of opportunity for babies who are born with muscle weakness, alignment problems, and fragility. Physical therapy should begin as soon as it is evident that the infant has weakness or motor skill delays when compared with other infants of the same age. This might be first noticed because the baby cannot hold up his or her head independently or possibly not sit without support until later than most other children. Treatments for such problems are often aimed at proper positioning, and placing children in a position to encourage their use of certain muscle groups. Proper positioning elicits specific antigravity muscular effort, which is the basis for learning to sit and stand later on. Babies with large heads will face additional challenges and limitations in developing the ability to move against gravity. An infant or child with weakness or motor skill delays should be working for brief periods daily or at least five days a week to improve muscle strength and motor skills such as coordination and balance. In the process, the child gains endurance and independence in self-care activities. Treatment should not be confined to 3 “therapy hours” only. Very short exercise efforts during the day, as short as five minutes, will often result in improvement more quickly than an hour-long session once or twice a week. Depending on the child’s age, the interventions will take several forms including positioning, specific exercises, and developmental activities such as standing in a standing device. Ideally, the activities are integrated naturally into the child’s day by family members and care providers. Playtime can be purposeful, but it should still be fun for parent and child. Children with OI can excel in the water, particularly if it is presented as an opportunity for recreation and independent exploration, rather than a situation where an adult places demands on the child to exercise. Water exercise can begin during infancy with backlying in 2 to 3 inches of warm water to promote independent kicking. Over time, the child can progress to independent activity in the water, first in a swim vest or other support, then swimming without support. Waterwalking may be possible for individuals who are not able to walk outside the pool. Water activities in childhood can be the foundation for a lifelong enjoyable fitness activity. Adults with OI can benefit from water activity, as well. It is an excellent form of aerobic conditioning, and may have some benefit with respect to strengthening. Because water activities do little to promote bone health, however, adults should also try to add walking or other weight-bearing exercise to their physical activity program. Safety:People of any age who have OI can safely exercise. Obstacles to consider when evaluating an activity include prior fracture history, degrees of bending of long bones, degree of muscle weakness, joint stiffness or laxity (looseness), joint alignment, poor exercise tolerance, and stamina. Inability to accomplish daily activities without specialized equipment can also affect which activities can be done safely. For instance, long-term sitting in a wheelchair may be associated with hip flexion contractures and compensatory back curvatures, often associated with back pain, joint stiffness, osteoporosis, and obesity. A safe physical activity program would include getting out of the chair and changing body positions at least every 2 hours when possible. People who have OI should avoid some activities. These include jumping, diving, and contact sports, as well as activities that promote falls, abrupt joint compressions or high rotary (twisting) forces on bones. Steps for Developing a Successful Exercise Program at Any Age:1. Determine the person’s capabilities by asking: “What can the child or adult do?”
2. Determine the objective you want to pursue by asking: “What is the child or adult trying to achieve?”
3. Determine the constraints or limitations to achieving the goal by asking: “Is limited range of motion, strength, alignment, or joint instability preventing successful performance?” These limitations may have to be addressed before the goal can be accomplished, perhaps by modifying the program itself.
4. Determine which equipment or treatments are available to help accomplish the goal. A wide range of devices exist to support improved function. Examples include bathroom safety equipment, walking aids, and devices for reaching objects in high or low places. A consultation with an occupational therapist may be necessary to help choose the best devices to accomplish a specific goal.

It’s NEVER Too Late To Begin: Adults and older children who do not have the exercise habit are encouraged to make a new commitment to a healthy lifestyle and become more physically active. Each person’s program should include exercises that will improve strength, balance, and endurance; be enjoyable; and promote socialization, if possible. Rehabilitation specialists or exercise specialists who are familiar with OI or osteoporosis can help design an appropriate program. Enjoyment plus improved function can be found through physical activity at every age**Pain Management in Osteogenesis Imperfecta:**For individuals with osteogenesis imperfecta (OI), the pain associated with multiple fractures can lead to needless suffering and, when untreated, may result in a chronic condition. Hip or back pain due to poor alignment is common in all types of OI, and chronic pain needs to be addressed. Since pain may impair a person’s ability to lead a productive life, chronic pain is a serious economic as well as a major health problem. Pain is the body’s way of responding to damaged tissue. When a bone breaks, nerves send pain messages through the spinal cord to the brain, where they are interpreted. How a person responds to pain is determined by many factors, including his or her emotional outlook. For example, depression seems to increase a person’s perception of pain and to decrease his or her ability to cope with it. Often, treating the depression treats the pain as well. If pain is not adequately treated, the transmission of pain impulses to the brain occurs more readily. Therefore, it is more effective to prevent pain than to treat it after it occurs. Acute pain is usually characterized by a short duration, normal functioning of both the peripheral and central nervous systems, a predictable course and, in most cases, a good outcome. Chronic pain is pain that lasts beyond the expected time for healing and interferes with normal life. The damaged tissues have healed, but the pain continues. The pain message may be triggered by muscle tension, stiffness, weakness, or spasms. Nonverbal responses to pain include elevated heart rate and blood pressure and immobilization of an injured part in order to avoid pain from movement. Whatever the cause of chronic pain, feelings of frustration, anger, and fear make the pain more intense. Chronic pain can often diminish the quality of a person’s life psychologically, socially, and physically. It is also the most frequent cause of suffering and disability in the world today. With OI, there can be associated conditions of acute and chronic pain resulting from multiple fractures, vertebral collapse, joint deformity, osteoarthritis, contractures, deformity/ malalignment of limbs, and recurrent abdominal pain. Pain management for OI, in both adults and children, requires adequate assessment and implementation of a regimen that should address the multifaceted presentation of acute and chronic pain. With the increased longevity of individuals with OI, the incidence of pain syndromes relating to degenerative changes becomes more likely. An interdisciplinary approach to pain management is best. The following information provides an overview of different options for pain management in OI. Individuals managing chronic pain associated with OI may wish to discuss the options listed below with a physician. Coping Strategies:*Physical Methods of Pain Management* 1. Heat and Ice: Heat in the form of warm showers or hot packs can relieve chronic pain or stiff muscles. Cold packs or ice packs provide pain relief by numbing the pain-sensing nerves in the affected area. Cold may also prevent swelling and inflammation. Heat or ice should be applied for 15 to 20 minutes at a time to the painful area, and a towel should be placed between the skin and the source of the cold or heat to protect the skin.
2. Warm towels or hot packs in the microwave provide a quick source of heat.
3. Frozen juice cans or bags of frozen vegetables make instant cold packs.
4. A damp towel that has been completely wrung out and frozen can provide relief from pain.
5. Freezing a plastic, resealable bag filled with water makes a good ice bag.

*Exercise or Physical Therapy:* Exercise or physical therapy, under the supervision of a health professional who understands the nature of OI, can be effective in strengthening muscles, improving stamina, and helping an individual have a more positive outlook on life. Because exercise raises the body’s level of endorphins (natural painkillers produced by the brain), pain can diminish. It is beneficial for children with OI to begin physical therapy as soon as possible. Physical therapists can teach proper positioning, posture, and exercises to strengthen muscles without injuring bones. Exercise can be as simple as moving a joint through its range of motion. Pool therapy is one of the best exercise techniques to gently improve muscle strength and reduce pain. Acupuncture and Acupressure: Acupuncture involves the use of special needles that are inserted into the body at specific points. These needles are believed to stimulate nerve endings and cause the brain to release endorphins. It may take several acupuncture sessions before the pain is relieved. Acupressure is direct pressure over trigger areas of pain. This technique can be self-administered after training with a certified instructor. Massage Therapy: Massage therapy can be a light, slow, circular motion with the fingertips or a deep and kneading motion that moves from the center of the body outward toward the fingers or toes. Massage relieves pain, relaxes stiff muscles, and smooths out muscle knots by increasing the blood supply to the affected area and warming it. The person doing the massage uses oils or powder so that his/her hands slide smoothly over the skin. Massage can also include gentle pressure over affected areas or hard pressure over trigger points in muscle knots. Special care must be taken when working on individuals with OI to avoid putting too much pressure on bones that might possibly fracture. *Psychological Methods of Pain Management*1. Relaxation training: Relaxation involves con-centration and slow, deep breathing to release tension from muscles and to relieve pain. Learning to relax takes a great deal of practice, but relaxation training can focus attention away from pain and release tension from all muscles.
2. Visual imagery or distraction:Imagery involves concentrating on mental pictures of pleasant scenes or events or mentally repeating positive sayings to reduce pain. Videotapes are also available to help visual imagery. Distraction techniques focus the person’s attention away from negative painful images to more positive thoughts. This technique may include watching television or a favorite movie, reading a book or listening to a book on tape, listening to music, or talking to a friend.

*Medications for Pain Management*To be most effective in alleviating pain, medications should be administered before pain appears and certainly before it worsens. Pain medications should be administered prior to painful procedures or before the performance of an activity that may be painful, such as deep breathing exercises after a spine fusion. As stated earlier, it is easier to prevent pain rather than to attempt to alleviate it. Pain is best prevented or treated by providing pain medication on a continuous or around-the-clock basis rather than as needed. Pain medications should be administered in an acceptable manner that is less uncomfortable than the pain itself. Ideally, they should be administered orally, intravenously, or under the tongue. Medications taken orally are generally the least expensive and most convenient alternative. Intramuscular injections are painful and may be a last resort. 1. Over-the-counter pain relievers: Aspirin, ibuprofen, naprosyn sodium, and acetaminophen can effectively relieve pain. While these medications are relatively safe, they can still cause side effects, such as stomach upset, and may result in complications from excessive dosing or prolonged administration. For this reason, these medications should be taken according to the doctor’s or manufacturer’s directions
2. Non-steroidal anti-inflammatory medications (NSAIDs): NSAIDs may be prescribed to treat mild to severe pain. These preparations block pain and treat inflammation. There are dozens of NSAIDs on the market, and a person may have to try several different brands to find the one that works best.
3. Topical Pain Relievers: A variety of topical creams may also relieve pain when they are rubbed directly into the painful area. While some of these creams are available by prescription only, others may be purchased over-the-counter.
4. Narcotic pain medication: Narcotics are powerful pain-relieving medications derived from opium or synthetic opium. Narcotics alter a person’s perception of pain and can also induce euphoria, mood changes, mental cloudiness, and deep sleep. These drugs may also cause nausea, lethargy, and constipation. People with OI must be especially careful when taking these medications. Narcotics can affect a person’s balance and increase the chance of falling. After repeated and prolonged use, some people may become dependent on or addicted to these medications, which are available only by prescription.
5. Antidepressant medications: People who suffer from chronic pain frequently suffer from chronic depression as well. Several studies using antidepressant medications have noted that these medications may not only improve depression but may also relieve or reduce the amount of pain a person feels. Additional research is needed to determine whether antidepressants can treat the chronic pain of OI and which antidepressants produce the best results.

**Nutrition in Osteogenesis Imperfecta (Brittle Bone Disease):** To promote bone development and optimal health, children with Osteogenesis Imperfecta (OI) should eat a balanced diet which is low salt and added sugar and contains a variety of vitamins and minerals. Some babies with OI display a weak sucking reflex and may require small, frequent feedings. Infants with severe forms of OI may have difficulty breathing and/or swallowing which will impair their ability to eat. The combination of small stature, feeding problems, and slow growth may be mistaken for failure to thrive. Burping an infant with OI should be done very gently, with soft taps, and possibly with padding over the hand, or by gently rubbing the baby’s back. Older infants and toddlers may require a food program designed by a clinical nutritionist or an oral motor therapist to help them make the transition to soft foods. http://archive.unu.edu/unupress/food2/UID06E/uid06e0x.gifHowever, there are some nutrition issues that are specific to children with OI. One of the aspects of OI is that children who are affected are unusually small. Severely affected patients may be short because of vertebral compression fractures, severe scoliosis, lower limb deformities, and disruption of growth plates.However, growth can also be delayed in the absence of these abnormalities. This can make it hard to assess for malnutrition in infants and children with OI. To assess for malnutrition in infants and children with OI, you should look for a slowing of linear growth Normal Linear GrowthslAbnormal/non-Linear GrowthIt’s also important that children with OI should avoid excessive weight gain, as this gain can increase pressure on load bearing bones, and cause fractures.Also, children with OI often have limited mobility, chronic pain and difficulty swallowing, which can lead to a low appetite. There are a few things that you can do to improve your child’s appetite and nutrition. It’s important to manage a child’s pain, this will make them more comfortable and relaxed. You can use medicine, such as paracetemol or ibuprofen, or non-medical techniques such as heat packs or distraction.If a child has difficulty swallowing, there are a few simple tricks you can use to help them swallow food safely without choking. Use thick fluids, like pureed or mashed meat/vegetables/fruit. The thicker the fluid, the easier it will be for the child to swallow. Also, if you take a spoon with a small amount of food on it, and press down on the infants tongue, this stimulates the swallowing reflex. Children with OI are also at a higher risk of constipation, which can be managed by providing the child with a high-fiber diet. Beans, brown rice, citrus fruits, apples, corn, sesame seeds, avocados, and whole meal/brown bread are all good sources of fiber. Its also important to drink plenty of water!Its very important that children with OI have a balanced diet. This includes protein (pork, chicken, beef, tofu, eggs), vitamins (found in most fruits and vegetables), minerals (found in green leafy vegetables such as spinach, mustard greens and morning glory) and dairy (milk, cheese, yoghurt). Foods that are high in calories are animal fats, vegetable oil, avocado, nuts and seeds, and processed meats. Vietnamese foods that good to give infants/toddlers with OI who are under/malnourished:1. Chau – made with meat, egg and vegetable oil. For added nutrition, you can chop up spinach, mustard greens or morning glory into very small pieces and add to the chau.
2. Mashed/pureed vegetables – pumpkin, carrot, potato, sweet potato, beans (black beans, pinto beans), green beans and peas, sweet corn, taro. These can be combined together in whatever combination you choose. You can also add pureed meat, for example beef and pumpkin puree.
3. Che/sweet soup – this dish is very high in calories, and children should only have small amounts
4. Mashed/pureed fruit – banana, avocado, apple, pear. Condensed milk can be added to these to add calories.
5. Always choose water or milk as a drink
6. Add yoghurt or cheese to the child’s diet
7. Hard boiled eggs
8. Soft mashed tofu/sweet tofu

You should give the child many small meals throughout the day, instead of three big meals. This will encourage the child to eat more, and is an easy way of increasing calories. For Example:Breakfast: Chau made with beef, eggs and spinach and using vegetable oil + bottle of milkMorning Tea: Yoghurt and/or mashed banana + waterLunch: Pureed beef and combined pumpkin, sweet potato, potato mash + waterAfternoon tea: Hard boiled egg + bottle of milkDinner: Soft mashed tofu with mashed green beans and peas + WaterDesert: Pureed apple *Weight Control:*Strategies for maintaining a healthy weight include: 1. Eat low-fat, high-nutrient foods,
2. Control portion size,
3. Exercise as much as safely possible,

*Calcium and Bone Health:* Calcium does not improve the basic collagen defects that cause OI. Even so, people with OI need to get adequate calcium in their diets to develop peak bone mass and prevent bone loss. Bone loss from any cause (calcium related, inactivity related, age related) makes OI bones even more fragile. In 1997, the National Academy of Sciences developed the following Recommended Dietary Intake for calcium: 1. Young children (1-3) 500 mg a day
2. Older children (4-8) 800 mg a day
3. Preteens/adolescents (9-18) 1,300 mg a day
4. Men and women (19-50) 1,000 mg a day
5. Men and women (50+) 1,200 mg a day

These guidelines were developed for people of average height and weight. A person with OI may have lower calcium needs. If you are unable to get the appropriate amount of calcium for your body size and age through your diet, a calcium supplement may be needed. Calcium supplements are also sometimes recommended for people taking certain medications such as bisphosphonates. In addition to dairy products, calcium is also found in foods such as broccoli, kale, some dried beans and nuts and soy-based products. Hypercalciuria is common in moderate and severe OI. Kidney stones affect only an estimated 20 percent of people with OI who have the condition. High-dosage calcium supplementation may contribute to kidney stones.*Vitamin D and Bone Health:*Vitamin D is necessary to help the body absorb calcium and make bone. New research suggests that it may play a role in the immune system, and that low levels may contribute to chronic pain. Most of the vitamin D in our bodies is made from sunlight absorbed through the skin. Vitamin D is also found in many fortified foods and in dietary supplements in the form called D-3. A blood test that measures 25(OH)D is the only way to tell if a person has adequate levels of Vitamin D in their system. Researchers recently began recommending that blood levels of vitamin D for children and adults on this test should be increased from a previous recommended level of 20 ng/ml to between 32 and 70 ng/ml. Studies suggest that many people have low vitamin D levels, especially in the winter. The amount of vitamin D in food or supplements is measured in terms of International Units (IU).The latest research supports the following guidelines for vitamin D for people with OI. See Note: Vitamin D Research in the Resources section for more information. Suggested Vitamin D-3 Intake for People with OI: Weight IU per day 1. 20 kg - 600-800
2. 40 kg - 1100-1600
3. 50 kg - 1200-2000
4. 70 kg and above 2000-2800

*Other Nutrients* Vitamin C has many functions in the body, including the production of healthy connective tissues, and the healing of wounds and fractures. Vitamin C is abundant in many fruits (such as citrus fruits, strawberries, and cantaloupe) and vegetables (including tomato, bell peppers, and sweet potato). It is fairly easy to get adequate Vitamin C through the diet. There is some evidence that Vitamin C tablets can increase the risk of kidney stones in people who already have high levels of calcium in the urine. Because 4 high urine calcium affects some people with OI, check with a physician before taking Vitamin C supplements. **Hearing Loss and Osteogenesis Imperfecta:**Significant hearing loss has been reported in approximately 50% of people with osteogenesis imperfecta (OI). While not everyone who has OI develops hearing loss, the incidence is much higher than in the general population. Sometimes visible deformities in the ossicles and inner ear can lead to hearing loss. Sometimes the cause of the loss is not visible. Environmental factors can contribute to hearing loss in a person with OI in the same way as for anyone else. These include loud noises, head injuries and infection. Hearing loss can start at any time but among people who have OI the loss often starts at an earlier age and the likelihood increases with age. Hearing loss can be treated with hearing aids and/or surgery depending on the type and severity of the loss.How Does Hearing Work? Our ability to hear depends on two factors—the mechanical translation of the sound waves into movement within the middle ear and then translation of that motion into a nerve transmission to the brain. In the first part, the sound waves make the tympanic membrane vibrate. That movement is transmitted through three small bones in the middle ear to the fluid in the inner ear. The movement of the fluid makes small hair cells move and that movement alters the signal along the hair cell nerve to the brain. Types of Hearing Loss There are two main types of hearing loss: conductive and sensorineural. Both types of hearing loss can occur with all types of OI. Hearing loss type can vary within a family just as much as between unrelated people. Sensorineural hearing loss tends to be more frequent as a person ages. Conductive loss generally occurs around age 20 or 30, but there are many exceptions. OI related hearing loss can occur at any age, including childhood.Conductive Hearing Loss: Usually results from a physical problem in the external or middle ear. It may occur as a result of ear infection, blockage of the middle ear, or from fixation or fracture of the stapes, one of three tiny bones in the middle ear that transmits the movement of the eardrum through the middle ear to the fluid in the inner ear. Sensorineural Hearing Loss: This type of hearing loss occurs when the inner ear is not transmitting the nerve signals normally to the brain. In addition, hearing losses are classified according to the degree of severity – mild, moderate, severe or profound. Hearing losses are also classified according to the sound frequency that can not be heard – low, high or all frequencies. Symptoms Some of the early signs of hearing loss are: 1. Difficulty understanding certain words or parts of words.
2. Frequently asking others to repeat themselves.
3. Difficulty understanding others on the telephone.
4. Turning up the sound on the television or radio to a level that is too loud for others in the room.
5. Difficulty hearing in noisy surroundings.

As it progresses, hearing loss can interfere with communication, performance at work or school, social activities, and personal relationships. Left untreated, hearing loss may even result in isolation and depression. Recommendations It is recommended that children with OI have a formal evaluation of hearing before they start school. Testing should begin at age 3-4 and be repeated every 3 years. Any 3 child with OI who demonstrates articulation problems, speech delays, recurrent ear infections, or whose parents suspect a hearing loss should have a formal audiologic assessment regardless of age. If borderline hearing is discovered, then yearly testing with a certified audiologist is recommended. Adults with borderline hearing should have yearly testing and follow up appointments similar to the schedule for children. Adults experiencing tinnitus (ringing in the ear) or symptoms of hearing loss should also have an audiologic assessment that includes a hearing test with air and bone conduction and speech reception threshold. This type of test can determine if the hearing loss is conductive or sensorineural. Adults who have an identified hearing loss should continue to see their audiologist or ENT on a regularly scheduled basis and whenever they feel their hearing has changed. Ear Protection Exposure to loud noises can lead to sensorineural hearing loss by damaging the hair cells (sound sensing cells) in the inner ear. The loudness and duration of the noise are the important factors determining damage, not the source of the noise. Power tools, chain saws, and loud music are but a few of the noises that can damage our hearing. Use of protection when working around loud noise or avoidance of the loud noise exposure can prevent this problem. Many different kinds of ear plugs and ear muffs are available to block out sound. The devices carry a rating of effectiveness on their packaging that tells how much sound is blocked out. When one must be around loud noise, preventive measures can help avoid permanent loss. Another important way to protect hearing is to keep the volume turned down whenever using a device that has personal headphones.Surgery:Surgical procedures such as stapedectomy or cochlear implant help some people with OI related hearing loss that is severe, conductive and progressively getting worse. In a stapedectomy, the fixed footplate of the stapes is replaced by a prosthesis, a copy of the bone, that allows the normal transmission of the bone movements to the inner ear. Using a laser to do the surgery improves accuracy and visibility during the surgery. Success of the surgery is highly dependent on the surgeon’s experience and on the person’s ear anatomy. The procedure itself takes less than an hour and the patient may be discharged from the hospital in less than 24 hours. Complications from stapedectomy surgery can include dizziness, change in taste to part of the tongue and worsening of the hearing loss rather than an improvement. Initially successful stapedectomies can fail for a number of reasons. There may be more problems in the ear than just with the stapes, the attachment of the prosthesis to the adjacent small bone called the incus may be faulty, the incus may be too thin to hold the prosthesis long term, or damage can occur to the inner ear leading to nerve loss or malfunction.A cochlear implant is an electronic device that can provide partial restoration of hearing that was lost because the very first part of the nerve transmission pathway is defective. A portion of the device is implanted behind the ear and electrodes are threaded into the cochlea inside the ear. This is an option for someone who has a sensorineural hearing loss. This surgery has successfully been performed on people with OI. Having a cochlear implant is significant ear surgery that may require a brief hospitalization. Complications can include dizziness and facial nerve injury. Since OI bone is not as solid or dense as bone in other people, the electrical signals from the device seem to travel a little more easily through the bone. This means that the facial nerve, which runs near the inner ear, may be stimulated from the implant. Stimulation of 5 the facial nerve can be corrected by turning off some of the electrodes. Hearing with a cochlear implant is variable. It requires a period of training and adjustment after the surgery. Hearing does not return to “normal,” but many people adapt .**Musculoskeletal Issues and Orthopaedic Management:** Scoliosis is a serious issue for some types of OI. The prevalence of spinal deformities among people with severe OI is high. Spine deformities increase with age. Rapid progression is common, and curves should be closely monitored. Bracing is of limited utility. It does not stop curve progression, and it may cause harm in more severely affected children. The use of customized braces for children with mild scoliosis and OI Type I is being studied. A rapidly progressing curve may require fusion surgery. Kyphosis (hunchback) affects many children with OI. Compression fractures of the spine occur in all types of OI. Families need instruction in how to avoid spine-jarring activities and how to protect the spine. *Orthopaedic management:* The goals of orthopaedic management include fracture care and the prevention or correction of bone deformities. 1. Bracing, splinting and orthotic supports
2. Intramedullary rodding with osteotomy is used to correct severe bowing of the long bones.
3. Plates and pins or screws are not recommended for the surgical repair of fractures in children with OI. These devices create a short, stiff segment with the bone. The bone is likely to break above or below the plate.
4. Immobilization due to surgery may increase osteopenia an dloss of muscle mass and functional strength. The recovery plan should include strategies for getting the child moving as soon as possible.

 *Rehabilitation, Physical Therapy, Occupational Therapy, and Physical Activity* Most children with OI benefit from physical activity programs. Treatment plans should promote and maintain optimal function. They should include early intervention, muscle strengthening, aerobic conditioning, and, when possible, protected ambulation. Infancy offers many opportunities to develop strength and avoid some of the deformities, such as torticollis, that are often seen in children with OI. Positioning is critical to avoid contracture and malformation. It is important not to leave a child with OI in a fixed position, either recumbent or sitting, for long periods. Immobilization reduces lean muscle mass and cardiovascular fitness, and it causes bone density to decline rapidly. Postfracture therapy is necessary to reduce the effects of immobilization on bone density and strength. The goal of physical therapy should be to improve function, fitness, and independent movement. Exercise should be prescribed based on the specific strengths and needs of each child. The focus should be on posture and stamina. Recreational activity can be a source of fun, socialization, and physical benefit to children with OI. Swimming and water therapy are highly recommended. Discrepancies in leg length can be caused by fractures or problems with growth plates. These discrepancies need to be evaluated to prevent the child from losing the ability to walk. **Infection in Osteogenesis Imperfecta:** Most people with OI can live long and healthy lives as long as they manage their condition appropriately. One important aspect of management in children with OI is infection control. Decreased chest volume and deformities may lead to restrictive pulmonary disorder in severe cases of OI. Pulmonary complications can occur due to rib fractures, kyphoscoliosis, muscle weakness of the chest wall, heart valve disorders, and chronic bronchitis or asthma. Lung collagen abnormalities have a negative impact on lung function. Respiratory complications, including pneumonia, are a significant cause of death among the OI population, especially among those with OI Type III. Supplemental oxygen, bilevel positive airway pressure, or bronchodilators for asthma may be indicated. Even when not caused by OI, lung problems can be more severe in people who have OI of any type. People with OI of any type have higher rates of asthma and pneumonia than people who do not have OI. It is important that all children with OI be seen by a doctor and treated as soon as they start displaying symptoms of a respiratory infection. These include:1. Fever
2. Cough – moist or dry
3. Runny nose
4. Headache

Children with OI are also more susceptible to ear infections such as otitis media. Ear infections can cause hearing loss in children with OI, who are already more susceptible to this condition. It is important that ear infections are also treated aggressively in children with OI. If a child displays any symptoms of ear infections, such as fever, pain in the ear or discharge from the ear they should be seen as soon as possible. **Medication in Osteogenesis Imperfecta:** *Bisphosphonates:*Bisphosphonates are analogous to compounds that naturally occur in the human body, but are metabolized in a different manner. There are a number of versions of these compounds, each having slightly different characteristics including mode of administration and potency. There are two main cell groups in the bone. The osteoblasts, which make bone, and the osteoclasts that break down the bone (bone resorption). Both types of cells are very active, even in the adult, and work together to keep the bone intact and responding to stress. Current knowledge suggests that bisphosphonates slow down the process of bone resorption by shortening the life of the osteoclasts and prolonging the life of the osteoblasts, thus tilting the balance towards the production of bone. The prolonged osteoblast still produces mutant collagen. Thus, the patient is still making "OI bone," but resorbing less of it. Early treatment studies have reported an increase in bone density, an increase in cortical bone width, a decrease in cortical bone porosity, and areduction in bone pain. It is still controversial as to whether this also results in a decrease in the number of fractures. Some animal studies of bisphosphonate treatment have shown that the femurs were less elastic, which would not be desirable in people with OI. It is also not clear at this time whether the bisphosphonates will have the same effect on the bone of the spine, which is mainly trabecular, and the bone of long bone, which is mainly cortical. Bisphosphonates are not metabolized in the body. Typically, treatment is repeated every 3-4 months for intravenous bisphosphonates, and weekly for oral bisphosphonates. Side effects could include a flu-like syndrome, decreased blood cells that return to normal values in 48-72 hours, gastric discomfort or severe burning of the esophagus, muscle pain, eye irritation and headaches. To date, there is no evidence that any of the bisphosphonates encourage growth, but neither do they seem to inhibit normal growth in children. Loose joints and tendon problems are not affected by bisphosphonate treatment. Some of the literature suggests that this therapy can be useful for the management of scoliosis in people who have OI. There is also relief of pain.  |

**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:** 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.

**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 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.

***This information has been adapted from the Osteogenesis Imperfecta Foundation*** [***www.oif.org***](http://www.oif.org)***.***