

Physical and Occupational Therapists *Guide to Treating Osteogenesis Imperfecta*



Key Principles and Therapeutic Strategies *for Infants, Children, Teenagers, and Adults Living with OI*

Safe Handling ▪ Adaptive Equipment ▪ Functional Assessments
Sport and Exercise Considerations ▪ Problem Solving for Self-Care Tasks

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What is Osteogenesis Imperfecta (OI)?

Osteogenesis imperfecta (OI) is a rare, complicated and variable disorder. Its major feature is a fragile skeleton, but many other body systems are also affected. OI is caused by a mutation (change) in a gene that affects bone formation, bone strength and the structure of other tissues. OI can be inherited from a person's parents in an autosomal dominant manner or occur via a new mutation in a gene that affects bone formation, bone strength, and the structure of body tissues containing collagen. OI occurs equally among males and females and in all racial and ethnic groups. It is estimated that approximately 25,000 to 50,000 people in the U.S have OI. The incidence of OI is approximately 1 in 15-20,000 live births.

People with OI experience broken bones from infancy through puberty. The frequency typically decreases in the young adult years but may increase again later in life. Respiratory problems including asthma are often seen. Short stature, rib cage deformities and spine curves make breathing problems more severe.



Other common medical characteristics and issues include:

- Bone deformity, and bone pain
- Hearing loss (present in more than 50% of people with OI)
- Brittle teeth (dentinogenesis imperfecta or DI) are seen in 50% of people who have OI
- Vision problems including myopia and risk for retinal detachment
- Loose joints, ligament laxity, and muscle weakness are common
- Cardiac issues
- Basilar Invagination (seen in some people with more severe forms of OI)

OI exhibits wide variation in appearance and severity. Severity is described as mild, moderate, or severe. The most severe forms may lead to early death. Clinical features (observable signs) vary widely not only between types, but within types, and even within the same family. Some features are age dependent. Children with milder OI, in particular, may have few obvious clinical features. Since the 1970's a list of numbered types has been used to describe the different forms of OI.

Major Types of OI

Below are some of the distinguishing features of the major types of OI.

Type I (Mild)

- Most common and mildest type of OI; few obvious clinical signs
- Typical or near-normal height versus age-matched peers and unaffected family members

Type II (Most Severe)

- Infants may die within weeks from respiratory or heart complications
- Numerous fractures and severe bone deformity are evident at birth
- Small stature with underdeveloped lungs, and low birth weight

Type III (Severe)

- Progressive bone deformity is often seen in long bones
- Fractures are present at birth, and x-rays may reveal healed fractures that occurred before birth
- Short stature
- Barrel-shaped rib cage
- Spinal curvature and compression fractures of vertebrae
- Triangular faces

Type IV (Moderate)

- Between Type I and Type III in severity and height
- Mild to moderate bone deformity
- Spinal curvature and compression fracture of vertebrae
- Barrel-shaped rib cage

Type V (Moderate)

- Similar to Type IV in appearance and symptoms of OI
- Large hypertrophic calluses form at fracture or surgical procedure sites
- Calcification of the membrane between the radius and ulna restricts forearm rotation

Type VI (Moderate)

- Extremely rare; similar to Type IV in appearance
- Distinguished by a characteristic mineralization defect seen in biopsied bone

Type VII (Severe)

- Recessive inheritance
 - Phenotype is moderate to severe
 - Rhizomelia (disproportional usually shorter proximal bone)

Type VIII (Very Severe)

- Similar to Type II but with recessive inheritance
- Severe growth deficiency and under mineralization of the skeleton

For a detailed list of OI Types including clinical signs, degree of severity and mutation, please see the OI Foundation's Medical Education website www.oif.org/meded.



The Role of Physical and Occupational Therapy in Managing OI

When working with individuals and families living with OI, therapists should keep these principles in mind:

- **Listen.** It is essential to listen to individuals with OI and their families and respect their input. Individuals and families are truly the experts on how OI 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. They have excellent solutions that you can share with other clients with OI.
- **Set Goals.** Make goals incremental, realistic and achievable. A successful plan of care includes the individual with OI and the family's personal goals.
- **Weakness** is a significant constraint to movement in OI. It is critical to remember that people with OI do not have impaired coordination, sensation or cognition and do not require the complex, neurologically based interventions used for people who do.
- **Fear of Fractures** is another serious constraint to movement. Establishing safe movement procedures, encouraging self-confidence and optimizing strength are strategies that can help resolve this issue. Passive range of motion is not recommended with new clients.
- **Expect Success.** With the proper environment and equipment, the majority of children and adults with OI can function well in many or most areas of daily life including but not limited to self-care, school and work.

The long-term goal for people with OI is independence or interdependence in all life functions with adaptive devices as needed at home, at school, in the workplace and in the community. In the case of very severely affected people, the goal becomes the ability to direct their own care.

Physical and occupational therapy are part of an interdisciplinary approach to treatment. The medical team may also include a primary care physician, orthopedist, geneticist, nutritionist, social worker, and psychologist. Children and adults with OI, especially those with spine curves which may affect pulmonary status, may regularly see a pulmonologist. Ideally planning ahead for rehabilitation is included in the preparation for surgery.

Maximizing a person's strength and function not only improves overall health and well-being, but also improves bone health, as mechanical stresses and muscle tension on bone help increase bone density. For example, deformities such as a flattened skull, a lordotic back, or tight hip flexor muscles can be prevented or minimized through therapy.

Approaches include:

- Exercise and recreational activities including weight bearing activities (braces may be needed), and low-impact activities such as swimming, once precautions are defined.
- Safe handling and encouraging different body positions and postures during the day to strengthen muscle groups and prevent deformities.
- Adaptive equipment. The individual with OI may need a variety of mobility aides depending on the environment (cane, walker, manual or power wheelchair).
- Environmental adaptations to the home, school or workplace.

Circumstances requiring intermittent or long-term physical and occupational therapy will include the following:

When a child with OI has delays or weakness in motor skills

Because of fractures, immobilization due to fracture, 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 and participation in peer and family centered activities. Large and small muscle groups may be affected. Therapy to promote achievement of developmental milestones should begin as soon as it is evident that an infant has muscle weakness or motor skill delay when compared with same age peers, and continue until a child reaches appropriate child/family centered therapy goals. The therapist often needs to address how muscle imbalances from bowing and/or muscle weakness affects forces across bones, postural alignment and movement. In some cases, an infant or young child may have delays, but after gaining sufficient strength, will be able to sit, stand, and walk. In other cases, certain motor skills may be unattainable due to weakness or skeletal deformities. For example, walking is not possible for some people with OI. When this is the case, the therapist works with the person to maximize function by developing other skills, and using adaptive equipment, energy conservation, and joint protection concepts. Fine motor skills can be delayed or diminished and make handwriting, typing, and using hand tools difficult.

When a child or adult with OI is recovering from a fracture, surgery or injury

Because fractures and surgery are frequent for many people with OI, it is particularly important for them to regain as much function as quickly as possible during recovery. It is beneficial to maintain strength in limbs that are not affected by the fracture or surgery. The therapist can work with the individual with OI during periods of fracture, surgery or injury to minimize the detrimental effects of immobilization such as decreased muscle mass, weakness, fear of movement or learned helplessness. Minimize immobilization time with physician approval. After recovery, additional intensive rehabilitation is often needed to assist relearning previously mastered skills, or to regain strength in the affected limb(s). Repetitive movement injuries are common among adults who have OI who use

mobility aids. Hands, wrists, shoulders and knees are often affected. The therapist can help the client develop alternate strategies, and learn to use appropriate equipment such as sliding boards to make self-care skills easier.

When a person with OI experiences fear of movement and trying new skills and activities.

In some cases, the biggest obstacle to independent function is fear. Children and adults who have had fractures may become fearful of moving or trying new things. Parental fears and concerns can complicate the situation. 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. Therapists must acknowledge these fears as understandable, but also suggest ways that new skills can be practiced in a safe environment. A successful strategy is breaking skills down into small, achievable steps. This allows the individual to succeed at something relatively easy and progress step by step until the skill is mastered. Encouraging active movement within a self-care activity can be both a strengthening exercise as well as empowering to the individual by engendering an "I can do it" attitude. Additional approaches include positioning, active movement, water therapy and the use of protective equipment (e.g., clamshell style ankle foot orthosis, or forearm supports on walkers). Encouraging the child to direct some of his or her own care, transfers, and handling will also build the child's confidence. This can start with family members, friends and teachers.

When a person with OI reaches a transition point in life, they must adapt to a new environment or require ADL training.

Many key activities of daily living—such as toileting, dressing, bathing, grooming, doing laundry 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 deformity, children and adults with OI often have to relearn how to do a task in an entirely different manner. Changes in the person's environment – attending day care, changing schools, moving out of the parental home, or new employment – may necessitate learning a new skill or improving stamina. Through a combination of strengthening activities, use of adaptive equipment, and creative problem solving, many obstacles to independent self-care can be overcome.

Key Principles of Therapeutic Strategies for Osteogenesis Imperfecta

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. Key therapeutic strategies include the following.

1. Skill Progression - 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. Adults may need to relearn a series of skills after a serious injury.
2. Protective handling, preventive positioning, and active movement with gradual progression contribute to safe development of motor skills.
3. Water therapy provides the opportunity for children with OI to develop skills in a reduced gravity environment before trying them on land. Adults often use water therapy to relearn or maintain motor skills. Water is a great place to start getting past the fear of movement.
4. 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.
5. Encouraging healthy living is an important part of the therapeutic relationship. Promoting general health, preventing obesity, and encouraging participation in recreational activities are important elements of achieving the goal of a lifestyle of wellness and greater independence.



Skills Progression

Meeting developmental milestones is challenging for many children who have OI. Some will not be met, but they can be compensated for by building related skills. For example, a particular child might not be able to crawl, but may develop other methods of floor mobility such as snaking or bottom scooting. A skate or scooter board can be used to eliminate the pull of gravity and encourage mobility. Introduce new positions and skills **gradually** to allow the person to feel safe as well as to promote gradual strengthening of muscles and bones. Provide adequate support to overcome weakness and prevent injury. See the **Developmental Progression Chart**

for **Children with OI** for more details. Older children and adults will need a similar progression of skills to learn or relearn activities of daily living.

Developmental Progression for Children with OI (Chart)

Progression	Possible Modifications Needed for People with OI
Supine Positioning	Provide frequent position changes (side lying) to prevent flat skull and arm/leg contractures. Keep arms/legs positioned with proper alignment to head/trunk to prevent tightness across the shoulders/elbows/hip/knee/ankle regions. If child cannot get hands to midline, use trough-shaped foam bed pad or small foam wedges to guide shoulders forward.
Side Lying	May need support under head and below axilla. Excellent position for infants; promotes hands to midline and relieves flattening of skull.
Prone Positioning	Use a small roll under the armpits to facilitate baby lifting his/her head; practice on parent's chest first. Be aware that many babies with OI due to barrel chest dislike prone position. However, even short periods in prone (2-5 minutes) are therapeutic.
Inclined Sitting	Blanket rolls along torso for support can also serve as arm rests. Position the child to decrease wide hip abduction and external rotation. Provide place for feet to rest flat. Use very wide straps or vest for trunk support.
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 try 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 infant car seat or positioning chair as with inclined sitting (above). Provide head and neck support at first. Slowly decrease support as head turning gets better and slowly decrease recline. Proper head, trunk, and pelvic alignment are encouraged. Firm infant seats, car seats, and rockers are recommended to provide a stable base of support and avoid a flexed spine posture.

Unsupported Sitting	<p>Start with ring sitting. Use corner sitting or nursing pillow initially. Once head and trunk control present, work toward sitting on a chair or bench with hips, knees, ankles at 90 degrees or short sitting (with close supervision). Due to bowing, arm length can be shortened in relation to the trunk; therefore, an infant or child's protective reactions of the arms to stop a fall when sitting can be inefficient. Use nursing pillows around infant or corner sitting for elevated support to the arms.</p>
Achieving Sitting	<p>Start in the pool with child beside the parent and side-leaning on the parent's thigh; then try to get to a sit. On land, side-lie on a wedge or pillow or parent/therapist thigh and forearm (not extended wrist). For adults, work on abdomen strength to use a sit-up method.</p>
Sit-pivot, sit-scoot	<p>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 mat. This position will be a transfer method from chair to bed to toilet during fractures and if legs are not strong enough to stand.</p>
Crawling	<p>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 laxity present and legs abduct, use "mermaid suit" of stretchy 6- to 8-inch wide tubigrip, old panty hose top or wide stockinet from child's waist to ankles. Consider a modified skate or scooter board with necessary padding and safety strap/support.</p>
Kneeling/Pulling to Stand	<p>High kneel for trunk development. In the pool or on land, 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.</p>

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.

Protective Handling

There are some basic principles of safe handling that are important to follow any time 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 trained in safe handling to prevent injury.

Safe Handling of Infants and Young Children with OI



- Ask the parents of infants with OI to demonstrate the safe handling techniques they have developed. It is often preferable to have the parent or caregiver do the handling at the beginning of a therapy relationship. Gradually over time, move into the therapist demonstrating specific techniques.
- Never pull, push, or twist a limb. Be very cautious with any passive rotation of the arms, legs, head or trunk.
- Lift an infant with OI with the widest base possible. Scoop the child up by placing one open hand underneath 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.
- Do not lift the buttocks by pulling on the ankles, especially during diapering. Consider scooping or side lying as an option.
- Use loose fitting clothing.
- Be aware of where the child's arms and legs are at all times to avoid awkward positions or getting a hand or foot caught in clothing or equipment.
- As soon as possible, encourage the family to let the child with OI assist with any of the positioning activities.

Safe Handling of Older Children and Adults with OI

- Explain each new movement. Before handling the person or moving a limb, state what you are going to do and how you are going to do it. Ask them how they normally perform an action. If they ask you to stop, stop! For young children, using a floppy doll to demonstrate a motion and to problem solve with the parent will make new transitions easier.
- Construct the environment to help the client achieve the task rather than using external caregiver hands-on assistance. Encourage clients to engage in analyzing tasks.

- Provide adequate support for the child or adult new to the standing position to minimize buckling and/or risk of fracture of the legs.
- Avoid positions and motions of great leverage that stress curved or bowed bones such as the pelvis, femur, tibia or spine. Examples of these positions include the following:
 - The “jack-knife” position when the person leans far forward while sitting stresses the hip.
 - The “bridging” exercise (lifting the buttocks with knees flexed while lying on the back) which stresses the tibia in the same plane in which they tend to bow.
 - “Straddle sitting” over an adult’s thigh. This stresses the femur at the same plane in which they tend to bow and can also lead to imbalances in the muscles of the hip.
- Avoid diagonal trunk rotation which stresses the spinal vertebrae and the ribs.

Preventive Positioning

A key method for helping a person with OI maximize strength and function is to encourage them 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 deformities that can limit mobility and increase pain. It is important to keep the hips and spine as aligned as possible, limit amount of time in supine to prevent flattening of the back of the head, and promote active head turning in both directions.

In many cases, everyday objects can be used to make different positions easier and safer. For example, towel rolls, swim noodles and padding can be used to encourage upright posture and avoid “frog-leg” positioning in a high chair, wheelchair, car seat, or stroller.

The need to frequently change positions does not end in childhood, and should be included in instruction provided to students and adults.

Active Movement

Therapists begin by assessing the person’s current functional abilities. The goal in therapy will be to gain the next level or improve within the lying, sitting, floor mobility and walking levels. Thus, the goal for a very severely affected person might be to sustain a supported and/or reclined sitting position to access a computer, table, or phone. For a severely affected person, learning to sit-scoot might enhance his or her self-care skills. More moderately affected people may gain walking skills, with or without braces or aids.



Very mildly affected people may function at the same level as their peers, with occasional modifications or limitations (such as no high-impact activities). Activity analysis helps to determine small increments which enhance progress. In some cases, young adults who have been wheelchair users decide to re-establish their walking abilities. Older adults may need therapy to learn how to effectively use walkers or canes after years of unassisted mobility. A series of videos developed at the Hospital for Special Surgery in New York City can help adults who use a wheelchair or who are more comfortable seated to begin a fitness program. The series titled “Wheelchair Based Exercises for People with Osteogenesis Imperfecta” can be seen on the hospital website (www.hss.edu) under information about osteogenesis imperfecta.

Water Therapy



The water provides an ideal environment for people with OI to practice water-supported limb and body movements and learn new skills. Water not only cushions bones and joints but its buoyancy protects the person from falls. Water also provides gentle resistance along the entire length of bones. This resistance helps strengthen bones and muscles, and also helps prevent fractures that can be caused when too much pressure is applied to an isolated area. Water activities can be used to improve cardiovascular function. Swimming and other water exercise often become favorite fitness activities for older children and adults with OI. Practicing the

developmental progressions listed previously in the water can help make transitions from one position to another easier, and also ease fears. Once the skills are mastered in the water, they can be tried on land. Examples of water therapy that can promote new skills or aid the post-surgery rehabilitation process include:

- Encourage the person to practice “shimmy-sitting” or scooting by sitting on steps in the water, and scooting from side-to-side or up and down the steps.
- Encourage 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 to 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 minimization of fracture risk, mobility, accessibility and promotion of independence.

Reduction of Fracture Risk

To help a person function safely and most efficiently, evaluate what tools and environmental adaptations might be needed so the client can accomplish common tasks without excessive strain or fatigue. Establishing work stations (such as a homework station, toothbrush station, hair-drying station, meal preparation 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 will minimize the effort needed to dress and undress.

Mobility

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

Accessibility

Physical environments (at home, school, or work) can be modified to allow maximum independence. While extensive structural changes are sometimes called for (such as building ramps or lowering kitchen and bathroom surfaces), some accessibility problems can be addressed with creative use of assistive devices, rearrangement of furniture and other equipment, and thoughtful consideration. An important by-product 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 chart "Commonly Used Adaptive Equipment" identifies many types of equipment and some factors to be considered when choosing appropriate equipment. Note that this list is only a starting point. More specific ideas and recommendations can be found in the section on self-care tasks.

Commonly Used Adaptive Equipment (chart)

Type of Equipment Common Considerations for People with OI

Walkers	Supported walking allows weight bearing in legs, which increases strength and bone density. Sufficient upper-body strength is needed to grasp/move walker. Posterior walkers may be useful for encouraging upright posture. Anterior walkers are used sometimes with very young or small children first starting to walk. Some people with OI report feeling more secure with an anterior walker. Walkers with seats may be indicated to encourage use of walker for longer periods as seat allows for short rest periods. Baskets attached to walker can help with independence. Platform attachments may be required for those with upper body deformities. Finding the appropriate size walker that has wheels, and hand brakes for a short stature adult is difficult and may require customizing a child sized walker.
Crutches and canes	Supported walking allows weight bearing in legs, which increases strength and bone density. More upper-body strength, balance and confidence are needed to use crutches as compared to a walker. 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. Both forearm crutches and standard crutches are used. Progressing to crutches or canes allows easier access to stairs, and narrow spaces. They can be attached to wheelchairs to encourage individuals to combine wheelchair use and walking.

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. Consider lightweight manual chairs which are easy to propel and turn. Power assist wheels as an add-on to manual chairs are a useful alternative to full power chairs. Power chairs allow people with frequent 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 severe OI. However power wheelchairs are more difficult to transport. For all mobility aids, consider the bigger picture for growth, development, transportation, school needs and recreation as the chair is generally “covered” by insurance every 5 years.



Other mobility aids

There are many options in this category including scooter boards, small floor level wheelchairs such as the Zipzac, riding toys, tricycles, etc. Tricycles or bikes with four wheels are best to prevent tipping/falls. Go Baby Go from the University of Delaware designs modified ride-on cars for children. Seat belts/safety harnesses are necessary. Seats wide enough to offer pressure to be dispersed 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

Braces should be used only if necessary to support weak muscles and should be lightweight. Long leg braces may be indicated post rodding surgery for children with inadequate quadriceps strength and help with alignment and promote standing and walking. They are used only as long as needed. Ankle foot or tibia orthosis (AFO's) with anterior portion (clamshell) are used when the gastrocnemius and soleus (calf muscle) is weak and unable to properly eccentrically control the anterior translation of the tibia over the planted foot on the ground while walking. The clamshell feature of the HAFO's with anterior shell can provide the client with an added protection from external forces that may strike their legs. Supramalleolar or ankle braces (SMO's), or shoe inserts are indicated if feet are pronating from laxity and cause poor alignment that could lead to deformity, pain or decreased walking endurance. 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. For example, lightweight forearm splints can be used to stabilize healing fractures. They may permit greater activity when worn during water therapy while a fracture is healing. Remember braces must fit perfectly.

Positioning aids

Pillows, bolsters, towel rolls, swim noodles, gel pads, etc. Promote 90/90/90 (+/- 10 degrees as some slight recline or precline may be necessary) position in car seat, high chair, wheelchair, stroller, etc., rather than "frog leg" position.

Standers

Standers are used to promote gradual 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. Often used post rodding to introduce standing. Tray attachments can allow a child to use stander while coloring, doing homework, games, working on computer, etc. For very small children consider a "garbage can" or "bucket" stander for initial standing with MD permission. This can be accomplished with a new 5 gallon bucket or garbage can filled with blankets or pillows to provide support and padding.

Infant/child car seats

Infant and child car seats must be used according to the height and weight guidelines. Infants with severe OI under 10 pounds in weight may benefit from an approved car bed. Look for breathable fabrics for padding and seat covers, as children with OI tend to overheat easily. Use approved positioners to keep the infant's head in mid-line. Many families affix a noticeable tag or sticker to the seat indicating the OI diagnosis, in case of a traffic accident. Special car seats which can accommodate casts are available for loan or purchase.



Self-care aids

This group includes transfer boards, bath chairs/benches, personal hygiene systems, grab bars, and reachers. Self-care aids can help overcome limitations due to weakness, short stature, or wheelchair use.

Remember a piece of adaptive equipment may mean the difference in achieving greater independence.

Encourage Healthy Living

People with OI and other physical disabilities must cope not only with the effects of their disorder but also with common illnesses, risks for cancer and diabetes, and the effects of aging. Research indicates that physical activity is an important part of a healthy lifestyle. Obesity is a

serious problem for people with OI. It limits movement, strains bones, adds to fatigue, and in older children and adults often causes loss of independent mobility. Therapists can facilitate the transfer of skills from therapy to recreational activities. Assistance may also be needed to transfer from the clinic to community or home based gyms or pools.

Strategies for Evaluation and Functional Assessment of Teens and Adults with OI

Exercise and fitness is as important for people with OI as it is for the general population. Recommendations regarding exercise will vary depending on the individual's medical diagnosis, fracture history, use of bisphosphonates, surgical intervention and bracing. Healthy forms of exercise are not only acceptable, but highly recommended by the American Heart Association and American Medical Association. Preparing a treatment plan for the teen or adult who has OI includes an assessment of their individual strengths, weaknesses, interests, goals, the physical characteristics that are specific to OI, as well as the age related characteristics shared with the general public.

Common characteristics specific to adults with OI include:

- Bone fragility- although fractures occur less often in adulthood, osteoporosis due to aging imposes an additional fracture risk
- Joint contractures due to increasing sedentary lifestyle or infrequent position changes
- Joint hypermobility and stress across the joints occur with weight gain and deconditioning
- Decreased general mobility (decreased ambulation/transfer)
- Decreased endurance and aerobic capacity
- Hearing loss and retinal detachment

The World Health Organization-International Classification of Function, Disability, and Health (WHO-ICF) format is particularly useful when evaluating an infant, child, or adult who has OI. The ICF model places an emphasis on health and function and assists with goal setting. The PT should be aware that although OI is a disorder of bone, besides the musculoskeletal system, OI can also affect the cardiopulmonary, neurological, visual, and auditory systems. Assessment of body structure and function impairments requires a multisystem approach.

Assessment Basics

- Obtain background information: this gives the PT valuable information regarding bone integrity, fragility, and the ease at which fractures occur or not. The PT may have to reach out to the doctors to gather this information.
 - Type of OI
 - Fracture history including date of last fracture
 - Bone density
 - Evidence of scoliosis and/or other spine curves
 - Radiographs/MRI
 - Surgical history including rodding.
 - Evidence of basilar impression
 - History of bisphosphonate or other bone medications
 - GI issues
 - Common co-morbidities seen in adults with OI
 - Hearing loss
 - Retinal detachment
 - Cardiac valve involvement
 - Obesity
- List all medications. This may include asthma related meds; bone building medications, vitamins and supplements (calcium or vitamin D).
- Obtain a social history. Include level of independence, community activities, employment, sports or hobbies and mobility.
- Select outcome measures that reflect improvements over time and use of assistive devices.
- Help adult with OI achieve personal goals.

Musculoskeletal Assessment

- Skeletal Alignment: Bowing of the upper extremities can affect overall reach, and bowing of lower extremities can affect how bone accepts weight bearing stresses. A shoe lift of correct height can reduce stress on the lower back for those with a leg length discrepancy.
- Muscle Tone
- Ligamentous Laxity: is common and can indicate joint instability.
- Range of Motion: Assessing active and active-assisted ROM is recommended as it allows the movement to be under the direction of the person with OI and decrease fracture risk. **Passive range of motion is not recommended due to fracture risk.**

- **Strength:** Strength testing to assess a muscle's strength against gravity, specifically hip extensors/abductors, quadriceps/hamstring, and gastrocnemius/anterior tibialis. Strength testing against light resistance in a functional context is also recommended. This allows strength of groups of muscles to be assessed such as a press up, leg press, step up or step down.
- **Muscle Imbalances:** PT's need to assess for muscle imbalances such as hip flexor/abductor/external rotator muscle dominance which can affect muscle flexibility, bowing, postural misalignments and increased stress across long bones. Some muscle imbalances may functionally benefit the patient. For example a tight gastrocnemius that only allows 5 degrees of active dorsiflexion may offer some stability in standing.

Other Assessments

- **Cardiopulmonary:** Monitoring of heart rate, blood pressure, Borg's Scale of Perceived exertion, and O₂ Saturation, during endurance and functional activities are recommended. Referral to the primary physician or specialist is recommended if there are atypical symptoms noted during the assessment.
- **Activity Limitations:** The PT assesses any activity limitations such as issues with transfers, balance, locomotion, and mobility.
- **Participation Restrictions:** The PT assesses for any participation restrictions such as issues with work.
- **Personal-fear-avoidance behavior** for physical activities, perceived ability to function pertaining to activities of daily living and work activities.
- **Environmental design** of home/school/work environment can be evaluated. Products, equipment and adaptive technology to support employment and activities of daily living can be suggested.

Nutrition and Body Mass Index

During the evaluation, stay conscious of the nutritional intake and the BMI of adults with OI as they age. Increased weight gain can add more stress to the joint, bones and muscles. Adults recovering from a fracture or muscle injury can be immobilized for a period of time and this cycle of immobilization can lead to weight gain and further compound the weakness and stress on the bone. Referral for a nutritional consult may be warranted. In your evaluation process of an adult who has lost function from an injury determine if they have gained a significant amount of weight after injury as this will impact your rehab process and you may have to help them reduce their BMI before you can get them into standing for weight bearing and weight bearing for their arms during transfer and scooting transfer.

Problem Solving for Specific Self-Care Tasks

Toileting, bathing/grooming, dressing, transferring skills and food preparation are five 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 would be to assist the individual to the next higher level of independence for the greatest freedom.

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. 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), stabilize a regular floor model potty seat in a large wooden base.
- 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. Alternatively adding a locking raised toilet seat may allow a person in a wheelchair to independently slide laterally or forward onto the toilet.
- A toilet-paper reacher (also referred to as a toilet aid) can extend the reach of the hand if a person’s arms are particularly short and unable to reach the privates. Alternatively, a hands-free personal hygiene system can be added to a regular toilet.
- A sliding transfer bench (commercial or customized) and/or grab bars located near the toilet will allow for easier same level transferring from wheelchair to toilet.

Bathing/Grooming. Bath time often provides a severely affected infant with some of his or her first experiences of independent movement 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 adaptations such as a sliding transfer bench into the tub, a shower seat, grab bars, and a hand-held shower head.

For grooming tasks, such as brushing teeth or styling hair, it is helpful to use the “work station” concept discussed previously. Long-handled brushes and special nail clippers are available.





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 onesies with snaps along both legs and the torso. For older children and adults with OI, simple, easy-on clothes are useful, such as pants with elastic waists. 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, to simplify clothing management at toileting or to accommodate a cast. For example, a seam can be cut, and hook-and-loop (Velcro) material sewn onto the seam so it can be opened and closed quickly and amply. Dressing tools, such as dressing sticks with hooks, reachers and sock donners, may be useful for some people.

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. A custom food preparation center can be made out of low tables, stacking cubes, or bookshelves. Commonly used items should be stored in low drawers or cabinets, in proximity to 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. 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.

Transferring Skills. Children from a young age need to learn how to move safely from their walkers or wheelchairs to bed, chair, toilet, tub or shower and car. It is best to first teach same level transfers to large surfaces (e.g. wheelchair to bed at same level) and progress gradually to more difficult transfers between surfaces at different heights or where a gap exists (e.g. moving from a walker or wheelchair to a car). Strong upper extremities and transfers boards are some of the strategies to ensure success.

Strategies: Exercise and Fitness for Teens and Adults with OI

The treatment plan for an adult with OI should include exercise and active recreation activities. The plan will be based not only on the functional assessment, health history and precautions, but also on the person's interests, preferences and goals.

Exercise Guidelines- "Less is More" when initiating an exercise program.

- Exercises should be performed in a slow and controlled manner with a focus on posture, form, and repetitions rather than increasing resistance.
- Use joint protection techniques with all movements and equipment with awareness of posture and body alignment. Light weights (i.e. weight of patient's shoe or braces or cuff weights) may be used positioned close to joints to shorten load and stress over long lever arm for joint protection.
- All movements should include trunk and core stabilization.
- Encourage proper alignment, posture, and body mechanics with all movements and exercises.
- Medical clearance should be acquired with all new exercise routines.
- Monitor tolerance to exercise with goal of a total of 60 minutes per day of activity. Teach the adult how to monitor his/her tolerance to activity for integration into community based daily exercise routines.
- Choose activities that the person enjoys and are meaningful.



Exercise Program Elements

- Aerobic Training can improve endurance during functional tasks. Some examples are aquatics/swimming, upper body ergometer, walking, or cycling (outdoor, recumbent, or stationary). When exercising in water, be conscious of the velocity of movement. Due to the degree of fragility of the patient, start with slow velocity and progress up from there but always be moving through the full available range of motion.
- Strength Training, including weight bearing activities on land and in water, improves bone density. Initially focus on form and repetitions rather than increasing resistance. Including dynamic strengthening using the person's own body weight as a source of resistance is a good way to begin functional training. If mobility requires transfers then target latissimus dorsi, triceps, and biceps muscles. Target deep abdominal and back extensor and gluteal muscles to support low back during mobility activities such as transfers, scooting, and ambulation.

- Flexibility Training is needed due to muscle imbalances, weakness, posture and movement impairments. The muscles to target are the upper back, pectoralis, low back, hip flexors/ external rotators, hamstring, and heel cord muscles. Other muscles may also be shortened and should be screened during PT examination. Positioning in prone and side lying are alternatives to typical seated or supine positioning.
- Emphasize trunk and core stabilization with all movements and exercises as well as proper alignment, posture and body mechanics while moving limbs, using equipment or performing functional training.

Specific Exercise & Sport Considerations for People with OI

- Safety is important: consider the risk level of the activity.
- Joint protection during exercise with focus on end range and mid-range muscle control due to ligamentous laxity, muscle imbalances in strength and flexibility.
- Despite the bony deformities that may be present analyze alignment, form, and muscle recruitment while performing exercises to minimize risk of injury. When using weights, consider how stress is transferred along the long bones and joints of an arm or leg that is bowed. Initially keeping the weight close to the joint may decrease the stress across the long lever arm of the bone and as the person demonstrates improved muscle strength and control during the movement, the weight can gradually be moved away from the joint as a progression of the strength training. Teach patients with OI how to stabilize the core with isometric muscle contraction of the deep abdominals and back extensors prior to any movement activity which will help stabilize the spine and reduce risk of injury. Use mirrors to encourage self-checking of alignment. PT's may recommend to the MD and patient the benefits of using braces or orthotics to support hypermobile joints and weak areas such as flat feet.
- Avoid exercises that involve excessive strain (jarring), jerking or heavy weight bearing, high impact or contact. Jogging and running activities may be appropriate if supported by the person's bone density, and MD.
- Fatigue-Muscles with low tone tend to fatigue faster and have a higher threshold for activation. Remember to allow for the muscle's latency of response to contract and allow the patient sufficient time to recover between sets.
- If an adult with OI wants to participate in a certain activity or sport, it is important for the PT to assess if the patients strength, flexibility, endurance, and movement patterns related to the activity or sport are sufficient to support safe participation. The standards and expectations of a sport must be considered. Participation in a sport may become a treatment goal.
 - Size and age should be considered when establishing benchmarks for performance.

- Using appropriately sized protective equipment is recommended. Customizing equipment to ensure proper fit and protection over bowed limbs will encourage success in the activity and minimize injury.

The Hospital for Special Surgery, New York, NY, developed a series of videos titled “Wheelchair Based Exercises for People with Osteogenesis Imperfecta.” They demonstrate a group of exercises that are based on the principles in this booklet and are appropriate for anyone beginning to exercise from a seated position. The videos can be seen on the hospital website www.hss.edu. Specific information is provided in the Reference Section.

Summary: Strategies for Physical and Occupational Therapy

- It is essential to listen to the client—child or adult-- and parents.
- The overall goal is independent function and integration into community.
- Make therapy goals incremental and achievable.
- Do not bend, twist, or pull a limb. **No passive range of motion** until the therapist and client know each other very well. Coach caregivers and adults to be proactive and warn healthcare and other providers about this.
- Issues to address: weakness, fatigue, poor cardio-pulmonary status, loose joints, flat feet, short stature, limb deformity, and obesity.
- Never force a person to do something they have been injured by in the past.
- Always begin by asking the caregiver or client how they perform a task/movement; have them demonstrate.

References

The Osteogenesis Imperfecta Foundation

The Osteogenesis Imperfecta (OI) Foundation welcomes inquiries from physical and occupational therapists, families living with OI, and others who have questions about the information included in this booklet or other issues related to osteogenesis imperfecta. The OI Foundation can also connect physical and occupational therapists with professionals who have experience working with people who have OI.

This booklet and many additional resources for healthcare professionals, families and people with OI can be found on the OI Foundation website.

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Books

Bleakney DA and Donohoe M. 2000. Osteogenesis Imperfecta. In Campbell, Suzann K., Darl W. Vander Linden, and Robert J. Palisano (eds.). *Physical Therapy for Children*. Second Edition. Philadelphia: W.B. Saunders. Pp. 320–338.

Chiasson R, Munns C, Zeitlin L, editors. *Interdisciplinary Treatment Approach for Children with Osteogenesis Imperfecta*. Montreal, QC, Canada: Shriners Hospital for Children (Canada); 2001.

Engelbert RH, Van Brussel M, Rameckers E. 2014. Chapter 51: Functional Outcome Measures in Children with Osteogenesis Imperfecta. In Shapiro JR, Byers PH, Glorieux FH, Sponseller PD, editors. *Osteogenesis*

Imperfecta: A Translational Approach to Brittle Bone Disease. Boston: Elsevier. Pp. 473-483.

Smith PA, Rauch F, Harris, GF. 2015. *Transitional Care in Osteogenesis Imperfecta: Advances in Biology, Technology, and Clinical Practice*. Chicago: Shriners Hospitals for Children – Chicago.

Trovato MK, Schultz SC, Joseph, C. 2014. Chapter 52: Rehabilitation for Adults with Osteogenesis Imperfecta. In Shapiro JR, Byers PH, Glorieux FH, Sponseller PD, editors. *Osteogenesis Imperfecta: A Translational Approach to Brittle Bone Disease*. Boston: Elsevier. Pp.485-491.

Booklets and Videos

Hospital for Special Surgery video series “Wheelchair Based Exercises for People with Osteogenesis Imperfecta,” can be viewed at www.hss.edu/conditions_wheelchair-based-exercises-osteogenesis-imperfecta.asp.

Shriners Hospitals for Children-Canada booklets

The following booklets are available by contacting the OI Foundation or they can be downloaded from the OI Foundation website.

- The Daily Care of Young Children with Osteogenesis Imperfecta
- Independence in Daily Life for Children with Osteogenesis Imperfecta
- Gross Motor Development of Infants with Osteogenesis Imperfecta
- Positioning, Seating and Mobility Devices for Children with Osteogenesis Imperfecta
- Exercise and Sports for Children with Osteogenesis Imperfecta

- Physiotherapy Rehabilitation for Children with Osteogenesis Imperfecta Following Femoral & Tibial Intramedullary Rodding

Equipment Sources

The following list is provided as a sample of the wide range of sources for assistive and adaptive equipment. Inclusion on this list does not imply an endorsement by the OI Foundation nor does exclusion suggest disapproval.

Abilities Expo

Annual exhibitions in different cities across the United States offer the opportunity to see the latest in adaptive equipment.

www.abilities.com

ActiveAid

Customized bathroom equipment

www.activeaid.com

Go Baby Go

Modified ride-on cars

sites.udel.edu/gobabygo

Hydro-Fit Aquatic Fitness Gear

Flotation vests and foam accessories for water therapy

www.hydrofit.com

Little People of America

Information on products to aid people with short stature

www.lpaonline.org

Maddak Inc.

ADL (activities of daily living) aids

www.maddak.com

North Coast Medical

ADL aids; splinting products

www.ncmedical.com

One Step Ahead

Child safety equipment; specialized clothing; toys

www.onestepahead.com

Rifton Equipment

Mobility aids; bathroom equipment

www.rifton.com

Wheelchairs

Panthera

Ultralight manual wheelchairs

www.panthera.se/en/produkt_x.html

Permobil

Power wheelchairs with seat elevator and tilt features

www.permobilus.com

Manual Wheelchairs

www.permobilus.com/products/manual-wheelchairs-by-Tilite/

Invacare Corporation/Alber USA

Rim assist and power add-on mobility

www.invacare.com

www.alber-usa.com

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