Stephen Sundberg, M.D., pediatric orthopedic surgeon
Kevin Sheridan, M.D., pediatric orthopedic surgeon

Osteogenesis imperfecta (OI), or “brittle bone” disease as it is sometimes called, is a hereditary condition that is best known for causing bones to be fragile and subject to frequent fractures. It affects six to seven people in 100,000, and an estimated 20,000 to 50,000 people in the U.S. have the condition.

The most common forms of OI result from a defect in the formation of Type I collagen (the building block of bones, ligaments, teeth, and middle ear bones). Children with OI may experience fractures that cause limb bowing or scoliosis. These bowed bones are then more likely to fracture again. Fracture of mid-dle ear bone can cause hearing loss as well.

At least eight different types of OI have been identified. Some of the newly described types do not have the classic collagen defect that is seen in the more common type of OI. Our discussion will concentrate on the classic Types I-IV, as described by Sillence.

Type I – Mild: This is the most common form of OI. Children with this mild form of OI suffer fractures more frequently than normal, and patients may demonstrate dentinogenesis imperfecta—an abnormality of tooth dentin. These children do not typically develop long bone bowing deformities and their height is usually normal.

Type II – Infantile: Also known as lethal OI, this type of OI is usually fatal within the first several days of life. Fractures result in moderately severe deformities at birth. With growth, further deformity typically develop long-bone bowing deformities and their height is usually normal.

Type III – Severe: Children with Type III OI have severe deformities and their height is usually below normal. They have bowing, scoliosis. These bowed bones are then more likely to fracture again. Fracture of middle ear bone can cause hearing loss as well.

Type IV – Moderate to severe: Although fractures are common with Type IV OI, typically bone deformities are not as severe. These children are more severely involved than those with Type II, but less than those with Type III.

The frequency of fractures in children with Type I OI may increase significantly during growth spurts. These children are at risk for short stature. Children with Type II OI have severe deformities and their height is usually below normal. They have bowing, scoliosis. These bowed bones are then more likely to fracture again. Fracture of middle ear bone can cause hearing loss as well.

Type III – Severely affected: Children affected with Type III OI have severe deformities and their height is usually below normal. They have bowing, scoliosis. These bowed bones are then more likely to fracture again. Fracture of middle ear bone can cause hearing loss as well.

Type IV – Moderate to severe: Although fractures are common with Type IV OI, typically bone deformities are not as severe. These children are more severely involved than those with Type II, but less than those with Type III.

Osteogenesis imperfecta affects six to seven people in 100,000, and at least eight different types of OI have been identified.

OI management centers on decreasing bone density and muscle mass, minimizing fractures, maintaining or restoring bone alignment, maximizing musculoskeletal function, and maintaining optimal growth and well-being.

Intravenous bisphosphonates help inhibit bone resorption and reduce the frequency of fractures in children who have OI.

When a child who has OI sustains a fracture, a lightweight cast or splint supports the limb while it heals. The period of immobilization is brief—often two to four weeks.

To stabilize fractures and realign deformed arm or leg bones, we use telescoping or solid rods within the deformed arm or leg bones, we use telescoping or solid rods. This may be done during childhood, adolescence, adulthood, or at any age.

To learn more about Gillette’s specialty centers and programs, visit www.gillettechildrens.org.

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After treatment with bisphosphonate therapy, some patients experience a reduction in bone pain within weeks. The use of bisphosphonates has also been shown to:

- Improve ambulation scores
- Decrease fracture incidence
- Maintain a more normal vertebral height
- Decrease cortical bone thickness

Other treatment modalities such as growth hormone, denosumab (a bone resorption inhibitor) and teriparatide (a bone anabolic medication) may provide alternatives to bisphosphonate treatment in the future. They are not yet widely used.

Orthopedic Management

Preventing and/or correcting bone deformities and treating fractures are the mainstay of orthopedic management in OI.

Braces, splints and orthotics may be used to help prevent fractures and to offer support while patients bear weight. When a child with OI sustains a fracture, a lightweight cast or splint supports the limb while it heals. Because many of these injuries occur from minor traumas, the period of immobilization is brief—often two to four weeks—in an attempt to reduce further bone loss that could occur with prolonged immobilization.

With OI, typical orthopedic fracture fixation implants (plates/screws) are not used to stabilize fractures, nor are they used in the correction of arm or leg bowing deformities, because the bone can break above or below the stiff metal plate that is placed on the bone at the time of surgery. Instead, we place telescoping or solid rods within the canal of the bone. This allows us to stabilize fractures and realign deformed arm or leg bones. Prior to 2005, nonexpandable rods were in widespread use, but they required replacement approximately every two years. Telescoping rods have been used more frequently in the past decade. These rods elongate within the bone as a child grows and require replacement less often—every five years is typical. Surgical deformity correction is required more commonly in the lower extremities than in the upper extremities.

The Role of Rehabilitation Medicine

The rehabilitation team (physicians, and physical and occupational therapists) work toward the common goal of supporting ongoing development, mobility and function in a safe way throughout the years.

Children who have OI commonly have delays in gross motor development. Interventions begin by educating the family and other caregivers about how to subtly lift and position the child to help facilitate normal development. As patients grow, we routinely evaluate their strength and development abilities; when needed, we recommend adaptive equipment to assist with them, sitting, standing or mobility. The equipment will vary with the type and severity of OI and the age of the child, but the goal is always to support children in attaining the highest level of independence and function possible.

Another important part of care is helping the child maintain strength and endurance through a carefully designed therapy program, particularly after periods of immobilization. To achieve those goals, pool therapies are often combined with therapies in the gym.

Conclusion

Although OI cannot be cured, multidisciplinary care can mitigate its effects. Treatment helps minimize the pain and deformity that can occur in children with OI and helps to maximize that child’s abilities. Gillette’s interdisciplinary OI team welcomes your questions. We look forward to the opportunity to become involved with your patient’s care.

Pre-op

Prior to surgery, this boy with Type III osteogenesis imperfecta had severe leg bowing.

Post-op

The patient’s bones have been straightened after he underwent expandable rod placement with realignment osteotomies.
Diagnosing OI

Findings from the physical exam and radiographs may initially suggest OI, but further testing is often required. Biochemical and genetic studies are used to identify OI. Some of these tests help to rule out other conditions such as hypophosphatemia, nutritional rickets, Cushings disease, calcium deficiency/malabsorption or nonaccidental injury. DNA analysis of COLIA/COLIA2 genes and protein-based analysis of collagen gathered through a skin biopsy may also be required. LEF81P and cartilage-associated protein (CRTAP) tests may suggest unusual recessive forms of OI. However, negative DNA or collagen tests do not rule out OI. A dual-energy X-ray absorptiometry (DXA) scan might also be ordered to quantify bone mass. On rare occasion a bone biopsy may be needed.

Treatment

Gillette Children’s Specialty Healthcare uses an interdisciplinary approach to managing OI. We focus on increasing bone density and muscle mass, minimizing fractures, maintaining or restoring bone alignment, maximizing muscular-skeletal function, and maintaining optimal growth and well-being. No current treatment cures OI, but the goal of treatment is to minimize the effects of the disorder.

Medical Regimen

Oral and intravenous bisphosphonates, such as alendronate, pamidronate and etidronate, are potent inhibitors of bone resorption that have been shown to reduce the frequency of fractures in children who have OI. Although oral bisphosphonates have been used, they are poorly absorbed. Additionally, they are difficult for children and families to administer; because patients must take the medication on an empty stomach and remain upright for 30 minutes before eating a meal. Therefore, many institutions use only intravenous bisphosphonates. Typically, the intravenous medications are administered over two to four hours for three days in a row. The regimen takes place every three to four months for a few years. The length of treatment is individualized but may continue for years.

Bisphosphonates may be indicated in a patient with Type III or Type IV OI who has experienced three to four fractures within two years. Prior to instituting such treatment, families have an extensive discussion with an endocrinologist to ensure that they understand the role the medications play in the management of OI, the course of treatment and the potential side effects.

After treatment with bisphosphonate therapy, some patients experience a reduction in bone pain within weeks. The use of bisphosphonates has also been shown to:

• Improve ambulation scores
• Decrease fracture incidence
• Maintain a more normal vertebral height
• Increase cortical bone thickness

Other treatment modalities such as growth hormone, denosumab (a bone resorption inhibitor) and teriparatide (a bone anabolic medication) may provide alternatives to bisphosphonate treatment in the future. They are not yet widely used.

Orthopedic Management

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Braces, splints and orthotics may be used to help prevent fractures and to offer support while patients bear weight. When a child with OI sustains a fracture, a lightweight cast or splint supports the limb while it heals. Because many of these injuries occur from minor trauma, the period of immobilization is brief—often two to four weeks—in an attempt to reduce further bone loss that could occur with prolonged immobilization.

With OI, typical orthopedic fracture fixation implants (plates/screws) are not used to stabilize fractures, nor are they used in the correction of arm or leg bowing deformities, because the bone can break above or below the stiff metal plate that is placed on the bone at the time of surgery. Instead, we place telescoping or solid rods within the canal of the bone. This allows us to stabilize fractures and realign deformed arm or leg bones. Prior to 2005, nonexpandable rods were in widespread use, but they required replacement approximately every two years. Telescoping rods have been used more frequently in the past decade. These rods elongate within the bone as a child grows and require treatment less often—every few years in typical. Surgical deformity correction is required more commonly in the lower extremities than in the upper extremities.

The Role of Rehabilitation Medicine

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Another important part of care is helping the child maintain strength and endurance through a carefully designed therapy program, particularly after periods of immobilization. To achieve those goals, post-treatment therapies are often combined with therapies in the gym.

Analysis

Although OI cannot be cured, multidisciplinary care can mitigate its effects. Treatment helps to minimize the pain and deformity that can occur in children with OI and helps to maximize that child’s abilities. Gillette’s interdisciplinary OI team welcomes your questions. We look forward to the opportunity to become involved with your patient’s care.

Conclusion

For children with Type IV OI who have experienced three to four fractures, maintennance treatment is often required. Patients experience a reduction in bone pain within weeks. After treatment with denosumab therapy, some patients experience a reduction in bone pain within weeks. The use of denosumab has also been shown to:

• Improve ambulation scores
• Decrease fracture incidence
• Increase cortical bone thickness

Another important part of care is helping the child maintain strength and endurance through a carefully designed therapy program, particularly after periods of immobilization. To achieve those goals, post-treatment therapies are often combined with therapies in the gym.

Biographies

Nanette Aldahondo, M.D.

Nanette Aldahondo, M.D. is board-certified in pediatric rehabilitation medicine and physical medicine and rehabilitation. She cares for patients who have cerebral palsy, acquired brain injury, spina bifida or other chronic disabling conditions. After receiving her medical degree from the University of Puerto Rico School of Medicine in Rio Piedras, Puerto Rico, she completed a residency at Tufts Medical Center in Boston and a fellowship in pediatric rehabilitation medicine at Gillette Children’s Specialty Healthcare. She is a member of the American Academy for Cerebral Palsy and Developmental Medicine and the American Academy of Physical Medicine and Rehabilitation.

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Understanding and Treating Osteogenesis Imperfecta

Stephen Sundberg, M.D., pediatric orthopedic surgeon
Kevin Sheridan, M.D., pediatrician specializing in endocrinology
Nanette Ackhondi, M.D., pediatric rehabilitation medicine specialist

Osteogenesis imperfecta (OI), or “brittle bone” disease as it is sometimes called, is a hereditary condition that is best known for causing bones to be fragile and subject to frequent fractures. It affects six to seven people in 100,000, and an estimated 20,000 to 50,000 people in the U.S. have the condition.

The most common forms of OI result from a defect in the formation of Type I collagen (the building blocks of bones, ligaments, teeth, and middle ear bones). Children with OI may experience multiple fractures that cause limb bowing or scoliosis. These bowed bones are then more likely to fracture again. Fracture of mid-rib bone can cause breathing loss as well.

At least eight different types of OI have been identified. Some of the newly described types do not have the classic collagen defect that is seen in the more common type of OI. Our discussion will concentrate on the classic Types IV and V, as described by Silenzo’s.

Type I – Mild: This is the most common form of OI. Children with this mild form of OI suffer fractures more frequently than normal, and patients may demonstrate desmogninemia imperfecta—an abnormality of skin and dentin. These children may not typically develop long bone bowing deformities and their height is usually normal.

Type II – Extremely severe/lethal: Children affected with Type II OI have severe birth deformities and usually do not survive past infancy. Those who do live may have only mild bone deformities.

Type III – Severe, progressive deformity: These children are born with fractures resulting in moderately severe deformities at birth. With growth, further deformity develops, leading to limb shortening and marked angulation. These deformities impair upper extremity motion and are often severe enough that children are unable to stand. Short stature and a triangular-shaped face are characteristic.

Type IV – Moderate to severe: Although fractures are common with Type IV OI, typical bone deformities are not as severe. These children are more severely involved than those with Type V, but less than those with Type III.

Osteogenesis imperfecta is a hereditary condition that is best known for causing bones to be fragile and subject to frequent fractures. It affects six to seven people in 100,000, and at least eight different types of OI have been identified.

Gillette offers an interdisciplinary OI Clinic that provides an interdisciplinary OI Clinic on one Friday every other month at our St. Paul Clinic. Our team (orthopedic surgeons, endocrinologists, geneticists and pediatric rehabilitation medicine specialists) evaluates and treats newly diagnosed patients in one location. Individual specialists collaborate as needed for follow-up care and ongoing management.

Cardiology Clinic Opens
In June, physicians from The Children’s Heart Clinic began offering a cardiology clinic every week at Gillette’s St. Paul Clinic.

New Videos and Brochures Explain Treatment Options for Cerebral Palsy
Gillette recently completed three videos about cerebral palsy. “Caring for a Child Who has Cerebral Palsy,” “Spasticity Treatment Options for Cerebral Palsy,” and “Orthopedic Care for Children Who Have Cerebral Palsy.” You and your patients can view them at gillettechildrens.org/CPVideo. We also have recently produced two brochures: About Cerebral Palsy and Managing Spasticity. Email Publications@gillettechildrens.com to request brochures.

KEY INSIGHTS

- Osteogenesis imperfecta (OI) affects six to seven people in 100,000, and at least eight different types of OI have been identified.
- OI management centers on increasing bone density and muscle mass, minimizing fractures, maintaining or restoring bone alignment, maximizing musculoskeletal function, and maintaining optimal growth and well-being.
- Intravenous bisphosphonates help inhibit bone resorption and reduce the frequency of fractures in children who have OI.
- When a child who has OI sustains a fracture, a lightweight cast or splint supports the limb while it heals. The period of immobilization is brief—often two to four weeks.
- To stabilize fractures and realign deformed arm or leg bones, we use telecasting or solid rods within the canal of the bone.
- Rehabilitation supports ongoing development, mobility and function.
Diagnosing OI

Findings from the physical exam and radiographs may initially suggest OI, but further testing is often required. Biochemical and genetic studies are used to identify OI. Some of these tests help to rule out other conditions such as hypophosphatasia, rickets, Cushing’s disease, calcium deficiency/malabsorption or nonaccidental injury. DNA analysis of COL1A1/COL1A2 genes and protein-based analysis of collagen gathered through a skin biopsy may also be required. LEFFER and cartilage-associated protein (CRTAP) tests may suggest unusual recessive forms of OI. However, negative DNA or collagen tests do not rule out OI. A dual-energy X-ray absorptiometry (DEXA) scan may also be ordered to quantify bone mass. On rare occasions a bone biopsy may be needed.

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Bisphosphonates may be indicated in a patient with Type III or Type IV OI who has experienced three to four major fractures within two years. Prior to instituting such treatment, families have an extensive discussion with an orthopedic and rehabilitation medicine specialist. She is a member of the American Academy for Cerebral Palsy and Developmental Medicine and the American Academy of Physical Medicine and Rehabilitation.

After treatment with bisphosphonate therapy, some patients experience a reduction in bone pain within weeks. The use of bisphosphonates has also shown to:

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The patient’s bones have been straightened after he underwent expandable rod placement with realignment osteotomies.

Pre-op

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Post-op

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Understanding and Treating Osteogenesis Imperfecta

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Type II – Extremely severe/fatal: Children affected with Type II OI have severe short stature, and they will develop long bone deformities due to intertrous fractures. Survival past infancy is rare.

Type III – Severe, progressive deformity: These children are born with fractures resulting in moderately severe deformities at birth. With growth, further deformity develops, leading to limb shortening and marked angulation. These deformities impair upper extremity motion and are often severe enough that children are unable to stand. Short stature and a triangular shaped face are characteristic.

Type IV – Moderate to severe: Although fractures are common with Type IV OI, typically bone deformities are not as severe. These children are more severely involved than those with Type I, but less than those with Type III.

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Gillette Offers Interdisciplinary OI Clinic
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