Symptoms or Behaviors

Signs and symptoms of OI may include:

- Frequent fractures
- Joint laxity
- Blue or gray whites of the eye
- Thin, smooth skin
- Easy bruising
- Spinal curvature
- Bowing of long bones
- Excessive sweating and heat intolerance
- Barrel-shaped rib cage (some)
- Triangular face (some)
- Dentinogenesis imperfecta (teeth affected, making them prone to cavities and cracking)
- Hearing loss (usually early adulthood, but some cases in childhood)

Osteogenesis Imperfecta (OI), commonly called brittle bone disease, is a genetic disorder in which bones break easily, often with little or no apparent cause. The term “osteogenesis imperfecta” means imperfect bone formation. Osteogenesis Imperfecta is caused by genetic defects in the structure of type I collagen. Type I collagen is the major component of the connective tissues in bones, ligaments, teeth and the white outer tissue of the eyeballs (sclera). There are at least four types, I, II, III and IV, of OI that researchers have identified (additional types continue to be identified). The signs and symptoms range from mild to severe. Individuals with Type I (mild) have half the normal amount of collagen, but it is all structurally normal. Those individuals with Types II, III and IV (severe and moderate) OI have structurally abnormal collagen. These defects lead to weak bones that fracture easily. Most cases of OI are caused by a dominant genetic defect. Some children with OI inherit the disorder from a parent. Some children born with OI have no family history of the disorder. In these children the genetic defect occurred as a spontaneous mutation. Individuals with OI have a 50% chance of passing it on to his or her child. The disorder occurs in one out of 20,000 to one out of 60,000 live births. Osteogenesis Imperfecta can affect males and females of all races.

Osteogenesis Imperfecta Traits:

**Type I**: This is the most common and mildest form of OI. Bones fracture easily, with most fractures occurring before puberty. Stature is normal to near normal. Joints are loose and muscle weakness is present. The whites of the eyes usually have a blue, purple or gray tint. The face has a triangular shape and there is a tendency toward spinal curvature. Bone deformity is absent or minimal. Brittle teeth and hearing loss beginning in the early 20’s and 30’s is possible.

**Type II**: This is the most severe form. Type II is frequently lethal at or shortly after birth, often due to respiratory problems. At birth there are often numerous fractures and severe bone deformity. Stature is small and lungs are underdeveloped.

**Type III**: Babies often have fractures present at birth. X-rays may reveal healed fractures that occurred before birth. Individuals with type III have short stature, sclera have a blue, purple, or gray tint, loose joints and poor muscle development in arms and legs. Barrel-shaped rib cage, triangular face and spinal curvature are also characteristic. Respiratory problems are possible and bone deformity is often severe. Brittle teeth and hearing loss is possible.

**Type IV**: This type falls between Type I and Type III in severity. Bones fracture easily with most fractures occurring before puberty. Stature is shorter than average with mild to moderate bone deformity. Sclera are white or near white (normal in color). There is a tendency toward spinal curvature, rib cage is barrel-shaped and face is triangular. Brittle teeth and hearing loss is possible.

Diagnosis of OI is often based on clinical features. Clinical geneticists can also perform biochemical (collagen) or DNA tests that can confirm a diagnosis of OI in some cases.

There is no cure for OI. Treatment is directed toward preventing or controlling the symptoms, maximizing independent mobility, developing bone mass and muscle strength. Care of fractures, extensive surgical and dental procedures and physical therapy are often needed. Use of wheelchairs, braces and other mobility aids are often used with individuals with more severe types. “Rodding” is a surgical procedure that involves inserting metal rods through the length of long bones to strengthen them and prevent or correct deformities. Several medications and other treatments continue to be explored.
Educational Implications
Children with a severe form of OI may have spent much of their early life lying on their back, either in the hospital or at home in casts. As a result, they have missed out on a range of “life experiences”. This may affect their confidence and early learning. Individuals with Osteogenesis Imperfecta usually have average or better academic abilities with the exception of physical education. Alternative PE activities and adaptive PE (DAPE) is often needed to develop safe and life-long leisure/recreational activities. Homebound instruction may be needed due to school absences from bone fractures that may result in hospitalization or home recovery. Children with a chronic disease should be monitored for depression, frustration and poor self concept. Staff and students may need support to deal with the fear of fractures which may result in over-protection. Since individuals with OI are generally cognitively capable, it is important that they are given every opportunity to practice social skills, make decisions for themselves and gradually learn to be independent individuals.

Instructional Strategies and Classroom Accommodations
Staff may need to provide:
- Homebound instruction or work with parents/hospital staff to make sure schoolwork continues if out of school for extended time due to fractures or surgical procedures
- Aids for mobility, and such services as adaptive physical education and physical therapy (to improve bone strength)
- Aids for writing (felt tip pen for writing due to inability to sustain pressure) and other fine motor tasks
- Accommodations/adjustments for delayed speed of writing (lax joints in hands increase effort and reduce stamina; may need to write with non-dominant hand due to fractures in other hand; in severe forms, may have short, bent arms reducing reach)
- School environment needs to reduce hazards such as items that could be tripped on or wet floors
- Appropriate times when a student may need help to avoid injuries such as recess, playground time, hallways, etc.
- Dismissal from class a few minutes early to avoid crowded hallways
- Assistance in restroom, especially for individuals with short arms and/or wheelchair users
- In-service to classmates and school personnel to understand OI and foster a considerate attitude and healthy peer relationships
- Familiarity with safe handling of children with OI. Fractures can occur if a part of the body is slightly twisted, pushed or pulled. Ask parents and physical/occupational therapists to demonstrate safe techniques
- Ongoing hearing screenings
- Develop emergency evacuation plan (fire/tornado)
- Develop Health Plan for any medical procedures and/or medications
- Information readily available for substitute teachers

Resources
Osteogenesis Imperfecta Foundation
804 W. Diamond Avenue
Suite 210
Gaithersburg, MD 20878
Telephone: 1-800-891-Bone
Internet Address: http://www.oif.org
E-mail: bonelink@oif.org

National Institutes of Health
Osteoporosis and Related Bone Diseases
National Resource Center
2 AMS Circle
Bethesda, MD 20892
Telephone: 1-800-624-BONE
Internet Address: http://www.osteo.org
E-mail: NIAMSBoneInfo@mail.nih.gov

Growing Up with OI: A Guide for Families and Caregivers
Osteogenesis Imperfecta Foundation
Gaithersburg, MD.
(15 chapter volume on OI)

For Children:
Jason’s First Day!
Available from OI Foundation

Resources for School (available from OI Foundation):
Plan for Success: An Educator’s Guide to Students with OI
(video and booklet)

Going Places
(video and discussion guide)

OI: A Guide for Nurses