

Osteogenesis Imperfecta

Background

Osteogenesis imperfecta (OI) is a rare, genetic bone disorder affecting bone formation. OI, also known as “brittle bone disease,” is characterized by bones that easily fracture or break, often from little to no apparent stress or injury.

OI is caused by a genetic mutation in the gene responsible for producing collagen; a protein that plays an important role in providing structure and strength. As a collagen-related disorder, lung and cardiac function, muscle strength, ligament flexibility, and dentition (teeth) may also be affected.

OI is generally subdivided into various types. OI Type 1 is the mildest form but occurs most frequently. Bones are prone to fracture, typically before puberty, often with a spiral fracture and minimal bone deformity. Signs and symptoms of OI include:

- Fracture
- Dislocation
- Bruising
- Muscle weakness
- Loose joints
- Scoliosis
- Hearing loss

Unique facial characteristics are also linked to OI including distinct blue or gray coloration of the sclera of the eyes, mildly triangular-shaped face, and brittle teeth.

Monitoring for pain and injury should occur promptly and include assessment of swelling, bruising, tenderness, and limited movement.

Fracture care is similar to other fractures and includes splinting, casting, or bracing to support bone stabilization and healing. For some fractures, surgical rod placement in long bones or “rodding” can be used to help with positioning and to strengthen and prevent fractures.

There is no cure for OI. Medications prescribed to treat OI include bisphosphonates, which help increase bone mineral density. Other treatments may include vitamin D and calcium supplements, pain management, and supplemental oxygen for individuals with respiratory involvement. A multidisciplinary treatment approach aims to prevent symptoms, develop muscle, strengthen bone mass, and maximize independence.



Top Takeaways for School

Osteogenesis imperfecta (OI) is a genetic bone disease characterized by bones that easily fracture or break, often for seemingly no reason.

Staff should be trained to implement precautions, follow activity restrictions, and respond appropriately to pain, injury or suspected fracture.

Changes in the student’s behavior could indicate an injury or fracture. The school nurse should be notified to assess the student.

Hearing loss is associated with OI. Classroom accommodations such as preferential seating, reduced background noise, and the use of supplementary visual aids should be considered.

Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnosis of risk for injury, pain (acute/chronic) and impaired physical mobility
- Use of specialized equipment, adaptive equipment and orthotics
- Activity, positioning, transferring (consider precautions and/or restrictions)
- Consider emergency action plans (EAPs) and emergency evacuation plans (EEPs) related to special health care needs, including staff education/training

Discussion Starters for the Educational Team

1. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
2. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
3. Does the student require activity precautions to prevent injury?
4. Is the physical school environment safely accessible for the student's mobility needs (e.g., entry and exit, ramps, location of classes, access to elevator, doorways)?
5. Does the classroom environment support the student's needs and/or equipment (e.g., desk/seating options, maneuverability space, electrical outlets, flash pass for bathroom or nurse)?
6. Will staff receive education/training to implement the student-specific emergency plan?

Resources

Kennedy Krieger Institute: Osteogenesis Imperfecta Clinic
[kennedykrieger.org/patient-care/centers-and-programs/osteogenesis-imperfecta-clinic](https://www.kennedykrieger.org/patient-care/centers-and-programs/osteogenesis-imperfecta-clinic)

Brittle Bone Society: Information for schools
[brittlebone.org/information-resources/information-for-schools/](https://www.brittlebone.org/information-resources/information-for-schools/)

Osteogenesis Imperfecta Foundation
[oif.org/](https://www.oif.org/)



For more information, please scan the QR code or visit: [KennedyKrieger.org/SHNIC](https://www.KennedyKrieger.org/SHNIC)

