Osteogenesis Imperfecta (OI)

OI is a congenital disorder caused by genetic mutations resulting in collagen abnormalities. Patients often present with multiple fractures, hearing loss, and/or scoliosis.

Pathophysiology

- Mutation in COL1A1 or COL1A2 genes causing collagen cross-linking abnormalities
- Autosomal dominant (severe) and recessive (mild) forms
- Results in decreased production of type 1 collagen

PRESENTATION

Orthopedic findings

- Bone fragility
- Multiple fractures
- Poor bone remodeling
- Ligament laxity
- Short stature
- Scoliosis

Non-orthopedic findings

- Blue sclera
- Hearing loss
- Cardiovascular abnormalities
- Decreased pulmonary function

PHYSICAL EXAM

All types: Multiple fractures
Types II and III:

- Bowing of long bones
- Scoliosis
- Short stature

INVESTIGATIONS

Radiographs findings:

- Thin bone cortices
- Osteopenia
- Anterior bowing of the tibia
- Coxa vara
- Wormian bones

Consult Orthopedics, and consider Audiology, Cardiology, ENT, Genetics and Endocrinology as needed.

SILLENCE CLASSIFICATION OF OSTEOGENESIS IMPERFECTA

Type	Inheritance	Sclera	Features
Type I	Autosomal dominant Quantitative collagen disorder	Blue	Non-deforming type. The most common type of OI. Often presents at 3-5 yrs. Hearing deficits and brittle teeth are common. ≤1 fracture per year.
Type II	Autosomal dominant Qualitative collagen disorder	Blue	**Deforming type**, potentially lethal within the perinatal period. 3+ fractures per year.
Type III	Autosomal dominant Qualitative collagen disorder	White	**Deforming type**, often present with fractures at birth and significantly short stature. Hearing loss is common. 3+ fractures per year.
Type IV	Autosomal dominant Qualitative collagen disorder	White	Non-deforming type. Long bone bowing, vertebral fractures, and hearing loss are common. ≤1 fracture per year.
Type V-XXII	Non-collagen mutation	White	Non-deforming type. Present with multiple fractures. Hearing loss is uncommon. ≤1 fracture per year.

Types I-IV are the most common. Types V-XXII are rare and not defined separately within this resource.

MANAGEMENT

Goals of Treatment: Maximize mobility, pain control, fracture prevention and management

Fracture Prevention	Acute Fracture Management	Deformity Correction
○ Bisphosphonates ○ Growth Hormone ○ Bracing ○ Physiotherapy	Fractures can be managed non-operatively (splint/cast) or operatively ○ Fractures management should follow the standard of practice in your jurisdiction.	Operative: Realignment osteotomy using rod fixation ○ Indicated when severe deformity is present ○ Telescoping or non-telescoping devices may be used

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Aspen Lillywhite (Medical Student, University of Alberta) and Dr. Elizabeth Pedersen (Pediatric Orthopedic Surgeon, Stollery Children's Hospital) for www.pedscases.com