Osteogenesis Imperfecta (OI)

- Postnatal growth deficiency with short stature
- Hypoplasia of dentin causing dentinogenesis imperfecta (gray-colored translucent teeth), late tooth eruption, caries
- Blue sclera, thin skin, easy bruising
- Bone fractures (25% in first year), deformations, joint hyperextensibility
- Cranial wormian bones, kyphosis, scoliosis
- Hearing impairment in 35% due to otosclerosis
- Genes
  - Most types are autosomal dominant due to mutations in one of two collagen genes: COL1A1 (17q21) or COL1A2 (7q21). Other genes, acting as autosomal dominant or recessive types, can be involved (see table).

Phenotype for the Genetic Types of OI

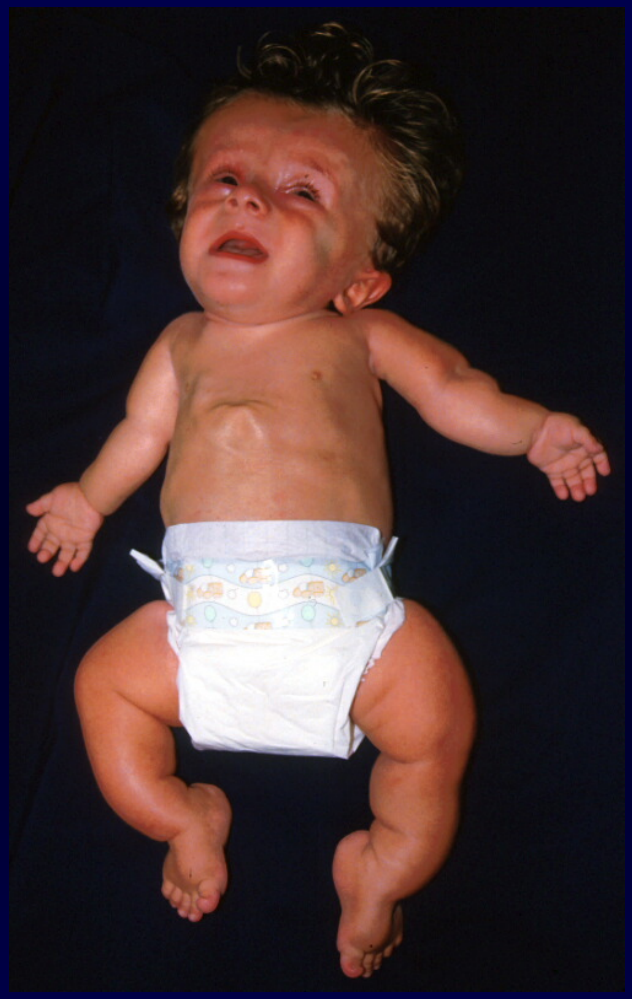
The International Nomenclature Group for Constitutional Disorders ICHG of the Skeleton 2009

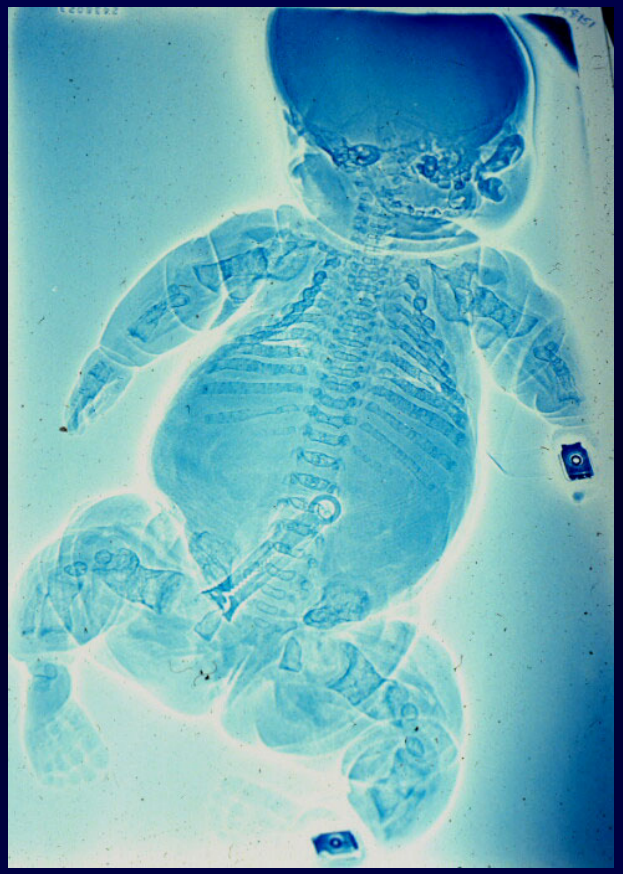
New OI classification/OI type	Phenotype
1/I	Mild, nondeforming
2/II	Severe, seen as perinatal and lethal forms
3/III, VI, VIII, IX, X, Bruck syndrome Type 1	Moderately severe, progressively deforming
4/IV, IV, VII, XI, XII, XIII	Moderate
5/V, osteoporosis-pseudoglioma syndrome, idiopathic juvenile osteoporosis, Bruck syndrome Type 1 and Type 2	Moderate, calcification of the interosseous membranes seen

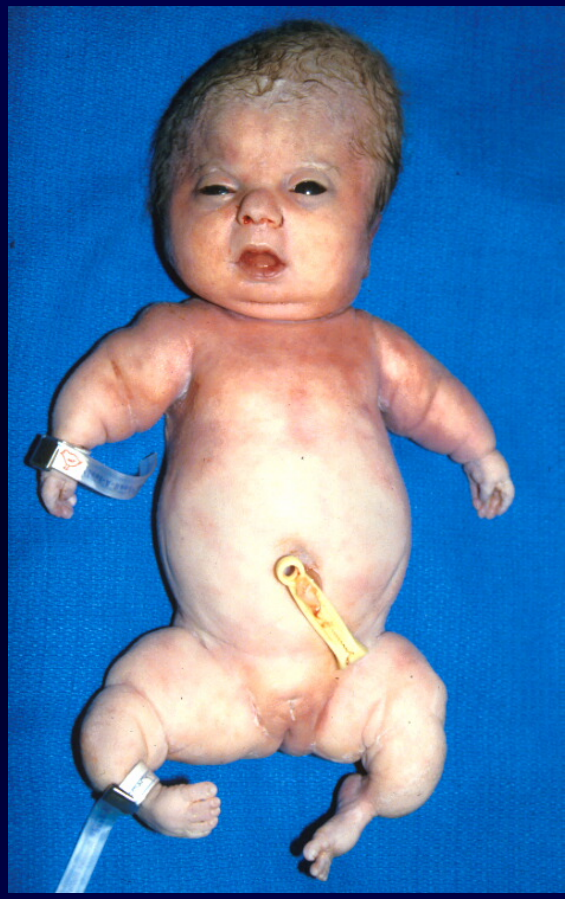
OI: Osteogenesis imperfect

Clinical Types of OI

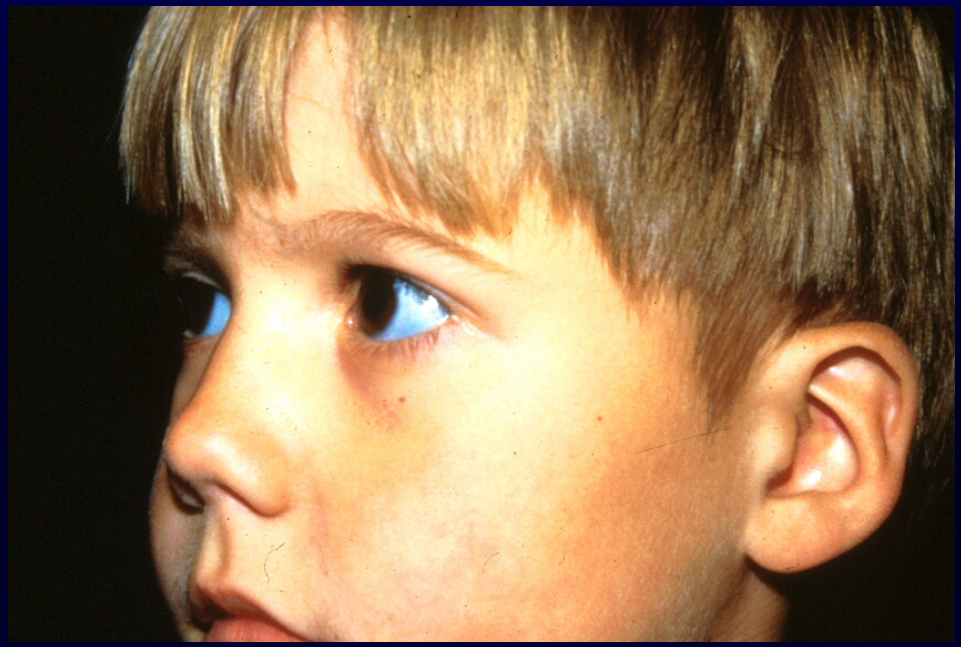
- I
  - (classical, most common type): fragile bones, blue sclera, dentinogenesis imperfecta, hyperextensibility, presenile deafness
- II
  - short broad long bones at birth, often lethal, blue sclera
- III
  - fractures at birth, progressive bone deformations, blue sclera, DI
- IV
  - femoral bowing , short stature, normal sclera, dentinogenesis imperfecta

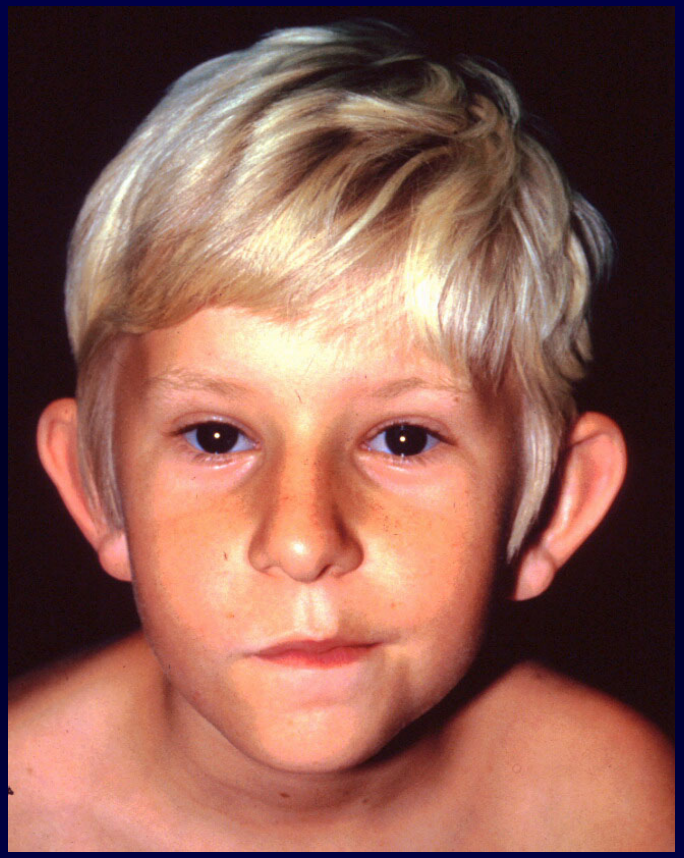




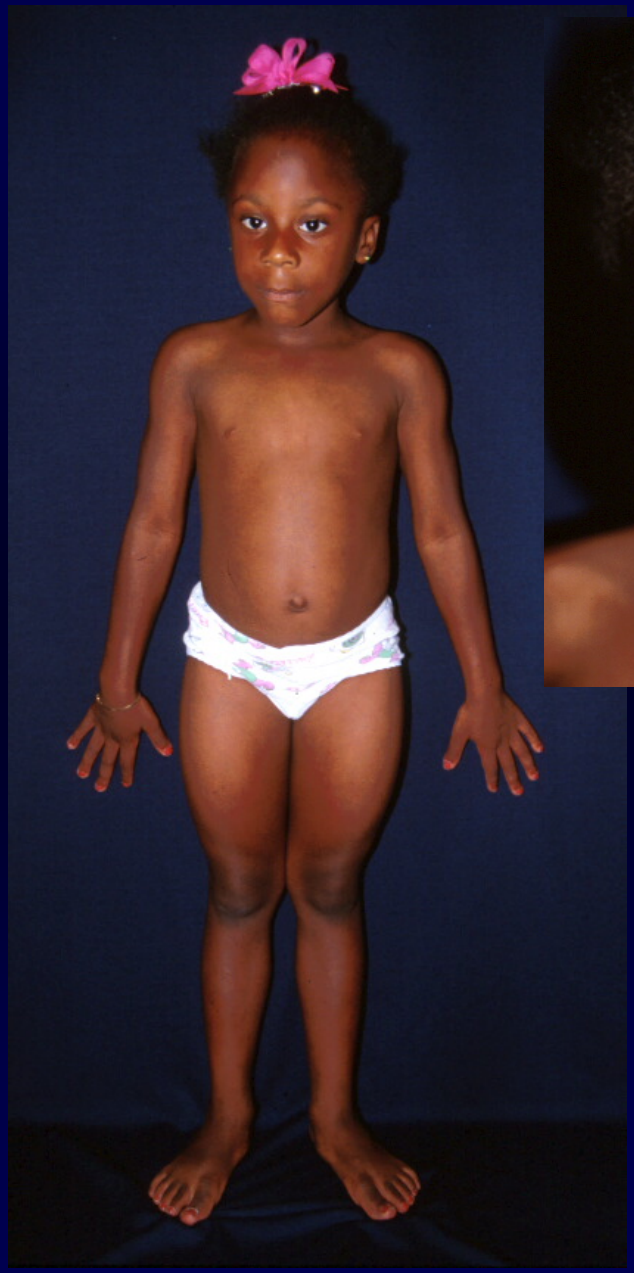


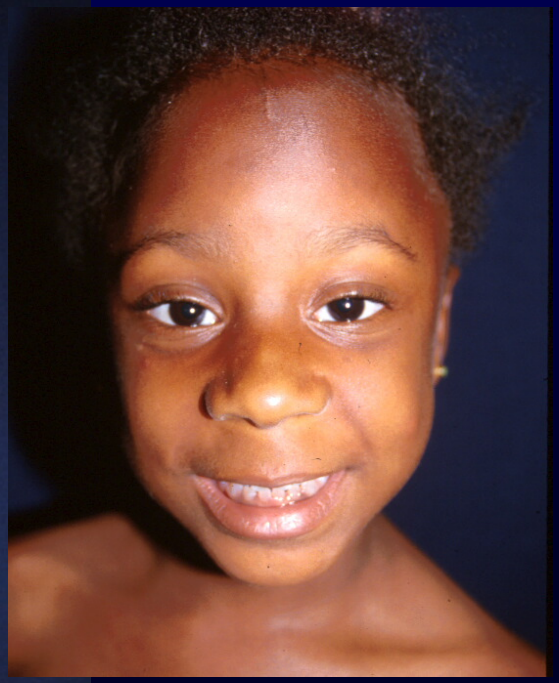
Severe involvement causes fetal demise or severe deformations and fractures in the neonate.

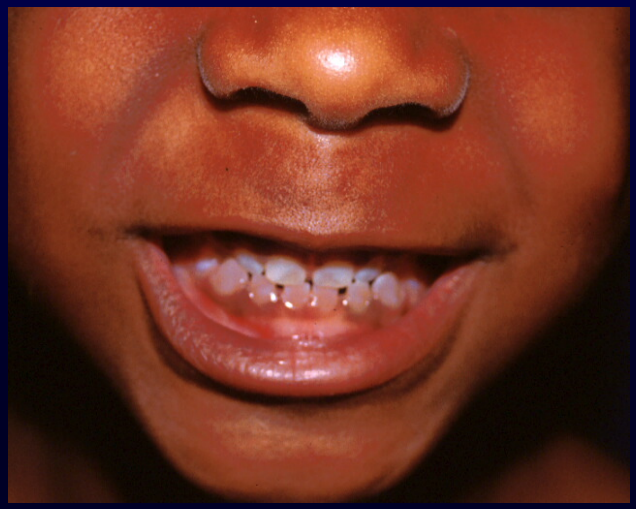




Blue sclera is commonly seen in O.I. but can also occur in normal children.







Dentinogenesis imperfecta manifests as gray, translucent teeth. This girl did not have fractures but was referred for evaluation of short stature and bowed femoral bones.

OI Mutations

OI is usually caused by mutations in one of two collagen genes, COL1A1 and COL1A2. Most cases (~ 90%) are due to COL1A1 mutations that thus affect two of the three collagen strands of the collagen fibril (as illustrated below).