

**Supplementary Table 2: Features used for diagnosis and classification of OI in the absence of biochemical or molecular studies**

**Clinical features**

Type	Inheritance	Severity	Fractures	Bone Deformity	Stature	DI	Sclerae	Hearing Loss
I	AD	Mild	Few to 100	Uncommon	Normal or slightly short for family	Rare	Blue	Present in about 50%
II	AD	Perinatal lethal	Multiple rib fractures, minimal calvarial mineralization, platyspondyly, marked compression of long bones	Severe	Severely short stature	+	Dark blue	—
III	AD	Severe	Thin ribs, platyspondyly, thin gracile bones with many fractures, "popcorn" epiphyses common	Moderate to severe	Very short	+	Blue	Frequent
IV	AD	Moderate to mild	Multiple	Mild to moderate	Variably short stature	+/-	Normal to grey	Some
V	AD	Moderate	Multiple with hypertrophic callus	Moderate	Variable	No	Normal	No
VI	AR	Moderate	Multiple	Rhizomelic shortening	Mild short stature	No	Normal	No
VII	AR	Moderate	Multiple	Yes	Short stature	No	Bluish	No

### Radiologic features

Type	Severity	Skull	Back	Extremities	Other
I	Mild	Wormian bones	Codfish vertebrae (adults)	Thin cortices	Osteopenia
II	Perinatal lethal	Undermineralization; plaques of calcification	Platyspondyly	Severely deformed; broad, crumpled, bent femurs	Small beaded ribs; findings are pathognomonic
III	Severe	Wormian bones	Codfish vertebrae; kyphoscoliosis	Flared metaphyses ("popcorn"-like appearance [childhood]), bowing, thin cortices	Thin ribs, severe osteoporosis
IV	Intermediate	± Wormian bones	Codfish vertebrae	Thin cortices	Protrusio acetabuli
V	Intermediate	?Wormian bones	?	Hypertrophic callus, usually of the femurs; mineralization of the interosseus membrane in the forearm	
VI	Intermediate	?Wormian bones	?	Similar to OI type IV	
VII	Intermediate	?Wormian bones	?	Similar to OI type IV	Rhizomelic shortening