severe with short stature and mild to moderate bone deformities. Types V and VI do not have a type 1 collagen defect and are clinically similar to type IV. Both types demonstrate a unique pattern to their bone. Individuals affected have hypertrophic callus formation at fracture sites, a radiodense metaphyseal band, and calcification of the interosseous membrane of the forearm. In type VI, bone has a characteristic mineralization defect or microscopic "fish scale" appearance with elevated alkaline phosphatase activity. Types VII through XII are rare, recessive forms of OI with different genetic defects being found. Clinical severity is variable and overlaps types II and III in relation to clinical features. Those who survive have white sclerae, short stature, and rhizomelia (Marini and Blissett, 2013).

Box 29-6

Classification of Osteogenesis Imperfecta*

Type I[*†]

A: Mild bone fragility; blue sclerae; normal teeth; hearing loss (occurs between 20 and 30 years old); autosomal dominant inheritance

B: Same as A except dentinogenesis imperfecta instead of normal teeth

C: Same as B but no bone fragility

Type II: Lethal; stillborn or die in early infancy; severe bone fragility, multiple fractures at birth; 10% of cases of OI; autosomal recessive inheritance

Type III: Severe bone fragility leading to severe progressive deformities; normal sclerae; marked growth failure; most autosomal recessive inheritance; few autosomal dominant inheritance