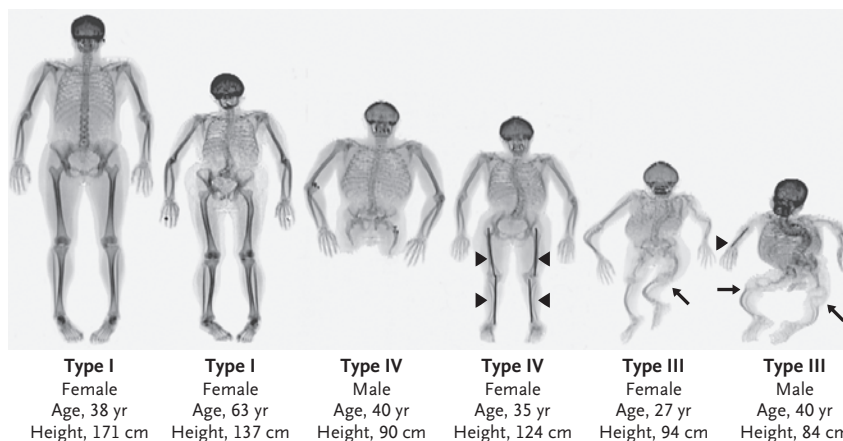


IMAGES IN CLINICAL MEDICINE

Adults with Osteogenesis Imperfecta



Janet Reeder, M.S., P.A.-C.
Eric Orwoll, M.D.

Oregon Health & Science University
Portland, OR 97239-3098

OSTEOGENESIS IMPERFECTA IS A HERITABLE DISORDER CAUSED BY MUTATIONS in the gene for type I collagen. The Sillence classification of osteogenesis imperfecta (types I through IV) is based on clinical characteristics. Whole-body images of adults with osteogenesis imperfecta who were recruited for a clinical trial were acquired with the use of dual-energy x-ray absorptiometry. A wide spectrum of skeletal manifestations is apparent, from the mild abnormalities in the 38-year-old woman with type I osteogenesis imperfecta to the severe deformities in the 40-year-old man with type III disease. Scoliosis is present in most of the adults shown. One has orthopedic rods in the femurs and tibia, and another has a rod in the radius (arrowheads). One underwent the amputation of both legs at 18 years of age owing to multiple fractures, and two have evidence of typical fragmentation of the epiphyseal growth plates (“popcorn epiphyses”) at the knees (arrows). Type II osteogenesis imperfecta is lethal in the perinatal period, owing to severe fractures and deformity. Patients with type III or type IV osteogenesis imperfecta survive fractures in infancy and childhood but have progressive deformity. Type I osteogenesis imperfecta is associated with an increased risk of fracture but is usually not deforming. Many mutations in the type I collagen gene have been shown to be associated with osteogenesis imperfecta. In general, the deforming types of the disease are the result of disruption of the helical stability of the molecule, whereas type I osteogenesis imperfecta is associated with the underproduction of structurally normal collagen. Bisphosphonate treatment reduces the risk of fracture during childhood, but information concerning its use in adults is limited.

Copyright © 2006 Massachusetts Medical Society.