

COLLAGEN DISEASES

Selected Collagen Disorders

| <i>Disorder</i> | <i>Collagen Defect</i> | <i>Clinical Manifestations</i> |
|---------------------------|--|--|
| Osteogenesis Imperfecta 1 | Decreased synthesis of type 1 collagen | Long bone fractures prior to puberty |
| Osteogenesis Imperfecta 2 | Point mutations & rearrangement of exons in triple helical regions | Perinatal death; soft, fragile & malformed bones |
| Ehlers-Danlos IV | Poor secretion, premature degradation of type III collagen | Translucent skin, easy bruising, rupture of arteries & colon |
| Ehlers-Danlos VI | Decreased hydroxylysine in types I & III collagen | Hyperextensive skin, joint hypermobility |
| Ehlers-Danlos VII | Type I procollagen accumulation, N-terminal propeptide not cleaved | Hypermobility & dislocation of joints |