Ehlers-Danlos/osteogenesis imperfecta syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ehlers-Danlos/osteogenesis imperfecta syndrome. ORPHA:230857

Ehlers-Danlos/osteogenesis imperfecta syndrome is an association of the features of Ehlers-Danlos syndrome and osteogenesis imperfecta, characterized by generalized joint hypermobility and dislocations, skin hyperextensibility and/or translucency, and easy bruising as the predominant clinical features, while being invariably associated with mild signs of osteogenesis imperfecta, including short stature, blue sclera, and osteopenia or fractures.