Ehlers-Danlos syndrome hypermobility (type III)

Genetics

-Genes unknown

-AD

Clinical findings/Dysmorphic features

-Joint hypermobility; recurrent joint dislocation/subluxation; chronic joint or limb pain

-Soft or velvety skin with normal/slightly increased elasticity --> Absence of skin or soft tissue fragility (vs. cEDS)

-Easy bruising, high narrow palate, dental crowding, and low bone density

Etiology

-Prevalence estimates ranging between 1:5,000 and 1:20,000

Pathogenesis

-Abnormal dermal elastic fibers

Genetic testing/diagnosis

-No biochemical or genetic tests clinically available

Others

-Least severe type of EDS