Ehlers-Danlos syndrome kyphoscoliotic (type VI)

Genetics

-PLOD1 (Procollagen-lysine,2-oxoglutarate 5-dioxygenase 1)

-AR; penetrance 100%

Clinical findings/Dysmorphic features

-Major criteria: congenital muscular hypotonia (--> progressive or non-progressive congenital or early-onset kyphoscoliosis), GJH with dislocations/subluxations (shoulders, hips, knees)

-Minor criteria: skin hyperextensibility, skin fragility (easy bruising, friable skin, poor wound healing, widened atrophic scarring), rupture/aneurysm of a medium-sized arteries, osteopenia/osteoporosis, blue sclerae, scleral/ocular fragility/rupture, pectus deformity, marfanoid habitus

-Life span may be normal, but risk for rupture of medium-sized arteries

Etiology

-Disease incidence of approximately 1:100,000 live births

Pathogenesis

-Enzyme deficiency leads to deficiency in hydroxylysine-based pyridinoline crosslinks in types I and III collagen

Genetic testing/diagnosis

-Sequencing of PLOD1 (67%), PLOD1 deletion/duplication analysis (33%)