Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency. ORPHA:1900

Ehlers-Danlos syndrome, kyphoscoliotic type (EDKT) is a form of Ehlers-Danlos syndrome characterized by severe hypotonia and kyphoscoliosis at birth, generalized joint hyperextensibility and ocular globe fragility.