X-linked Ehlers-Danlos syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked Ehlers-Danlos syndrome. ORPHA:75497

Ehlers-Danlos syndromes (EDS) form a heterogeneous group of hereditary connective tissue diseases characterized by joint hyperlaxity, cutaneous hyperelasticity and tissue fragility. EDS type V is characterised by hyperextensible skin but tissue fragility and joint hyperlaxity are mild. This form of EDS is very rare and has been described in only two families so far. Other reported features include congenital heart disease, hernias and short stature. Transmission is X-linked recessive.