Dermatosparaxis Ehlers-Danlos syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dermatosparaxis Ehlers-Danlos syndrome. ORPHA:1901

Ehlers-Danlos syndromes (EDS) form a heterogeneous group of hereditary connective tissue diseases characterized by joint hyperlaxity, cutaneous hyperelasticity and tissue fragility. The dermatosparaxis type (formerly called EDS type VIIC) is marked by extremely fragile tissues, hyperextensible skin and easy bruising. Facial skin contains numerous folds, as in the cutis laxa syndrome. Umbilical or inguinal hernias have also been described. Dermatosparaxis is extremely rare and few cases only have been reported. The disease is transmitted as an autosomal recessive trait. It is due to N-terminal procollagen I peptidase deficiency causing abnormal maturation of the alpha1 (I) and alpha2 (I) collagen I pro-chains, in which the aminoterminal propeptide is incorrectly cleaved. The causative gene, ADAMTS2, has been localised to 5q23. The homozygous mutation Q225X was present in 80% of cases subjected to molecular analysis. There is no specific treatment available for this disease, but symptomatic management should be offered in a specialised centre.