Opis choroby *

Definicja

A rare systemic disease characterized by a severe phenotype in all male patients, combining abnormality of connective tissue typical for Ehlers-Danlos syndrome (including joint hypermobility, scoliosis, soft and doughy skin, hyperextensible skin, abnormal scarring, facial peculiarities, and generalized hypotonia, among others) and eventually lethal congestive heart failure due to polyvalvular disease. Female carriers are affected to a variable degree.

Dane

Klasyfikacja
Choroba

Synonimy
EDS V
Sprzężony z płcią zespół Ehlersa i Danlosa
Ehlers-Danlos syndrome type 5
X-linked EDS

Kod ORPHA
75497

Kod OMIM
314400

Kod ICD10
Q79.6

Kod ICD11
LD28.1Y

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*Źródło

orphanet