DISEASE:
X-linked Ehlers-Danlos syndrome

NAME:	X-linked Ehlers-Danlos syndrome
DESCRIPTION:	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.
ORPHACODE:	75497
SYNONYMS:	EDS V Ehlers-Danlos syndrome type 5 X-linked EDS
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	FLNA
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