DISEASE:
SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome

NAME:	SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome
DESCRIPTION:	A form of spondylodysplastic Ehlers-Danlos syndrome (EDS) due to variants in the SLC39A13 gene and characterized by the presence of thin and finely wrinkled skin of the hands and feet, hypermobile distal joints, characteristic facial features (downslanting palpebral fissures, mild hypertelorism, prominent eyes with a paucity of periorbital fat, blueish sclerae, microdontia or oligodontia), muscular hypotonia, associated with significant short stature of childhood-onset, ocular findings (myopia and keratoconus) and, more rarely, vascular complications. Mild radiographic changes were observed, among which platyspondyly is a useful diagnostic feature.
ORPHACODE:	157965
SYNONYMS:	SCD-EDS SLC39A13-related spEDS SLC39A13-related spondylodysplastic EDS Spondylocheirodysplastic Ehlers-Danlos syndrome spEDS-SLC39A13
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SLC39A13

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