DISEASE:
Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency

NAME:	Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency
DESCRIPTION:	A rare subtype of kyphoscoliotic Ehlers-Danlos syndrome characterized by congenital muscle hypotonia, congenital or early-onset kyphoscoliosis (progressive or non-progressive), and generalized joint hypermobility with dislocations/subluxations (in particular of the shoulders, hips, and knees). Additional common features are skin hyperextensibility, easy bruising of the skin, rupture/aneurysm of a medium-sized artery, osteopenia/osteoporosis, blue sclerae, umbilical or inguinal hernia, chest deformity, marfanoid habitus, talipes equinovarus, and refractive errors. Subtype-specific manifestations include congenital hearing impairment (sensorineural, conductive, or mixed), follicular hyperkeratosis, muscle atrophy, and bladder diverticula. Molecular testing is obligatory to confirm the diagnosis.
ORPHACODE:	300179
SYNONYMS:	Ehlers-Danlos syndrome with kyphoscoliosis, myopathy, and deafness Ehlers-Danlos syndrome with kyphoscoliosis, myopathy, and hearing loss FKBP14-related EDS FKBP22-deficient EDS Kyphoscoliotic EDS due to FKBP22 deficiency kEDS-FKBP14

XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	FKBP14
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- FKBP prolyl isomerase 14

Related Gene Panels

- Ehlers-Danlos syndrome -UGent
- Recessive Ehlers-Danlos Syndrome (11 genes) - UGent