DISEASE:
B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome

NAME:	B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome
DESCRIPTION:	A form of spondylodysplastic Ehlers-Danlos syndrome due to variants in B3GALT6 and characterized by short stature, variable degrees of muscle hypotonia, joint hypermobility, especially of the hands, bowing of limbs and congenital or early onset, progressive kyphoscoliosis. Additional features include the typical craniofacial gestalt (prominent forehead, sparse hair, mid-face hypoplasia, blue sclerae, proptosis and abnormal dentition), hyperextensible, soft, thin, translucent and doughy skin, delayed motor and/or cognitive development, characteristic radiographic findings (spondyloepimetaphyseal dysplasia, platyspondyly, anterior beak of vertebral body, short ilia, elbow malalignment and generalized osteoporosis), joint contractures and ascending aortic aneurysm.
ORPHACODE:	536467
SYNONYMS:	B3GALT6-related spEDS B3GALT6-related spondylodysplastic EDS Beta3GalT6-deficient EDS Ehlers-Danlos syndrome progeroid type 2 spEDS-B3GALT6
XREF(S):	Orphanet OMIM ICD-10

ANALYTE(S):	B3GALT6
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RELATED CONTENT

Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)
- Osteogenesis imperfecta / Osteoporose (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- beta-1,3-galactosyltransferase 6

Related Gene Panels

- Ehlers-Danlos syndrome -UGent
- Osteogenesis imperfecta and Osteoporosis (43 genes) - UGent