DISEASE:
B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome

NAME:	B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome
DESCRIPTION:	A form of spondylodysplastic Ehlers-Danlos syndrome due to variants in B4GALT7 and characterized by short stature, variable degrees of muscle hypotonia, joint hypermobility, especially of the hands, and bowing of limbs. Additional features include the typical craniofacial gestalt (mid-face hypoplasia, round, flat face, proptosis and narrow mouth), hyperextensible skin that is soft, thin, translucent and doughy, delayed motor and/or cognitive development, characteristic radiographic findings (such as radio-ulnar synostosis, radial head subluxation or dislocation, metaphyseal flaring and osteopenia) and ocular abnormalities.
ORPHACODE:	75496
SYNONYMS:	B4GALT7-related spondylodysplastic EDS EDS progeroid type 1 EDS with short stature and limb anomalies spEDS-B4GALT7
XREF(S):	Orphanet OMIM MeSH ICD-10
ANALYTE(S):	B4GALT7
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/2892

RELATED CONTENT

Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- beta-1,4-galactosyltransferase 7

Related Gene Panels

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