Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
B3GALT6-related spEDS
B3GALT6-related spondylodysplastic EDS
Beta3GalT6-deficient EDS
Ehlers-Danlos syndrome progeroid type 2
spEDS-B3GALT6
B3GALT6-related spEDS
B3GALT6-related spondylodysplastic EDS
Beta3GalT6-deficient EDS
Ehlers-Danlos syndrome progeroid type 2
spEDS-B3GALT6

Kod ORPHA
536467

Kod OMIM
615349

Kod ICD10
Q79.6

Kod ICD11
LD28.1Y

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*Źródło

orphanet