

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

B4GALT7-related spondylodysplastic EDS
B4GALT7-CDG
Defekt biosyntezy siarczynu proteodermatanu
EDS, typ progeroidalny
Niedobór 4-beta-galaktozylotransferazy
ksylozylproteiny
Niedobór galaktozylotransferazy I
Niedobór XGPT
PDS
EDS progeroid type 1
EDS with short stature and limb anomalies
spEDS-B4GALT7

Kod ORPHA

75496

Kod OMIM

130070

Kod ICD10

Q79.6

Kod ICD11

LD28.1Y

[*Źródło](#)

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