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	Synonimy
Podtyp kliniczny	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	**Kod OMIM**	**Kod ICD10**
75496	130070	Q79.6

Kod ICD11
LD28.1Y

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*Źródło

orphanet