Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by the association of developmental delay, variable intellectual disability, skeletal dysplasia, and in many cases T-cell immunodeficiency and other immunologic abnormalities. Skeletal findings include short stature, anomalies of the long bones, hands and feet, and pelvis, platyspondyly, cervical malformation, and pectus excavatum. Dysmorphic facial features, such as coarse face, hypertelorism, and broad nasal tip, may be present. Additional reported manifestations are seizures, hyperreflexia, nystagmus, and muscular hypotonia, as well as multiple liver cysts.

Dane

Klasyfikacja
Choroba

Synonimy
EXTL3-related neuro-immuno-skeletal dysplasia syndrome
Zespół dysplazji neuro-immuno-szkieletowej związanej z EXTL3
Zespół dysplazji neuro-immuno-szkieletowej wywołanej niedoborem EXTL3
Neuro-immuno-skeletal dysplasia syndrome due to EXTL3 deficiency

Kod ORPHA
508533

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet