Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by moderate intellectual disability, dysmorphic facial features (such as prominent glabella, synophrys, and prognathism), generalized hirsutism, bilateral single palmar creases, and seizures. Additional reported manifestations include slowly progressive neurological deterioration with muscular weakness and impaired gait and balance, as well as hypogammaglobulinemia with specific absence of plasma and/or secretory IgA, among others. Brain imaging may show mild cerebellar atrophy and thin corpus callosum.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
85317

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet