Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by severe to profound intellectual disability, muscular hypotonia in childhood, delayed walking, delayed or minimal/absent speech, behavioral abnormalities including aggressiveness, agitation, and self-injurious behavior, and dysmorphic facial features (such as triangular face with high forehead, prominent ears, and small, pointed chin). Additional reported manifestations include microcephaly, short stature, and seizures, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
85329

Kod OMIM
304340

Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet