

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

*Źródło

orphanet