Opis choroby *

Definicja

A rare X-linked intellectual disability characterized by marked neonatal hypotonia, progressive quadriparesia, severely delayed developmental milestones (walking at 3 years of age), gastroesophageal reflux, stereotypic movements of the hands, esotropia and infantile autism.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
85277

Kod OMIM
300912

Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet