Opis choroby *

Definicja

A rare X-linked syndromic intellectual disability characterized by global developmental delay and severe intellectual disability, seizures, and recurrent lower respiratory tract infections, resulting in premature death in affected males. Additional reported manifestations include mild dysmorphic facial features (such as epicanthic folds, high nasal bridge, or small mouth), gait disturbances, brisk tendon reflexes, delayed bone age, and tapering fingers. No evident heterozygous manifestation has been reported in females.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
85322

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
LD90

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*Źródło

orphanet