Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability disease characterized by global developmental delay, microcephaly, mild to moderate intellectual disability, truncal ataxia, trunk and limb, or generalized, choreiform movements, and elevated serum creatine kinase levels. Variably associated features include mild cerebral atrophy, muscular weakness or hypotonia in early childhood, and/or seizures. Ocular abnormalities (e.g. exophoria, anisometropia, amblyopia) have been reported.

Dane

Klasyfikacja
Choroba

Kod ORPHA
369847

Kod OMIM
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Kod ICD10
G25.5

Kod ICD11
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*Źródło

orphanet