

Opis choroby *

Definicja

An X-linked syndromic intellectual disability characterized by congenital ataxia and generalized hypotonia, global developmental delay with intellectual disability, myoclonic encephalopathy, progressive neurological deterioration, macular degeneration, and recurrent bronchopulmonary infections.

Dane

Klasyfikacja

Choroba

Kod ORPHA

85334

Kod OMIM

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Kod ICD10

G31.8

Kod ICD11

LD90

*Źródło

orphanet