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

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*Źródło

orphanet