

Opis choroby *

Definicja

An X-linked syndromic intellectual disability characterized by a few months of normal development, followed by progressive neurodegenerative course with gradual loss of vision, development of spastic tetraplegia, convulsions, microcephaly, failure to thrive, and early death.

Dane

Klasyfikacja

Choroba

Kod ORPHA

85336

Kod OMIM

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Kod ICD10

G31.8

Kod ICD11

LD90.Y

*Źródło

orphanet