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

Kod OMIM

Kod ICD10

G31.8

85336

Kod ICD11

LD90.Y

*Źródło

orphanet