

Opis choroby *

Definicja

Late infantile neuronal ceroid lipofuscinoses (LINCLs) are a genetically heterogeneous group of neuronal ceroid lipofuscinoses (NCLs; see this term) typically characterized by onset during infancy or early childhood with decline of mental and motor capacities, epilepsy, and vision loss through retinal degeneration.

Dane

Klasyfikacja

Choroba

Synonimy

Jansky-Bielschowsky disease

Choroba Jansky'ego i Bielschowsky'ego

LINCL

Późnodziecięca NCL

LINCL

Late infantile NCL

Kod ORPHA

168491

Kod OMIM

256730

Kod ICD10

E75.4

Kod ICD11

5C56.1

*Źródło

orphanet