

Opis choroby *

Definicja

Congenital neuronal ceroid lipofuscinosis (CNCL) is a severe form of neuronal ceroid lipofuscinosis (NCL; see this term) with onset at birth characterized by primary microcephaly, neonatal epilepsy, and death in early infancy.

Dane

Klasyfikacja

Choroba

Synonimy

Congenital NCL

Wrodzona NCL

Kod ORPHA

168486

Kod OMIM

610127

Kod ICD10

E75.4

Kod ICD11

5C56.1

[*Źródło](#)

orphanet