

Opis choroby *

Definicja

Corpus callosum agenesis-neuronopathy syndrome is a neurodegenerative disorder characterized by severe progressive sensorimotor neuropathy beginning in infancy with resulting hypotonia, areflexia, amyotrophy and variable degrees of dysgenesis of the corpus callosum. Additional features include mild-to-severe intellectual and developmental delays, and psychiatric manifestations that include paranoid delusions, depression, hallucinations, and 'autistic-like' features. Affected individuals are usually wheelchair restricted in the second decade of life and die in the third decade of life. The disease is inherited as an autosomal recessive trait.

Dane

Klasyfikacja

Choroba

Synonimy

Andermann syndrome
Choroba Charlevoix
Zespół Andermanna
Charlevoix disease

Kod ORPHA

1496

Kod OMIM

218000

Kod ICD10

G60.0

Kod ICD11

LD20.Y

*Źródło

orphanet