

## 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

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#### \*Źródło

orphanet