Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability disorder characterized by variable degrees of intellectual disability, behavioral problems (including attention deficit and hyperactivity disorder, autism spectrum disorder, and aggressiveness), an altered sleeping pattern, and delayed speech and language development associated with disruption of ankyrin-3 (ANK3 gene). Additional features observed may include muscular hypotonia and spasticity. Epilepsy, chronic hunger, and dysmorphic facial features have been reported.

Dane

Klasyfikacja
Choroba

Kod ORPHA
356996

Kod OMIM
615493

Kod ICD10
G93.8

Kod ICD11
-

---

*Źródło

orphanet