

Opis choroby *

Definicja

X-linked cerebral-cerebellar-coloboma syndrome is a rare, genetic syndrome with a cerebellar malformation as major feature characterized by cerebellar vermis hypo- or aplasia, ventriculomegaly, agenesis of corpus callosum and abnormalities of the brainstem and cerebral cortex in association with ocular coloboma. Clinically, patients show hydrocephalus at birth, neonatal hypotonia with abnormal breathing pattern, ocular abnormalities with impaired vision, severe psychomotor delay, and seizures.

Dane

Klasyfikacja

Choroba

Synonimy

X-linked intellectual disability, Kroes type

Niepełnosprawność intelektualna sprzężona z chromosomem X, typ Kroesa

Kod ORPHA

163961

Kod OMIM

300864

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet