

Opis choroby *

Definicja

A rare, genetic, neurological disorder characterized by horizontal gaze palsy, sensorineural deafness, central hypoventilation, developmental delay, and intellectual disability, described in persons of Athabascan American Indian heritage. Swallowing dysfunction, vocal cord paralysis, facial paresis, seizures, internal carotid artery, and cardiac outflow tract anomalies may be additionally observed. No dysmorphic facial features are associated.

Dane

Klasyfikacja

Choroba

Synonimy

ABSD

ABSD

Zespół pnia mózgu Navajo

Athabascan brainstem dysgenesis syndrome

Navajo brainstem syndrome

Kod ORPHA

69739

Kod OMIM

601536

Kod ICD10

G96.8

Kod ICD11

LD2H.Y

*Źródło

orphanet