

Opis choroby *

Definicja

Isolated cerebellar vermis hypoplasia is a rare, non-syndromic cerebellar malformation characterized by an underdeveloped cerebellar vermis. Patients may present a variable phenotype ranging from normal neurodevelopment to motor and/or language delay, variable degrees of cognitive impairment, hypotonia, equilibrium disturbances, static/dynamic ataxia, oculomotor abnormalities, epilepsy and/or clumsiness. Behavioral disorders such as attention deficit hyperactivity disorder and generalized anxiety have also been reported. Brain MRI may reveal diffuse or selective (mostly posterior) vermian cerebellar hypoplasia and EEG may show focal paroxysms.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA	Kod OMIM	Kod ICD10
199630	-	Q04.3
Kod ICD11		
-		

*Źródło

orphanet