Opis choroby *

Definicja

A rare, syndromic intellectual disability characterized by macrocephaly, short stature, intellectual disability, variable degree of spastic paraplegia, central nervous system malformations (hydrocephalus, Dandy-Walker malformation), and dysmorphic features, such as high and broad forehead, midface hypoplasia, and small and broad hands and feet. There have been no further descriptions in the literature since 1993.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Volcke-Soekarman syndrome
Zespół Volcke i Soekarmana

Kod ORPHA
2427

Kod OMIM
-

Kod ICD10
Q87.1

Kod ICD11
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*Źródło

orphanet