Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by severe intellectual deficit, Dandy-Walker malformation, macrocephaly, severe myopia, brachytelephalangy with short and broad fingernails, and dysmorphic facial features (such as thick eyebrows, synophrys, epicanthal folds, low-set ears, short philtrum, and high-arched palate). Additional reported manifestations include seizures and skeletal and genital anomalies, among others. There have been no further descriptions in the literature since 1989.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1970

Kod OMIM
220219

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet