Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability, characterized by macrocephaly, intellectual disability, seizures, dysmorphic facial features (including tall forehead, downslanting palpebral fissures, hypertelorism, depressed nasal bridge, and macrostomia), megalencephaly, and small thorax. Other reported features are umbilical hernia, muscular hypotonia, global developmental delay, autistic behavior, and café-au-lait spots, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
MINDS syndrome
Zespół MINDS
Smith-Kingsmore syndrome

Kod ORPHA
457485

Kod OMIM
616638

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet