

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 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