

Opis choroby *

Definicja

A rare, genetic, neurological disease characterized by association of macrocephaly, dysmorphic facial features and psychomotor delay leading to intellectual disability and autism spectrum disorder. Facial dysmorphism may include frontal bossing, hypertelorism, midface hypoplasia, depressed nasal bridge, short nose, and long philtrum.

Dane

Klasyfikacja

Choroba

Kod ORPHA

210548

Kod OMIM

605309

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet