## 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