Opis choroby *

Definicja

A rare, autosomal recessive, multiple congenital anomalies/dysmorphic syndrome characterized mainly by developmental delay, variable intellectual disability, microcephaly, cerebellar hypoplasia, dysmorphic features (central incisors macrodontia and slender fingers), short stature and variable congenital anomalies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Cerebellofaciodental syndrome
Cerebellofaciodental syndrome

Kod ORPHA
444072

Kod OMIM
616202

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet