Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability disorder characterized by craniofacial features, global developmental delay, intellectual disability and variable neurobehavioral abnormalities (autism spectrum disorder, aggressiveness, and self-injury). Additional features include vision abnormalities and variable sensorineural hearing loss, as well as short stature, hypotonia and gastrointestinal manifestations (e.g. poor feeding, gastroesophageal reflux, constipation).

Dane

Klasyfikacja
Choroba

Synonimy
Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome
Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome

Kod ORPHA
468678

Kod OMIM
616364

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet