

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
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Kod ORPHA

468678

Kod OMIM

616364

Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet