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## Intellectual disability-microcephalystrabismus-behavioral abnormalities syndrome

**INSERM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome</u>. ORPHA:468678

Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome is a rare, genetic, syndromic intellecutal disability disorder characterized by craniofacial dysmorphism (microcephaly, hypotonic facies, strabismus, long and flat malar region, posteriorly rotated ears, flat nasal bridge with broad nasal tip, short philtrum, thin vermillion border, open mouth with down-turned corners, high arched palate, pointed chin), global developmental delay, intellectual disability and variable neurobehavioral abnormalities (autism spectrum disorder, aggressivness, 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).

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