Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome. ORPHA:352577

Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome is rare, genetic, syndromic intellectual disability disorder with a variable phenotypic presentation typically characterized by microcephaly, severe feeding difficulties, failure to thrive, severe global development delay that frequently results in absent/poor speech, moderate to profound intellectual disability, hypotonia and a distinctive facies that includes prominent forehead, high-arched, thin eyebrows, hypertelorism, downslanting palpebral fissures, long, tubular nose with broad tip and prominent nasal bridge and wide mouth with full, everted lower lip.