Ring chromosome 13 syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ring chromosome 13 syndrome. ORPHA:96176

Ring chromosome 13 is a chromosomal anomaly of chromosome 13 characterized by a widely variable phenotype (ranging from mild to severe) principally characterized by intrauterine growth retardation, developmental delay, short stature, moderate to severe intellectual deficit, microcephaly, facial dysmorphism (i.e. upslanting palpebral fissures, hypertelorism, abnormal ears, broad nasal bridge, high arched palate, micrognathia, small mouth, and thin lips), hands and feet anomalies, and genital abnormalities. Additional features reported include behavioral problems, hearing and speech disorders, congenital heart defects, cerebral malformations, and anal atresia.