Ring chromosome 16 syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ring chromosome 16 syndrome. ORPHA:96178

Ring chromosome 16 is a rare chromosomal anomaly syndrome, resulting from the partial deletion of chromosome 16, characterized by pre- and postnatal growth delay, severe developmental delay, intellectual disability, speech delay, and craniofacial dysmorphism (e.g. microcephaly, hypertelorism, downslanted palpebral fissures, ptosis, telecantus, low set and dysmorphic ears, broad flat nasal bridge, down-turned mouth corners, high palate, retrognathia). Patients may also present congenital cataract, mild synophrys, hypotonia, and poor social contact. Congenital heart anomalies (e.g. ventricular septal defect, patent ductus arteriosus) have also been reported.