Ring chromosome 5 syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ring chromosome 5 syndrome. ORPHA:251043

Ring chromosome 5 syndrome is a rare chromosomal anomaly syndrome, with high phenotypic variability, principally characterized by a neonatal mewing cry, severe developmental delay and intellectual disability, short stature, hypotonia, dysmorphic features (incl. microcephaly, facial asymmetry, hypertelorism, epicanthal folds, abnormal ears, micro/retrognathia), congenital cardiac anomalies (such as atrial and ventricular septal defect, tricuspid insufficiency, hypoplastic aorta) and skeletal abnormalities (e.g. hypoplastic thumbs, anomalous ulna/radius, dysplastic metacarpals and phalanges).