# Ring chromosome 19 syndrome

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ring chromosome 19 syndrome. ORPHA:1443*

Ring chromosome 19 syndrome is a rare chromosomal anomaly syndrome with a highly variable phenotype that may range from normal to patients with profound intellectual disability, developmental delay, learning disability (esp. speech) and mild dysmorphism (incl. micro/macrocephaly, prominent forehead, low-set and posteriorly rotated ears, hypertelorism, high nasal bridge, prominent philtrum, retro/micrognathia). Mild hypotonia and autistic-like mannerisms (e.g. hand opening and closing, head banging) may also be associated. Other anomalies, such as cutis laxa, hearing loss, syndactyly, digital hypoplasia, and talipes equinovarus, have also been reported.