Ring chromosome 20 syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ring chromosome 20 syndrome. ORPHA:1444

Ring chromosome 20 syndrome is marked by a characteristic seizure phenotype. Depending on the amount of chromosomal loss and associated mosaicism, ring(20) can be associated with macrocephaly, mild to moderate intellectual deficit, or behavioural problems. In rare cases, brain, kidney or heart malformations may be present.