Tetraploidy

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Tetraploidy. ORPHA:3305

Tetraploidy is an extremely rare chromosomal anomaly, polyploidy, when an affected individual has four copies of each chromosome, instead of two, resulting in total of 92 chromosomes in each cell. The phenotype is severe with multiple congenital anomalies, including central nervous system, ocular, cardiac, renal, and/or genital malformations and limb defects. Most patients show severe intrauterine groth retardation, hypotonia, failure to thrive and developmental delay. It is usually associated with miscarriage.