3q13 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 3q13 microdeletion syndrome. ORPHA:1621

3q13 microdeletion syndrome is a rare chromosomal anomaly syndrome resulting from a partial deletion of the long arm of chromosome 3. Phenotype can be highly variable, but it is primarily characterized by significant developmental delay, postnatal growth above the mean, muscular hypotonia and distinctive facial features (such as broad and prominent forehead, hypertelorism, epicantic folds, anti-mongloid slanted eyes, ptosis, short philtrum, protruding lips with a full lower lip, high arched palate). Abnormal hypoplastic male genitalia and skeletal abnormalities are frequently present.