Xq12-q13.3 duplication syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Xq12-q13.3 duplication syndrome. ORPHA:314389

Xq12-q13.3 duplication syndrome is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the long arm of chromosome X, characterized by global developmental delay, autistic behavior, microcephaly and facial dysmorphism (including down-slanting palpebral fissures, depressed nasal bridge, anteverted nares, long philtrum, down-slanting corners of the mouth). Seizures have also been reported in some patients.