9p13 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 9p13 microdeletion syndrome. ORPHA:324313

9p13 microdeletion syndrome is a rare chromosomal anomaly syndrome, resulting from a partial interstitial deletion of the short arm of chromosome 9, characterized by mild to moderate developmental delay, hand tremors, myoclonic jerks, attention deficit-hyperactivity disorder and a social personality. Patients also present bruxism, short stature and minor facial dysmorphic features (e.g., bilateral epicantic folds, broad, flat nasal bridge, anteverted nares, low-set ears micro/retro-gnathia).