1p31p32 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 1p31p32 microdeletion syndrome. ORPHA:401986

1p31p32 microdeletion syndrome is a rare chromosomal anomaly syndrome, resulting from the partial deletion of the short arm of chromosome 1, characterized by developmental delay, corpus callosum agenesis/hypoplasia and craniofacial dysmorphism, such as macrocephaly (caused by hydrocephalus or ventriculomegaly), low-set ears, anteverted nostrils and micrognathia. Urinary tract defects (e.g. vesicoureteral reflux, urinary incontinence) are also frequently associated. Other reported variable manifestations include hypotonia, tethered spinal cord, Chiari type I malformation and seizures.