17q11 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 17q11 microdeletion syndrome. ORPHA:97685

17q11 microdeletion syndrome is a rare severe form of neurofibromatosis type 1 (NF1; see this term) characterized by mild facial dysmorphism, developmental delay, intellectual disability, increased risk of malignancies, and a large number of neurofibromas.