8q21.11 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 8q21.11 microdeletion syndrome. ORPHA:284160

8q21.11 microdeletion syndrome encompasses heterozygous overlapping microdeletions on chromosome 8q21.11 resulting in intellectual disability, facial dysmorphism comprising a round face, ptosis, short philtrum, Cupid's bow and prominent low-set ears, nasal speech and mild finger and toe anomalies.