Monosomy 22q13

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Monosomy 22q13. ORPHA:48652

Monosomy 22q13 syndrome (deletion 22q13.3 syndrome or Phelan-McDermid syndrome) is a chromosome microdeletion syndrome characterized by neonatal hypotonia, global developmental delay, normal to accelerated growth, absent to severely delayed speech, and minor dysmorphic features.