Distal monosomy 17q

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Distal monosomy 17q. ORPHA:1597

A partial deletion of the long arm of chromosome 17 characterized by hypotonia, growth delay, severe global developmental delay, microcephaly, seizures, congenital heart anomalies, hand and foot anomalies (syndactyly, symphalangism) and dysmorphic facial features, including round face, hypertelorism, upslanting palpebral fissures, and micrognathia. Reported deletions involve regions 17q21-q24.