

[Open Peer Review on Qeios](#)

Non-distal monosomy 12q

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Non-distal monosomy 12q. ORPHA:96160*

Non-distal monosomy 12q is a partial autosomal monosomy characterized by variable combination of developmental delay, intellectual disability, ectodermal, genitourinary and minor cardiac anomalies, and specific dysmorphic features (prominent forehead and low-set ears). Specific combination depends on the size and breakpoints of deleted regions.