# Monosomy 22

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Monosomy 22. ORPHA:96123*

A rare autosomal anomaly syndrome, with a highly variable phenotype, typically characterized by short length, joint abnormalities (e.g. dysplasia, hyperextensibility, contractures, dislocation), congenital cardiac defects, and craniofacial dysmorphism (incl. microcephaly, a high, prominent, narrow and/or hairy forehead, epicanthus, upward-slanting and/or small palpebral fissures, broad, high or depressed nasal bridge and malformed ears). Delayed motor development and intellectual disability is observed in patients not presenting early demise.