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Atrioventricular defect-blepharophimosis-radial and anal defect syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Atrioventricular defect-blepharophimosis-radial and anal defect syndrome. ORPHA:1352

Atrioventricular defect-blepharophimosis-radial and anal defect syndrome is a rare, genetic multiple congenital anomalies syndrome characterized by atrioventricular septal defects and blepharophimosis, in addition to radial (e.g. aplastic radius, shortened ulna, fifth finger clinodactyly, absent first metacarpal and thumb) and anal (e.g. imperforate or anteriorly placed anus, rectovaginal fistula) defects.