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Preaxial polydactyly-colobomata-intellectual disability syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Preaxial polydactyly-colobomata-intellectual disability syndrome. ORPHA:2921*

Preaxial polydactyly-colobomata-intellectual disability syndrome is characterised by growth retardation, intellectual deficit, preaxial polydactyly and colobomatous anomalies. It has been described in one pair of sibs (brother and sister). The mode of transmission is thought to be autosomal recessive.