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Autosomal dominant primary microcephaly

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal dominant primary microcephaly. ORPHA:2514

Autosomal dominant primary microcephaly is a rare, genetic, non-syndromic, developmental defect during embryogenesis malformation syndrome characterized by a congenital, non-progressive, occipitofrontal head circumference that is 2 or more standard deviations below the mean for age, gender and ethnicity which is associated with normal brain architecture and uncomplicated by other abnormalities. Borderline to moderate intellectual disability, as well as early psychomotor delay, may or may not be associated.