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Microcephaly-deafness-intellectual disability syndrome

INSERM

Source

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

Microcephaly-deafness-intellectual disability syndrome. ORPHA:2533

Microcephaly-deafness-intellectual disability syndrome is characterised by microcephaly, deafness, intellectual deficit and facial dysmorphism (facial asymmetry, prominent glabella, low-set and cup-shaped ears, protruding lower lip, micrognathia). It has been described in a mother and her son. The mode of inheritance is probably autosomal dominant.