Miller-Dieker syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Miller-Dieker syndrome. ORPHA:531

Miller-Dieker Syndrome (MDS) is a contiguous gene deletion syndrome of chromosome 17p13.3, characterised by classical lissencephaly (lissencephaly type 1) and distinct facial features. Additional congenital malformations can be part of the condition.