Open Peer Review on Qeios

MEHMO syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. MEHMO syndrome. ORPHA:85282

MEHMO syndrome is characterised by severe intellectual deficit, epilepsy, microcephaly, hypogenitalism, and obesity. Growth delay and diabetes are also present. To date, it has been described in seven boys, all of whom died within the first two years of life. The causative gene has been localised to the 21.1-22.13p region of the X chromosome and the syndrome appears to result from mitochondrial dysfunction.