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DPM1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. DPM1-CDG. ORPHA:79322

The CDG (Congenital Disorders of Glycosylation) syndromes are a group of autosomal recessive disorders affecting glycoprotein synthesis. CDG syndrome type 1e is characterised by psychomotor delay, seizures, hypotonia, facial dysmorphism and microcephaly. Ocular anomalies are also very common.