Open Peer Review on Qeios

COG2-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. COG2-CDG. ORPHA:435934

A rare, congenital disorder of glycosylation caused by mutations in the COG2 gene and characterized by normal presentation at birth, followed by progressive deterioration with postnatal microcephaly, developmental delay, intellectual disability, seizures, spastic quadriplegia, liver dysfunction, hypocupremia and hypoceruloplasminemia in the first year of life. Diffuse cerebral atrophy and thin corpus callosum may be observed on brain MRI.