ALG3-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG3-CDG. ORPHA:79321

ALG3-CDG is a form of congenital disorders of N-linked glycosylation characterized by severe neurological involvement, including hypotonia, developmental delay, intellectual disability, postnatal microcephaly, and progressive brain and cerebellar atrophy. Epilepsy with hypsarrythmia is frequently reported. Additional features that may be observed include failure to thrive, arthrogryposis multiplex congenita (AMC, see this term), vision impairment (optic atrophy, iris coloboma) and facial dysmorphism (hypertelorism with a broad nasal bridge, large and thick ears, thin lips, micrognathia). ALG3-CDG is caused by loss of function mutations of the gene ALG3 (3q27.3).