ALG1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG1-CDG. ORPHA:79327

ALG1-CDG is a severe form of congenital disorders of N-linked glycosylation characterized by severe developmental and psychomotor delay, muscular hypotonia, intractable early-onset seizures, and microcephaly. Additional features include altered blood coagulation with a high probability of hemorrhages or thromboses, nephrotic syndrome, ascites, hepatomegaly, cardiomyopathy, ocular manifestations (strabismus, nystagmus), and immunodeficiency. The disease is caused by loss-of-function mutations in the gene ALG1 (16p13.3).