ALG9-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG9-CDG. ORPHA:79328

ALG9-CDG is a form of congenital disorders of N-linked glycosylation characterized by progressive microcephaly, hypotonia, developmental delay, drug-resistant infantile epilepsy, and hepatomegaly. Additional features that may be observed include failure to thrive, pericardial effusion, renal cysts, skeletal dysplasia, facial dysmorphism (frontal bossing, hypertelorism, depressed nasal bridge, low-seated ears, large mouth) and hydrops fetalis (see this term). The disease is caused by loss-of-function mutations in the gene ALG9 (11q23).