ALG8-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG8-CDG. ORPHA:79325

ALG8-CDG is a form of congenital disorders of N-linked glycosylation that is characterized by gastrointestinal symptoms (diarrhea, vomiting, feeding problems with failure to thrive, protein-losing enteropathy), edema and ascites (including hydrops fetalis; see this term), hepatomegaly, renal tubulopathy, coagulation anomalies due to thrombocytopenia, brain involvement (psychomotor delay, seizures, ataxia), facial dysmorphism (low-set ears and retrognathia), pes equinovarus, and muscular hypotonia. Cataracts may also be observed. Prognosis is usually poor. The disease is caused by loss-of-function mutations in the gene ALG8 (11q14.1), resulting in a block in the initial step of protein glycosylation.