ALG11-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG11-CDG. ORPHA:280071

ALG11-CDG is a form of congenital disorders of N-linked glycosylation characterized by facial dysmorphism (microcephaly, high forehead, low posterior hairline, strabismus), hypotonia, failure to thrive, intractable seizures, developmental delay, persistent vomiting and gastric bleeding. Additional features that may be observed include fat pads anomalies, inverted nipples, and body temperature oscillation. The disease is caused by mutations in the gene ALG11 (13q14.3).