ALG12-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG12-CDG. ORPHA:79324

ALG12-CDG is a form of congenital disorders of N-linked glycosylation characterized by facial dysmorphism (prominent forehead, large ears, thin upper lip), generalized hypotonia, feeding difficulties, moderate to severe developmental delay, progressive microcephaly, frequent upper respiratory tract infections due to impaired immunity with decreased immunoglobulin levels, and decreased coagulation factors. Additional features include hypogonadism with or without hypospadias in males, skeletal anomalies, seizures and cardiac anomalies in some cases. ALG12-CDG is caused by loss of function mutations of the gene ALG12 (22q13.33).