ALG2-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG2-CDG. ORPHA:79326

ALG2-CDG is a form of congenital disorders of N-linked glycosylation characterized by iris coloboma, cataract, infantile spasms, developmental delay and abnormal coagulation factors. The disease is caused by loss-of-function mutations in the gene ALG2 (9q31.1). Transmission is autosomal recessive.