ALG6-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. ALG6-CDG. ORPHA:79320

ALG6-CDG is a form of congenital disorders of N-linked glycosylation characterized by feeding problems, mild-to-moderate neurologic involvement with hypotonia, poor head control, developmental delay, ataxia, strabismus, and seizures, ranging from febrile convulsions to epilepsy. Retinal degeneration has also been reported. A minority of patients show other manifestations, particularly intestinal (such as protein-losing enteropathy) and liver involvement. The disease is caused by loss of function mutations of the gene ALG6 (1p31.3).