DPAGT1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. DPAGT1-CDG. ORPHA:86309

DPAGT1-CDG is a form of congenital disorders of N-linked glycosylation characterized by hypotonia, intractable seizures, developmental delay, microcephaly and severe fetal hypokinesia. Additional features that may be observed include apnea and respiratory deficiency, cataracts, joint contractures, vermian hypoplasia, dysmorphic features (esotropia, arched palate, micrognathia, finger clinodactyly, single flexion creases) and feeding difficulties. The disease is caused by loss-of-function mutations in the gene DPAGT1 (11q23.3).