SLC35A2-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. SLC35A2-CDG. ORPHA:356961

SLC35A2-CDG is a congenital disorder of glycosylation characterized by severe or profound global developmental delay, early epileptic encephalopathy, muscular hypotonia, dysmorphic features (coarse facies, thick eyebrows, broad nasal bridge, thick lips, inverted nipples), variable ocular defects and brain morphological abnormalities on brain MRI (cerebral atrophy, thin corpus callosum).