Pediatric Neurology Part III: Chapter 171. Glucide metabolism disorders (excluding glycogen myopathies) (Handbook of Clinical Neurology)

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Glucide metabolism comprises pathways for transport, intermediate metabolism, utilization, and storage of carbohydrates. Defects affect multiple organs and present as systemic diseases. Neurological symptoms result from hypoglycemia, lactic acidosis, or inadequate storage of complex glucide molecules in neurological tissues. In glycogen storage disorders hypoglycemia indicates hepatic involvement, weakness and muscle cramps muscle involvement. Hypoglycemia is also the leading neurological symptom in disorders of gluconeogenesis. Disorders of galactose and fructose metabolism are rare, detectable by neonatal screening, and manifest following dietary intake of these sugars. Rare defects within the pentose metabolism constitute a new area of inborn metabolic disorders and may present with neurological symptoms. Treatment of these disorders involves the avoidance of fasting, dietary treatment eliminating specific carbohydrates, and enzyme replacement therapy in individual glycogen storage diseases.GLUT1 deficiency syndrome, a specific disorder of glucose transport into brain, results in global developmental delay, early-onset epilepsy, and a complex movement disorder. Treatment with a high-fat, low-carbohydrate ketogenic diet provides ketones as an alternative fuel to the brain and restores brain energy metabolism. Recently paroxysmal exertion-induced dyskinesia and stomatin-deficient cryohydrocytosis have been identified as an allelic disorder to GLUT1 deficiency equally responding to a ketogenic diet.

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