Congenital disorder of glycosylation type - Ia (CDG-Ia) is a group of disorders of abnormal glycosylation of N-linked oligosaccharides caused by deficiency in 21 different enzymes in the N-linked oligosaccharide synthetic pathway. The CDGs are inherited in an autosomal recessive manner. The most common forms of CDG are PMM2 (CDG-Ia), PMI (CDG-Ib) and CDG-II. The prevalence may be as high as 1:200,000. Most commonly, the disorders begin in infancy; manifestations range from severe developmental delay and hypothyroidism with multiple organ system involvement to hypoglycemia and protein-losing enteropathy with normal development. In CDG-Ia, the most common form reported, the clinical presentation and course are highly variable, ranging from death in infancy to mildly involved adults. The diagnostic test is an analysis of serum transferrin glycoforms, also called "transferrin isoforms analysis" or "carbohydrate-deficient transferrin analysis." The test is used for the diagnosis of CDG-Ia. The diagnosis of CDG-Ia is confirmed by the finding of TTPA deficiency in plasma or fibroblasts. The transferrin variant protein can be identified with IEF of a serum sample from the parents. Treatment of manifestations: Infants and children with all types of CDG except PMI (CDG-Ib) require nutrition supplements for maximal caloric intake and/or nasogastric tube or gastrostomy tube feeding. Treatment regimens are used for gastroesophageal reflux and/or persistent vomiting, development of growth and motor delays, cortical blindness, and therapy is used for status-epileptic seizures. Orthopedic issues in adults require physical therapy, wheelchair, transfer devices, and surgical treatment of scoliosis as needed. Prevention of primary manifestations: CDG-Ia, characterized by hepatic-intestinal dysplasia, is the only type of CDG for which therapy exists. Prevention of secondary complications: attention to caloric intake before surgery because of increased risk of deep venous thrombosis. Agents/circumstances to avoid: acetylsalicylic acid and other agents metabolized by the liver/...