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CLINICAL SYMPTOMS IN 11 CHILDREN WITH CDG SYNDROME

T. Honzik, H. Hansikova, E. Flachsova, V. Malinova, E. Hruba, A. Puchmajerova, H. Poupetova, M. Hrebicek, P. Martasek, J. Zeman
Department of Paediatrics and Center for Integrated Genomics, Charles University, Prague, Czech Republic
honzikt@seznam.cz

Congenital disorders of glycosylation-CDG syndrome represent a heterogeneous group of inherited metabolic disorders. We present the results of clinical, biochemical and molecular analyses in eleven patients with CDG syndrome I. Material and methods: The CDG syndrome type I was recognised in 11 children from 8 families. The nonglycosylated and hypoglycosylated transferrins were measured in serum using turbidimetric immunoassay and isoelectric focusing. The genes for phosphomannomutase 2 (PMM2) and 1,3-glucosyltransfere (ALG 6) were sequenced. Results: In ten children, the disease manifested in infancy with failure to thrive, muscle hypotonia, epilepsy, microcephaly and psychomotor retardation. In last patient, failure to thrive, diarrhoea and malabsorption developed in infancy. In most patients, strabismus and inverted nipples were present. Hypoplasia of the cerebellum was found in six patients and cyclic pericardial effusion in one boy. In all patients profound coagulopathy and increased amount of hypo- and nonglycosylated tranferrins were observed. Molecular analyses in two siblings revealed compound heterozygosity for the mutation G422A and C357A and in one girl for the mutation G422A and C338T in gene for PMM2. Three other patient are heterozygotes for the mutation G422A. The second mutation has not been found yet. Conclusion: The prognosis of children with CDG I syndrome is unfavourable and except patients with CDG syndrome Ib there is no treatment available. Enzymatic and molecular studies are necessary for genetic counseling and the prenatal diagnosis. Supported by GAUK 26 2003 C