OPEN ACCESS Freely available online

Health Care: Current Reviews

The clinical course of disorders of glycosylation of proteins in combination with numerous signs of dysembryogenesis

Dmytro Tokariev

Amel Dental Clinic, Ukraine

Abstract

Recently, significant practical interest is caused by genetically determined diseases, mainly with autosomal recessive type of inheritance, caused by disorders of the process of glycosylation of proteins - congenital disorders of glycosylation (CDG).

According to current data, the leading clinical manifestations of these diseases are various neurological disorders - convulsions, psychomotor developmental delay, various, quite severe somatic disorders - respiratory disorders, manifestations of cardiomyopathy, gastrointestinal disorders, including protein-losing diarrhea, anemia and hematological manifestations. , hemorrhagic syndrome, immunodeficiencies in the form of manifestations of threatening bacterial infections and others, often against the background of numerous stigmas of dysembryogenesis (small anomalies of development), with the onset of manifestations since the neonatal period.

Multiorgans, multisystem disorders in these diseases are caused by a violation of one of the key biological processes - glycosylation, due to which the carbohydrate part joins the polypeptide chain and synthesizes a fully functional molecule.

Given the fact that most of the currently known CDG is characterized by the presence of various somatic and neurological disorders in combination with small anomalies of development, it is possible to assume the relationship between clinical and phenotypic manifestations and the nature of glycosylation disorders of certain proteins, including alpha-1-acid glycoprotein.

For this, it was proposed to use a specially developed scale for assessing the phenotype for the early diagnosis of genetically determined diseases, in particular, CDG.

Biography

Dmytro Tokariev received a medical degree at the age of 23 from the Dnepropetrovsk State Medical Academy, Ukraine. He was a neonatologist at the Regional Neonatal Center, Dnipro, Ukraine, and medical director of the research company Arensia Exploratory Medicine Ukraine., Kiev, Ukraine. Now he is a pediatrician in Amel Dental Clinic, Dnipro, Ukraine. He has publications devoted to the problems of neonatology, pediatrics, clinical genetics, in particular to the problems of CDG. He is the co-author (another author is Alexandr Kulikov) of the medical diagnostic program Neo Genetics (Interactive database including clinical information on more than 2,500 genetic syndromes with an innovative search engine using about 8,000 symptoms.

4th World Summit on Neonatology Nursing and Health Care | March 24-25, 2021

Citation: Dmytro Tokariev, The clinical course of disorders of glycosylation of proteins in combination with numerous signs of dysembryogenesis, Neonatology Nursing2021, 4th World Summit on Neonatology Nursing and Health Care, March 24-25, 2021, 01