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THE SPECTRUM OF HEREDITARY METABOLIC DISORDERS THAT ARE ASSOCIATED WITH GASTROINTESTINAL PATHOLOGY

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The number of patients with Hereditary Metabolic Diseases (HMD) has been steadily increasing, but only 50% of them receive the necessary assistance. To overcome these problems, we strive to replace the "specialized" approach to the patient with the systemic and personalized approach.

Objective: To determine the spectrum of hereditary metabolic diseases that are associated with gastrointestinal pathology based on the systemic pheno- and genotypic assessment of the proband and his family.

The Medical Genetic Center for 20 years (1999-2018) has been conducting a systematic assessment of families, carrying out the monitoring. During this period, we had 628,971 families (primary-7,269-18,746 annually, data of 2018). 30,392 consultations were conducted, 9892 of them were primary. There were 1,457 children and 332 adults with HMD.

The spectrum of gastrointestinal disorders was represented by nausea, vomiting, typical and untypical reflux syndromes, cyclical vomiting syndrome, recurrent vomiting, abdominal pain, acute gastroenteritis, chronic diarrhea, celiac disease, malabsorption, Hirschsprung's disease, hepatomegaly, hepatosplenomegaly, chronic pancreatitis etc. The difference in the frequency of gastrointestinal pathology was established based on complaints (63%) and based on objective assessment of patient (87%) using clinical, biochemical, molecular genetic biomarkers and somato-genetic research.

Nosology spectrum associated with gastrointestinal disorders includes urea cycle defects, phenylketonuria, hypervalinemia, galactosemia, hyperglycemia, isopathic calcinemia, renal tubular acidenemia, disorders of ornithine, methionine, cobalamin metabolism, maple syrup diseases, methylmalonic academic, lysinic protein intolerance, Leigh syndrome, MELAS syndrome, Kearns-Sayre syndrome, neuro-gastrointestinal encephalopathy, congenital lysine intolerance, etc.

Conclusions: A systematic approach based on clinical genomics is the only effective way to provide adequate medical care to patients who have gastrointestinal disorders as the first signs of HMD.

BIOGRAPHY

Olena Grechanina has completed her PhD at the age of 24 years from National Medical University (department of general medicine) and postdoctoral studies from National Medical University (department of obstetrics and gynecology). She is the general director of Kharkiv Interregional Specialized Medical Genetic Center – Center of Rare (Orphan) Diseases, and is the member-correspondent of National Academy of Medical Sciences and professor of department of medical genetics.

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