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The single consolidated system for helping families with a hereditary pathology in the modern world: 50 years of experience

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Introduction: A powerful stream of new knowledge, evolutionary changes, loss of communication language between the genome and the environment – a short list of modern world features that entails comorbidity phenomenon. Kharkiv Interregional Specialized Medical Genetic Center – builds a single system of assistance for families with hereditary pathology, combining the efforts of scientists, doctors, nurses, technical personnel, regional authorities.

Aim: The aim of this studies the epidemiological, clinical, genetic and epigenetic characteristics of the population for the early and effective care of families.

Materials & Methodology: epidemiological, clinical-genetic, biochemical, cytogenetic, molecular-genetic.

Results: KISMGC-CR(O)D includes Medical Genetic Center, the Department of Medical Genetics and Ukrainian Institute of Clinical Genetics, KNMU, 160 employees (an average age is 30). Admitted in 2008 - 34699, 2017 - 37726 (for 10 years - 374929); primary - 2008 - 18018, 2017 - 11603 (for 10 years - 132309); established nosological forms in 2008 - 553, 2017 - 585 (for 10 years - 5622). A high incidence of mesodermal and ectodermal dysplasias, hamartoses, defects and diseases of the nervous system, hereditary connective tissue dysplasias, cardiovascular diseases, mitochondrial dysfunctions, hereditary metabolic diseases (metabolic disorders of methionine, heme, amino acids, purines, pyrimidines, organic acids, neurotransmitters, vitamins and cofactors). A unified system of assistance has been upbuilt: used "Harper principle" - the lifelong communication between a geneticist and a family; Principles Hardi I. - "doctor-nurse-family"; "1+5 Principles" - the diagnosis of expert team; collegiality principle - a council of experts, social workers, government officials; the principle of "there are no somebody else's children" have made it possible to involve representatives of the regional parliament and social services, parents associations in the fate of children.

Conclusions: The effectiveness system results are significant, which is reflected in early diagnosis, socially provided rehabilitation and treatment, the demand for the Center in Ukraine and other CIS countries.

Biography

Olena Grechanina has completed her PhD at the age of 24 years from National Medical University (the Department of General Medicine) and postdoctoral studies from National Medical University (the Department of Obstetrics and Gynecology). She is the General Director of Kharkiv Interregional Specialized Medical Genetic Center – Center of Rare (Orphan) Diseases, the Member-Correspondent of National Academy of Medical Sciences, Professor of The Department of Medical Genetics, MD.

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