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The experience of preconceptional care and pre and postnatal specifying diagnosis as a consistent system of inborn inherited pathology prevention

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Preconceptional care, prenatal programming, prenatal fetal education, neonatal screening and specifying diagnosis in families with genetically caused disorders of the reproductive function, the treatment of intrauterine fetus makes a basis of the scientific and practical work of author for the last 50 years in Medical Genetic Center and at Gynecology and Obstetrics Departments. Consistent system of prenatal medical genetic consultation and prenatal diagnosis has been developed, which includes specifying diagnosis of the inherited pathology in parents and a fetus by classical genetic methods. The families are included in a life-long monitoring that has given the possibility to consult three generations in many cases. 10 precepts of preconceptional care have been made and introduced. Population studies of mitochondrial dysfunction frequency, folate-methionine cycle disorders have been conducted and the distribution of hetero and homozygous compounds have been determined in MTHFR (C676T, A1298C, G1793A; MTRR (A66G); RFC1 (G80A) in Ukraine. The spectrum of the expected inherited pathology has been described on this basis. The system to help yet unborn child in families, which are burdened by thrombotic conditions, which has a high positive result in thrombophilic complications, intrauterine fetal infections and the disorders of the central nervous system. Study of the efficacy of neonatal and selective screening of PKU, MPS, hypothyroidism, adrenal-genital syndrome, Fabry disease, Gauche disease takes a special place, their efficacy is being increased by a life-long monitoring of a family. The phenomenon of comorbidity, pheno-genotypic syntropy, which are currently important, are being studied in present days.

Biography

Olena Crechanina has completed her PhD at National Medical University, Department of General Medicine and Postdoctoral studies at 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, the Member-Correspondent of National Academy of Medical Sciences and the Professor of Department of Medical Genetics.

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