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Fund “Nauka” Project № 24022 Resume – Autumn Competition-based Session 2024:

“Bleeding phenotype in girls and women, heterozygotes for the Hemophilia A and Hemophilia B gene”

Project leader: assoc. prof. doctor Milena Ivanova Belcheva, PhD

The study aims to clinically analyze the bleeding phenotype in girls and women heterozygous for the hemophilia A/ hemophilia B gene, its correlation with underlying genetic mutations, and to define algorithms for assessing the risk of significant bleeding and for clinical management of individuals in this target group.

The main objectives of the study are:
(1) Conduction of a comparative analysis of bleeding manifestations in hemophilia carriers (HC) and healthy controls;
(2) Determination of the correlation between bleeding phenotype and coagulation status, plasma factor VIII/IX levels and genetic mutation in carriers of hemophilia;
(3) Joint status assessment;
(4) Analysis of the impact of bleeding on the quality of life of HC;
(5) Development of an algorithm for hemorrhagic risk assessment and an algorithm for prophylactic and therapeutic management of HC.

The study enrolls 50 carriers of the hemophilia gene and 40 age-matched healthy controls.

Study methods include bleeding assessment questionnaires and instruments, quality of life questionnaire, laboratory, genetic and imaging tests.

The results will be used to develop predictive models of bleeding risk in HC and to individualize prophylactic and therapeutic care.

In Bulgaria there is no established program for diagnosis and follow-up of hemophilia carriers. This study will create an opportunity to unify the behavior towards women from the families of patients with hemophilia, will create conditions for the introduction of a multidisciplinary approach in their care and will raise awareness about this specific problem.

Очаквани резултати от проекта:
(1) Development of risk assessment tools of bleeding in girls and women heterozygous for the Hemophilia A/ Hemophilia B gene and an algorithm for individualized prophylactic and therapeutic management of this specific group;

(2) Enriched knowledge of the correlation between the bleeding phenotype and the type of genetic mutation in hemophilia carriers;
(3) Raised awareness among participants and their relatives about the nature of the carrier, the need for follow-up and counselling about possible bleeding risks;
(4) Creating conditions for optimizing the multidisciplinary approach in the care of patients with hemophilia and their families in the Expert Centers for Coagulopathies.