DNA Technologies in Medicine and Sport

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Research Trend

- Structural and functional organization of genomes in plants, animals, microorganisms and human;
- Genetic and cell engineering;
- Development of DNA-technologies for agriculture, health protection, sport and environmental control;
- Biosafety problems.
It is known that

- **70-80%** of «personality» is defined by hereditary information (DNA, genotype);
- **20-30%** is defined by of life style (environmental factors).
- **Personal technology** - a choice of therapeutic modalities, based on genetic characteristics of a person;

- **Predictive technology** – predicting the development of future human diseases before first symptoms appear;

- **Preventive (precautionary) technology** – carrying out preventive measures (treatment) in regard to potential, predicted diseases;

- **Participative technology** – active participation of a patient in the prevention of possible diseases and their treatment.
Personalized medicine

The goal of modern medicine is not to cure disease, but to treat a patient. Drugs should be administered according to genotypes (at present there are more than 50 medications that require genotyping for their use).
At present, anticoagulant Warfarin is widely used as a preventive measure for Ischemic stroke and thromboembolia of the pulmonary artery.

In 2004 Warfarin was used in the USA after myocardial infarction for 31 million people. **Complications, including deaths, were observed in 16% of cases.**
Частота в популяции

Высокая чувствительность к варфарину

«Нормальная» чувствительность к варфарину

Resistance to Warfarin

Поддерживающая доза варфарина, мг/сутки

Warfarin maintenance dose (milligram per day)
Predictive Medicine is based on the risk detection of multifactorial diseases before first symptoms appear.

We investigate genetic mechanisms of some dangerous and widespread multifactor pathologies and we use the most informative genes for genetic predisposition evaluation.
Technologies for DNA-diagnostics of genetic predisposition to multifactorial diseases were worked out.

- Cardiovascular diseases (thrombophilia, ischemic disease of heart, myocardial infarction, etc.);
- Venous thromboses (thrombophlebites);
- Diseases of respiration organs (bronchial asthma, allergoses);
- Endocrine diseases (second-type diabetes, obesity);
- Diseases of bone tissue metabolism (osteoporosis, rheumatoid arthritis);
- Disturbances in normal physiological process of pregnancy.

Technologies for DNA-diagnostics of congenital or hereditary hypoacusis, mitochondrial pathologies, hemochromatosis (disturbance of iron metabolism) were worked out.

In all 10 000 persons were subjected to DNA-diagnostics.
In big cities, more than 300 cases of a heart attack (infarction) per every 100 thousand inhabitants are registered every year. Myocardial infarction was considered a disease of the elderly, but an alarming trend has been traced in recent years - a heart attack is getting "younger".

We study 21 the most informative genes for heart attack genetic risk detection.
Using modern sequencing technologies, mutation carriers were found in the LMNA gene associated with the development of **dilated cardiomyopathy** and associated life-threatening syndromes.

For the first time ever, the Arg190Pro mutation of the LMNA gene was described in the human genome. Its phenotypic manifestation was studied and pathogenicity was identified.

The asymptomatic carriers (children) of mutations in the patients’ families were determined that allows to carry out the therapeutic correction of unfavorable disease outcomes.

NGS sequencing based on Illumina MiSeq

Institute of Genetics and Cytology, NAS of Belarus
The problem of habitual miscarriage is getting more and more acute.

There are a lot of reasons for pregnancy loss but it is very difficult to recognize one of them - inherited thrombophilia.
We have tested more than 3000 women with undetected miscarriage reasons. 90% of the examined women had from 1 to 4 risk factors (unfavorable genes) in their genotypes.

Genotype frequency distribution with miscarriage risk factors.
Detection of genetic predisposition to inherited thrombophilia allows to recommend appropriate preventive therapy, resulting in the normal pregnancy course without complications.

Hundreds of women, we examined a year ago or earlier, have become mothers thanks to predicted and preventive medicine. Many women are in their late pregnancy, which also allows to hope for a successful birth of babies.
GENETIC TECHNOLOGIES IN SPORTS
DNA-testing of Top-qualification Athletes

More than 500 representatives of 30 Olympic & National teams of the Republic of Belarus in various sports have been tested:

• cyclic sports;
• complex coordination sports;
• playing sports (team sports).
«DNA-TESTING SYSTEM FOR ATHLETES»
DEVELOPED

- **Cardiovascular system genes:**
  - Gene subgroup of blood-vessel endothelium growth;
  - Thrombosis risk factor subgroup.

- **Oxygen transport system genes;**

- **Fragility of bone genes;**

- **Carbohydrate-adipose genes;**

- **Psychomotorial state genes.**
Received results lead to the conclusion that in order to become an outstanding athlete you should have corresponding genetic potential.
Risk Detection of Genetic Professional Pathologies in Athletes

It is a known fact that sport is often fraught with grave consequences for a human organism: football and hockey players frequently have leg venous thrombosis, boxers - brain injuries, gymnasts suffer from tears of connective tissues and bone fractures, development of cardiac hypertrophy in athletes and even sudden death syndrome.
Knowing genetic risks of these pathologies, it is possible to carry out their prevention before the onset of first clinical symptoms and thus preventing the development of these diseases in athletes.
THE REPUBLICAN CENTRE FOR GENETIC MARKING AND CERTIFICATION OF PLANTS, ANIMALS, MICROORGANISMS AND HUMANS

at the Institute of Genetics and Cytology of the NASB

Put into operation in 2011

- Accredited by ISO 17025-2007
- Licence of the Ministry of Health to the right to carry out medical activities
FIELD OF ACCREDITATION

- Detection of genetically modified ingredients (GMIs) in alimentary raw material and food products
- Detection of GMIs in agricultural products, feeds and seed material
- Determination of DNA markers for identification and certification of agricultural crop varieties
- Identification of genes responsible for economically valuable traits and hereditary diseases of animals
- Identification of genes responsible for various individual characteristics of a person
- Species identification of animal and plant origin ingredients
- Molecular genetic verification of origin, species and pedigree belonging of animals
Contracts were concluded with:
UAB Carzap Baltic, Lithuania – carrying out of genetic certification

Onorach Baltics (clinical trials), Latvia – determination of the risk of miscarriage

Customers of genetic testing services: citizens of Russia, Ukraine, Spain, Estonia, Italy, Moldova, Tajikistan, Denmark, Lithuania, China, Belgium, Turkey, Libya, Macedonia, Mauritius, Latvia, etc.
A project proposal is being prepared for the Horizon-2020 program

**Theme of the project:** “Interrelation of the Intestinal Microbiome, Diet and Health”

**Subproject:** “Genetic Basis of Active Ageing and Longevity in Belarus and Sardinia”

**Partner from Sardinia:** Università degli studi di Cagliari
Areas of cooperation

Development of DNA-testing technologies for genetic predisposition of humans to diseases.

Genetic markers of monogenic cardiological diseases (channelopathies).

Study in the polymorphism of genes encoding the proteins of the xenobiotics biotransformation system; immune, sympathetic-adrenal and renin-angiotensin systems in children with atopic pathology.

Diagnostic mutations’ search in patients with hypertrophic cardiomyopathy.

In vitro study of the functional characteristics of A/C lamins carrying a mutation that was first identified in patients with dilated cardiomyopathy.

Identification of genetic predisposition to high sporting achievements and resistance to physical stress.
THANK YOU FOR YOUR ATTENTION