



ТОМСКИЙ НИИ  
НИИ  
МЕДИЦИНСКОЙ  
ГЕНЕТИКИ

November 2022

ТОМСК

2022

conference.medgenetics.ru



## The program of the XIII Scientific Conference "Human Genetics and Pathology"

NOVEMBER 2022  
T O M S K  
2022



20 November 2022

## 10.00 - 11.20 Symposia

### Aud. 1

Omics technologies in medical genetics

#### Chairpersons:

Nazarenko M.S. (Tomsk),  
Lagarkova M.A. (Moscow)

10.00-10.20

**Nazarenko M.S. (Tomsk)**

Omics approach to molecular profiling of  
atherosclerotic plaque

10.20-10.40

**Cherdyntseva N.V. (Tomsk)**

Molecular genetic aspects of drug therapy of  
malignant tumors

10.40-11.00

**Trifonova E.A. (Tomsk)**

Preeclampsia: genomics and transcriptomics  
perspectives on disease pathogenesis

11.00-11.20

**Seitova G.N. (Tomsk)**

Fabry disease: a modern view of the problem

### Aud. 2

Genetics and epigenetics of development  
(Symposium in memory of V.S. Baranov,  
RAS Corresponding Member, and S.A.  
Nazarenko, RAS Corresponding Member)

#### Chairpersons:

Lebedev I.N. (Tomsk),  
Glotov A.S. (St.Petersburg)

10.00-10.20

**Glotov A.S. (St.Petersburg)**

Baranov V.S. Department of Genomic Medicine:  
from the past to the future

10.20-10.40

**Lebedev I.N. (Tomsk)**

Progress in cytogenetics of embryo  
development and advances in reproductive  
medicine (In memory of Nazarenko S.A.)

10.40-10.55

**Pendina A.A., Efimova O.A. (St. Petersburg)**

In memory of Baranov V.S. Ode to chromosomes:  
from cytogenetics to cytogenomics (online)

10.55-11.05

**Pchelina S.N., Gorbunova V.N. (St. Petersburg)**

In memory of our friends and colleagues:  
Vladislav S. Baranov and Evgeny I. Schwartz  
in the foundation of medical genetics  
in St. Petersburg

11.05-11.20

**Maslennikov A.B. (Novosibirsk)**

Contribution of Baranov V.S. and Nazarenko S.A.  
to the organization of interaction between  
federal centers and regional medical genetic  
services

## 11.20 - 11.30 Coffee break

## 11.30 - 13.05 Symposia

11.30-11.45

**Beliaeva S.A., Goncharova I.A. (Tomsk)** Genetic  
and epigenetic factors of comorbidity of  
ascending aortic aneurysm and coronary artery  
atherosclerosis

11.30-11.50

**Vasilyev S.A. (Tomsk)**

Epigenetics and cytogenetics of the placenta

20.11.2022

## 11.30 - 13.05 Symposia

### Aud. 1

11.45-12.00

**Yalaev B.I., Khusainova R.I. (Ufa)**

Study of genetic and epigenetic factors in the development of primary osteoporosis in men and women of the Volga-Ural region

12.00-12.15

**Davydova Yu.D. (Ufa)**

Association of DNA methylation and polymorphic loci of hypothalamic-pituitary-adrenal genes with phenotypic variations in the level of depression (online)

12.15-12.35

**Freidin M.B. (London, UK)**

Genetic and omics studies of pain syndromes (online)

12.35-13.00

**Aulchenko Yu.S. (Novosibirsk)**

Genetic control of population diversity of N-glycosylation of human proteins (online)

13.00-13.05

**Discussion**

### Aud. 2

11.50-12.05

**Demeneva V.V. (Tomsk)**

LINE-1 retrotransposon methylation level in the placenta of the first trimester of pregnancy

12.05-12.25

**Shevchenko A.I. (Novosibirsk)**

Epigenetic regulation of X chromosome inactivation

12.25-12.40

**Fonova E.A. (Tomsk)**

A modifying effect of X chromosome inactivation on CNVs clinical manifestation

12.40-13.00

**Moshkin Yu. M. (Geneva, Switzerland)**

IVF problems and destabilization of prenatal development (online)

13.00-13.05

**Discussion**

## 13.05 - 14.00 Lunch

## 14.00 - 14.45 Grand opening of the conference

**Chairpersons:** Stepanov V.A.,  
Puzyrev V.P.

## 14.45 - 15.30 Plenary lecture. Puzyrev V.P. (Tomsk)

Origins and the Soviet period in the history of the Tomsk Institute of Medical Genetics

## 15.45 - 17.20 Symposia

### Genetics of common diseases

#### **Председатели:**

Puzyrev V.P. (Tomsk),  
Polonikov A.V. (Kursk)

15.45-16.05

**Bragina E.Yu. (Tomsk)**

Genetic aspects of the phenomenon of comorbid diseases

### Cytogenetics and chromosomal diseases

#### **Председатели:**

Vasilyev S.A. (Tomsk),  
Rubtsov N.B. (Novosibirsk)

15.45-16.05

**Rubtsov N.B. (Novosibirsk)**

Peculiarities of the diagnostics of balanced chromosome rearrangements

20.11.2022



## Aud. 1

16.05-16.25

**Ivanova S.A., Fedorenko O.Yu., Bokhan N.A. (Tomsk)**

Genetics and pharmacogenetics of mental disorders

16.25-16.45

**Moshkin M.P. (Novosibirsk)**

Circadian desynchronosis in the polar regions inhabitants and genetic variability (online)

16.45-17.05

**Moskalev E.A. (Erlangen, Germany)**

The role of non-canonical mitotic inheritance in the development of oncological diseases (online)

17.05-17.20

**Voronin S.V. (Moscow)**

Current aspects of diagnosis and therapy of hypophosphatasia (with the sponsorship of Astra Zeneca company)

## Aud. 2

16.05-16.20

**Gridina M.M. (Novosibirsk)**

Detection of structural variants in the human genome using 3C-methods

16.20-16.40

**Kashevarova A.A. (Tomsk)**

Pleiotropic effects of CNV in human ontogenesis

16.40-16.50

**Nikonov A.M. (Barnaul)**

Mucopolysaccharidosis type II: Diagnosis and treatment

16.50-17.05

**Belyaeva E.O., Fonova E.A. (Tomsk)**

Exome sequencing in patients with CNV: problems of clinical significance interpretation of microstructural chromosomal variants

17.05-17.20

**Minzhenkova M.E. (Moscow)**

Complex genomic rearrangements in the etiology of the "chromosomal phenotype"

## 17.20 - 17.35 Coffee break

17.35-17.55

**Polonikov A.V., Azarova Iu.E. (Kursk)**

Genetic determinants of glutathione metabolism disorders in multifactorial pathology: focus on cardiometabolic diseases

17.55-18.15

**Solodilova M.A., Medvedeva M.V. (Kursk)**

Contribution of gene polymorphisms of vascular endothelial growth factors and their receptors to the development of coronary heart disease (online)

18.15-18.35

**Rzhetsky A. (Chicago, USA)**

Perspectives on multifactorial diseases through the prism of big clinical data (online)

18.35-18.40

**Discussion**

17.35-17.50

**Chernykh V.B., Oparina N.V. (Moscow)**

Gonosomal anomalies and mosaicism: variability in the system of X and Y coordinates

17.50-18.05

**Tolmacheva E.N. (Tomsk)**

Contribution of X-linked CNVs to intellectual disorders formation

18.05-18.20

**Markova Zh.G. (Moscow)**

Common polymorphic variants in 17q21.31: pitfalls of chromosomal microarray analysis

18.20-18.35

**Yurchenko D.A. (Moscow)**

Wolf-Hirschhorn Syndrome: Perspectives on Etiopathogenesis "in the retrospect of time"

18.35-18.40

**Discussion**

## 18.45 - 19.30 Evening lecture. Tarabykin V.S. (Nizhny Novgorod)

My Road in Science

20.11.2022

## 16.00 - 20.10 Round Table with the Heads of Medical Genetic Services

### Medical care for patients with hereditary diseases

(Genetics Clinic, Moskovsky Trakt 3)

**Chairpersons:** Nazarenko L.P. (Tomsk),  
Bushueva T.V. (Moscow)

16.00-16.15

**Minaycheva L.I. (Tomsk)**

Phenylketonuria. Peculiarities in adolescents and in pregnancy management

16.15-16.30

**Maslennikov A.B. (Novosibirsk)**

Molecular and genetic peculiarities of phenylketonuria in the Siberian Federal District

16.30-16.45

**Seitova G.N. (Tomsk)**

Use of sapropterin in clinical practice

16.45-17.00

**Nazarenko L.P. (Tomsk)**

Difficult patient: Niemann-Pick disease A, B (with the sponsorship of Sanofi company)

17.00-17.30

**Maksimova Yu.V. (Novosibirsk)**

Workshop. Diagnostics of spinal muscular atrophy in an adult patient  
(with the sponsorship of Johnson & Johnson company) (online)

17.30-17.45

**Kondakova O.B. (Moscow)**

Hereditary diseases caused by primary disorders of neurotransmitter synthesis

17.45-18.30

**Discussion.** Preparation of subjects of the Russian Federation for advanced neonatal screening

18.30-20.10

**Associated School:**

"Organization of medical care for patients with phenylketonuria"  
(with the sponsorship of Biomarin company)

20.11.2022

### **Associated School:**

"Organization of medical care for patients with phenylketonuria"  
(with the sponsorship of Biomarin company)

**Meeting with experts:** Modern approaches to the pharmacotherapy of phenylketonuria

**Chairpersons:** Nazarenko L.P., Seitova G.N. (Tomsk), Bushueva T.V. (Moscow)

**Seitova G.N., Bushueva T.V.**

Opening speech of the chairpersons (5 min)

**Shestopalova E.A. (online)**

Board Certified in Genetics, Research Centre for Medical Genetics (Moscow)

Data of FKU audit (25 min)

**Bushueva T.V.**

Ph.D., Leading Researcher, National Medical Center for Children's Health (Moscow)

Pharmacotherapy of PKU - for which category of patients and why it is important (20 min)

**Seitova G.N.**

Ph.D., Chief Physician of Genetics Clinic of Research Institute of Medical Genetics, Tomsk Scientific Center (Tomsk)

Routing of a patient with PKU. Performing the stress test in the hospital and the possibility of outpatient testing (20 min)

**Kuzmicheva I.A.**

Head of Genetics Department, Kaluga Regional Hospital (Kaluga)

New strategies in the treatment of PKU. Start of enzyme replacement therapy for PKU in Russia.

Portrait of a patient (25 min)

### **Discussion**



## 21 November 2022

### 09.00 - 11.20 Symposia

#### Aud. 1

##### Clinical genetics and orphan diseases

#### Chairpersons:

Nazarenko L.P. (Tomsk),  
Zaklyazminskaya E.V. (Moscow)

9.00-9.20

**Zaklyazminskaya E.V., Sadekova M.A.,  
Dzemeshevich S.L. (Moscow)**

Open questions in the genetics of hypertrophic cardiomyopathy

9.20-9.40

**Pchelina S.N. (St. Petersburg)**

The role of lipid metabolism in the development of Parkinson's disease

9.40-10.00

**Izhevskaya V.L. (Moscow)**

Genomic testing: a cross-talk of medical geneticist and patient

10.00-10.20

**Zakharova E.Y. (Moscow)**

Modern state and problems of diagnostics of hereditary metabolic disorders (online)

10.20-10.40

**Larionova V.I. (St. Petersburg)**

Molecular and metabolic basis of drug selection for treatment of hereditary diseases. What should a pediatrician know?

10.40-10.55

**Bushueva T.V. (Moscow)**

New strategies in the treatment of phenylketonuria (with the sponsorship of Biomarin company)

#### Aud. 2

##### Evolutionary and population genetics

#### Chairpersons:

Stepanov V.A. (Tomsk),  
Yankovsky N.K. (Moscow)

9.00-9.30

**Yankovsky N.K. (Moscow)**

Results of the Union State Program "DNA Identification 2017-2021"

9.30-9.50

**Kharkov V.N. (Tomsk)**

Population genomics of the indigenous population of Southern and Western Siberia

9.50-10.20

**Kim Hie Lim (Singapore)**

Prehistory human migration between Sundaland and South Asia driven by rapid sea-level rise (online)

10.20-10.40

**Lavryashina M.B. (Kemerovo)**

Dynamics of the surname pool of Siberian Tatars: four ethnographic groups in three generations

10.40-10.55

**Valikhova L.V. (Tomsk)**

Structure of the gene pool of Tomsk Tatars according to Y-chromosome markers

### 11.00 - 11.15 Coffee break

11.15-11.35

**Maksimova N.R., Sukhomyasova A.L. (Yakutsk)**

Genomic studies of genetically isolated Yakut population (online)

11.35-11.55

**Zhalsanova I.Zh. (Tomsk)**

Spectrum of mutations in ATP7B gene in patients with Wilson's disease in Siberian region

11.15-11.30

**Kolesnikov N.A. (Tomsk)**

Directional selection signals in indigenous populations of Central Asia

11.30-11.45

**Golubenkov M.V. (Tomsk)**

Peculiarities of mtDNA variability in terms of adaptation to environmental conditions and diseases susceptibility

## **Aud. 1**

11.55-12.10

**Zaripova A.R. (Ufa)**

Spectrum and frequency of pathogenic changes in patients with Osteogenesis Imperfecta from the Republic of Bashkortostan

12.10-12.25

**Kadnikova V.A. (Moscow)**

Spastic Paraplegia Type 47 in Russian patients

12.25-12.40

**Ismagilova O.R. (Moscow)**

Molecular genetic analysis of the Rubinstein-Taybi syndrome in the Russian Federation

12.40-12.55

**Postrigen A.E. (Tomsk)**

Molecular genetic diagnosis of long QT syndrome

12.55-13.10

**Kazarian R. (Moscow)**

Differential diagnosis of cystic fibrosis and Shwachman-Diamond syndrome

13.10-13.15

**Discussion**

## **Aud. 2**

11.45-12.00

**Nazarenko L.P. (Tomsk)**

Genetic aspects of Pompe disease with clinical cases review  
(with the sponsorship of Sanofi company)

12.00-12.15

**Serebrova V.N. (Tomsk)**

Replicative studies of hereditary thrombophilia and endothelial dysfunction factors in pre-eclampsia development

12.15-12.30

**Babovskaya A.A. (Tomsk)**

Identification of population characteristics of the decidual cell transcriptome in severe preeclampsia

12.30-12.45

**Gavrilenko M.M. (Tomsk)**

Landscape of alternative splicing in placental decidual cells in Russians

12.45-13.00

**Nazarenko L.P. (Tomsk)**

Alagille syndrome. Clinical and genetic diagnosis  
(with the sponsorship of FarmaMondo company)

13.00-13.10

**Krasnobaeva A. (Novosibirsk)**

High-quality reagents means a reliable result

13.10-13.15

**Discussion**

## **13.15 - 14.00 Lunch**

## **14.00 - 14.45 Plenary lecture. Stepanov V.A. (Tomsk)**

Population genomics, medical genetics and evolutionary medicine

## **15.00 - 17.30 Symposia**

### **Clinical genetics and orphan diseases**

#### **Chairpersons:**

Seitova G.N. (Tomsk),

Larionova V.I. (St. Petersburg)

15.00-15.20

**Polyakov A.V. (Moscow)**

Preparation for validation phase of advanced neonatal screening

### **Prenatal and preimplantation genetic testing**

#### **Chairpersons:**

Lebedev I.N. (Tomsk),

Svetlakov A.V. (Krasnoyarsk)

15.00-15.20

**Vermeesch J. (Leuven, Belgium)**

Mapping the genomic landscape to improve prenatal care (online)

**21.11.2022**

## **Aud. 1**

15.20-15.40

**Kondratyeva E.I. (Moscow)**

Cystic fibrosis as a model for diagnosis and personalized therapy

15.40-15.55

**Kovalskaya V.A. (Moscow)**

Genotype verification in cystic fibrosis as an important step in preparation for targeted therapy with Orcambi (online)

15.55-16.10

**Pinegina Iu.S. (Novosibirsk)**

Compliance to therapy in patients with cystic fibrosis. Experience of the Novosibirsk region

16.10-16.25

**Stepanova A.A., Polyakov A.V. (Moscow)**

Selective screening of patients with hereditary retinal dystrophies to form a target group for gene therapy

16.25-16.40

**Orlov D.S. (Tomsk)**

Experience of introducing the tandem mass spectrometry into the practice of selective homocystinuria screening

16.40-16.55

**Odinokova O.N. (Tomsk)**

Molecular diagnosis of Duchenne muscular dystrophy

16.55-17.10

**Zinina E.V. (Moscow)**

Study of the spectrum of mutations in the *DMD* gene as a basis for pathogenetic therapy planning in Russian patients with Duchenne/Becker muscular dystrophy

17.10-17.25

**Chausova P.A. (Moscow)**

Merosin-deficient congenital muscular dystrophy. From diagnosis to treatment perspectives (online)

17.25-17.30

**Discussion**

## **Aud. 2**

15.20-15.40

**Liehr T. (Jena, Germany)**

Limits and possibilities of non-invasive prenatal testing (NIPT) (online)

15.40-16.00

**Shilova N.V. (Moscow)**

Towards cellular NIPT: Detection of trophoblast cells in maternal cervical specimens

16.00-16.15

**Zhigalina D.I. (Tomsk)**

Digital karyotyping of blastocysts based on RNA sequencing

16.15-16.30

**Stepanchuk Ya.K. (Novosibirsk)**

Hi-C for pre-implantation genetic testing

16.30-16.45

**Skleimova M.M., Solovieva E.V. (Tomsk)**

Preimplantation genetic testing for spinocerebellar ataxia type 1

16.45-17.00

**Gayner T.A. (Novosibirsk)**

Prenatal diagnostics: 10 years of experience (online)

17.00-17.10

**Kornikova E. (Moscow)**

Illumina technologies: the news and the perspectives

17.10-17.15

**Discussion**

**17.30 - 17.45 Coffee break**

**17.45 - 18.30 Evening lecture. Kozlov A.P. (St. Petersburg)**

New biological theory - carcino-evo-devo theory, its non-trivial predictions and relations with other biological theories

**19.00 Furshet**

21.11.2022

## 22 November 2022

### 09.00 - 11.35 Symposia

#### Aud. 1

##### Modeling of hereditary diseases

##### Chairpersons:

Nikitina T.V. (Tomsk),  
Serov O.L. (Novosibirsk)

9.00-9.20

**Serov O.L., Shnaider T.A. (Novosibirsk)**

Novel role of CNTN6 in human neurogenesis

9.20-9.40

**Tarabykin V.S. (Nizhny Novgorod)**

Mouse models of neurological diseases

9.40-10.00

**Lagarkova M.A. (Moscow)**

Pluripotent stem cells. 15 years of IPSC

10.00-10.15

**Zakharova I.S. (Novosibirsk)**

Creation of cell models of familial  
hypercholesterolemia

10.15-10.30

**Nazarenko L.P. (Tomsk)**

Aromatic L-amino acid decarboxylase deficiency  
(AADCD) (with the sponsorship of PTC pharma  
company)

10.30-10.45

**Kopatova A.E., Emelyanov A.K. (St. Petersburg)**

Development of targeted therapy for  
Parkinson's disease using 3D modeling

10.45-11.00

**Savchenko R.R. (Tomsk)**

Influence of differential expression of  
*ADAMTS1* and *THBS1* genes on  
radiation-induced response of human somatic  
cells *in vitro*

11.00-11.15

**Vasiliev F.F. (Yakutsk)**

Study of VPS33A protein functions in  
mucopolysaccharidosis-plus syndrome (online)

11.15-11.30

**Nuriddinov M.A. (Novosibirsk)**

Modeling of the spatial organization of  
chromatin with chromosomal rearrangements

11.30-11.35

**Discussion**

#### Aud. 2

##### Bioinformatics

##### Chairpersons:

Skryabin N.A. (Tomsk),  
Fishman V.S. (Novosibirsk)

9.00-9.30

**Kolchanov N.A. (Novosibirsk)**

Bioinformatics and medicine in the context of big  
genetic data (online)

9.30-9.50

**Ryzhkova O.P. (Moscow)**

Peculiarities of clinical interpretation of NGS  
results

9.50-10.10

**Fishman V.S. (Novosibirsk)**

Interpretation of genomic variants in non-coding  
DNA sequences using machine learning methods

10.10-10.30

**Pomaznov M.Yu. (Novosibirsk)**

Processing of sequencing data of patients with  
monogenic and oncological diseases using the  
NGS Wizard

10.30-10.50

**Kononov F.A. (Moscow)**

Detection of chromosomal rearrangements by  
routine exome and genome sequencing (online)

10.50-11.10

**Zarubin A.A. (Tomsk)**

The dragon may be small, but it's still a dragon.  
Experience of using DRAGON on NextSeq 2000

11.10-11.30

**Khozyainova A.A. (Tomsk)**

Multimic analysis of single cell sequencing data:  
from theory to practice

11.30-11.35

**Discussion**



## 11.35 - 11.45 Coffee break

## 11.45 - 13.05 Symposia

### Aud. 1

#### Modeling of hereditary diseases

##### Chairpersons:

Nikitina T.V. (Tomsk),  
Serov O.L. (Novosibirsk)

11.45-12.00

**Skryabin N.A. (Tomsk)**

Modeling of Wilson's disease using cell technologies

12.00-12.15

**Goldshtein D.V. (Moscow)**

Technologies of application of intestinal organoids for diagnostics and selection of cystic fibrosis therapy (online)

12.15-12.30

**Demchenko A.G. (Moscow)**

Lung organelles and cultures of ciliated epithelial cells as models for the diagnosis and treatment of hereditary lung diseases (online)

12.30-12.45

**Skoblov M.Yu. (Moscow)**

Genome-wide screening of complex splice alleles

12.45-13.00

**Salakhov R.R. (Tomsk)**

Functional analysis of variants in non-coding sites of MYBPC3 gene in hypertrophic cardiomyopathy

13.00-13.05

Discussion

### Aud. 2

#### Cytogenetics and chromosomal diseases

##### Chairpersons:

Kashevarova A.A. (Tomsk),  
Shilova N.V. (Moscow)

11.45-12.00

**Fedotov D.A. (Tomsk)**

CNV structure in disorders of psychomotor development and miscarriages

12.00-12.15

**Drozdov G.V. (Tomsk)**

CNV incidence and spectrum in early embryonic death

12.15-12.30

**Tonyan Z.N. (St. Petersburg)**

Analysis of chromosome segregation in carriers of reciprocal translocations (online)

12.30-12.45

**Krivoshchapova Ya.V. (Chelyabinsk)**

Influence of chronic radiation exposure on telomeric chromosome segments in human peripheral blood T-cells

12.45-13.00

**Vozilova A.V. (Chelyabinsk)**

Cytogenetic diagnostics of chronic lymphoproliferative diseases using locus-specific fluorescent probes

13.00-13.05

Discussion

## 13.05 - 14.00 Lunch

## 14.00 - 17.30 Symposia

#### Clinical genetics and orphan diseases

##### Chairpersons:

Skryabin N.A. (Tomsk),  
Polyakov A.V. (Moscow)

14.00-14.15

**Voronkova A.Yu. (Moscow)**

Primary ciliary dyskinesia. (online)

#### Genetics and epigenetics of multifactorial diseases

##### Chairpersons:

Kucher A.N. (Tomsk),  
Pchelina S.N. (St. Petersburg)

14.00-14.15

**Bocharova A.V. (Tomsk)**

Intergenic interactions in human disorders with cognitive impairment

22.11.2022

## **Aud. 1**

14.15-14.30

**Kotalevskaya Yu.Yu. (Moscow)**

Rare syndromal epidermolysis bullosa.  
Clinical cases

14.30-14.45

**Orlova A.A. (Moscow)**

Ultra-rare form of hereditary cardiomyopathy:  
the first presentation of a patient from the  
Russian Federation

14.45-15.00

**Valiakhmetov N.R. (Tomsk)**

Variability of sarcomeric proteins genes in  
myocardial hypertrophy of various genesis

15.00-15.15

**Kuzenkova L.M. (Moscow)**

Alpha-mannosidosis - a rare disease, the  
frequent symptoms (with the sponsorship of  
Chiesi company)

15.15-15.30

**Pobedinskaya A.I. (Chelyabinsk)**

Clinical case of congenital sucrase-isomaltase  
deficiency

## **Aud. 2**

14.15-14.30

**Nikolaev M.A. (St. Petersburg)**

Glucocerebrosidase dysfunction and  
alpha-synuclein accumulation is a  
pathophysiological duo in GBA-associated  
Parkinson's disease

14.30-14.45

**Usenko T.S. (St. Petersburg)**

Selective inhibition of LRRK2 kinase activity as an  
approach for therapy of Parkinson's disease,  
associated with GBA mutations

14.45-14.55

**Seitova G.N. (Tomsk)**

Therapy for hereditary angioedema

14.55-15.10

**Miroshnikova V.V. (St. Petersburg)**

Extracellular vesicles of adipose tissue: the search  
for new biomarkers of lipid and carbohydrate  
metabolism dysfunction in obesity

15.10-15.25

**Sirotkina O.V. (St. Petersburg)**

MicroRNAs miR-221 and miR-223 in blood plasma  
microvesicles – promising biomarkers of  
post-thromboembolic syndrome severity

## **15.30 - 15.45 Coffee break**

15.45-16.00

**Yakovleva A.E. (Yakutsk)**

Genotype-Phenotype analysis in patients with  
multiple exostose chondrodysplasia in the Yakut  
population

16.00-16.15

**Mikhalechuk K.A. (Moscow)**

Spectrum of minor variants of the SMN locus  
(online)

16.15-16.30

**Shestak A.G. (Moscow)**

Representation of the phenomenon of "allelic  
dropout" in sequencing results

16.30-16.40

**Bondar V. (Moscow)**

Fundamentals and possible applications of the  
droplet digital PCR

16.40-16.55

**Ogorodova N.Yu. (Moscow)**

The variety of skeletal dysplasias, caused by *SLC26A2*  
mutations

15.45-16.00

**Sleptcov A.A. (Tomsk)**

T-cell landscape in atherosclerosis

16.00-16.15

**Koroleva Iu.A. (Tomsk)**

Methylation of regulatory elements of microRNA  
genes in unstable atherosclerosis

16.15-16.30

**Babushkina N.P. (Tomsk)**

Methylation status of the regulatory regions of  
the *ATM* and *MLH1* genes depending on their  
polymorphisms in age-dependent pathologies

16.30-16.45

**Rykova E.Yu. (Novosibirsk)**

Genome-wide search for SNPs in microRNA target  
sites associated with individual drug sensitivity

### **Aud. 1**

16.55-17.10

**Shatokhina O.L. (Moscow)**

Secondary findings identified by exome sequencing (online)

17.10-17.25

**Zabnenkova V.V. (Moscow)**

When the patient is a mystery: combined exome sequencing findings (online)

17.25-17.30

**Discussion**

### **Aud. 2**

16.45-17.00

**Gervas P.A. (Tomsk)**

Molecular genetic testing for prescribing targeted therapy for malignant tumors

17.00-17.20

**Salumets A. (Tartu, Estonia)**

Genomic evaluation of endometrial tissue at assisted reproduction (online)

17.20-17.25

**Discussion**

## **17.30-18.00 Closing of the conference**