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Winter School on Cytogenomics

In the early 2000s, the possibilities of classical and molecular cytogenetics in the study of chromosomal abnormalities in humans were significantly expanded by the emerging technologies of genomic analysis. The use of chromosomal microarray analysis, various modifications of next-generation sequencing, chromosome conformation capture, and optical genome mapping marked the beginning of a new cytogenomic era in cytogenetics. The combination of cytogenetic and genomic technologies has opened up new perspectives in the diagnosis of complex and submicroscopic chromosomal aberrations. The Winter School on Cytogenomics, held in Tomsk on November 25–29, 2024, was devoted to discussing these current trends.

The event was organized by the Research Institute of Medical Genetics of the Tomsk National Research Medical Center of the Russian Academy of Sciences. It was dedicated to the memory of Corresponding Member of the Russian Academy of Medical Sciences, Professor Sergey Nazarenko – founder and first head of the Laboratory of Cytogenetics at the Institute, and also coincided to the 25th Anniversary of the Medical Genetics Division of the Siberian State Medical University. Lectures were delivered by Russian and foreign experts in the field of clinical cytogenetics and cytogenomics – N. Rubtsov (Novosibirsk), I. Lebedev (Tomsk), N. Shilova (Moscow), V. Chernykh (Moscow), V. Fishman (Novosibirsk), T. Liehr (Germany), M. Zamani-Esteki (Netherlands), P. Li (USA), J. Vermeesch (Belgium). Workshops were held on chromosomal microarray analysis (CMA), fluorescent *in situ* hybridization (FISH), quantitative real time PCR, as well as on the clinical interpretation of the CMA results. Within the framework of the School, a competition for talks by young scientists was organized, the participants of which and their colleagues had the opportunity to present their research on the pages of this Issue in the section “Medical cytogenomics”.

The Issue opens with an article by V.P. Pushkarev and co-authors “Molecular genetic study of triploidy and the hydatidiform mole in pregnancy loss: analysis of 10,000 consecutive cases”. By studying a significant samples of spontaneous abortions using quantitative fluorescent PCR, the frequency of complete hydatidiform mole was determined. This pathology arises due to genomic imprinting effects caused by abnormal combination of parental haploid genomes in the zygote – specifically two paternal genomes in the absence of a maternal one. The frequency was estimated as 0.11 % and it was close to the epidemiological data typical for European populations.

The article by A.S. Iakovleva and co-authors presents a case involving a combination of low-level mosaicism for trisomy 9 and uniparental disomy of the same chromosome. The use of a CMA and FISH made it possible to describe the mosaic karyotype in detail and demonstrate that such a combination of chromosomal abnormalities is a consequence of postzygotic trisomy rescue. It is noteworthy that trisomy in the zygote arose as a result of an error in the meiosis II, as indicated by the alternation of iso- and heterodisomy sites in uniparental disomy.

The article by A.E. Kopytova and co-authors presents a family case of Xq28 chromosome duplication. The area of revealed copy number gain overlaps with the region of chromosome Xq28 duplication syndrome. However, both brothers and their mother carry a rare, non-classical duplication. That is why the use of common algorithms for classifying the clinical significance of duplication defines it as a variant of unknown clinical significance. However, given the asymmetric inactivation of the X chromosome in a mother, healthy carrier of the duplication, the authors reasonably suggest considering this duplication as pathogenic.

The article by G.D. Moskvitin and co-authors presents a case of interstitial deletion 6p22.3-p24.3 in monozygotic twins from Yakutia with severe speech delay, intellectual disability and congenital malformations. The study highlights the importance of CMA in diagnosis of chromosomal disorders and discusses the challenges in establishing genephenotype correlations.

In their article, M.M. Antonova and co-authors examine the meiotic segregation features of the paracentric inversion inv(7)(q11.23q22), one of the most frequent in the human karyotype. FISH analysis of spermatozoa in the inversion carrier allowed to establish the predominant and almost equally proportions of gametes with inversion and intact chromosome 7. Recombinant chromosomes were noted in 0.7 % of gametes only, confirming the presence of crossing over in the inversion loop.

Finally, the article by A.V. Vozilova and co-authors describes the segregation of a balanced translocation t(3;10)(p25;p15) across seven family members spanning three generations. The structure of chromosomal rearrangement and the products of its meiotic segregation, including unbalanced translocations, were investigated using CMA and clinical exome sequencing. The mechanisms of formation and clinical features of segmental aneuploidy in the terminal regions of chromosomes 3 and 10 are discussed.

The articles in the section "Medical cytogenomics" of this Issue demonstrate the potential of current cytogenomic technologies in diagnosing of chromosomal disorders and reproductive abnormalities. It is important that these technologies are available in national research centers that have presented their results at the Winter School on Cytogenomics.

I.N. Lebedev, Corresponding Member
of the Russian Academy of Sciences,
Doctor of Biological Sciences,
Chairman of the Program Committee
of the Winter School on Cytogenomics,
Executive editor of the Issue