OBITUARY

Svetlana G. Vorsanova (1945–2021)

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Les vivants ne sont que des morts qui ne sont pas encore entrés en fonction.  
Marcel Proust

On 31 August, 2021, Professor Svetlana Grigorievna Vorsanova (Fig. 1), an irreplaceable member of the Molecular Cytogenetics Editorial Board, passed away. It is to note that she was among the founders of Molecular Cytogenetics and was intrinsically involved in the developing journal’s aims, scope and original policy [1]. Once the journal was launched, she modestly took the role as a member of the Editorial Board. Still, the impact of her involvement in Molecular Cytogenetics is hard to estimate.

As a true child of her times, Svetlana lived her professional and personal life as an idealistic scientist and reflective practitioner. Even her sudden passing away due to COVID-19 confirms this statement. ‘Until the very end’, she worked on a textbook dedicated to one of the most sensitive problems of current cytogenetics, the role and applications of classical cytogenetics in current biomedicine [2]. Her life was almost completely devoted to cytogenetics as a science and practical discipline. It was not an easy life. She struggled a lot… Overcoming numerous obstacles on the road to demonstrating the importance of molecular cytogenetic diagnosis and validity of original cytogenomic/cytogenetic theories, she became a beautiful example of an intrinsic scientist paying real attention to practical issues of her science. Probably, Antonio Gramsci’s dictum — ‘pessimism of the intellect, optimism of the will’ — is the best laconic description of her modus operandi and vivendi.

Looking back at Svetlana’s walk of life, I prefer to describe briefly the professional journey of this brilliant researcher and bright person by thematic landmarks to provide her profound, albeit non-explicit, impact on biomedicine. Certainly, she would be happy, if her life’s description would be used by forthcoming generations of cytogeneticists and biomedical scientists for understanding the true meaning of their mission.

Svetlana started the long and winding road to her professional achievements by nursing. Difficult times of her adolescence obliged to combine biological education with the hard and responsible work of a surgical nurse.
In fact, her employment history began when she was 14 years old. Her work was repeatedly found remarkable by the surgeons in charge, but her ultimate purpose was to study bioscience. Eventually, she succeeded by graduating from biological faculty of Lomonosov Moscow State University 'working in parallel.'

She started her cytogenetic quest in the pre-banding era [3]. Then, Svetlana used her cytogenetic skills to analyze chromosomal behavior and variation in cells (nuclei) during cultivation. The striking outcome of these studies was uncovering progressive accumulation of chromosomal instability (suggested aneuploidization) in senescent cells or aged cell populations proposing a new cytogenetic paradigm for aging research [4, 5]. Although these findings and idea was dogmatically misunderstood, open-minded researchers found these data applicable for discovering biomarkers of cancer [6]. Now we know that these findings (discoveries) are valid, inasmuch as chromosome instability and aneuploidization are confirmed biomarkers of cellular aging and senescence. Moreover, this is valid for post-mitotic cells, as well [7]. Regardless of being a local (but important) story, it is to note that introducing of cytogenetic prenatal diagnosis in USSR is largely the result of Svetlana’s hard work [8, 9]. Here, it is to mention the way she introduced cytogenetic prenatal diagnosis in our country, because it picturesquely describes her attitude to her work. Within the short time, she was able to introduce cytogenetic techniques for prenatal diagnosis without any local technological background using her enthusiasm and assistance of a technician. On the other hand, previously, a whole laboratory specifically established to achieve the purpose of introducing parental diagnosis in USSR was not able to succeed. Additionally, Svetlana never forgot to use her practical achievements for basic studies, i.e. analysis of DNA replication in cultured amniotic fluid cells [10]. Furthermore, her true enthusiasm in opening new opportunities for cytogeneticists to uncover chromosome abnormalities led to introducing in situ hybridization methods to human cytogenetics. Unfortunately, the paper about these developments was published two years later after the submission along with other articles reporting similar data [11]. Thus, the molecular cytogenetic era began.

Interphase cytogenetics is another field of bioscience benefited from Svetlana’s work [12]. Started in the early 1990s, Svetlana’s odyssey in interphase cytogenetics resulted in proposing numerous molecular cytogenetic techniques, which were the essence of true discoveries in biomedical science [13–15]. Alternatively, this body of research was a basis for a successful book dedicated to human interphase chromosomes, in which basic research and diagnostic applications are intimately linked to each other [16]. The second edition of this book is certainly the demonstration of the success [17].

The success of Svetlana’s research in interphase molecular cytogenetics was the result of original technological developments in the field. These may be divided in two essential parts: (i) centromeric one-color/two-color/multicolor fluorescence in situ hybridization (FISH), which was used to uncover aneuploidies, to decipher marker chromosomes and to analysis of structural chromosome abnormalities involving centromeric DNAs [18–21] and (ii) specific developments in interphase chromosome analysis, i.e. quantitative FISH and interphase chromosome-specific multicolor banding (a historical help of Professor Thomas Liehr (Jena, Germany) is gratefully appreciated), which factually led to rebirth of interphase cytogenetics in the 2000s [22–28].

The former was used as a basis for molecular cytogenetics discoveries in germ-line cells [29], spontaneous abortions (e.g. discovery of intrinsic rates of chromosomal mosaicism in pregnancy losses) [30, 31], and analyzing mutagenic activity of environmental factors [32]. The latter was a basis for numerous discoveries in human genetics and neuroscience [33–42]. The most notable among these are the role of chromosome instability and aneuploidy in human neurodevelopment [43] as well as the causes and consequences of chromosomal mosaicism and instability in psychiatric diseases [44–52], neurodegeneration and aging [53–62].

A specifically important area of Svetlana’s research was Rett syndrome. During the late 1990s, she became an internationally recognized Rett syndrome researcher. More precisely, epigenetic mechanisms, genotype–phenotype correlations, mechanisms of the syndrome in males, cytogenomic causes of mutation-negative cases and the occurrence among children with intellectual disability were the essential results of her Rett syndrome studies [63–74]. These discoveries in Rett syndrome biology were the basis for the decision of holding VIII World Rett Syndrome Congress & Symposium of rare diseases in Russia [75]. Other genetic diseases, which were the focus of successful Svetlana’s research, were disorders associated with trisomy 21 (Down syndrome, mosaic trisomy 21 etc.) [76–78] and with subtelomeric deletions [79, 80].

As a true cytogeneticist, Svetlana thought and worked a lot on chromosomal heteromorphisms. Her contributions to the field are hard to overestimate (e.g. analysis of alpha- and classical satellite DNAs in situ, quantification of heteromorphisms by FISH, analysis of heteromorphisms in children with neurodevelopmental diseases) [47, 81–85].

Another local story of Svetlana’s contribution to diagnostic research that resulted in important contribution
to biomedicine was related to introducing cytogenomic microarray analysis of copy number variations (CNVs) in Russia. Svetlana was among the leaders of the research group, which was the first one that introduced microarray analysis of CNVs in Russia [86, 87]. These cytogenomic studies resulted in several discoveries, i.e. new causative CNVs for neurodevelopmental diseases and long contiguous stretches of homozygosity spanning shortly the imprinted loci as a cause for neurodevelopmental diseases [88–91]. Moreover, the term ‘cytogenomics’ in its actual meaning was introduced by Svetlana and her colleagues [92]. Finally, these cytogenomic studies opened a new big chapter of Svetlana’s research, which was roughly defined as cytogenetic/cytogenomic bioinformatics or in silico molecular cytogenetics.

Svetlana’s research merging bioinformatics and molecular cytogenetics (in silico molecular cytogenetics) was dedicated to unraveling mechanisms of diseases associated with CNVs or chromosome abnormalities, to determine causes and consequences of chromosomal and/or genomic instability, to simulate consequences of genomic variations and to interpret CNVs [93–97]. These studies were successful enough to re-consider the basis of genetic disease suggesting adding the pathway-based classification to the generally acknowledged (gene-centric) classification [98]. Additionally, CNVariome, a model for analysis of the whole set of individual or disease-specific CNVs, was developed [99]. Furthermore, the combination of advanced molecular cytogenetic technologies and bioinformatic analysis allowed to uncover a new type of chromosome instability (chromohelkosis) and to propose the ‘cytogenetic theory of everything’ [100]. In total, the body of Svetlana’s bioinformatic research yielded new biomedical direction of systems cytogenomics [101]. Moreover, these studies provided for ground-breaking achievements in medical cytogenetics (cytogenomics) — the treatment of chromosomal disorders, which are presumably incurable genetic conditions [102, 103]. Finally, paying a tribute to our time, the latest bioinformatic research of Svetlana was focused on the impact of COVID-19 on biomedical publishing [104] and cellular genomes [105]. The results of these studies became a glorious finish of bioinformatic chapter of Svetlana’s research and... life.

The latest interests of Svetlana were related to ‘syndrome-specific’ chromosomal mosaicism in neurodevelopmental diseases. Thus, Turner’s syndrome mosaicism [106] and Klinefelter syndrome mosaicism [107] were found to have an important contribution to pathogenesis of neurodevelopmental disorders. She finally analyzed the analysis of autosomal mosaics in the same large cohort of children with neurodevelopmental diseases (n ~ 10,000), but she did not managed to finish the manuscript describing the study.

Svetlana made serious efforts in elevating the educational level in cytogenetics. She received numerous thanks and acknowledgements of Russian-spoken specialists from all over the world because of her co-authored books (textbooks) [2, 108–114], which are almost the unique ones dedicated to classical cytogenetics, molecular cytogenetics and cyto(post)genomics in Russian. Her original ideas about current role of cytogenetics in bioscience were repeatedly expressed. She consistently signaled numerous problems in the field and proposed the ways to solve these problems [115–123]. It is certain that her ideas should not be forgotten.

It is not a big secret that Svetlana’s family is the core of research groups performed such a body of biomedical research. Her son, Dr. Ilia V. Soloviev, a pioneer of molecular cytogenetic and cytogenomic research (the results of his brilliant work and his ideas formed our research directions), tragically passed away in the 1990s. Her husband Professor Yuri B. Yurov passed away five years ago [124]. Unacceptable loss of Svetlana is also a grievous personal loss for me and her two granddaughters. It is hard to imagine the professional life of our two laboratories (one is named after Prof. YB Yurov and another one is named after Svetlana) without her. The only thing our labs can do is to multiply and to share the legacy of two outstanding researchers, Professors Yuri B. Yurov and Svetlana G. Vorsanova.

Declarations

Consent for publication
Authors’ original submitted files for images.

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References

1. Yurov YB, Liehr T, Shaffer LG, Iourov IY, Vorsanova SG. A new open access journal for a rapidly evolving biomedical field: introducing molecular cytogenetics. Mol Cytogenet. 2008;1:1.
2. Vorsanova SG, Iourov IV, Yurov YB. Classical clinical cytogenetics (textbook). Moscow: The publishing house of The Russian Academy of Natural History, 2021.
3. Revazov AA, Derilo TG, Vorsanova SG. Enlargement of the long arm of a B-group chromosome (Bq+) in a boy aged nine years. Sov Genet. 1973;7(8):1086–9.
4. Vorsanova SG. Dynamics of changes in anomalous human cells during prolonged cultivation in the stationary phase Trisomy 7 cells. Bull Eksp Biol Med. 1977;83(6):742–4.
5. Vorsanova SG. Stationary populations of genetically different homo- nuclear strains of human cells. Biull Eksk Biol Med. 1978;85(2):225–7.

6. Tatarinov YS, Kalashnikov VV, Vasiliev MY, Voloshuk SG, Kraevsky NA, Vorsanova SG. Human embryonic prealbumin as a marker for tumours and fibrolasts. Lancet. 1978;2(8100):1122–3.

7. Iourov IY, Yurov YB, Vorsanova SG, Kutsev SI. Chromosome instability, aging and brain diseases. Cells. 2021;10(5):1256.

8. Vorsanova SG. Method of culturing amniotic fluid cells. Biull Eksk Biol Med. 1979;87(6):625–7.

9. Vorsanova SG. Prenatal diagnosis of hereditary diseases. Med Sestra. 1988;47(7):22–5.

10. Iurov IuB, Vorsanova SG. DNA replication in cultured amniotic fluid cells. Biull Eksk Biol Med. 1981;92(9):349–52.

11. Vorsanova SG, Yurov YB, Alexandrov IA, Demidova IA, Mitkevich SP, Tirskaja AF. 18p- syndrome: an unusual case and diagnosis by in situ hybridization with chromosome 18-specific alphoid DNA sequence. Hum Genet. 1986;77(2):185–7.

12. Vorsanova SG, Yurov YB, Iourov IY. Human interphase chromosomes: a review of available molecular cytogenetic technologies. Mol Cytogenet. 2010;3:1.

13. Vorsanova SG, Iourov IuB, Deriagin GV, Solovev IV, Bytsenska GA. Method of diagnosing aneuploidies and in situ hybridization: analysis of interphase nuclei. Biull Eksk Biol Med. 1991;112(10):413–5.

14. Iourov IY, Vorsanova SG, Yurov YB. Recent patent on molecular cytogenetics. Recent Pat DNA Gene Seq. 2008;2(1):6–15.

15. Yurov IY, Vorsanova SG, Solovev IV, Iourov IB. Molecular cytogenetic methods for studying interphase chromosomes in human brain cells. Genetika. 2010;46(9):1171–4.

16. Yurov YB, Vorsanova SG, Iourov IY. Human Interphase Chromosomes: Biomedical Aspects. New York: Springer, 2013.

17. Iourov IY, Vorsanova SG, Yurov YB. Human Interphase Chromosomes: biomedical aspects (second edition). New York: Springer; 2020.

18. Vorsanova SG, Yurov YB, Passarge I, Schmidt A, Zerova TE, Demidova IA, Buzhiyevskaya TI. Identification of marker chromosomes by in situ hybridization technique using alpha and “classical” satellite DNA probes with relative chromosomal specificity. Tsitol Genet. 1994;28(3):67–88.

19. Vorsanova SG, Yurov YB, Solovev IV, Demidova IA, Malek P. Rapid identification of marker chromosomes by in situ hybridization under different stringency conditions. Anal Cell Pathol. 1994;7(3):251–8.

20. Yurov YB, Laurent AM, Marcais B, Vorsanova SG, Roizes G. Analysis of pericentromeric chromosome 21 specific YAC clones by FISH: identification of new markers for molecular-cytogenetic application. Hum Genet. 1995;95(3):287–92.

21. Yurov YB, Solovev IV, Vorsanova SG, Marcais B, Roizes G, Lewis R. High resolution multicolor fluorescence in situ hybridization using cyanine and fluorescein dyes: rapid chromosome identification by directly fluorescently labeled alphoid DNA probes. Hum Genet. 1995;96(4):672–88.

22. Iourov IY, Solovev IV, Vorsanova SG, Monakhov VV, Yurov YB. An approach for quantitative assessment of fluorescence in situ hybridization (FISH) signals for applied human molecular cytogenetics. J Histochem Cytogenet. 2005;53(3):401–8.

23. Iourov IY, Liehr T, Vorsanova SG, Kolotii AD, Yurov YB. Visualization of interphase chromosomes in postmitotic cells of the human brain by multicolour banding (MCB). Chrom Res. 2006;14(1):223–9.

24. Iourov IY, Liehr T, Vorsanova SG, Yurov YB. Interphase chromosome-specific multicolor banding (ICS-MCB): a new tool for analysis of interphase chromosomes in their integrity. Biomed Eng. 2007;24(4):415–7.

25. Iourov IY, Vorsanova SG, Yurov YB. Fluorescence intensity profiles of in situ hybridization signals depict genome architecture within human interphase nuclei. Tsitol Genet. 2008;42(5):3–8.

26. Iurov IuB, Vorsanova SG, Solovev IV, Iurov Iu. Instability of chromosomes in human nerve cells (normal and with neuromental diseases). Vestn Ross Akad Med Nauk. 2010;67(9):45–53.

27. Iourov IY, Zelenova MA, Vorsanova SG, Voinova VV, Yurov YB. 4q21.2q21.3 duplication: molecular and neuropyschological aspects. Curr Genomics. 2018;19(3):173–8.

28. Yurov YB, Vorsanova SG, Iourov IY, Zelenova MA, Korostelev SA, Yurov YB. Genomic copy number variation affecting genes involved in the cell cycle pathway: implications for somatic mosaicism. Int J Genomics. 2015;2015:757680.
50. Yurov YB, Vorsanova SG, Demidova IA, Kravets VS, Vostrikov VM, Soloviev IV, Urunava NA, Iourov IV. Genomic instability in the brain: chromosomal mosaicism in schizophrenia. Zh Nevrol Psikhiatr Im S S Korsakova. 2016;116(1):86–91.
51. Vorsanova SG, Zelenova MA, Yurov YB, Iourov IV. Behavioral variability and somatic mosaicism: a cytogenomic hypothesis. Curr Geno. 2018;19(3):158–62.
52. Yurov YB, Vorsanova SG, Demidova IA, Kolotii AD, Soloviev IV, Iourov IV. Mosaic brain aneuploidy in mental illnesses: an association of low-level post-zygotic aneuploidy with schizophrenia and comorbid psychiatric disorders. Curr Geno. 2018;19(3):163–72.
53. Iourov IV, Vorsanova SG, Yurov YB. Ataxia telangiectasia paradox can be explained by chromosomal instability at the subtelomere level. Med Hypoth. 2007;68(3):716.
54. Iourov IV, Vorsanova SG, Liehr T, Yurov YB. Aneuploidy in the normal, Alzheimer’s disease and ataxia-telangiectasia brain: differential expression and pathological meaning. Neurobiol Dis. 2009;34(2):212–20.
55. Iourov IV, Vorsanova SG, Liehr T, Kolotii AD, Yurov YB. Increased chromosome instability dramatically disrupts neural genome integrity and mediates cerebellar degeneration in the ataxia-telangiectasia brain. Hum Mol Genet. 2009;18(4):2656–69.
56. Yurov YB, Iourov IV, Vorsanova SG. Neurodegeneration mediated by chromosome instability suggests changes in strategy for therapy development in ataxia-telangiectasia. Med Hypoth. 2009;73(6):1075–6.
57. Yurov YB, Vorsanova SG, Iourov IV. GIN’n’CIN hypothesis of brain aging: deciphering the role of somatic genetic instabilities and neural aneuploidy during ontogeny. Mol Cyto. Genet. 2009;2:23.
58. Iourov IV, Vorsanova SG, Yurov YB (2011) Genomic landscape of the hypoth. 2007;68(3):716.
59. Yurov YB, Vorsanova SG, Iourov IV. Chromosome instability in the neuro-degeneration of post-replicated cells with site-specific cosmid and cosmid contig probes. Prenat Diagn. 1995;15(3):237–48.
60. Vorsanova SG, Yurov YB, Kolotii AD, Demidova IA, Novikova IM. 16q subtelomeric deletion in proband with congenital malformations and mental retardation. Am J Hum Genet Suppl. 1998;65:A154.
61. Vorsanova SG, Yurov YB, Kolotii AD, Demidova IA, Novikova IM. 16q subtelomeric deletion and mental retardation. Tselis Genet. 2005;104(10):73–80.
62. Vorsanova SG, Yurov YB, Kravets VS, Soloviev IV, Urunava NA, Iourov IV. Genomic instability in the brain: chromosomal mosaicism in schizophrenia. Zh Nevrol Psikhiatr Im S S Korsakova. 2015;105(7):4–11.
63. Vorsanova SG, Yurov YB, Kolotii AD, Demidova IA, Soloviev IV. Chromosome instability and somatic mosaicism: a cytogenomic hypothesis. Curr Geno. 2018;19(3):163–72.
64. Vorsanova SG, Demidova IA, Ulas VIu, Soloviev IV, Kravets VS, Beresheva AK, Novikov PV, lurov IuV. Epigenetic study of Rett’s syndrome as an adequate model for autistic disorders. Zh Nevrol Psikhiatr Im S S Korsakova. 2005;105(7):4–11.
65. Vorsanova SG, Iurov IV, Vorsanova SG, Zelenova MA, Demidova IA, Giunti L, Guivabuccu-Uzzielli ML, Budilov AV, Beresheva AK, Novikov PV, lurov IuV. Epigenetic analysis of Rett’s syndrome in a Russian cohort of girls with Rett syndrome. Zh Nevrol Psikhiatr Im S S Korsakova. 2012;113(10):63–8.
66. Iourov IV, Vorsanova SG, Voimova IV, Urunava NA, Zelenova MA, Demidova IA, Yurov YB. Xq28 (MECP2) microdeletions are common in mutation-negative females with Rett syndrome and cause mild subtypes of the disease. Mol Cyto. Genet. 2013;6(1):53.
67. Iourov IV, Yurov YB, Bertrand T. VIII world rett syndrome congress & symposium of rare diseases, Kazan. Russia Mol Cyto. Genet. 2018;11:61.
68. Soloviev IV, Uroev YB, Vorsanova SG, Fayet F, Roizes G, Malet P. Prenatal detection of trisomy 21 using interphase fluorescence in situ hybridization of post-replicated cells with site-specific cosmid and cosmid contig probes. Prenat Diagn. 1999;19(3):237–48.
69. Vorsanova SG, Iourov IV, Beresheva AK, Demidova IA, Monakhov VV, Kravets VS, Bartseva OB, Goiko EA, Soloviev IV, Yurov YB. Non-disjunction of chromosome 21: alpha satellite DNA variation, and somatic genetic instabilities and neural aneuploidy during ontogeny. Mol Cyto. Genet. 2009;2:23.
70. Iourov IV, Vorsanova SG, Yurov YB (2011) Genomic landscape of the Alzheimer’s disease brain: chromosome instability—aneuploidy, but not tetraploidy—mediates neurodegeneration. Neurodegener Dis. 2009;8(1–2):35–7; discussion 18–40.
71. Yurov YB, Vorsanova SG, Iourov IV. The DNA replication stress hypothesis of Alzheimer’s disease. Scientific World J. 2011;11:2602–12.
72. Yurov YB, Vorsanova SG, Liehr T, Kolotii AD, Iourov IV. X chromosome aneuploidy in the Alzheimer’s disease brain. Mol Cyto. Genet. 2014;71(20).
73. Yurov YB, Vorsanova SG, Iourov IV. Chromosome instability in the neuro-degenerating brain. Front Genet. 2019;10:892.
74. Vorsanova SG, Yurov YB, Iourov IV. Dynamic nature of somatic chromosomal mosaicism, genetic-environmental interactions and therapeutic opportunities in disease and aging. Mol Cyto. Genet. 2020;20:13:16.
75. Vorsanova SG, Demidova IA, Ulas VIu, Soloviev IV, Kazantseva LZ, Yurov Yub. Cytogenetic and molecular-cytogenetic investigation of Rett syndrome: analysis of 31 cases. Neuroreport. 1996;8(1):187–9.
76. Vorsanova SG, Demidova IA, Ulas VIu, Soloviev IV, Kravets VS, Kazantseva LZ, Iurov IuB. Cytogenetic and molecular genetic diagnostics of Rett syndrome in children. Zh Nevrol Psikhiatr Im S S Korsakova. 1998;98(4):53–6.
77. Vorsanova SG, Ulas VIu, Demidova IA, Kravets VS, Iurov IuB. Contemporaneous views on Rett’s syndrome: clinical, cytogenetic and molecular studies. Zh Nevrol Psikhiatr Im S S Korsakova. 1999;99(3):61–9.
78. Vorsanova SG, Yurov YB, Kolotii AD, Soloviev IV. FISH analysis of replication and transcription of chromosome X loci: new approach for genetic analysis of Rett syndrome. Brain Dev. 2001;23(Suppl 1):S191–5.
79. Vorsanova SG, Yurov YB, Ulas VIu, Demidova IA, Sharanov VO, Kolotii AD, Gorbatchevskaiia NL, Beresheva AK, Soloviev IV. Cytogenetic and molecular-cytogenetic studies of Rett syndrome (RTT): a retrospective analysis of a Russian cohort of RTT patients (the investigation of 57 girls and three boys). Brain Dev. 2001;23(Suppl 1):S196–201.
80. Yurov YB, Vorsanova SG, Kolotii AD, Iourov IV. Molecul-cytogenetic investigation of skewed chromosome X inactivation in Rett syndrome. Brain Dev. 2001;23(Suppl 1):S214–7.
81. Vorsanova SG, Ulas VIu, Iurov IuB, Giovannucci-Uzzielli ML, Demidova IA, Giunti L, Villard L, Iurov IuB, Beresheva AK, Novikov PV. Genotype-phenotype correlations in Rett syndrome: the study of Russian cohort of patients. Zh Nevrol Psikhiatr Im S S Korsakova. 2002;102(10):23–9.
82. Iurov IuV, Iurov IuB, Iurov IuB. Nervous and mental diseases in boys and mutations of MECP2 gene. Zh Nevrol Psikhiatr Im S S Korsakova. 2004;104(10):73–80.
83. Vorsanova SG, Iourov IV, Yurov YB. Neurological, genetic and epigenetic features of Rett syndrome. J Pediatr Neurol. 2004;2:179–90.
89. Iourov IY, Vorsanova SG, Korostelev SA, Zelenova MA, Yurov YB. Long contiguous stretches of homozygosity spanning the imprinted loci are associated with intellectual disability, autism, and/or epilepsy. Mol Cytogenet. 2015;8(77).

90. Iourov IY, Vorsanova SG, Korostelev SA, Vasin KS, Zelenova MA, Kurinnaia OS, Yurov YB. Structural variations of the genome in autistic spectrum disorders with intellectual disability. Zh Nevrol Psikhiatr Im S S Korsakova. 2016;116(7):50–4.

91. Iourov IY, Vorsanova SG, Zelenova MA, Vasin KS, Kurinnaia OS, Korostelev SA, Yurov YB. Epigenetic variations manifesting as a loss of heterozygosity affecting imprinted genes represent a molecular mechanism of autism spectrum disorders and intellectual disability in children. Zh Nevrol Psikhiatr Im S S Korsakova. 2019;119(5):91–7.

92. Iourov IY, Vorsanova SG, Yurov YB. Molecular cytogenetics and cytogenomics of brain diseases. Curr Genomics. 2008;9(7):452–65.

93. Iurov II, Vorsanova SG, Saprina EA, Iurov IB. Identification of candidate genes of autism on the basis of molecular cytogenetic and in silico studies of the genome organization of chromosomal regions involved in unbalanced rearrangements. Genetika. 2010;46(10):1348–51.

94. Iourov IY, Vorsanova SG, Yurov YB. In silico molecular cytogenetics: a bioinformatic approach to prioritization of candidate genes and copy number variations for basic and clinical genome research. Mol Cytogenet. 2014;7:98.

95. Yurov YB, Vorsanova SG, Iourov IY. Network-based classification of molecular cytogenetic data. Curr Bioinform. 2017;12:27–33.

96. Vorsanova SG, Yurov YB, Iourov IY. Neurogenomic pathway of autism spectrum disorders: linking germline and somatic mutations to genetic-environmental interactions. Curr Bioinform. 2017;12:19–26.

97. Zelenova MA, Yurov YB, Vorsanova SG, Iourov IY. Laundering of CNV data for candidate process prioritization in brain disorders. Mol Cytogenet. 2019;12:54.

98. Iourov IY, Vorsanova SG, Yurov YB. Pathway-based classification of genetic diseases. Mol Cytogenet. 2019;12:4.

99. Iourov IY, Vorsanova SG, Yurov YB. The varome concept: focus on CNVariome. Mol Cytogenet. 2019;12:52.

100. Iourov IY, Vorsanova SG, Yurov YB, Zelenova MA, Kurinnaia OS, Vasin KS, Kutsiev SI. The cytogenomic “theory of everything”: chromohelkosis may underlie chromosomal instability and mosaicism in disease and aging. Int J Mol Sci. 2020;21(21):8328.

101. Iourov IY, Vorsanova SG, Yurov YB. Systems cytogenomics: are we ready yet? Curr Genomics. 2021;22(2):75–8.

102. Iourov IY, Vorsanova SG, Voinova VY, Yurov YB. 3p22.1p21.31 microdeletion identifies CCX as a genetic anchor point for neurodegeneration in preterm infants: genomic anomalies detection. Res Results Biomed. 2019;5(1):25–51.

103. Iourov IY, Liehr T, Vorsanova SG, Mendez-Rosado LA, Yurov YB. The applicability of interphase chromosome-specific multicolor banding (ICS-MCB) for studying neurodevelopmental and neurodegenerative disorders. Res Results Biomed. 2019;5(3):4–9.

104. Kolotii AD, Vorsanova SG, Yurov YB, Kurinnaia OS, Zelenova MA, Yurov YB. Molecular genetic study of Turner’s syndrome mosaicism in girls with delay, congenital malformations and chromosome instability manifesting as low-level aneuploidy. Springer. Berlin. 2015:616.

105. Iourov IY, Zelenova MA, Vorsanova SG. COVID-19: a crash test for biomedical publishing? medRxiv. 2020;https://doi.org/10.1101/2020.06.13.20130310.

106. Iourov IY, Vorsanova SG. COVID-19 and aging-related genome (chromosome) instability in the brain: another possible time-bomb of SARS-CoV-2 infection. Front Aging Neurosci. 2022;14:766364.

107. Vorsanova SG, Demidova IA, Kolotii AD, Kurinnaia OS, Kravets VS, Soloviev IV, Yurov YB. Turner’s syndrome mosaicism in girls with neurodevelopmental disorders: a cohort study and hypothesis. Mol Cytogenet. 2021;14(1):9.

108. Vorsanova SG, Demidova IA, Kolotii AD, Kurinnaia OS, Kravets VS, Soloviev IV, Yurov YB. Klinefelter syndrome mosaicism in boys with neurodevelopmental disorders: a cohort study and an extension of the hypothesis. Mol Cytogenet. 2022;15(1):8.

109. Vorsanova SG, Yurov YB, Chernyshov VN. Medical cytogenetics. Moscow: Medpraktika; 2006.

110. Vorsanova SG, Iourov IY, Soloviev IV, Yurov YB. Heterochromatic regions of human chromosomes: medical and biological aspects. Moscow: Medpraktika; 2008.

111. Vorsanova SG, Iourov IY, Demidova IA, Kravetz VS, Yurov YB. Cytogenetics and molecular cytogenetics of autism. Moscow: The publishing house of The Russian Academy of Natural History; 2016.

112. Vorsanova SG, Iourov IY, Kurinnaia OS, Yurov YB. Idiopathic intellectual disability in children: cytogenetic and molecular cytogenetic aspects. Moscow: The publishing house of The Russian Academy of Natural History; 2017.

113. Iourov IY, Voinova VY, Vorsanova SG, Yurov YB. Molecular and clinical basis of inherited diseases (textbook). Moscow: The publishing house of The Russian Academy of Natural History; 2018.

114. Iourov IY, Vorsanova SG, Voinova VY, Churnosov MI, Yurov YB. Cytogenetics, molecular and clinical basis of genetic diseases (textbook). Moscow: The publishing house of The Russian Academy of Natural History; 2019.

115. Iourov IY, Vorsanova SG, Yurov YB. Somatic Genome Variations. In: eLS, Wiley, Hoboken, 2012; https://doi.org/10.1002/9780470015902.a0023889.

116. Iourov IY, Vorsanova SG, Liehr T, Yurov YB. Mosaik im Gehirn des Menschen. Diagnostische Relevanz in der Zukunft. Medizinische Genetik. 2014;26:342–5.

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