Human Interphase Chromosomes

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We dedicate this work to the memory of Ilia V. Soloviev, who will not be forgotten. He was a talented young researcher and a pioneer of molecular cytogenetics and of genome and chromosome research. His prodigious work and original ideas have formed our current research directions.

Dr. Ilia V. Soloviev

Preface

The study of human chromosomes in the context of an interphase nucleus is biologically most meaningful for understanding eukaryotic DNA expression and reproduction inasmuch as the interphase comprises essential periods of normal cell activity. To determine the architectural organization of chromosomes inside the nuclear space is thereby important for understanding how the genome functions during the cell cycle. Moreover, variations in chromosome number and structure in humans, who possess more than 200 types of cells, the majority of which are usually in interphase, cannot be properly addressed without using interphase cytogenetics (an umbrella term covering techniques for analysis of interphase chromosomes). The latter is often viewed as an esoteric discipline that only concerns a few specialists trying to implement single-cell approaches to genome biology and medicine. However, studying interphase chromosomes is relevant to numerous fields of life sciences, including, but not limited to, molecular and cell biology, biomedicine, genetics (including medical genetics), neuroscience, evolution, oncology, and genomics.

The beginning of experimental interphase cytogenetics can be attributed to significant advances in human molecular genetics and cytogenetics. As the consequence of experimental and theoretical research at the interface between cellular and molecular levels of chromosomal organization and function, high-resolution techniques for chromosomal analysis (molecular cytogenetic techniques) have become available. Molecular cytogenetics is a branch of biomedical sciences that explores chromosomes at molecular and single-cell resolutions at all stages of the cell cycle. It also comprises the techniques that operate with either the entire genome or specific DNA sequences to analyze genomic structural and functional variations at the chromosomal level. In the postgenomic era, molecular cytogenetics has appreciably transformed and has given rise to a new field of genomics, called cytogenomics. As a result, new opportunities have emerged for analysis of human interphase chromosomes in almost all cell types and states at unprecedented resolution. In this volume, we have attempted to provide an overview of current developments in the study of human interphase chromosomes with special attention to available molecular cytogenetic technologies for basic and clinical chromosome research.

The main body of the book is composed of 12 chapters. Chapter 1 (by Prof. Y.B. Yurov, Prof. S.G. Vorsanova, and Prof. I.Y. Iourov) is devoted to the basics of interphase molecular cytogenetics and cytogenomics in historical perspective. Chapter 2 (by Prof. J. Bridger and associates) considers contemporary views on interphase chromosome behavior in normal and diseased cells. The relationship between nuclear architecture and occurrence of chromosome aberrations is discussed in Chap. 3 (by Prof. G. Folle and Drs. Tomaso, Lafon-Hughes, and Liddle). The role of nuclear chromosome positioning, chromatin organization sensitivity to environmental exposures, genetic damage in metaphase and interphase, DNA replication and chromatin remodeling, and their involvements in the generation and localization of primary genetic damage are discussed. The unique possibility to visualize the interphase chromosome of the human brain and analyses of chromosome (genome) instability in postmitotic neuronal and glial cells are discussed in Chap. 4 (by Prof. I.Y. Iourov, Prof. S.G. Vorsanova, and Prof. Y.B. Yurov). In this chapter, mosaic aneuploidy is defined as a new feature of the normal human brain; increased chromosome instability in the developing and adult human brain is shown to be associated with neurodevelopmental and neurodegenerative genetic brain disorders (autism, schizophrenia, ataxia-telangiectasia, Alzheimer's disease); and interphase molecular cytogenetics is demonstrated to be the way for future studies of somatic genome instability and etiology (and pathogenesis) of genetic brain diseases. Taking into account the increased interest in somatic cell evolution mediated by genome alteration and its clinical significance, Dr. J. Stevens and Prof. H. Heng (Chap. 5) review mechanisms of chromosome fragmentation and premature chromosome condensation. They also discuss the mechanisms and definition of premature chromosome condensation and its applications to basic and clinical research. Chapter 6 (by Prof. E. Volpi) reviews the association between pathology, large-scale chromatin organization, and nuclear architecture in an enigmatic chromosome instability syndrome (ICF syndrome: a rare epigenetic disorder caused by autosomal recessive mutations, often fatal in childhood). Chapter 7 (by Prof. D. Griffin and Drs. Fonseka, Tempest, Thornhill, and Ioannou) overviews interphase cytogenetics of human embryos, highlighting the progress and contentious pitfalls that it encounters. Because interphase cytogenetics has important applications in prenatal medicine, other chapters outlined in this book pave the way for a range of exciting new studies that, potentially, might emerge on human embryos and show FISH as a still useful tool for rapid, low-cost, and robust cell-by-cell information. Chapter 8 (by Drs. O.S. Mudrak, L. Solovjeva, and V. Chagin) describes experimental data of studies dedicated to human spermatozoa and discusses the implications of sperm chromosome organization for male reproductive health. Chromosomes in human sperm nuclei adopt a hierarchy of structures from protamine toroids (the elementary units of DNA packaging) to the higher-order organization (chromosome territories), suggesting that chromatin organization in sperm may have functional significance. The intention of Chap. 9 (by Prof. I.Y. Iourov, Dr. T. Liehr, Prof. S. Vorsanova, and Prof. Y. Yurov) is to present the basics of interphase chromosome-specific multicolor banding (ICS-MCB) and to list its applications in different biomedical fields. Chapter 10 (by Dr. T. Liehr and his collaborators) is focused on technical limitations

in biomedical research of interphase chromosomes in their integrity. To overcome these limitations, the authors have proposed a new technology based on three-dimensional suspension fluorescence in situ hybridization (3D S-FISH) with microdissection-based engineered DNA probes and multicolor chromosome banding (MCB). Chapter 11 (by Prof. S.G. Vorsanova and her collaborators) describes technological aspects and numerous approaches of interphase molecular cytogenetic, which are all useful for chromosomal analysis in almost all human cell types. Regardless of numerous technological difficulties encountered during studying human interphase chromosomes in health and disease, molecular cytogenetics or cytogenomics ("chromosomics") does provide for high-resolution single-cell analysis of genome organization, structure, and behavior at all stages of the cell cycle. Finally, the editors provide a list of references to websites containing regularly updated information on molecular cytogenetics and cytogenomics, including useful links to relevant websites (see Appendix).

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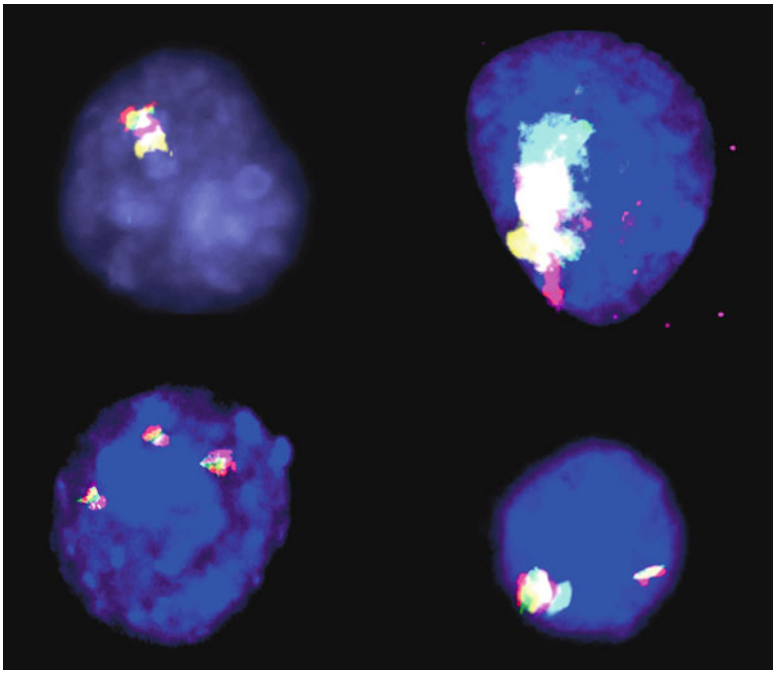

Molecular cytogenetic analyses of the developing and adult human brain by ICS-MCB. Upper row (*left*) - loss of chromosome 16 (monosomy) in a cell isolated from the prefrontal cortex of the normal human brain; (*right*) - loss of chromosome 1 (monosomy) in a cell isolated from the prefrontal cortex of the schizophrenia brain. Bottom row (*left*) - gain of chromosome 21 (trisomy) in a cell isolated from the prefrontal cortex of an Alzheimer's disease brain; (*right*) - chromosome instability in the cerebellum of the ataxia-telangiectasia brain manifesting as the presence of normal and a rearranged chromosome14 order (14)(14pter->14q12:)

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