

Appendix: Internet Links for Interphase Molecular Cytogeneticists

Abstract In the final chapter, “Appendix,” we have provided a list of Internet sites that are highly useful for those working in the field of interphase molecular cytogenetics. The collection includes sites dedicated to cytogenetics (molecular cytogenetics), clinical aspects of diseases studied by molecular cytogenetic techniques, retrieving scientific (biomedical) literature, and bioinformatic analysis (including resources allowing gene prioritization and pathway analysis, genome browsers, and analyzing tools). The inclusion was performed according to our own experience and reported relevance to genome and chromosome research based on data acquired during molecular cytogenetic analyses. It is clearly impossible to list all the links that are of importance for a scientist whose activity is related to studying genome (chromosome) structure and behavior in interphase. In this instance, we encourage the readers to follow the links provided by the listed resources.

Introduction

To list Internet sites relevant to the field of interphase molecular cytogenetics, we preferred to subdivide the list according to the main modes of application. We also provide references to papers describing the resource (if applicable) and to journals that are publishing papers dedicated to interphase chromosome analyses. However, one has to keep in mind that the provided sites are not all specifically dedicated to interphase cytogenetics. On the other hand, these are almost indispensable to be addressed when a study of genome (chromosome) structure and behavior in interphase at a high technological level is performed. A brief example of the use of *in silico* (bioinformatic) methods in a study almost completely dedicated to interphase chromosome behavior and variations and its relevance to more general biological processes can be found in Iourov et al. (2009). Finally, these sites are included to the present chapter (Appendix) according to our own experience and reported relevance to genome and chromosome research based on data acquired during molecular cytogenetic analyses. All the links were tested in November 2012.

Cytogenetics: Chromosome Abnormalities (Including Cancer Cytogenetic Databases)

- arrayMap (genomic arrays for copy number profiling in human cancer) (Cai et al. 2012): <http://www.arraymap.org/cgi-bin/amHome.cgi>
- Atlas of genetics and cytogenetics in oncology and haematology (Huret et al. 2012): <http://atlasgeneticsoncology.org/>
- Chromosomal Variation in Man or Borgaonkar DS. Chromosomal Variation in Man: A Catalog of Chromosomal Variants and Anomalies: Online NLM Version. Bethesda (MD): National Center for Biotechnology Information (US); 1975: <http://www.ncbi.nlm.nih.gov/books/NBK105441/> and <http://www.wiley.com/legacy/products/subject/life/borgaonkar/access.html>
- Chromosome Anomaly Collection (managed by Dr John Barber): <http://www.ngrl.org.uk/wessex/collection/>
- CyDAS (drawing derivative chromosomes online) (Hiller et al. 2005): <http://www.cydias.org/OnlineAnalysis/>
- Cytogenetic Gallery (a scholar website for cytogeneticists): <http://www.pathology.washington.edu/galleries/Cytogallery/>
- DECIPHER—database of unbalanced chromosome aberrations (Firth et al. 2009): <http://decipher.sanger.ac.uk/>
- ECARUCA European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations (Feenstra et al. 2006): <http://umcccaruca01.extern.umcn.nl:8080/ecaruca/ecaruca.jsp>
- Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer: <http://cgap.nci.nih.gov/Chromosomes/Mitelman>
- NCI and NCBI's SKY/M-FISH and CGH Database (2001): <http://www.ncbi.nlm.nih.gov/sky/sky-web.cgi>
- Progenetix (database of cytogenetic abnormalities in cancer) (Baudis 2007): <http://www.progenetix.de/progenetix/index.html>
- Small supernumerary marker chromosomes (sSMC) homepage (managed by Dr. T. Liehr): <http://www.med.uni-jena.de/fish/sSMC/00START.htm>

Clinical Databases

- Autism Chromosome Rearrangement Database (Marshall et al. 2008): <http://projects.tcag.ca/autism/>
- GeneReviews™ or Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews™. Seattle, WA: University of Washington, Seattle; 1993: <http://www.ncbi.nlm.nih.gov/books/NBK1116/>
- Genetics Home Reference (consumer-friendly information about the effects of genetic variations on human health): <http://ghr.nlm.nih.gov/>
- MedGen (organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution): <http://www.ncbi.nlm.nih.gov/medgen/>
- OMIM (online Mendelian inheritance in man): <http://www.omim.org/>
- ORPHANET (the portal for rare diseases and orphan drugs): <http://www.orpha.net>
- The Phenotype-Genotype Integrator (PheGenI): <http://www.ncbi.nlm.nih.gov/gap/PheGenI>
- SFARI Gene/AutDB (a publicly available, curated, web-based, searchable database for autism research) (Basu et al. 2009): <http://www.mindspec.org/autdb.html>
- UNIQUE (rare chromosome disorder support group): <http://www.rarechromo.org/html/home.asp>

FISH and Array CHG-Based Techniques (Including Pages Containing Information About DNA Probes)

- A general compilation of links to molecular cytogenetics resources and beyond (the last chapter of the *Fluorescence In Situ Hybridization (FISH): Application Guide*, Edited by Dr. T. Liehr): <http://www.springer.com/life+sciences/cell+biology/book/978-3-540-70580-2>
- ArrayCyGHt (a web-based application tool for analysis and visualization of array-CGH data) (Kim et al. 2005): <http://genomics.catholic.ac.kr/arrayCGH/>
- CHORI BACPAC resources (Children's Hospital & Research Center Oakland; Dr. P. De Jong): <http://bacpac.chori.org/about.htm>
- CCAP BAC Clones (Cancer Genome Anatomy Project, NCI): http://cgap.nci.nih.gov/Chromosomes/CCAP_BAC_Clones
- e-FISH (an in-silico FISH simulation tool): <http://projects.tcag.ca/efish/>
- Genomic Clone Database (The Centre for Applied Genomics and The Hospital for Sick Children, Toronto): <http://projects.tcag.ca/gcd>
- The International Standards for Cytogenomic Arrays (ISCA) Consortium: <https://www.iscaconsortium.org/>
- Multicolor FISH database (managed by Dr. T. Liehr): <http://www.fish.uniklinikum-jena.de/mFISH.html>
- NCBI Probe/Probe Database (a public registry of nucleic acid reagents designed for use in a wide variety of biomedical research applications, with information on reagent distributors, probe effectiveness, and computed sequence similarities): <http://www.ncbi.nlm.nih.gov/probe>
- Resources for molecular cytogenetics: <http://www.biologia.uniba.it/rmc/>
- Scitable by Nature Education—Cytogenetics—FISH: <http://www.nature.com/scitable/topic/chromosomes-and-cytogenetics-7> <http://www.nature.com/scitable/topicpage/fluorescence-in-situ-hybridization-fish-327>
- University of Kansas Medical Center compilation of links to cytogenetic (molecular cytogenetic) resources: <http://www.kumc.edu/gec/prof/cytogene.html>

Genome, Epigenome and Pathway Analyzing Tools (Including Genome Browsers, Bioinformatic Tools for Gene Prioritization and Pathway Analysis)

- AmiGO browser (Gene Ontology project browser and search engine): <http://amigo.geneontology.org/cgi-bin/amigo/go.cgi>
- BioGPS (a free extensible and customizable gene annotation portal, a complete resource for learning about gene and protein function; The Scripps Research Institute) (Wu et al. 2009): <http://biogps.org>
- A Catalog of Published Genome-Wide Association Studies (NHGRI) (Hindorf et al. 2009): <http://www.genome.gov/gwastudies/>
- ENDEAVOUR (a gene prioritization tool) (Tranchevent et al. 2008): <http://homes.esat.kuleuven.be/~bioiuser/endeavour/tool/endeavourweb.php>
- Ensembl Genome Browser: <http://www.ensembl.org/index.html>
- Gene Expression Omnibus (GEO): <http://www.ncbi.nlm.nih.gov/geo/>
- The Gene Wiki (Wikipedia-based gene annotation portal) (Huss et al. 2010): http://en.wikipedia.org/wiki/Portal:Gene_Wiki
- Gene Wanderer (a gene prioritization tool using interactome) (Köhler et al. 2008): <http://compbio.charite.de/genewanderer/GeneWanderer>

KEGG or Kyoto Encyclopedia of Genes and Genomes (source for understanding high-level functions and utilities of the biological system): <http://www.genome.jp/kegg/>

NCBI BioSystems Database (access to biological systems and their component genes, proteins, and small molecules, as well as literature describing those biosystems and other related data): <http://www.ncbi.nlm.nih.gov/biosystems>

NCBI Build 37.1/NCBI Map Viewer (*Homo sapiens*, Annotation Release 104): http://www.ncbi.nlm.nih.gov/projects/mapview/map_search.cgi?taxid=9606

NCBI Gene (a record may include nomenclature, RefSeqs, maps, pathways, variations, phenotypes, and links to genome-, phenotype-, and locus-specific resources worldwide): <http://www.ncbi.nlm.nih.gov/gene/>

Pathway Commons (maintained by Memorial Sloan-Kettering Cancer Center and the University of Toronto.): <http://www.pathwaycommons.org/pc/>

REACTOME (manually curated and peer-reviewed pathway database) (Vastrik et al. 2007): www.reactome.org/

UCSC Genome Browser: <http://genome.ucsc.edu/>

UniHI (interactome analysis tool) (Chaurasia et al. 2007): <http://www.unihi.org/>

Genomic Variation Databases

Database of Genomic Variants (hosted by the Centre for Applied Genomics): <http://dgvbeta.tcag.ca/dgv/app/home?ref=GRCh37/hg19>

Database of genomic structural variation (dbVar) <http://www.ncbi.nlm.nih.gov/dbvar/>

Human Genome Structural Variation Project (Structural Variation Database) (managed by Eichlerlab) <http://humanparalogy.gs.washington.edu/structuralvariation/>

Human Genome Variation Society: <http://www.hgvs.org/dblist/dblist.html>

Biomedical Literature (Including Websites Allowing Acquiring Scientific Information Through the Entire World Wide Web)

PubMed: <http://www.ncbi.nlm.nih.gov/pubmed>

ScienceDirect—Elsevier: (>11 million full-text journal articles and book chapters): <http://www.sciencedirect.com/>

SCIRUS (for scientific information only; searches more than 545 million science-specific web pages): <http://scirus.com/>

Scopus (easy and comprehensive resource to support the research needs in the scientific, technical, medical, and social sciences fields): <http://www.scopus.com/home.url>

Springer (contains nearly six million resources): <http://link.springer.com/>

Wiley online library: <http://onlinelibrary.wiley.com/>

HighWire Stanford University: <http://highwire.stanford.edu/>

BioMed Central The Open Access Publisher: <http://www.biomedcentral.com/>

Web of Knowledge (Thompson Reuters/impact factor): <http://apps.webofknowledge.com>

Google Scholar: <http://scholar.google.com/>

To this end, we found pertinent to provide a list of journals that publish research in interphase molecular cytogenetics. The inclusion was made according to ratio of numbers of interphase cytogenetics papers to the overall number of papers. Journals are sorted by relevance; those journals that have ceased to publish are excluded.

Molecular Cytogenetics: <http://www.molecularcytogenetics.org/>

Chromosoma: <http://link.springer.com/journal/412>
 Chromosome Research: <http://link.springer.com/journal/10577>
 Journal of Cell Biology: <http://jcb.rupress.org/>
 Journal of Cell Science: <http://jcs.biologists.org/>
 Genes, Chromosomes and Cancer: <http://onlinelibrary.wiley.com/journal/10.1002/%28ISSN%291098-2264;jsessionid=14F5A0A05993373860006F087881472C.d01t04>
 Experimental Cell Research: <http://www.journals.elsevier.com/experimental-cell-research/>
 Molecular Biology of the Cell: <http://www.molbiolcell.org/>
 Molecular and Cell Biology: <http://mcb.asm.org/>
 Cytogenetic and Genome Research: <http://content.karger.com/ProdukteDB/produkte.asp?Aktion=JournalHome&ProduktNr=224037>
 Journal of Histochemistry and Cytochemistry: <http://jhc.sagepub.com/>
 Human Genetics: <http://www.springer.com/biomed/human+genetics/journal/439>
 The American Journal of Human Genetics: <http://www.cell.com/AJHG/>
 Human Molecular Genetics: <http://hmg.oxfordjournals.org/>
 BioDiscovery: <http://www.biodiscoveryjournal.co.uk/>
 Current Genomics: <http://www.benthamscience.com/cg/>
 Methods in Molecular Biology: <http://www.springer.com/series/7651>
 PLoS One: <http://www.plosone.org/>
 Cell: <http://www.cell.com/home>
 Nature Cell Biology: www.nature.com/ncb/
 Prenatal Diagnosis: <http://onlinelibrary.wiley.com/journal/10.1002/%28ISSN%291097-0223>
 Mutation Research: <http://www.elsevier.com/journals/mutation-research-full-set/FS00-0289>
 Journal of Cellular Physiology: <http://onlinelibrary.wiley.com/journal/10.1002/%28ISSN%291097-4652>
 Journal of Medical Genetics: <http://jmg.bmj.com/>
 Genes & Development: <http://genesdev.cshlp.org/>
 PNAS USA: <http://www.pnas.org/>
 Cancer Research: <http://cancerres.aacrjournals.org/>

References

- Basu SN, Kollu R, Banerjee-Basu S (2009) AutDB: a gene reference resource for autism research. *Nucleic Acids Res* 37:D832–D836
- Baudis M (2007) Genomic imbalances in 5918 malignant epithelial tumors: an explorative meta-analysis of chromosomal CGH data. *BMC Cancer* 7:226
- Cai H, Kumar N, Baudis M (2012) ArrayMap: a reference resource for genomic copy number imbalances in human malignancies. *PLoS One* 7(5):e36944
- Chaurasia G, Iqbal Y, Hänig C, Herzel H, Wanker EE, Futschik ME (2007) UniHI: an entry gate to the human protein interactome. *Nucleic Acids Res* 35:D590–D594
- Feenstra I, Fang J, Koolen DA, Siezen A, Evans C, Winter RM et al (2006) European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations (ECARUCA): an online database for rare chromosome abnormalities. *Eur J Med Genet* 49(4):279–291
- Firth HV, Richards SM, Bevan AP, Clayton S, Corpas M, Rajan D et al (2009) DECIPHER: database of chromosomal imbalance and phenotype in humans using Ensembl resources. *Am J Hum Genet* 84(4):524–533
- Hiller B, Bradtke J, Balz H, Rieder H (2005) CyDAS: a cytogenetic data analysis system. *Bioinformatics* 21(7):1282–1283
- Hindorf LA, Sethupathy P, Junkins HA, Ramos EM, Mehta JP, Collins FS, Manolio TA (2009) Potential etiologic and functional implications of genome-wide association loci for human diseases and traits. *Proc Natl Acad Sci U S A* 106(23):9362–9367

- Huret JL, Ahmad M, Arsaban M, Bernheim A, Cigna J, Desangles F et al (2012) Atlas of genetics and cytogenetics in oncology and haematology in 2013. *Nucleic Acids Res* 41(D1):D920–D924. doi:[10.1093/nar/gks1082](https://doi.org/10.1093/nar/gks1082)
- Huss JW 3rd, Lindenbaum P, Martone M, Roberts D, Pizarro A, Valafar F et al (2010) The gene Wiki: community intelligence applied to human gene annotation. *Nucleic Acids Res* 38:D633–D639
- Iourov IY, Vorsanova SG, Liehr T, Kolotii AD, Yurov YB (2009) Increased chromosome instability dramatically disrupts neural genome integrity and mediates cerebellar degeneration in the ataxia-telangiectasia brain. *Hum Mol Genet* 18(14):2656–2669
- Kim SY, Nam SW, Lee SH, Park WS, Yoo NJ, Lee JY, Chung YJ (2005) ArrayCyGHt: a web application for analysis and visualization of array-CGH data. *Bioinformatics* 21(10):2554–2555
- Köhler S, Bauer S, Horn D, Robinson PN (2008) Walking the interactome for prioritization of candidate disease genes. *Am J Hum Genet* 82(4):949–958
- Marshall CR, Noor A, Vincent JB, Lionel AC, Feuk L, Skaug J et al (2008) Structural variation of chromosomes in autism spectrum disorder. *Am J Hum Genet* 82(2):477–488
- Tranchevent LC, Barriot R, Yu S, Van Vooren S, Van Loo P, Coessens B et al (2008) ENDEAVOUR update: a web resource for gene prioritization in multiple species. *Nucleic Acids Res* 36:W377–W384
- Vastrik I, D'Eustachio P, Schmidt E, Gopinath G, Croft D, de Bono B et al (2007) Reactome: a knowledge base of biologic pathways and processes. *Genome Biol* 8(3):R39
- Wu C, Orozco C, Boyer J, Leglise M, Goodale J, Batalov S et al (2009) BioGPS: an extensible and customizable portal for querying and organizing gene annotation resources. *Genome Biol* 10(11):R130

Index

A

- Alpha-satellite DNA, 195
- Alzheimer's disease (AD)
 - abnormal DNA replication, 71
 - cell death, 69
 - disomy 21 and trisomy 21, 72
 - replication stress hypothesis, 72–73
- Aneuploidy
 - brain (*see* Brain, neural aneuploidy)
 - and DNA copy number alterations, 25
 - human embryos development
 - aCGH, 128
 - incidence, 126
 - PGS, efficacy of, 127–128
 - spermatozoa/sperm nucleus, 152–153
- Apoptotic cell, 89
- Array comparative genome hybridization (array CGH)
 - molecular cytogenetic diagnosis, 198
 - whole-genome analysis, 5, 6
- Ataxia-telangiectasia (AT)
 - CIN, 62, 66
 - FISH/QFISH and ICS-MCB, 67–70
- Autism
 - chromosome-enumeration DNA probes, 64
 - chromosome X aneuploidy rates, 65
 - prevalence, 61
 - somatic genome instability, 61
- 5-azacytidine, 109

B

- BCL2*, 20
- β -globin genes, 16, 115
- Biomphalaria glabrata*, 26
- Blastomeres, 131–132

- Brain, neural aneuploidy
 - in aging human brain, 75–77
 - Alzheimer's disease
 - abnormal DNA replication, 71
 - cell death, 69
 - disomy 21 and trisomy 21, 72
 - replication stress hypothesis, 72–73
 - ataxia-telangiectasia
 - CIN, 66
 - FISH/QFISH and ICS-MCB, 67–70
 - autism
 - chromosome-enumeration DNA probes, 64
 - chromosome X aneuploidy rates, 65
 - prevalence, 61
 - somatic genome instability, 61
 - chromosome/genome instabilities, 62–63
 - developing human brain, 55–58
 - embryonic development, 74–75
 - history, 54–55
 - ICS-MCB, 58–59
 - interphase chromosomes and genome organization, 77–78
 - normal adult human brain, 58–60
 - origins, 74–75
 - prenatal development, 55–56
 - schizophrenia, 65–66
- Burkitt's lymphoma, 23, 41

C

- Cancer
 - chromosome positioning, 19–20
 - interphase nuclei behavior, 18–19
 - ionizing radiation, 23
- CDK1, 93

- Chicken DT40, 41
- Chinese hamster ovary (CHO), 36, 45–46
- Chromatin compaction levels, 37
- Chromatin dynamics
 - DNA replication and genetic damage, 43, 45–48
 - gene expression and genetic damage, 42–43
- Chromosomal aberrations (CA)
 - banding techniques, 36
 - cytogenetic evidence, 40
 - DDR, 39
 - differentially expressed transcripts, 41–42
 - DSB, 38
 - gH2AX, 39
 - metaphase, 36
 - nonrandom organization, of CTs, 40–41
 - tissue-specific CT organization, 41
 - translocations, 41–42
- Chromosome fragmentation (C-Frag)
 - cellular stress and death, 91
 - diverse mechanisms, 96–97
 - genomic instability, 90–91
 - implications and utility, 91–92
 - mitotic catastrophe, 99
 - mitotic cell death
 - apoptotic cells, 89
 - giemsa and late stages, 88
 - mitosis stages, 88
 - morphology and mechanism, 87
 - morphological characterization, 89–90
 - pulverization and shattering, 98–99
 - response to stress, 90
 - vs. PCC, 95, 97–98
- Chromosome instability (CIN)
 - aneuploidy, in aging human brain, 75–76
 - ataxia-telangiectasia (AT), 66
 - in brain disorders, 62–63
- Chromosome 1 juxtacentromeric heterochromatin, 114
- Chromosome positioning
 - cancer, 19–20
 - chromosomes 18 and 19, 13
 - and gene expression, 15–17
 - proliferating and nonproliferating cells, 14
- S-FISH
 - (*see* Suspension-based fluorescence in situ hybridization (S-FISH))
- spermatozoa/sperm nucleus
 - intranuclear location, 149
 - longitudinal position, 149–150
 - MSCI, 150
 - polarity, 150–151
- Chromosome pulverization and shattering, 98–99
- Chromosome translocations
 - chromosome size, 24
 - clinical evidence, 22
 - DSB repair, 21–22
 - ionizing radiation, 23
 - nucleoli, 23–24
- Chromothripsis, 98
- CT/IC model. *See* Interchromatin compartment (IC)
- D**
- DNA copy number alterations, 25
- DNA damage
 - chromosome translocations, 41–42
 - cytogenetic evidence, 40
 - DDR, 39
 - differentially expressed transcripts (DET), 41–42
 - DSB, 38
 - gH2AX, 39
 - nonrandom organization, of CTs, 40–41
 - tissue-specific CT organization, 41
- DNA damage response (DDR), 39
- DNA double-strand break (DSB), 21–22, 38
- DNA hypomethylation, 117
- DNA methylation, ICF syndrome
 - 5-azacytidine, 109
 - DNMT3B*, 110–111
 - satellite 2 DNA, 109
 - ZBTB24*, 111
- DNA packaging, in spermatozoa/sperm nucleus
 - donut-loop model, 141
 - histones replacement, 141–142
 - protamines, 140–141
 - sperm chromatin, 142
 - toroidal structures, 141
- DNA replication
 - and genetic damage
 - CHO cells, 45–46
 - CHO9 metaphase, 47
 - chromosome breakpoints model, 45–46
 - confocal z-sections, 47
 - replicating chromatin, 45
 - SCNA, 48
 - topoisomerase II (Topo II), 46
 - ICF syndrome, 117–118

- I-FISH**
 alpha-satellite DNA, 195
 with cosmid probe, 194
 diploid cells, 192
 PHA-stimulated lymphocytes, 196
 sex chromosome aneuploidy, 194–195
 trisomy 21 identification, 193–194
 DNA replication stress, 72–73
DNMT3B, 110–111, 113
 Double-strand breaks (DSB), 21–22
- E**
 Embryos development. *See* Human embryos development
 Envelope, nuclear, 11–12
 Epigenomic instability, 65–66
 Euchromatin, 38
- F**
 Fast-FISH with microwave activation, 189–191
 Fiber FISH, 188–189
 Fluorescence in situ hybridization (FISH)
 aneuploidy in adult human brain, 58–59
 autism, 64
 with chromosome-enumeration DNA probes, 57, 64
 history and development, 4–5
 human embryos, 129
 ICF syndrome diagnosis, 112
 and immunocytochemistry, 5
 metaphase, DNA in, 37
 multicolor banding (*see* Interphase chromosome-specific multicolor banding (ICS-MCB))
 nucleus, 11
 sperm genome architecture, 143–144, 146
 technical basis, 6
 translocation detection, 124–125
FOXP1B, 67, 68
- G**
 Gamma H2AX (gH2AX) foci, 39, 97
 Gene expression
 and chromosome positioning
 3D structure, 16
 genes target, 15
 Hutchinson-Gilford progeria syndrome, 17–18
 loop formation, 15–16
 ncRNA, 16–17
 radial position changes, 15
 and genetic damage
 RIDGEs, 42–43
 tumor-deregulated genes (TDRG), 43
 ICF syndrome, 113–114
 Gene repositioning
 interphase nuclei, 20–21
 translocation event, 24–25
 Genetic damage, gene expression and colocalization of clusters, 45
 human idiograms, 44
 RIDGEs, 42–43
 tumor-deregulated genes (TDRG), 43
 Genome architecture abnormalities, 151–152
 Genome chaos, 92
 Genome instabilities (GIN)
 aneuploidy in aging human brain, 75–76
 ataxia-telangiectasia (AT), 66
 in brain disorders, 62–63
 chromosome fragmentation, 90–91
 schizophrenia, 65–66
- H**
 H2AX, 39
 Heterochromatin, 38, 117
HLXB9, 24
 Human embryos development
 aneuploidy assessment
 aCGH, 128
 incidence, 126
 PGS, efficacy of, 127–128
 24-color FISH screen assay, 130
 FISH analysis, 129
 interphase cytogenetics, future of, 129–130
 nuclear organization
 blastomeres, 131–132
 chromosome loci, 131, 133
 totipotency, 130
 sex chromosomes detection, 124
 translocation detection, 124–125
 Hutchinson-Gilford progeria syndrome (HGPS)
 characteristics, 17
 definition, 14
LMNA gene, 17
 Hyperacetylated histone H4 (H4^{ac}), 42

I

ICF syndrome

- characteristics, 107–108
- gene expression and regulation, 113–114
- methylation defects and genetic heterogeneity
 - 5-azacytidine, 109
 - DNMT3B*, 110–111
 - satellite 2 DNA, 109
 - ZBTB24*, 111
- nuclear architecture
 - chromosome 1 juxtacentromeric heterochromatin, 114
 - DNA hypomethylation, 117
 - DNA replication, 117–118
 - interphase gene-heterochromatin, 117
 - intranuclear positionin, 116
 - long-range heterochromatin-gene, 115
 - original interphase studies, 111–112

Immuno-FISH (I-FISH)

- advantages and limitations, 191–192
- AT cerebellum cells, 77
- chromosome architecture, 196–197
- chromosome-specific DNA probes
 - QFISH, 183
 - resolution related assays, 184
- DNA replication analysis
 - alpha-satellite DNA, 195
 - with cosmid probe, 194
 - diploid cells, 192
 - PHA-stimulated lymphocytes, 196
 - sex chromosome aneuploidy, 194–195
 - trisomy 21 identification, 193–194
- fast-FISH with microwave activation, 189–191
- fiber FISH, 188–189
- interphase chromosome-specific MCB, 187–188
- molecular cytogenetic identification, 197–198
- site-specific probes, 184–186, 193
- two- and three-color, 185
- wcp probe and MFISH/SKY, 186–187

Integral membrane proteins (IMPs), 11–12

Interchromatin compartment (IC), 38

Interphase chromosomes, history of, 2–4

Interphase chromosome-specific multicolor banding (ICS-MCB)

- advantages and disadvantages, 165
- applications
 - chromosome abnormalities identification, 165
 - DNA-based structure definition, 166–167
 - human brain, 166

on human interphase nuclei, 164

procedure

- chromosome spatial organization, 163
- DNA-specific banding, 162–163
- fluorescence intensity ratios, 163

Interphase nuclei

- aneuploidy, 25
- cancer, 18–19
- chromosome positioning
 - cancer, 19–20
 - chromosomes 18 and 19, 13
 - and gene expression, 15–17
 - proliferating and nonproliferating cells, 14
 - translocation, 14
- chromosome translocations
 - chromosome size, 24
 - clinical evidence, 22
 - DSB repair, 21–22
 - ionizing radiation, 23
 - nucleoli, 23–24
- gene repositioning, 20–21, 24–25
- Hutchinson-Gilford progeria syndrome, 17–18
- neurons and glial cells, 26
- nonrandom chromosome positioning, 26
- nuclear structure
 - chromatin movement, 13
 - envelope, 11–12
 - FISH, 11
 - genes transcription, 11
 - lamina, 12
 - nucleolus, 10–11
- Intracytoplasmic sperm injection (ICSI), 152–153

L

Lamin proteins, 12–13

Leukemia, S-FISH, 175–176

LMNA, 12, 17

LMNB1, 12

Locus control region (LCR), 16, 115

Long interspersed repeated elements (LINEs), 36

M

Male infertility, 151–152

MFISH/SKY, 186–187

Microdeletion syndromes, 195

Minichromosomes (MICs), 41

Mitosis-promoting factor (MPF), 93, 94

Mitotic catastrophe, 99

- Mitotic cell death
 apoptotic cells, 89
 giemsa and late stages, 88
 mitosis stages, 88
 morphology and mechanism, 87
- Molecular cytogenetics
 advantages, 180
 conventional cytogenetics, 182
 FISH, 181
 history, 2–4, 180
 identification, 197–198
 I-FISH
 (*see* Immuno-FISH (I-FISH))
 interphase chromosomes, 181–182
- Mosaic neural aneuploidy, 75
- Multicolor banding (MCB). *See* Interphase chromosome-specific multicolor banding (ICS-MCB)
- N**
- Nonclonal chromosome aberration (NCCA), 92, 100
- Noncoding RNAs (ncRNA), 16–17
- Nuclear architecture
 chromatin compaction levels, 37
 euchromatin, 38
 heterochromatin, 38
 ICF syndrome
 chromosome 1 juxtacentromeric heterochromatin, 114
 DNA hypomethylation, 117
 DNA replication, 117–118
 interphase gene-heterochromatin, 117
 intranuclear positioning, 116
 long-range heterochromatin-gene, 115
- Nuclear organization
 chromatin movement, 13
 envelope, 11–12
 FISH, 11
 genes transcription, 11
 human embryos
 blastomeres, 131–132
 chromosome loci, 131, 133
 totipotency, 130
 lamina, 12
 nucleolus, 10–11
- P**
- Phosphoinositide 3-kinaserelated kinase (PIKK), 39
- pKi67, 14
- Premature chromosome condensation (PCC)
 application, 95
 definition, 93
 historical perspectives, 93–94
 mitotic catastrophe, 99
 pulverization and shattering, 98–99
vs. C-Frag, 95, 97–98
- Protamines, 140–141
- Q**
- QFISH, 183
- R**
- Radiation breakpoint clusters (RBPC), 43
- Rett syndrome (RTT), 195
- S**
- Satellite 2 DNA, 109
- Schizophrenia, 65–66
- Sex chromosome positioning, 152–153. *See* also Spermatozoa/sperm nucleus
- Sex chromosomes detection, 12
- S-FISH. *See* Suspension-based fluorescence in situ hybridization (S-FISH)
- Short interspersed repeated elements (SINES), 36
- Somatic copy number alterations (SCNA), 48
- Somatic genome variation, 61, 74
- Spermatozoa/sperm nucleus
 aneuploidy, 152–153
 chromosome organization, 149
 chromosome positioning
 intranuclear location, 149
 longitudinal position, 149–150
 MSCI, 150
 polarity, 150–151
 DNA packaging
 donut-loop model, 141
 histones replacement, 141–142
 protamines, 140–141
 sperm chromatin, 142
 toroidal structures, 141
 genome architecture abnormalities, 151–152
 sex chromosome positioning, 152–153
 sperm genome architecture
 centromeres clustering into chromocenter, 146–147
 FISH, 143–144
 hairpin configuration of chromosomes, 147–148
 longitudinal position, 144–145
 radial position, 145–146
 telomeres, 147

- Sperm genome architecture
- centromeres clustering into chromocenter, 146–147
 - FISH, 143–144
 - hairpin configuration of chromosomes, 147–148
 - longitudinal position, 144–145
 - radial position, 145–146
 - telomeres, 147
- Suspension-based fluorescence in situ hybridization (S-FISH)
- additional chromosomal fragments, tissues with, 174–175
 - DNA probes, 173–174
 - female tissues and X chromosome, 175
 - human sperm, 174
 - leukemia and chromosomes 8 and 21, 175–176
 - method, 173
- T**
- Toroidal structures, 141
- Translocation
- chromosomal aberrations, 41–42
 - chromosome positioning, 14
 - DNA damage, 41–42
 - fluorescence in situ hybridization (FISH), 124–125
 - gene repositioning, 24–25
 - human embryos, 124–125
- Tumor-deregulated genes (TDRG), 43
- X**
- X chromosome, 175. *See also* Spermatozoa/sperm nucleus
- Z**
- ZBTB24, 111
- ZMPSTE24, 17