

Contents

1	Historical Introduction.....	1
1.1	1956–1984.....	1
1.2	1985–1995.....	3
1.3	1996–2004.....	4
1.4	2005–2009.....	5
1.5	2010–2013.....	5
1.6	2014–2016.....	6
2	Normal Chromosomes	7
2.1	Introduction.....	7
2.2	Chromosome Number and Morphology.....	7
2.2.1	Non-Banding Techniques	7
2.2.2	Banding Techniques.....	8
2.2.3	X- and Y-Chromatin.....	9
2.3	Chromosome Band Nomenclature	9
2.3.1	Identification and Definition of Chromosome Landmarks, Regions, and Bands..	9
2.3.2	Designation of Regions, Bands, and Sub-Bands	11
2.4	High-Resolution Banding	12
2.5	Molecular Basis of Banding	14
3	Symbols and Abbreviated Terms	34
4	Karyotype Designation.....	37
4.1	General Principles	37
4.2	Specification of Breakpoints.....	40
4.3	Designating Structural Chromosome Aberrations by Breakpoints and Band Composition	40
4.3.1	Short System for Designating Structural Chromosome Aberrations.....	41
4.3.1.1	Two-Break Rearrangements.....	41
4.3.1.2	Three-Break Rearrangements.....	41
4.3.1.3	Four-Break and More Complex Rearrangements.....	42
4.3.2	Detailed System for Designating Structural Chromosome Aberrations.....	42
4.3.2.1	Additional Symbols	43
4.3.2.2	Designating the Band Composition of a Chromosome.....	43
4.4	Derivative Chromosomes	44
4.5	Recombinant Chromosomes.....	46

5	Uncertainty in Chromosome or Band Designation	47
5.1	Questionable Identification	47
5.2	Uncertain Breakpoint Localization or Chromosome Number.....	48
5.3	Alternative Interpretation.....	48
5.4	Incomplete Karyotype	48
6	Order of Chromosome Abnormalities in the Karyotype	50
7	Normal Variable Chromosome Features.....	52
7.1	Variation in Heterochromatic Segments, Satellite Stalks, and Satellites	52
7.1.1	Variation in Length.....	52
7.1.2	Variation in Number and Position	53
7.2	Fragile Sites	53
8	Numerical Chromosome Abnormalities.....	54
8.1	General Principles	54
8.2	Sex Chromosome Abnormalities.....	55
8.3	Autosomal Abnormalities.....	56
8.4	Uniparental Disomy.....	57
9	Structural Chromosome Rearrangements.....	58
9.1	General Principles	58
9.2	Specification of Structural Rearrangements	59
9.2.1	Additional Material of Unknown Origin.....	59
9.2.2	Deletions.....	60
9.2.3	Derivative Chromosomes	60
9.2.4	Dicentric Chromosomes.....	65
9.2.5	Duplications	67
9.2.6	Fission	67
9.2.7	Fragile Sites	67
9.2.8	Homogeneously Staining Regions	68
9.2.9	Insertions	68
9.2.10	Inversions.....	69
9.2.11	Isochromosomes	70
9.2.12	Marker Chromosomes.....	70
9.2.13	Neocentromeres.....	72
9.2.14	Quadruplications	72
9.2.15	Ring Chromosomes.....	72
9.2.16	Telomeric Associations	74
9.2.17	Translocations.....	75
9.2.17.1	Reciprocal Translocations	75
9.2.17.2	Whole-Arm Translocations.....	77
9.2.17.3	Robertsonian Translocations	78
9.2.17.4	Jumping Translocations	79
9.2.18	Tricentric Chromosomes	79
9.2.19	Tripllications	79
9.3	Multiple Copies of Rearranged Chromosomes.....	79

10	Chromosome Breakage.....	81
10.1	Chromatid Aberrations	81
10.1.1	Non-Banded Preparations	81
10.1.2	Banded Preparations	82
10.2	Chromosome Aberrations	82
10.2.1	Non-Banded Preparations	82
10.2.2	Banded Preparations	83
10.3	Scoring of Aberrations	83
11	Neoplasia.....	84
11.1	Clones and Clonal Evolution	84
11.1.1	Definition of a Clone.....	84
11.1.2	Clone Size.....	85
11.1.3	Mainline.....	85
11.1.4	Stemline, Sideline and Clonal Evolution	86
11.1.5	Composite Karyotype	88
11.1.6	Unrelated Clones	89
11.2	Modal Number	90
11.3	Constitutional Karyotype.....	90
12	Meiotic Chromosomes	92
12.1	Terminology	92
12.1.1	Examples of Meiotic Nomenclature	93
12.1.2	Correlation between Meiotic Chromosomes and Mitotic Banding Patterns.....	94
13	In situ Hybridization	100
13.1	Introduction.....	100
13.2	Prophase/Metaphase <i>in situ</i> Hybridization (ish)	101
13.2.1	Use of dim and enh	105
13.2.2	Subtelomeric Metaphase <i>in situ</i> Hybridization	106
13.3	Interphase/Nuclear <i>in situ</i> Hybridization (nuc ish)	106
13.3.1	Number of Signals.....	106
13.3.2	Relative Position of Signals	109
13.3.2.1	Single Fusion Probes	111
13.3.2.2	Single Fusion with Extra Signal Probes.....	111
13.3.2.3	Dual Fusion Probes	111
13.3.2.4	Break-Apart Probes	111
13.4	<i>In situ</i> Hybridization on Extended Chromatin/DNA Fibers (fib ish).....	112
13.5	Reverse <i>in situ</i> Hybridization (rev ish)	113
13.5.1	Chromosome Analyses Using Probes Derived from Sorted or Microdissected Chromosomes.....	113
13.6	Chromosome Comparative Genomic Hybridization (cgh).....	113
13.7	Multi-Color Chromosome Painting.....	114
13.8	Partial Chromosome Paints.....	114
14	Microarrays.....	115
14.1	Introduction.....	115
14.2	Examples of Microarray Nomenclature.....	116
14.2.1	Nomenclature Specific to SNP Arrays.....	121
14.2.2	Complex Array Results	122

15	Region-Specific Assays	123
15.1	Introduction.....	123
15.2	Examples of RSA Nomenclature for Copy Number Detection	123
15.3	Examples of RSA Nomenclature for Balanced Translocations or Fusion Genes ...	124
16	Sequence-Based Assays.....	125
16.1	Introduction.....	125
16.2	General Principles	125
16.3	Examples of Sequence-Based Nomenclature for Description of Chromosome Rearrangements	127
16.3.1	Deletions.....	127
16.3.2	Derivative Chromosomes	127
16.3.3	Duplications	128
16.3.4	Insertions	128
16.3.5	Inversions.....	128
16.3.6	Ring Chromosomes	128
16.3.7	Translocations.....	129
17	References	130
18	Members of the ISCN Standing Committee and Advisors.....	132
19	Appendix	134
20	Index	136