

Contents

Foreword	xiii
Bernard DUJON	
Introduction	xv
Guy-Franck RICHARD	
Chapter 1. Whole-Genome Duplications, a Source of Redundancy at the Entire-Genome Scale.	1
Elise PAREY and Camille BERTHELOT	
1.1. Prevalence of polyploids in the tree of life	2
1.1.1. Whole duplications in eukaryotes	2
1.1.2. Polyploidies in prokaryotic organisms.	6
1.1.3. Polyploid cells in normal and pathological physiology.	7
1.2. Mechanisms for the appearance of whole-genome duplications.	7
1.2.1. Non-separation of chromosomes after replication	7
1.2.2. Autopolyploidization, a perfect genome redundancy	9
1.2.3. Allopolyploidization, an overlapping of genomes of similar species	9
1.3. Cellular consequences of whole-genome duplications	11
1.3.1. Disruption of cell and nucleus organization	11
1.3.2. Modifications in the expression of genes and transposons.	13
1.3.3. Unstable meiosis	15
1.4. Rediploidization: evolutionary reduction in genetic redundancy	16
1.4.1. Resolution of meiosis by karyotype rearrangement	16

1.4.2. Evolutionary divergence of duplicated sequences	18
1.4.3. Bias and dominance during rediploidization	20
1.4.4. Incomplete and lineage-specific rediploidizations	21
1.5. Functions and evolution of duplicated genes	22
1.5.1. Redundancy and subfunctionalization	23
1.5.2. Neofunctionalization and evolutionary innovations	24
1.5.3. Gene repertoire bias	26
1.5.4. Regulatory blocks and splitting of regulatory regions	29
1.6. Whole-genome duplications and evolutionary diversification	32
1.6.1. Association with geological crises	32
1.6.2. Evolutionary speciations and radiations	33
1.7. Perspectives and conclusions	34
1.8. References	35
Chapter 2. Segmental Duplications and CNVs: Adaptive Potential of Structural Polymorphism	47
Patricia BALARESQUE and Franklin DELEHELLE	
2.1. The multiple facets of genetic polymorphism	48
2.2. From Segmental Duplications to Copy Number Variants: terminology	49
2.3. SDs: a general overview	49
2.3.1. Background	49
2.3.2. SDs: more than a category of sequences, superstructures	50
2.3.3. SD and CNV: study biases related to the attractiveness of subjects as well as to the technological developments of the moment	51
2.3.4. SD: characteristics in human and non-human primates	52
2.4. Methodologies for detecting structural variation in genomes	53
2.4.1. In vitro methods	54
2.4.2. Methods on reads	54
2.4.3. Post-assembly methods	54
2.5. The molecular mechanisms at the origin of structural variation	56
2.5.1. Homologous recombination mechanisms	56
2.5.2. Non-homologous recombination mechanisms	57
2.6. Regions rich in SDs/LCRs favor the creation of CNVs: insertions/duplications, deletions and inversions	58
2.6.1. Insertions/duplications and deletions	58
2.6.2. Inversions	60
2.7. From SDs to CNVs in humans and primates	61
2.7.1. General overview	61
2.7.2. Delineating regions of interest	61

2.7.3. Heterogeneity in the distribution of intra- and interchromosomal SDs.	62
2.7.4. Intrachromosomal and interchromosomal SDs: what do they teach us about the evolutionary history and origin of SDs?	62
2.7.5. Intra- and interchromosomal SDs: the specific case of sex chromosomes.	66
2.7.6. SDs: an association with specific sequences?	66
2.8. SDs in little-studied species: general genomic profiles	66
2.8.1. Twelve genomes under study	68
2.8.2. Distribution and characteristics of SDs in genomes	70
2.9. SD content: impact of a duplicated environment on sequences that make up the SDs.	70
2.9.1. SDs and non-coding sequences: the case of microsatellites	71
2.9.2. SDs and coding genes: the fate of genes in SDs	72
2.10. SDs and epigenetic modifications	75
2.11. The adaptive potential of SDs: between the benefit of innovation and the cost of pathology	78
2.11.1. The organism's defense: immune system	79
2.11.2. Nutrient/food assimilation	80
2.11.3. Sensory perception of the environment	80
2.11.4. Neurological processes	82
2.11.5. Reproduction and the X and Y chromosomes: true SD concentrates	83
2.12. SDs and associated CNVs: their roles in species adaptation to changes in environments.	86
2.12.1. SDs: a link between genomic architecture, adaptive potential and environmental changes?	86
2.12.2. Adaptation to global environmental stress.	86
2.12.3. Adaptation to nutrient-poor surroundings	88
2.12.4. Adaptation to low and high temperatures	88
2.12.5. Heavy-metal adaptation	89
2.12.6. Antibiotics and drugs	90
2.12.7. Pesticide resistance	90
2.12.8. Domestication and post-domestication of plant and animal species.	91
2.12.9. Competition and evolutionary success: invasive species and hybridization	93
2.13. Conclusion	94
2.14. Glossary of terms.	95
2.15. References.	96

Chapter 3. Transposable Elements: Parasites that Shape Genome Evolution	117
Amandine BONNET, Karine CASIER, Clément CARRÉ, Laure TEYSSET and Pascale LESAGE	
3.1. Transposable elements in eukaryotic genomes	117
3.1.1. TEs: essential components of eukaryotic genomes	118
3.1.2. Acquisition of new TEs by horizontal transfer	119
3.2. Classification of TEs and transposition mechanisms	120
3.2.1. Class I retrotransposons	120
3.2.2. Class II DNA transposons	123
3.3. TE self-regulation	123
3.3.1. Spatio-temporal regulation of TE expression	124
3.3.2. Self-regulation of transposition efficiency	125
3.3.3. Selective integration to better protect the genome	125
3.4. TE restriction by the host	129
3.4.1. Transcriptional repression of genomic copies	129
3.4.2. TE transcripts: choice targets for multiple restrictions	132
3.4.3. The Swiss knives of TE restriction: piRNAs	134
3.4.4. Reverse transcription of retroelements: a key step to inhibit	139
3.5. The impact of transposition events on genomes	140
3.5.1. The structural and functional consequences of TE activity on the genome	140
3.5.2. Pathologies associated with TE activity	144
3.5.3. The impact of TEs on the evolution of the host	148
3.6. Conclusion	155
3.7. References	156
Chapter 4. Insights Into the Evolutionary Diversity of Centromeres	181
Nuria CORTES-SILVA, Aruni P. SENARATNE and Ines A. DRINNENBERG	
4.1. The centromere	181
4.1.1. Definition and historical background	181
4.1.2. Two main types of centromeric architectures	183
4.2. Monocentromeres	184
4.2.1. The diversity of monocentric architectures across fungi	184
4.2.2. Animal and plant models contain long repetitive regional centromeres	190
4.3. Holocentromeres	192
4.3.1. Nematodes	193

4.3.2. Plants	195
4.3.3. Insects.	196
4.4. Open questions.	198
4.5. Acknowledgments.	198
4.6. References	198
Chapter 5. Evolution and Functions of Telomeres	207
Arturo LONDOÑO-VALLEJO	
5.1. Primary structure of telomeres.	207
5.1.1. Origin and evolution of telomeres	210
5.1.2. Nucleoprotein structure of telomeres	212
5.2. A telomere specific higher order structure: the T-loop	215
5.2.1. Telomere replication, a fundamental mechanism for telomere maintenance	215
5.3. Telomere lengthening mechanisms.	220
5.4. Telomere length homeostasis	222
5.5. Telomeres and genome organization and function.	225
5.6. Cell senescence, aging and disease	226
5.7. Conclusion	227
5.8. Acknowledgments.	227
5.9. References	227
Chapter 6. G-quadruplexes: Structure, Detection and Functions	239
Emilia Puig LOMBARDI	
6.1. From guanine-guanine base-pairing to a secondary structure	239
6.1.1. G-quartets	239
6.1.2. Folding into a G-quadruplex structure.	241
6.2. The G4 structure: variations on a theme	243
6.2.1. RNA G-quadruplexes (rG4).	245
6.2.2. Exceptions to the rule(s): non-canonical G-quadruplexes	245
6.3. Finding G-quadruplexes in a genome	246
6.3.1. Experimental methods for G-quadruplex detection	247
6.3.2. Computational methods	250
6.4. Biological roles of G-quadruplexes.	257
6.4.1. First role attributed to quadruplexes: their formation in telomeres.	257
6.4.2. Predictions based on bioinformatic analyses	259
6.5. Perspective: G-quadruplexes as anticancer therapeutic targets.	261
6.6. References	264

Chapter 7. Satellite DNA, Microsatellites and Minisatellites	273
Wilhelm VAYSSE–ZINKHÖFER and Guy-Franck RICHARD	
7.1. Satellite DNAs, origin and definition.	273
7.1.1. Minisatellites.	274
7.1.2. Microsatellites	274
7.2. From semantics to biology.	275
7.2.1. Distribution of satellite DNAs in genomes	275
7.2.2. Polymorphic genetic markers	277
7.2.3. Trinucleotide repeat expansions	281
7.2.4. Microsatellites regulate gene expression	283
7.2.5. Minisatellites are important in cell adhesion	285
7.2.6. Function of megasatellites.	287
7.2.7. Centromeric satellite DNA, complexity of structure–function studies	288
7.3. The evolutionary mechanisms of tandem repeats	289
7.3.1. Historical model of slippage during replication	290
7.3.2. Slippage during DNA repair	292
7.3.3. Repeat expansions and contractions during homologous recombination	292
7.4. Microsatellites in human diseases.	297
7.4.1. Triplet repeat expansion disorders	297
7.4.2. Colorectal cancers and the mismatch repair system.	298
7.4.3. Fragile sites	299
7.5. De novo formation and evolution of tandem repeats.	300
7.5.1. Birth and death of microsatellites	300
7.5.2. Formation of minisatellites	304
7.6. Perspectives	307
7.6.1. Inadequacy of software tools	307
7.6.2. The importance of definitions in biology	310
7.7. Acknowledgments.	311
7.8. References	311
Chapter 8. CRISPR-Cas: An Adaptive Immune System	319
Marie TOUCHON	
8.1. A brief history of the discovery of CRISPR-Cas systems.	319
8.2. General characteristics of CRISPR-Cas systems	323
8.2.1. Diversity of repeats	324
8.2.2. Diversity and origin of spacers	325
8.2.3. Diversity and evolutionary classification of cas genes	327

8.2.4. Origin of CRISPR-Cas systems	329
8.3. Evolution of CRISPR-Cas systems	330
8.3.1. Scattered distribution of CRISPR-Cas systems	330
8.3.2. Massive transfer of CRISPR-Cas systems	331
8.3.3. Commonly lost systems	332
8.3.4. Evolutionary dynamics of CRISPR arrays	333
8.4. An adaptive immune system.	334
8.4.1. A three-stage immune response.	334
8.4.2. Diversity of CRISPR-Cas molecular mechanisms.	337
8.4.3. Self- and non self-discrimination: avoiding self-targeting by CRISPR	340
8.5. Phage escape mechanisms	341
8.5.1. Genomic modifications	341
8.5.2. Anti-CRISPR proteins	343
8.6. Biological cost of CRISPR-Cas systems.	344
8.6.1. Cost of expression	344
8.6.2. Cost of autoimmunity	345
8.6.3. The genetic background of the host	346
8.6.4. Limiting horizontal gene transfer.	347
8.6.5. Naïve and primed adaptation	348
8.7. Importance in nature: impact of ecological factors.	349
8.7.1. Phage diversity – mutation rate.	349
8.7.2. Phage diversity – population size.	350
8.7.3. Infectious risk – alternative strategies	350
8.8. Conclusions and perspectives	351
8.9. References	353
List of Authors	361
Index	363