
Contents

Dedication v

In Memorium vii

Preface ix

Contributors..... xv

PART I INTRODUCTION

1 The CMT1A Duplication: *A Historical Perspective Viewed
 From Two Sides of an Ocean* 3
 James R. Lupski and Vincent Timmerman

PART II GENOMIC STRUCTURE

2 *Alu* Elements 21
 Prescott Deininger

3 The Impact of LINE-1 Retrotransposition on the Human
 Genome 35
 **Amy E. Hulme, Deanna A. Kulpa, José Luis Garcia Perez,
 and John V. Moran**

4 Ancient Transposable Elements, Processed Pseudogenes,
 and Endogenous Retroviruses 57
 Adam Pavlicek and Jerzy Jurka

5 Segmental Duplications 73
 Andrew J. Sharp and Evan E. Eichler

6 Non-B DNA and Chromosomal Rearrangements 89
 Albino Bacolla and Robert D. Wells

7 Genetic Basis of Olfactory Deficits 101
 Idan Menashe, Ester Feldmesser, and Doron Lancet

8 Genomic Organization and Function of Human
 Centromeres 115
 Huntington F. Willard and M. Katharine Rudd

PART III GENOME EVOLUTION

9 Primate Chromosome Evolution 133
 Stefan Müller

10 Genome Plasticity in Evolution: *The Centromere Repositioning* 153
 Mariano Rocchi and Nicoletta Archidiacono

PART IV GENOMIC REARRANGEMENTS AND DISEASE TRAITS

- 11 The CMT1A Duplication and HNPP Deletion 169
Vincent Timmerman and James R. Lupski
- 12 Smith-Magenis Syndrome Deletion, Reciprocal Duplication
dup(17)(p11.2p11.2), and Other Proximal
17p Rearrangements 179
Paweł Stankiewicz, Weimin Bi, and James R. Lupski
- 13 Chromosome 22q11.2 Rearrangement Disorders 193
Bernice E. Morrow
- 14 Neurofibromatosis 1 207
Karen Stephens
- 15 Williams-Beuren Syndrome 221
Stephen W. Scherer and Lucy R. Osborne
- 16 Sotos Syndrome 237
Naohiro Kurotaki and Naomichi Matsumoto
- 17 X Chromosome Rearrangements 247
Pauline H. Yen
- 18 Pelizaeus-Merzbacher Disease and Spastic Paraplegia Type 2 263
Ken Inoue
- 19 Y-Chromosomal Rearrangements and Azoospermia 273
Matthew E. Hurles and Chris Tyler-Smith
- 20 Inversion Chromosomes 289
*Orsetta Zuffardi, Roberto Ciccone, Sabrina Giglio,
and Tiziano Pramparo*
- 21 Monosomy 1p36 As a Model for the Molecular Basis
of Terminal Deletions 301
Blake C. Ballif and Lisa G. Shaffer
- 22 inv dup(15) and inv dup(22) 315
Heather E. McDermid and Rachel Wevrick
- 23 Mechanisms Underlying Neoplasia-Associated Genomic
Rearrangements 327
Thoas Fioretos

PART V FUNCTIONAL ASPECTS OF GENOME STRUCTURE

- 24 Recombination Hotspots in Nonallelic Homologous
Recombination 341
Matthew E. Hurles and James R. Lupski
- 25 Position Effects 357
Paweł Stankiewicz

PART VI GENOMIC DISORDERS: MODELING AND ASSAYS

- 26 Chromosome-Engineered Mouse Models 373
Pentao Liu
- 27 Array-CGH for the Analysis of Constitutional Genomic
Rearrangements 389
*Nigel P. Carter, Heike Fiegler, Susan Gribble,
and Richard Redon*

PART VII APPENDICES

- Appendix A: Well-Characterized Rearrangement-Based
Diseases and Genome Structural Features at the Locus 403
Paweł Stankiewicz and James R. Lupski
- Appendix B: Diagnostic Potential for Chromosome
Microarray Analysis 407
Paweł Stankiewicz, Sau W. Cheung, and Arthur L. Beaudet

- Index 415
- About the Editors 427

Genomic Disorders

The Genomic Basis of Disease

Lupski, J.R.; Stankiewicz, P.T. (Eds.)

2006, XVIII, 426 p. 112 illus., 1 illus. in color., Hardcover

ISBN: 978-1-58829-559-0

A product of Humana Press