Table of Contents

Preface xviii
Contributors xix

CHAPTER 1  The Organismal Prospect  1
Mark Pagel and Andrew Pomiankowski

Evolutionary Genetics in the Time of Genomics  1
The Organism in the Genome  3
Complexity and Regulation  4
The Origins of Novelty  6
Robustness and Evolvability  8
Genome Immunity  9
Identifiying Selective Change  9
What If Evolutionary Genomic and Proteomic Scientists Succeed?  10

CHAPTER 2  Evolutionary Systems Biology  11
Eugene V. Koonin and Yuri I. Wolf

Systems Biology: The New Big Science of “Omes”  11
Systems Biology in the Light of Evolution: The Quest for the Link between Genomic and Phenotypic Evolution  13
Correlations between Variables Characterizing Genome Evolution and Functioning  16
The Multidimensional Space of Omics  20
Glimpses of Biology behind the Correlations  22
Some Generalizations: Evolutionary Systems Biology—a Nascent Field in Turmoil  24
Acknowledgments  25
CHAPTER 3 The Origin of New Genes 27
Chuanzhu Fan, J. J. Emerson, and Manyuan Long

Introduction 27
Recent Discoveries of New Genes 27
Mechanisms to Generate New Genes 30
Evolutionary Forces for New Gene Retention 32
The Location and Movement of New Genes 33
Inferring the Functionality of New Genes 33
General Methods to Detect New Genes 35
  Early findings 35
  Comparative molecular cytogenetic analyses 35
  Computational genomic analysis 35
Comparative Genomic Hybridization to Detect New Genes 36
  Challenges in using array-based CGH in new gene studies 39
Outlooks and Perspectives 42

CHAPTER 4 Lateral Gene Transfer 45
W. Ford Doolittle, Camilla L. Nesbo, Eric Bapteste, and Olga Zhaxybayeva

Introduction 45
Defining terms 45
A Very Brief History of Lateral Genomics 46
Mechanisms of Lateral Gene Transfer 46
  Transduction 47
  Conjugation 48
  Transformation 50
Detecting LGT 51
  Compositional methods 51
  Unusual phyletic similarity patterns 51
  Phylogenetic incongruence 52
  Patchy (anomalous) gene distribution among phyla and species 52
  Strain-specific genes (patchy distribution within species) 53
  Physical methods 57
Ever-Present Alternatives 58
Quantifying LGT 61
  Genome-centric approaches 61
  Gene-centric methods 61
An LGT-Resistant Core 63
  The complexity hypothesis 64
CHAPTER 5  Evolution of Genomic Expression  81
Bernardo Lemos, Christian R. Landry, Pierre Fontanillas, Susan C. P. Renn, Rob Kulathinal, Kyle M. Brown, and Daniel L. Hartl

Introduction  81
The Complex Regulation of Genomic Expression  81
Classical Transcriptional Regulation in Cis and Trans  83
Promoters, enhancers, repressors, transcription factors, and regulatory proteins  83
Epigenetic Regulation and Chromatin Modifications  86
DNA methylation  87
Histone modifications  88
Chromosome territories and nuclear architecture  89
Post-Transcriptional Regulation  91
Measuring Attributes of Genomic Expression with Experimental and Computational Tools  93
Experimental approaches  93
Computational approaches  99
The Evolution of Genomic Expression: What Do We Know?  103
Genomic expression and morphological evolution  104
Molecular evolution of regulatory sequences  105
Stabilizing selection, positive selection, and neutrality of gene expression levels  107
Genomic attributes and rates of gene expression evolution  109
Inheritance of gene expression levels: Regulatory variation in cis and trans  111
Genotype-by-environment interactions, sex-biased genes, and epistasis  115

© Sinauer Associates, Inc. This material cannot be copied, reproduced, manufactured or disseminated in any form without express written permission from the publisher.
viii Contents

Gene Regulatory Networks, Subnetworks, and Modules 117
Conclusion 118

CHAPTER 6 The Evolution of Proteome Complexity and Diversity 119
László Patthy

Introduction 119
Evolution of Protein Complexity 120
Evolution of Proteome Complexity 123
Multidomain Proteome Complexity 126
Proteome Interaction Networks 127
Multidomain Proteins and Organismic Complexity 133
Factors Favoring the Formation of Multidomain Proteins 136
Conclusion 137
Acknowledgments 139

CHAPTER 7 Genomic Redundancy and Dispensability 141
Laurence D. Hurst and Csaba Pá1

Introduction 141
Are “Dispensable” Genes Really Dispensable? 143
Nonessential genes are typically under strong purifying selection 146
By What Mechanisms Are Nontrivially Dispensable Genes Dispensable? 148
Network architecture and distributed robustness 151
How do duplicates provide backup? 153
Backup by paralogs or by distributed robustness: which is more important? 156
Why Did Dispensability Evolve? 157
General conditions for the evolution of redundancy 158
Specific models for dispensability support the “side-consequence” hypothesis 160
Discussion 164

CHAPTER 8 Genome Defense 167
Christopher B. Schaefer, Mary Grace Goll, and Timothy H. Bestor

Introduction 167
Transposable elements 167
Transposons compromise genomic stability 169
Transposable elements are a greater threat to sexual hosts 170
Genome defense systems must continuously evolve 171
Contents

Transposons and the expansion of genomes 172
Mechanisms of Genome Defense 173
RNA interference 173
Cosuppression 176
Quelling 176
Meiotic silencing by unpaired DNA 177
DNA elimination 177
Transcriptional gene silencing 178
Systemic silencing 180
Cytosine Methylation 181
Restriction modification in bacteria 181
5-Methylcytosine and heritable silencing in eukaryotes 181
DNA methylation in vertebrates 183
Methylation in insects 185
Methylation in plants 185
Methylation in N. crassa 186
Editing as a Form of Defensive Mutagenesis 187
Restriction of retroviruses by G-to-A hypermutation 187
A-to-I editing of SINE elements 189
Conclusion 189

CHAPTER 9  Sex-Biased Genomic Expression 193
Brian Oliver

Introduction 193
One Species, Two Optimal Genomes 194
Y Chromosomes 195
X Chromosomes 198
Comparing Mammalian, Drosophila, and C. elegans X Chromosomes 201
Moving Forward 204

CHAPTER 10  Sex Chromosome Origins and Evolution 207
D. Charlesworth

Introduction: The Diversity of Sex Chromosomes 207
The Origins of Sex-Determining Loci and Recombination Suppression around the Sex-Determining Locus Region 213
Evolution of an initial sex-determining region 213
Recombination suppression 214
Evolution of separate sexes and proto–sex chromosomes 214

© Sinauer Associates, Inc. This material cannot be copied, reproduced, manufactured or disseminated in any form without express written permission from the publisher.
Evolution in Young and Proto–Sex Chromosomes: Sexually Antagonistic Genes 217
Determining the Age of Sex Chromosomes 218
   Phylogenetic evidence 218
   Divergence between X- and Y-linked sequences 218
Degeneration and Loss of Adaptation of Genes on the Nonrecombining Sex Chromosome 219
   Degeneration due to deleterious mutations 221
   Interference with the spread of favorable Y-linked mutations 223
   Selective sweeps 223
   Evidence for degeneration processes 223
Do Y Chromosomes Always Degenerate, and What Determines Which Genes Are Retained? 225
   Transposable element accumulation 227
   Low gene density of MSY regions 227
How Is Recombination Suppressed? 228
   Chromosome inversions 228
   Progressive recombination suppression 228
Further Chromosome Rearrangements 231
Neo–Sex Chromosome Formation 231
   Molecular evolutionary evidence for genetic degeneration in neo–sex chromosomes 232
   Neo–sex chromosomes in species other than Drosophila 234
Evolution and Adaptation of Ancient Sex Chromosomes: Evolution of Dosage Compensation and of Changed Gene Content 235
   Dosage compensation 235
Discussion: Adaptation of Sex Chromosomes 238

CHAPTER 11 Molecular Signatures of Adaptive Evolution 241
Alan Filipski, Sonja Prohaska, Sudhir Kumar

Introduction 241
Codon Usage Bias and the Estimates of Selection Ratio (ω) 243
Hypermutability of CpG Dinucleotides and the Estimates of Selection Ratio 247
Using Pseudogenes to Estimate Nonsynonymous Mutation Rates 248
Rate of Intron Divergence as a Proxy for Nonsynonymous Mutation Rates 250
Direct Estimates of Mutation Rates in the Laboratory 252
Conclusion 253
Acknowledgments 254

© Sinauer Associates, Inc. This material cannot be copied, reproduced, manufactured or disseminated in any form without express written permission from the publisher.
CHAPTER 12  Gene Networks and Natural Selection  255
Andreas Wagner

Introduction  255
Is There a Network Biology? 256
Natural Selection and Network Parts  257
Natural Selection and Small-Scale, Local Network Features  261
Natural Selection and Global Network Structure  263
  Natural selection and the degree distribution  264
  Global features of transcriptional regulation networks  266
  Alternative pathways in transcriptional regulation networks  266
Conclusion  269

CHAPTER 13  Human Evolutionary Genomics  271
Ines Hellmann and Rasmus Nielsen

Introduction  271
Mutations—the Sources of Genome Variability  272
  Point mutations  272
  Insertions, deletions, and inversions  274
Recombination  276
Transposable Elements  278
Evolution through Gene Duplication  280
Analyses of Protein-Coding Regions  281
  Comparative data  283
  Detecting selective sweeps from SNP data  286
Human-Specific Genetic Adaptations  288
Selection on Noncoding Sequences  290
  Regulatory sequences  292
Expression Data  293
Amount of Selection Affecting the Human Genome
  and the Genetic Load of Humans  295
Conclusion  297

References  299
Index  337