

Contents

Part I	Genetic Mapping	1
1	Mapping Populations and Principles of Genetic Mapping	3
	<i>Katharina Schneider</i>	
	Overview	3
	Abstract	3
1.1	Introduction	4
1.2	Mapping Populations	6
1.2.1	Mapping Populations Suitable for Self-fertilizing Plants	7
1.2.1.1	F ₂ Populations	7
1.2.1.2	Recombinant Inbred Lines	8
1.2.1.3	Backcross Populations	9
1.2.1.4	Introgression Lines: Exotic Libraries	10
1.2.1.5	Doubled Haploid Lines	11
1.2.2	Mapping Populations for Cross-pollinating Species	12
1.2.3	Two-step Strategies for Mapping Mutants and DNA Fragments	12
1.2.4	Chromosome-specific Tools for Mapping	13
1.2.5	Mapping in Natural Populations/Breeding Pools	14
1.2.6	Mapping Genes and Mutants to Physically Aligned DNA	15
1.2.7	Specific Mapping Problems	15
1.3	Discussion	17
	Acknowledgments	18
	References	19
2	Molecular Marker Systems for Genetic Mapping	23
	<i>Henry T. Nguyen and Xiaolei Wu</i>	
	Abstract	23
2.1	Introduction	23
2.2	DNA-based Markers Popularly Used in Genetic Mapping	25
2.2.1	RFLP	25
2.2.1.1	Conventional RFLP Analysis	26
2.2.1.2	PCR-RFLP	26
2.2.1.3	Mismatch PCR-RFLP	27

2.2.2	RAPD	28
2.2.3	SSR Markers	29
2.2.3.1	Conventional SSR Analysis	30
2.2.3.2	ISSR	34
2.2.3.3	STMP	36
2.2.4	AFLP	36
2.2.4.1	Conventional AFLP Analysis	36
2.2.4.2	f-AFLP	39
2.2.4.3	cDNA-AFLP and HiCEP	40
2.2.4.4	TE-AFLP	42
2.2.4.5	MEGA-AFLP	42
2.2.4.6	MITE-AFLPs	44
2.2.4.7	AFLP Conversion	45
2.2.5	REMAP and IRAP	47
2.2.5.1	IRAP	48
2.2.5.2	REMAP	48
2.2.6	SRAP	48
2.3	Discussion	49
	References	50
3	Methods and Software for Genetic Mapping	53
	<i>James C. Nelson</i>	
	Overview	53
	Abstract	53
3.1	Introduction	54
3.1.1	Methods and Tools for Genetic Linkage Mapping in Plants	54
3.1.1.1	Statement of the Problem	54
3.1.2	Locus Grouping	55
3.1.3	Locus Ordering	55
3.1.4	Multilocus Distance Estimation	57
3.1.5	Using Variant and Mixed Cross Designs	57
3.1.5.1	Outbreeding Species	57
3.1.5.2	Autopolyploid Species	58
3.1.5.3	Combining Datasets	58
3.1.6	Linkage-mapping Software Availability, Interfaces, and Features	58
3.2	Methods and Tools for QTL Mapping in Plants	59
3.2.1	Statement of the Problem	59
3.2.2	Single-marker Association	60
3.2.2.1	Metric Traits	60
3.2.2.2	Categorical Traits	60
3.2.3	Interval Mapping: Simple (SIM)	61
3.2.3.1	ML Methods	61
3.2.3.2	Least-squares (Regression) and Nonparametric Methods	61
3.2.4	Interval Mapping: Composite (CIM)	63
3.2.5	Significance Testing	63

3.2.6	Interval Mapping: Multiple-QTL Model Building	64
3.2.6.1	Stepwise and Exhaustive-search Methods for Building Multiple-QTL Models	64
3.2.6.2	Markov Chain Monte Carlo (MCMC) Methods	65
3.2.6.3	Genetic Algorithms	65
3.2.7	Multiple-trait (MT) QTL Mapping	66
3.2.8	Multiple-cross (MC) QTL Mapping	66
3.2.9	Computational Optimization Methods	66
3.3	Future Directions in Mapping Methods and Tools	67
3.3.1	Future of Linkage and QTL Mapping	67
3.3.2	Adequacy of Software Tools for Plant Mapping	68
3.3.2.1	Software Merit Criteria	68
3.3.2.2	Analytical Scope	68
3.3.2.3	Ease of Learning and Use	68
3.3.2.4	Accessibility and Extensibility	69
3.3.3	A Development Model for Public Genetic Mapping Software	69
	References	70
4	Single nucleotide Polymorphisms: Detection Techniques and Their Potential for Genotyping and Genome Mapping	75
	<i>Günter Kahl, Andrea Mast, Nigel Tooke, Richard Shen, and Dirk van den Boom</i>	
4.1	Introduction	75
4.2	Selected Techniques	80
4.2.1	SNP Analysis I: The Invader Technology	80
4.2.1.1	Introduction	80
4.2.1.2	Cleavase Enzymes	80
4.2.1.3	Oligonucleotides and Structure	80
4.2.1.4	Probe Cycling and Signal Amplification	81
4.2.1.5	DNA Format	81
4.2.1.6	RNA Format	81
4.2.1.7	Alternate Detection Formats	82
4.2.1.8	Specificity	83
4.2.1.9	Robustness	85
4.2.1.10	Invader Applications	85
4.2.1.11	Conclusion	85
4.2.2	SNP Analysis II: Pyrosequencing	85
4.2.2.1	Introduction	85
4.2.2.2	SNP Genotyping Using Pyrosequencing Technology	86
4.2.2.3	Allele Frequency Quantification	88
4.2.2.4	Haplotyping	90
4.2.3	SNP Analysis III: A Scalable High-multiplex SNP Genotyping Platform	91
4.2.3.1	Introduction	91
4.2.3.2	The Illumina Genotyping Platform	91
4.2.3.3	Genotyping Data	95

4.2.3.4	Conclusion	96
4.2.4	SNP Analysis IV: High-throughput SNP Analysis by MALDI-TOF MS	96
4.2.4.1	Introduction	96
4.2.4.2	SNP Analysis Using the Massarray Platform	97
4.3	Conclusions and Perspectives	103
	Acknowledgments	104
	References	104
5	Breeding By Design: Exploiting Genetic Maps and Molecular Markers Through Marker-assisted Selection	109
	<i>Johan D. Peleman, Anker P. Sørensen, and Jeroen Rouppe van der Voort</i>	
	Abstract	109
5.1	Introduction	110
5.2	Marker-assisted Selection	111
5.2.1	Genetic Distance Analysis, Variety Identification, and Seed Purity Analysis	113
5.2.2	Indirect Selection	113
5.2.2.1	Monogenic Traits	113
5.2.2.2	Polygenic (Quantitative) Traits	114
5.2.2.3	Marker-assisted Backcrossing	116
5.3	The Creation of Novel Varieties (Marker-assisted Breeding)	116
5.3.1	Removal of Linkage Drag	117
5.3.2	Pyramiding Resistance Genes	117
5.3.3	Marker-assisted Breeding of Polygenic Traits	119
5.3.4	Introduction of Novel Characteristics	120
5.3.5	Effective Exploitation of (Exotic) Germ Plasm	120
5.4	Breeding by Design	121
5.4.1	Mapping Loci Involved in All Agronomically Relevant Traits	121
5.4.2	Assessment of the Allelic Variation at the Loci Associated With Agronomically Relevant Traits	124
5.4.3	Breeding by Design	125
5.4.4	Future of Breeding by Design	127
	Acknowledgments	127
	References	128
Part II	Physical Mapping	131
6	Physical Mapping of Plant Chromosomes	133
	<i>Barbara Hass-Jacobus and Scott A. Jackson</i>	
6.1	Introduction	133
6.2	Classical Physical Mapping Techniques	134
6.2.1	Knob Mapping in Maize	134
6.2.2	Deletion/Aneuploid/Substitution Mapping	134
6.2.3	Polytene Chromosomes in <i>Drosophila</i>	136
6.2.4	Chromosome Banding	136

6.3	Molecular Physical Mapping Techniques	139
6.3.1	CHEF Gel Mapping	139
6.3.2	Radiation Hybrid Mapping	139
6.3.4	Large-insert Clone Libraries (YACs, BACs, Cosmids)	140
6.3.5	Fluorescent <i>in situ</i> Hybridization	142
6.3.6	Mapping Gene Space	144
6.4	Discussion	145
	References	146
7	Chromosome Flow Sorting and Physical Mapping	151
	<i>Jaroslav Doležel, Marie Kubaláková, Jan Bartoš, and Jiří Macas</i>	
	Overview	151
	Abstract	151
7.1	Introduction	152
7.2	Development of Flow Cytogenetics for Plants	153
7.2.1	The Uses of Flow Karyotyping	155
7.2.2	Applications of Flow-sorted Chromosomes	155
7.3	Methodologies and Techniques	157
7.3.1	Cell Cycle Synchronization and Metaphase Accumulation	157
7.3.2	Chromosome Isolation	158
7.3.3	Chromosome Analysis	158
7.3.3.1	Detection of Structural Chromosome Changes	160
7.3.3.2	Quantitative Detection of Numerical Chromosome Changes	160
7.3.3.3	Chromosome Sorting	160
7.3.3.4	Cytogenetic Mapping on Flow-sorted Chromosomes	162
7.3.3.5	Physical Mapping Using PCR	163
7.3.3.6	Chromosome and Chromosome Arm-specific DNA Libraries	165
7.3.3.7	Targeted Isolation of Molecular Markers	166
7.4	Discussion	167
	Acknowledgments	168
	References	168
8	Genomic DNA Libraries and Physical Mapping	173
	<i>Chengwei Ren, Zhanyou Xu, Shuku Sun, Mi-Kyung Lee, Chengcang Wu, Chantel Scheuring, and Hong-Bin Zhang</i>	
	Overview	173
	Abstract	173
8.1	Introduction	174
8.2	Methodologies and Techniques	176
8.2.1	Bacteria-based Large-insert DNA Clones	176
8.2.1.1	BAC	178
8.2.1.2	PAC	179
8.2.1.3	BIBACs	180
8.2.1.4	PBC	181
8.2.1.5	TAC	182

8.2.2	Genomic DNA Library Quality and Genome Physical Mapping	182
8.2.2.1	Insert Sizes	183
8.2.2.2	Clone Genome Coverage	184
8.2.2.3	Representation for Genome	185
8.2.2.4	The Importance of Binary Vectors in Plant Physical Mapping	187
8.2.3	Construction of Bacteria-based Large-insert Genomic DNA Libraries	188
8.2.3.1	Megebase-size Nuclear DNA Preparation	188
8.2.3.2	Vector Preparation	191
8.2.3.3	Library Construction	196
8.2.4	Applications of Bacteria-based Large-insert Genomic DNA Libraries in Genome Physical Mapping	202
8.2.4.1	Physical Mapping by Fingerprint Analysis	203
8.2.4.2	Physical Mapping by Fluorescent <i>in situ</i> Hybridization	204
8.2.4.3	Physical Mapping by Optical Mapping	204
8.2.4.4	Physical Mapping by Iterative Hybridization	205
8.2.4.5	Physical Mapping by Other Techniques	206
8.3	Discussion	208
	Acknowledgments	209
	References	210
9	Integration of Physical and Genetic Maps	215
	<i>Khalid Meksem, Hirofumi Ishihara, and Tacco Jesse</i>	
9.1	Introduction	215
9.2	Colony Hybridization Techniques for the Integration of DNA Sequences Into a Physical Map	216
9.3	Integrating Gene-rich Sequences into the Physical Map Via Overgo Hybridization	218
9.3.1	Overlapping Oligo Labeling	218
9.3.2	Hybridization	219
9.3.3	Washing	220
9.3.4	Autoradiography	220
9.4	Pooling Strategy for the Integration of DNA Sequences Into a Physical Map	220
9.4.1	DNA Pooling	220
9.4.2	PCR Multiplexing	222
9.4.3	DNA Isolation from Pooled Large-insert Clones	222
9.5	Forward and Reverse Integrated Physical-Genetic Mapping: Targeted DNA Marker Mapping	223
9.5.1	Integrated AFLP Mapping	224
9.5.2	Targeted SSR Mapping	225
9.5.2.1	Subcloning of Large-insert DNA Clones	226
9.5.2.2	Colony Hybridization	226

9.6	Bioinformatic Tools for an Integrated Physical-Genetic Map: Genome Browser	227
9.6.1	GBrowse	227
9.6.2	iMap	229
9.7	Discussion	230
	References	231
10	Positional Cloning of Plant Developmental Genes	233
	<i>Peter M. Gresshoff</i>	
	Overview	233
	Abstract	234
10.1	Introduction	235
10.2	Gene Discovery Through Insertional Mutagenesis	238
10.3	Technology Requirements for Map-based Cloning	240
10.4	Positional Cloning Successes in Legume Nodulation Genes	245
10.4.1	Nodulation Biology	245
10.4.2	Non-nodulation Genes	247
10.4.3	Autoregulation of Nodulation Genes	249
10.5	Conclusions	252
	Acknowledgments	253
	References	253
11	Whole-genome Physical Mapping: An Overview on Methods for DNA Fingerprinting	257
	<i>Chengcang Wu, Shuku Sun, Mi-Kyung Lee, Zhanyou Xu, Chengwei Ren, Teofila S. Santos, and Hong-Bin Zhang</i>	
	Overview	257
	Abstract	257
11.1	Introduction	258
11.1.1	Inception and Development of DNA Fingerprinting	258
11.1.2	Clone-based Whole-genome Physical Mapping	259
11.1.3	Source DNA Libraries for Whole-genome Physical Mapping	260
11.2	Techniques and Methodologies	261
11.2.1	Preparation of DNA from Bacteria-based Large-insert Clones	263
11.2.1.1	Isolation of DNA from Bacteria-based Large-insert Clones Using the Clone-by-Clone Approach	263
11.2.1.2	Isolation of DNA from Bacteria-based Large-insert Clones in 96-well Format Using a Manual Approach	264
11.2.1.3	Isolation of DNA from Bacteria-based Large-insert Clones in 96-well Format Using the AutoGenprep 960 Robotic Workstation	265
11.2.2	Fingerprinting the Clone DNA Using Restriction Enzymes	265
11.2.2.1	Agarose Gel-based, Restriction Fingerprinting Method	265
11.2.2.2	Polyacrylamide (Sequencing) Gel-based, Restriction Fingerprinting Method	268

11.2.2.3	Automatic Sequencing Gel- and Capillary Electrophoresis-based Restriction Fingerprinting Methods	271
11.2.3	Applications of DNA Fingerprinting for Genome Physical Mapping	275
11.2.3.1	Whole-genome Physical Maps Generated from Large-insert Bacterial Clones by Fingerprint Analysis	275
11.2.3.2	Comparison of Different Fingerprinting Methods	277
11.3	Discussion	280
	Acknowledgements	282
	References	282
12	Software for Restriction Fragment Physical Maps	285
	<i>William Nelson and Carol Soderlund</i>	
	Overview	285
12.1	Introduction	285
12.1.1	Review of Agarose Fingerprinting and FPC	285
12.2	HICF Techniques	288
12.2.1	Generalities	288
12.2.2	Approaches Using Type IIS Enzymes	289
12.2.3	SNaPshot HICF	290
12.3	Processing HICF Data	291
12.3.1	Peak Scoring	291
12.3.2	Sizing of Fragments	292
12.3.3	Quality and Contamination Screening	293
12.3.4	Vector and Repeat Screening	294
12.3.5	Packaging HICF Data for FPC	295
12.3.6	GenoProfiler	295
12.4	Building HICF Maps in FPC	296
12.4.1	Creating a New Project, Loading the Fingerprints, and Screening Vectors	297
12.4.2	Tolerance and Gel Length	298
12.4.3	Cutoff	299
12.4.4	Building	300
12.4.5	Build Quality, Q Clones, and the DQer	300
12.4.6	multiFPC	301
12.5	Theoretical Aspects	302
12.5.1	Simulations	302
12.5.2	Overlap Equations	303
	Acknowledgments	304
	References	304

- 13 Reduced Representation Strategies and Their Application to Plant Genomes 307**
Daniel G. Peterson
Overview 307
- 13.1 Introduction 308
- 13.2 Reduced Representation Techniques 311
- 13.2.1 EST Sequencing 311
- 13.2.2 Methylation Filtration 311
- 13.2.3 Cot-based Cloning and Sequencing 313
- 13.2.4 *De Novo* Polymorphism Discovery 316
- 13.2.4.1 Reduced Representation Shotgun (RRS) Sequencing 317
- 13.2.4.2 DOP-PCR 318
- 13.2.4.3 SSR Capture 318
- 13.3 Other Reduced Representation Techniques 320
- 13.4 Discussion 320
- 13.4.1 Repeat Sequence Enrichment 320
- 13.4.2 Sequencing Gene space 322
- 13.4.2.1 EST Sequencing 322
- 13.4.2.2 Methylation Filtration 323
- 13.4.2.3 Cot-based Cloning and Sequencing 325
- 13.4.2.4 Integration of Reduced Representation Strategies 327
- 13.4.3 Polymorphism Discovery 328
- 13.4.3.1 RRS Sequencing and DOP-PCR 328
- 13.4.3.2 Microsatellite Isolation 328
- 13.5 Conclusions 329
- Acknowledgments 330
- References 330
- 14 Large-scale DNA Sequencing 337**
Christopher D. Town
Abstract 337
- 14.1 Introduction 337
- 14.2 Motivation for Sequencing and Choice of Strategy 338
- 14.3 EST Sequencing 338
- 14.4 Large-scale Sequencing BAC-based Projects 339
- 14.5 Chromosome-based Sequencing 340
- 14.6 Whole-genome Shotgun Sequencing 341
- 14.7 Selective Genome Sequencing by Differential Cloning Strategies 341
- 14.8 Low-coverage Sequencing 341
- 14.9 BAC End Sequencing 343
- 14.10 The Sequencing Process Itself 343
- 14.10.1 Library Production 343
- 14.10.2 Template Production 344
- 14.10.3 Sequencing Reactions and Analysis 344
- 14.10.4 Sequencing Capacity and Costs 345

14.10.5	Post-sequencing Data Processing	345
14.10.5.1	Quality Trimming	345
14.10.5.2	Sequence Assembly	346
14.10.5.3	Scaffolding	346
14.10.5.4	Sequence Editing and Gap Closure	347
14.11	Conclusions	347
	Acknowledgments	348
	References	348

Glossary 353

Subject Index 371