

Index

a

acceptor-tagged nucleotide 99
acrylamide-based copolymers 154
acrylamide gel 59
– matrix 61
– monomer 58
affymetrix genome-wide human SNP array 206
agarose networks 156
allele-specific color tagging 125
amplicon library method 45
amplicon variant analyzer software 48
anatomically modern human (AMH) 191
Arabidopsis thaliana transcriptome 221
array technology 205

b

BAC fingerprinting 113
BAC libraries 50
bacterial cells 58
bacterial colonies 58
basic local alignment search tool (BLAST) 210
– algorithm 210
– method 81
– score 84
BCR-ABL translocation 171
bead-or filter-based methods 195
bead purification 31
BEAMing method 61, 64
biological systems 3
biological warfare agents 98
biomedical ethics 245, 248
bisulfite-treated DNA 211, 212
blunt-ended tags 237

c

Caenorhabditis elegans 217
CAP adapter 30

capillary array electrophoresis (CAE) 5, 6, 110, 153
– development of 153
– instrument 38, 157
– system(s) 154, 157
CCD cameras 141
cDNA fragments 169
cDNA libraries 219
cDNA sequences 114
ChIA-PET experiments 177
chromatin immunoprecipitation (ChIP) 16, 29, 39, 202
– derived fragments 204
– experiments 206
– method 173, 174, 203, 205
– paired end ditagging 169
– reactions 39
– SAGE 204
– technique 202
chromatin immunoprecipitation sequencing (ChIP-seq) 26, 201, 202, 208, 209
– challenges 209
– experiments 212
– history 202
– introduction 201
– medical applications 209
– method 26, 202, 203, 208, 209, 211, 212
– protocols 26, 201, 202, 208, 209
chromatin interactions 175, 176, 177
chromosomal conformation capture (3C) method 175
chromosome sequencing 72
clone-based genome sequencing 113
copy number variations (CNVs) 23
cross-hybridization noise 170
cross-linked polyacrylamide, *see* agarose networks
custom-written software 120

- cytosine 187
- hydrolytic deamination of 187

d

- data analysis, subsequent 86
- data processing software pipeline 46
- Deep SAGE 238
 - analysis experiment 231
 - data 235
 - procedure 232
 - protocol 237
- de novo* genome sequencing 36, 112
- dideoxy-based sequencing methods 207
- digital gene expression (DGE) 21
- digital karyotyping 75
- direct linear analysis (DLA) technique 120
- disease-related mutations 120
- ditags 232
- DNA (deoxy ribose nucleic acids) 15, 19, 26, 45, 104, 211
 - A-form 107
 - amplification 6, 58, 156
 - bisulfite-treated 19
 - double-stranded 107, 135
 - methylation of 211
 - sequencing of 26
 - single-stranded 105
- DNA backbone(s) 125, 143, 146
- DNA barcoding method 118, 119, 129, 131
 - DNA mapping 117
 - molecular haplotyping 117
- DNA chip 206, 230
- DNA-coated beads 59, 64
- DNA electrophoresis 157, 160
- DNA fragment(s) 5, 10, 15, 16, 18, 45, 118, 127, 128, 155, 168
 - blunt ended 16, 18
 - libraries 17, 204
- DNA hybridization technique 205, 230
- DNA-labeled beads 65
- DNA ladders 153
 - single-stranded 153
- DNA mapping method 118, 120, 121
- DNA methylation 23, 38
- DNA microarrays 170, 173, 230, 237
 - analysis 230
 - fabrication 170
- DNA miscoding lesions 187
- DNA molecular templates 134
- DNA molecules 58, 103, 107, 129, 134, 135, 137, 184, 202
- DNA polymerase(s) 4, 18, 19, 109, 119, 140, 141
 - *Bst* 140

- *Taq* 140
- *Tth* 140
- DNA polymorphisms 111
- DNA–protein interaction 173, 203, 207
- DNA replication process 99, 105
- DNA sample 127
 - electrophoretic processing of 117
 - single- molecule templates 133
- DNA separation device 5
- DNA separation matrix 159
- DNA sequence 5, 29, 34, 153
 - alignment algorithms 156
 - analysis 92, 113
 - assemblies 92
- DNA sequencing 18, 57, 97, 103, 159
 - high-throughput 57
 - libraries 31
 - platforms 167
 - real-time 97
 - technologies 43, 89, 90, 94, 111, 167
- DNASTAR 91, 92
 - solution 91–94
- DNA strand(s) 3, 4, 99, 100, 110
 - double-stranded molecules 104, 119, 130
 - synthesis 3, 4
- DNA template 4, 31, 45, 135, 147
- dual CPU Linux PC 85
- dye-labeled oligonucleotides 29, 30
- dynamic programming matrix (DPM) 80

e

- Efficient local alignment of nucleotide data (ELAND) 210
- EGFR mutations 50
- electrophoretic separation and detection 154
- end sequence profiling (ESP) 40
- enzyme-based chain-termination method 43
- enzyme-digested DNA 15
- epigenetic modifications 173
- ESP, *see* end sequence profiling
- EST, *see* expressed sequence tags
- eukaryotic cells 173
- eukaryotic genomes 50
- expressed sequence tags (ESTs) 229
 - analysis 229
 - sequencing 230

f

- false discovery rate (FDR) 238
- FASTA format data 79
- fluorescence signals 67, 230
- fluorescent tags 110
- fluorochrome intensities 146

fluorochrome-labeled nucleotides 135, 140
 fluorochrome-labeled PCR products 144
 Fourier transformation 107
 full width-at-half-maximum (FWHM) 143
 fusion transcripts 172

g

gapping nick sites 139
 Gaussian fit 145
 Gaussian-like distribution 209
 Gaussian parameters 143
 gel-based restriction mapping 130
 gel electrophoresis 16, 108, 147
 gene expression 36
 – analysis 229
 – array-based 36
 – profiles 238
 – tag-based 36
 gene Hit method, *see* SeqID Hit method
 gene identification signature (GIS) 169, 170
 genetic information 117
 – haplotype 117
 – RNA splicing pattern 117
 genome analyzer 15, 19, 20, 23, 26
 – instrument 20
 – pipeline software 20
 – software tools 21
 genome information 245–247
 genome methylation analysis 38
 genome/phenome associations 246
 genome protein/DNA interaction analysis 175
 genome resequencing 38
 genome sequencing 9, 23, 57
 – applications 23
 – epigenomics 23
 – multiplexing 26
 – protein–nucleic acid interactions 26
 – transcriptome analysis 23
 – impact of 9
 genome-tiling arrays 230
 genome-wide association studies (GWAS) 246
 genome-wide DNA interactions 209, 212
 genomic DNA fragments 119
 genomic DNA molecules 118, 135
 genomic DNA Mounting/Overlay 139
 genomic material 155
 genomic sequence assembly 120
 gigabase-sized genome 3, 50
 glass fabrication techniques 154
 GMAT, *see* ChIP-SAGE
 GS FLX system 44, 46, 50

h

Haemophilus influenzae 6
 haploid genomes 114
 haplotype analysis 114
 haplotype barcode(s) 125, 128
 heterochromatic knobs 8
 high-performance polymeric materials 160
 high-throughput DNA Sanger sequencing equipment 230
 high-throughput sequencing 246
 hippocratic database 247
 histone 24
 – ChIP-seq analysis 24
 histone-DNA interactions 203
 Hit method 82, 83
 human adenovirus 123, 124
 human/animal genome sequencing 50, 89, 114
 human diploid genome 103
 human genome project (HGP) 6, 9, 57, 117
 human transcript lengths 114
 hybridization-based gene arrays 205
 hybridization-based tag sequencing method 205
 hybridization chips 134
 hybridization probes 62
 hybridization techniques 9, 206
 hydrophobically modified polyacrylamides 159

i

Illumina's bead array technology 38
 Illumina genome analyzer II system 15–21
 – cluster creation 17–19
 – data analysis 20–21
 – library preparation 15–17
 – paired end reads 19–20
 – sequencing 19
 Illumina/Solexa and applied biosystems platforms 222
in situ hybridization 119
in situ template amplification 133
 Institute for Genomic Research 3
 Institutional Review Board (IRB) 248
 integrated microfluidic devices 154

k

Klenow fragment 188

l

lab-on-a-chip systems 156
 lambda DNA 121
 – sequence motif maps 121

- templates 148
- large-scale research ethics 249–250
- laser-induced fluorescence (LIF) 154, 156
- library preparation 15, 221
- DNA fragmentation 15–16
- end repair and ligation 16–17
- ligation-based sequencing 29
- ligation-mediated amplification (LMA) 175
- linear amplification 105
- linear DNA mapping techniques 120
- linear polyacrylamide (LPA) solution 157
- linker-flanked tags, *see* ditags
- linker-tag molecule 232, 237
- liquid-handling platforms 5
- locus control regions (LCRs) 175
- long-range PCR fragments 118
- LPA matrix 157

m

- mammalian genes 114
- mammalian transcriptomes 168
- massive parallel signature sequencing (MPSS) 66, 133, 221, 222, 234
- Mathies lab device 154
- Maxam–Gilbert's method 10
- microarray technologies 39
- microchip systems 154
- microfluidic devices 135, 153, 154, 156, 160, 231
- Agilent's Bioanalyzer 231
- PDMS 139
- microRNAs (miRNAs) 39, 217, 225
- background 217
- cloning frequencies of 225
- coding genes 217
- discovery 217, 223
- expression profiling 217, 225
- identification 218
- pathway model 218
- sized fragments 221
- molecular haplotyping, technology 125
- mRNA population, *see* mammalian transcriptome
- mRNA tag library sequencing 57
- mtDNA genomes 183
- *Anomalopteryx didiformis* 183
- *Dinornis giganteus* 183
- *Emeus crassus* 183
- multiplex sequencing of paired end ditags (MS-PET) 168

n

- nanosequencing machines 99
- neuron-restrictive silencer factor (NRSF) 208

- next-generation sequencing (NGS) 79, 83, 86
- data analysis 79, 86
- data sets 83
- projects 86
- sequence analysis, strategies 84
- system 27
- technologies 177
- next-generation software, DNASTAR's 89
- nicked/gapped templates 135
- Northern blot 223, 225, 230
- NRSF binding motif 208
- NRSF binding site 208
- nucleic acid sequence 133
- nucleotide pairs 104

o

- Olympus IX-71 microscope 120
- optical sequencing 133–140, 148
- microscope 137
- reactions 140
- surfaces 137
- organelle genome 192

p

- padlock probe ligation 119
- paired end ditag (PET) sequencing 167–170
- approach 170
- mapping regions 174
- methodology 168
- strategy 167
- technology 168, 179
- paleogenomic DNA 194
- paleogenomics 183
- parallel sequencing technologies 117
- personal genome project (PGP) 250
- phiX174 bacteriophage genome 3
- photopolymerized monolith 155
- plus and minus method 3
- point spread function (PSF) 120, 141
- Poisson distribution 31
- polony karyotyping 75
- polony sequencing method 57, 59, 61, 62, 66, 69, 70
- applications 57
- polony SAGE 73
- transcript characterization 73
- transcript profiling 73
- history 57–58
- system 67
- technology 57
- poly electrolyte multilayer (PEM)-modified glass surface 119
- polyacrylamide gel (PAG) 5
- polyadenylation sites (PAS) 168

- polymerase chain reaction (PCR) 10, 30, 31, 45, 58, 60, 109
 - amplicon 44, 141
 - amplification process 31, 43, 63, 108
 - amplified templates 33
 - based ultra-deep sequencing 47
 - chamber 156
 - inhibiting molecules 156
 - primers 34, 44, 124, 125, 176
 - primer sites 64
 - products 8, 143
 - reagents 58
 - sequencing analysis 184
 - polymer networks 156
 - polymorphic alleles tagged 125
 - direct haplotype determination 127
 - localization of 125
 - primer extension reactions 147
 - protein–DNA interaction 39, 201, 203, 204, 205, 209, 212
 - mapping of 201
 - sites 212
 - protein–nucleic acid interactions 21, 26
 - protein–protein interactions 203
 - prototype technique, *see* plus and minus method
- q**
- Q-PCR, *see* Northern blot
- r**
- rapid amplification of cDNA ends (RACE) reactions 230
 - RDBMS model 86
 - reaction chamber setup 137–139
 - real-time DNA sequencing 97, 100
 - real-time PCR method 62
 - repressor element-1 silencing transcription factor (REST) 208
 - restriction enzyme 235
 - *BsmFI* 235
 - *DpnII* 235
 - *NlaIII* 235
 - *Sau3A* 235
 - reverse transcription 124
 - rhinovirus genomes 123
 - ribosomal RNA band 231
 - RNA-degrading RNases 231
 - RNA-induced silencing complex (RISC) 217
 - RNA splicing patterns 130
 - RNA virus 124
- s**
- SABE, *see* serial analysis of binding elements
 - SAGE, *see* serial analysis of gene expression
 - Sanger DNA sequencing 5, 6, 10, 50, 91, 133, 153, 156
 - approaches 133
 - basics of 3
 - capillary electrophoresis 133
 - dideoxy-based tag sequencing 203, 204, 205
 - instruments 36
 - limitation and opportunities 7
 - method 4, 10, 90, 117, 153
 - principle of 4
 - metagenomic libraries 183
 - SeqID Hit method 82
 - SeqMan genome assembler (SMGA) 91
 - sequence assembly tools 21
 - sequence-based expression profiling 226
 - sequence-based techniques 40
 - sequence searching, strategies 80
 - sequence-specific probes 119
 - sequencing-by-synthesis (SBS) 133, 206
 - sequencing factories 5
 - sequencing library 63
 - sequencing technology 3, 15, 69, 109
 - ultrahigh-throughput 69
 - serial analysis of binding elements (SABE) 204
 - serial analysis of gene expression (SAGE) 113, 230
 - library 235
 - protocol 231
 - technique 204, 237
 - short-read sequencing technology 211
 - shotgun cloning 10
 - signal-to-noise (S/N) ratio 146
 - silica sol-gel monolith, *see* photopolymerized monolith
 - single fluorescent dye molecules 125
 - single-molecule sequencing technologies 110
 - single-nucleotide polymorphisms (SNPs) 3, 8, 72, 89
 - single-pair fluorescent resonance energy transfer (spFRET) 133
 - SMART technology (Clontech) 221
 - SMGA assembly projects 91
 - Smith–Waterman alignment algorithm 210
 - sodium dodecyl sulfate (SDS) 159
 - Solexa sequencing 233–235
 - SOLiD system 29, 30, 35–38, 40
 - applications 35
 - library generation 30–31
 - overview of 29
 - performance of 29, 33
 - technology of 29, 39
 - SSAHA-based methods 84

t

- tag-based sequencing 21, 36, 167
 - platforms 167
- tag-based transcriptome analysis methods 229, 238
- tag-based transcriptome profiles 235
- Tamra dye signals 121
- total internal reflectance fluorescence (TIRF) 99
 - microscopy 118
 - system 120
- transcriptional elements 202
- transcription factor-binding elements 39
- transcription factor binding sites (TFBS) 167, 168, 169, 173, 177
 - genome mapping 167, 168
- transcription regulatory circuits 173
- transcription regulatory networks 179
- transcription start sites (TSS) 168
- transcriptome 23, 229
 - high-resolution map 24
- transcriptome analysis methods 23, 37, 113, 170, 229, 230
 - DNA microarrays 230
 - serial analysis of gene expression (SAGE) 230
- transcriptome profiling 238
- transcriptome sequencing 19
- transient entanglement coupling (TEC) 158
- transmission electron microscopes (TEMs) 103
 - analysis 104
 - based sequencing 109

- image 104
- instrument 107
- sequencing technology 110
- substrate 106, 107
- technology 111
- visualization 107
- T7 exonuclease 148

u

- ultradeep sequencing 47
- ultrahigh-speed DNA sequencing 97

v

- viral genomes 123
- VisiGen's core technology 101
 - approach 100

w

- Western blotting methods 39
- whole genome shotgun (WGS) 5, 6

x

- xenon lamp 34

y

- yeast telomeric heterochromatin 204

z

- Z-labeled nucleotides (Z-dNTPs) 107
- Z-modified nucleotides (Z-dNTPs) 104, 105, 108
- ZS genetics (ZSG) 103, 111
- Z-substituted DNA molecules 103
- Z-tagged nucleotides 108