

## Index

- achondroplasia, 147
- aggressive behavior, 122
- agriculture, 18, 42
- alleles, 3
- alternative splicing, 74, 75
- Angelman syndrome, 220
- Avery, 51
  
- Bateson, 27
- Beadle, 45
- Beckwith, 257
- Benzer, 55
- BRCA1*, 89, 233, 236
- BRCA2*, 233, 236
- Brenner, 58
- Brno, 18
  - socio-economic context of, 18
- Brooks, 21, 28
  
- cancer development, 146
- cancers, 144, 223
- carcinogens, 145
- CDK6*, 120
- cell potency, 221
- Central Dogma
  - Crick's version, 58
  - 1970s version, 64
  - reconceptualization, 227
- Chargaff, 46
- Chase, 51
- chromatin, 70
  - types of, 70
- chromosomes, 70
- cistron, 57
- classical gene, 44, 66, 67, 68
- Clinton, 106
- Collins, 84, 107f5.1, 155, 229, 231
- conceptions, 2
- concepts, 2
  - scientific, 2
- Correns, 25
- Crick, 46, 49, 51, 57, 58
- crossing over, 34–40
- CSI, 96, 245
  
- Darwin, 21
- de Vries, 22, 25
- deCODEme, 158
- development, 172, 176
- developmental plasticity, 183, 206
- developmental robustness, 183, 206
- direct-to-consumer genetic tests, 98, 155
  - user's perspectives, 103
- disease, 130
  - complex diseases, 151
- DNA decoding, 105
- DNA fingerprinting, 245
- DNA methylation, 215
  - hypermethylation, 224
  - hypomethylation, 223
- DNA replication, 51
- DNA sequencing, 105
- Doppler, 18
- double helix model of DNA, 46, 48
- Driesch, 174
  
- East, 42
- ENCODE Project, 84, 210, 211
- enhancers, 63
- epigenetic landscape, 221
- epigenetic reprogramming, 222
  - and cancer, 226
- epigenetics, 215
- epigenome, 217
- epistasis, 29
- essentialism, 94
- euchromatin, 72
- eugenics, 43
  - negative, 44
  - positive, 44
- exons, 73
- expressivity, 166
  
- false negative result, 239
- false positive result, 239
- Familial hypercholesterolemia, 135
- FDA, 159
- FGFR3*, 148
- Franklin, 46

## 306 INDEX

- Galton, 21, 25, 28, 43, 191  
 Gärtner, 18  
 gene  
   blueprint metaphor, 91, 172  
   differential concept of, 32  
   ENCODE definition of, 84, 87  
   original definition, 30  
   school definition, 3  
 gene expression, 63  
   complexities of, 67  
 gene nomenclature, 10  
*Gene Partner*, 102  
 gene regulation, 63, 67  
 gene symbols, 10  
 gene-knockout, 164  
 genes  
   causal role of, 163  
   as difference makers, 32, 189, 203  
   as discrete parts of  
     chromosomes, 34  
     origami metaphor, 178  
   “genes for”, 1, 5, 33, 109,  
     129, 154  
   genetic code, 58, 59  
   genetic determinism, 6, 33, 92, 93, 172,  
     185, 189  
   genetic diagnosis, 236  
   genetic essentialism, 6, 95, 185, 189  
   genetic mapping, 34  
   genetic material  
     definition of, 87  
   genetic reductionism, 6, 185, 189  
   genetic screening, 236  
 GeneticHealth, 100  
 genetics  
   definition in this book, 9  
 genome mosaicism, 241  
 genomic imprinting, 217, 219  
 genomic medicine, 230  
 Griffith, 46  
 GWAS, 115, 116, 118, 120, 131, 132, 150, 157, 160
- Haeckel, 21  
 Haldane, 42  
 hallmarks of cancer, 145  
*HBB*, 131, 132, 204  
*HBS1L*, 134  
 hemophilia, 42  
*HERC2*, 113  
 heredity, 20  
 heritability, 196, 197, 198, 205  
   distinguish from inheritability, 206  
 Hershey, 51
- heterochromatin, 72  
 heterozygote, 27  
*HHIP*, 120  
 histone acetylation, 216  
 histone methylation, 217  
 histones, 70, 216  
*HMG2*, 118  
 homozygote, 27  
 human eye color, 111  
 Human Genome Project, 80,  
   171, 189  
 human height, 118  
 hybrid corn, 42  
 hybridization, 17  
 hyperphenylalaninemia, 140
- iGENEA*, 101  
 information  
   encoding of, 87  
   metaphor of, 58  
 inheritance, epigenetic, 215  
 inheritance, genetic  
   definition of, 20  
 introns, 73  
 Ioannidis, 242  
 IQ, 194
- Jacob, 59, 210  
 Jensen, 194  
 Johannsen, 30  
 “junk” DNA, 208
- Kölreuter, 18
- Lander, 231  
 lcRNAs, 214  
*LDLR*, 136  
 Lewontin, 172, 196  
 linkage-disequilibrium, 116  
 Linnaeus, 18  
*LMBR1*, 69
- MacLeod, 51  
*MAOA*, 128  
 Matthaei, 59  
 McCarty, 51  
 McClintock, 40, 62  
 Mendel, 3, 11, 24f1.5  
   experiments with peas, 18  
   law of independent assortment, 13  
   law of segregation, 13  
   life of, 13  
   “rediscovery” of, 26  
 Meselson, 51

- metaphors, 252
  - about genes, 6
  - limits of, 255
- miRNAs, 214
- molecular gene, 65, 67, 68
- Monod, 59, 210
- monogenic characters, 109
- Morgan, 31, 32, 34, 36
- mRNA, 59, 64
- Muller, 39
- mutation, 110
  - driver mutations, 148
  - germline, 147
  - passenger mutations, 148
  - somatic, 147
- Mutations, 52
- muton, 56
- MYB, 134
- Myriad Genetics, 100
  
- Nägeli, 21, 22
- Napp, 18
- nature, 191, 206
- Navigenics, 158
- Nirenberg, 59
- nurture, 191, 206
  
- Obama, 231
- OCA2, 113
- one gene-one enzyme hypothesis, 45
- overlapping genes, 79
  
- PAH, 140
- Pathway Genomics, 99
- Pauling, 46, 55
- PCSK9, 137
- Pearson, 28
- penetrance, 166
- personalized medicine, 230
- phenylketonuria, 138
- piRNAs, 214
- pleiotropy, 167
- Prader-Willi syndrome, 220
- promoters, 62
  
- recon, 56
- reductionism
  - metaphysical (ontological), 172
  - methodological, 172
- risk
  - absolute, 156
  - lifetime, 232
  - relative, 156
- RNA editing, 80
- RNA splicing, 73
- Roux, 174
- rRNA, 59, 64
  
- scientific models, 260
- sex chromosomes, 36
- Shull, 42
- sickle cell anemia, 55
- siRNAs, 214
- SNPs, 111, 115, 118
- Spencer, 20
- SRY, 188f9.7, 203
- Stahl, 51
- statistical association and cause-effect
  - relation, 162
- Sturtevant, 34
  
- Tatum, 45
- TCOF1, 182
- test sensitivity, 239
- test specificity, 239
- transcription, 64
- transcription factors, 62
- translation, 64
- trans-splicing, 77
  - intergenic, 78
  - intragenic, 78
- tRNA, 59, 64
- Tschermak, 25
- 23andMe, 98, 158, 159t8.2
- twins, 192
  
- Unger, 18
  
- validity
  - analytical, 159, 231
  - clinical, 159, 231
- variants of unknown significance, 237
- Venter, 84, 107f5.1
  
- Waddington, 215, 221
- Watson, 46, 49, 51, 83
- Weismann, 22, 25
- Weldon, 28
- Wilkins, 46
  
- ZBTB38, 120
- β-thalassemia, 131