

Principles of  
**GENETICS**  
EIGHTH EDITION

# **Principles of** **GENETICS**

**EIGHTH EDITION**

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The eighth edition  
is dedicated to the memory of

**Elton John Gardner**

who skillfully guided this textbook  
through seven editions

# PREFACE

The eighth edition is a continuing attempt to provide a true "principles of genetics" text that is up-to-date, readable, and challenging. Its major goal is to emphasize the central role that genetics plays in the lives of all organisms—plant, animal, and microbe—on planet Earth. In our complex modern society, with the very survival of our planet at stake, it is imperative that citizens be informed about scientific issues. Thus, our goal has been to write this text for students with only a minimal background in biology and chemistry. It is suitable for one-quarter, one-semester, or two-quarter courses in genetics that are designed for biology majors or for students from other disciplines. We have attempted to provide balanced coverage of many areas in genetics—classical, molecular, and population—but we recognize that each instructor will favor some topics over others. Consequently, this book has been written to allow instructors flexibility in the choice of topics and in the depth of coverage.

Genetics and related sciences have grown explosively in recent years, generating a large body of new and important information. As a result, the eighth edition has expanded considerably when compared with previous editions. A new introductory chapter attempts to convey the excitement of genetics today and to emphasize the central role that it plays in our lives. Two other new chapters cover the major developments in our knowledge of transposable genetic elements and the rapidly advancing methodologies of genetic engineering. Other chapters have been revised and expanded to incorporate new information about the structure and function of genes and their roles in development.

Compared with its predecessor, this edition has been substantially reorganized. The material on the structure and replication of DNA and chromosomes has been divided into two chapters, one focusing on prokaryotes and the other on eukaryotes. The analysis of genetic fine structure is included in two chapters, one dealing with classical studies and the other with molecular approaches, including cloning and sequencing. Also included in this molecular chapter are such important techniques as Southern, northern, and western blotting, PCR amplification of DNA, and oligonucleotide-directed site-specific mutagenesis. Regulation, which was treated in a single chapter in the previous edition, is now treated in four; two of these deal with the basic features of regulation, one focusing on prokaryotes and the other on eukaryotes, and two deal with special eukaryotic phenomena—the immune response and the genetic control of cell division. This last chapter includes an expanded discussion of the structure and function of oncogenes and protooncogenes.

Other changes have been made in the eighth edition to enhance its readability and focus. Multiple alleles are now treated in the same chapter as basic Mendelian genetics. The

chapters on quantitative genetics and population and evolutionary genetics have been rewritten to provide more coherence to these somewhat difficult subjects. New material has been added to several other chapters; for example, the chapter on gene expression now contains sections on RNA splicing and the spliceosome, and the chapter on extrachromosomal inheritance contains new sections on the organization of mitochondrial and chloroplast genomes. As in previous editions, each chapter ends with an extensive set of problems and questions of varying difficulty, with the answers provided at the end of the book.

We believe that the eighth edition presents the basic principles of genetics in a clear and logical sequence, but we realize that many instructors will prefer other sequences. As mentioned earlier, this edition begins with a new introductory chapter that attempts to (1) convey some of the excitement that has resulted from recent developments in the field of genetics and (2) emphasize the important role that genetics plays in many aspects of our lives and, indeed, in the very survival of life on our planet. Chapter 2 describes the classic work of Gregor Mendel and the basic principles of Mendelian genetics that resulted from his work. Cells, chromosomes, and the process of meiosis that provide the basis for Mendel's laws are described in Chapter 3. The chromosomes and their behavior during gametogenesis are related to sex determination and sex-linked patterns of inheritance in Chapter 4. The molecular structure of genes and their modes of replication in prokaryotic chromosomes are introduced in Chapter 5. The complex structure and mechanism of replication of eukaryotic chromosomes are the subjects of Chapter 6. Chapter 7 covers the classical genetic topics of linkage, chromosome mapping, tetrad analysis, gene conversion, and the mechanism of crossing over. Recombination mechanisms unique to bacteria are covered in Chapter 8, and the structure and function of transposable genetic elements are the subjects of Chapter 9.

After considering the structure and recombination of genetic elements in Chapters 5 through 9, the focus shifts to the function of genetic elements in Chapter 10 (Gene Expression), which covers transcription, RNA processing ("splicing"), translation, and the genetic code. The mechanisms by which genes change or mutate are covered in Chapter 11. Chapters 12 and 13 dissect genes by both classical (Chapter 12) and molecular (Chapter 13) approaches. Chapter 13 also provides background information on the powerful tools of molecular genetics that are used in these dissections: cloning vectors, Southern, northern, and western blots, PCR, and site-specific mutagenesis. Chapters 14 through 17 describe our present understanding of the various mechanisms by which gene expression and cell division are regulated. Control of gene expression in prokaryotes is the topic covered in Chapter 14, whereas Chapter 15 focuses on the regulation of



gene expression and development in eukaryotes. Although the genetic control of development in higher eukaryotes is much too complex to be covered in detail in an introductory genetics text, we hope the information presented in this chapter will give students an insight into the exciting developments in this field. Although much remains to be learned, molecular pictures of the genetic control of development in *Drosophila melanogaster* and the worm *Caenorhabditis elegans* have rapidly begun to emerge during the last few years.

The genetic control of the vast repertoire of antibodies that can be produced by higher animals was an enigma for decades. In the last few years, many of the mysteries of the immune system have been replaced by fascinating pictures of the genetic control of antibodies and T cell receptors; these new breakthroughs are described in Chapter 16. In Chapter 17, the genetic control of cell division is discussed, with emphases on the roles of protooncogenes and on the perturbations caused by oncogenes. The next two chapters discuss mutation in the broad sense of gross changes in chromosome structure (Chapter 18) and chromosome number (Chapter 19). In Chapter 20, the behavior of genes that are not located on the main nuclear chromosomes of the organism is considered, with an emphasis on the structure of the chromosomes of mitochondria and chloroplasts. Chapter 21 discusses those traits that are controlled by large numbers of genes and thus exhibit quantitative patterns of inheritance. In Chapter 22, the analysis of genes and their effects is extended to the level of the population; major emphases are the distributions of alleles in populations and the changes in these distributions (evolution) over time in response to factors such as mutation, genetic drift, and selection. Chapter 23 provides a brief introduction to the genetic factors that influence behavior.

Chapter 24 introduces the student to some of the present and future applications of genetic engineering. We realize that most introductory genetics classes will cover few, if any, of the topics discussed in Chapter 24. However, we believe that this chapter will be of considerable interest to many students, and we anticipate that those students who find genetics to be fascinating and challenging will read Chapter 24 on their own. Some instructors will probably choose to integrate certain topics from Chapter 24 into their courses. For example, a class with a plant genetics emphasis might appropriately include the material on *Agrobacterium*

*tumefaciens* and plant transformation via the Ti plasmid. Other classes may choose to include the section on somatic-cell gene therapy, and so on. A glossary of important genetic terms, the answers to all the problems in the book, and an index complete the text.

Our thanks to students, teachers, and colleagues who have suggested improvements for the book. We especially thank Rayla Temin, R. H. Whalen, Linda Kosturko, Clint Magill, Peter Weijksnora, Charles Rodell, Hugh Stanley, Larry Puckett, Robert M. Fineman, Irene Uchida, Scott R. Woodward, John R. Simmons, and Franklin D. Enfield, who have all made valuable contributions. We also thank the following reviewers for their valuable suggestions and comments: Judith Van Houten, The University of Vermont; Robert Peters, Pennsylvania State University; Darrell S. English, Northern Arizona University; Romesh C. Mehra, Indiana University at South Bend, Glenn Wolfe, Kansas University; Paul J. Homsher, Old Dominion University; Paul A. Roberts, Oregon State University; Asim Esem, Virginia Polytechnic Institute and State University; Howard Laten, Loyola University; Charles H. Green, Glassboro State College; Alice S. Hunter, College of the Pacific; Martin Bard, Purdue University; John B. Jenkins, Swarthmore College; Kandy D. Baumgardner, Eastern Illinois University; Dwight E. Wilson, Rensselaer Polytechnic Institute; Carter Denniston and William R. Engels, University of Wisconsin, Madison; H. James Price, Texas A & M University; Peter Dawson, Oregon State University; Barry Bean, Lehigh University; Dean Whited, North Dakota State University.

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D. PETER SNUSTAD

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# BRIEF CONTENTS

Chapter 1		Chapter 14	
Introduction	1	Regulation of Gene Expression in Prokaryotes	390
Chapter 2		Chapter 15	
Mendelian Genetics	20	Regulation of Gene Expression and Development in Eukaryotes	410
Chapter 3		Chapter 16	
Cell Mechanics	52	Genetic Control of the Immune Response	448
Chapter 4		Chapter 17	
Sex Determination and Sex Linkage	69	Genetic Control of Cell Division: Oncogenes and Protooncogenes	465
Chapter 5		Chapter 18	
Genetic Material: Properties and Replication	91	Variations in Chromosome Structure	488
Chapter 6		Chapter 19	
Structure and Replication of Eukaryotic Chromosomes	130	Variations in Chromosome Number	511
Chapter 7		Chapter 20	
Linkage, Crossing-over, and Chromosome Mapping	158	Extrachromosomal Inheritance	535
Chapter 8		Chapter 21	
Recombination in Bacteria	205	Quantitative Genetics	552
Chapter 9		Chapter 22	
Transposable Genetic Elements	230	Population and Evolutionary Genetics	566
Chapter 10		Chapter 23	
Gene Expression	247	Genetics of Behavior	591
Chapter 11		Chapter 24	
Mutation	288	Genetic Engineering and the Future	604
Chapter 12		GLOSSARY	G1
Genetic Fine Structure: Classical Genetics	320	ANSWERS TO PROBLEMS	A1
Chapter 13		INDEX	11
Genetic Fine Structure: Gene Cloning and Sequencing	348		

# CONTENTS

## Chapter 1

### Introduction 1

#### THE WORLD IS A COLOSSAL GENETICS LABORATORY 3

Genetics and the Phenotype of Our Biosphere 5

Genetics and Food Production 8

Genetics and Health 9

Genetics, Politics, and Law 12

Genetic Engineering 13

#### THE BIRTH OF GENETICS 15

#### GROWTH OF A SCIENCE: FROM MENDEL TO GENETIC ENGINEERING 17

#### SUMMARY 18

#### REFERENCES 19

## Chapter 2

### Mendelian Genetics 20

#### MENDEL'S EXPERIMENTS 21

#### PRINCIPLE OF SEGREGATION 21

#### SYMBOLS AND TERMINOLOGY 22

#### MONOHYBRID CROSSES 24

Dominance 24

Recessiveness 26

Codominance 26

Semidominance 26

Lethals 27

#### PRINCIPLE OF INDEPENDENT ASSORTMENT 28

Dihybrid Ratios 29

Trihybrid Ratios 30

Forked-Line Method for Genetic Problems 31

Mathematical Method for Genetic Problems 31

Gene Interaction 33

Epistasis 34

#### GENETIC VERSUS ENVIRONMENTAL EFFECTS 36

#### MULTIPLE ALLELES 37

ABO Blood Type Alleles in Humans 38

Rh Factor Alleles in Humans 39

#### PROBABILITY IN MENDELIAN INHERITANCE 40

Fitting Results from Crosses to Hypotheses 41

Chi-square 41

Independent Assortment and Probability 43

Expansion of a Binomial 44

#### PROBABILITY IN PEDIGREE ANALYSIS 45

#### MODERN EVALUATIONS OF MENDEL'S CONCLUSIONS 47

#### SUMMARY 47

#### REFERENCES 47

#### PROBLEMS AND QUESTIONS 48

## Chapter 3

### Cell Mechanics 52

#### CELL CYCLE 54

Mitosis 55

Meiosis 57

#### GAMETE FORMATION IN ANIMALS 57

Spermatogenesis 57

Oögenesis 63

#### SPORE FORMATION IN PLANTS 65

Plant Fertilization 66

#### MEIOSIS AND MENDEL'S PRINCIPLES 66

#### SUMMARY 67

#### REFERENCES 67

#### PROBLEMS AND QUESTIONS 68

## Chapter 4

### Sex Determination and Sex Linkage 69

#### MECHANISMS OF SEX DETERMINATION 70

Simple Mechanisms: One or a Few Genes 70

Identification of Sex Chromosomes 70

XX–XY Mechanism of Sex Determination 72

Species with Heterogametic Females 73

The Y Chromosome and Sex Determination in Mammals 73

The Balance Concept of Sex Determination in *Drosophila* 74

Haplodiploidy and Sex Determination in Hymenoptera 76

Mosaics and Gynandromorphs 76

#### ENVIRONMENTAL FACTORS AND SEX DETERMINATION 77

#### SEX DIFFERENTIATION 78

Sex Chromatin Bodies 78



Dosage Compensation	78
"Fragile X" Chromosome and Mental Retardation in Humans	80
Hormones and Sex Differentiation	81
SEX-INFLUENCED DOMINANCE	81
SEX-LIMITED GENE EXPRESSION	82
SEX-LINKED INHERITANCE	82
Morgan's Discovery of Sex Linkage in <i>Drosophila</i>	82
Patterns of Inheritance of Sex-Linked Genes	83
X-Linked Traits in Humans	84
Deleterious Recessive Sex-Linked Genes in Humans	88
Human Y Chromosome: Dominant Male Determinants, But Few Other Genes	88
SUMMARY	89
REFERENCES	89
PROBLEMS AND QUESTIONS	89

<b>Chapter 5</b>	
<b>Genetic Material: Properties and Replication</b>	<b>91</b>
DNA, THE GENETIC MATERIAL	92
Transformation in <i>Pneumococcus</i>	92
Proof That the "Transforming Principle" Is DNA	93
The "Hershey-Chase Experiment"	94
RNA as Genetic Material in Small Viruses	96
DNA STRUCTURE	97
The Watson and Crick DNA Double Helix	99
Conformational Flexibility of DNA Molecules	103
SEMICONSERVATIVE REPLICATION OF DNA	104
The "Meselson-Stahl" Experiment	105
Autoradiography of Replicating Bacterial Chromosomes	107
Unique Origins and Bidirectional Replication	110
DNA Polymerases and <i>In Vitro</i> DNA Synthesis	114
The "Growing-Point Paradox" and Discontinuous DNA Synthesis	117
Initiation and the "Primer Problem"	118
The Complete "Replication Apparatus" Is Complex	119
Phage $\Phi$ X174 and "Rolling Circle" Replication	122
PROKARYOTE NUCLEOID STRUCTURE	124
SUMMARY	127
REFERENCES	128
PROBLEMS AND QUESTIONS	128

<b>Chapter 6</b>	
<b>Structure and Replication of Eukaryotic Chromosomes</b>	<b>130</b>
GENOME COMPLEXITY	131
CHEMICAL COMPOSITION OF EUKARYOTIC CHROMOSOMES	131
ONE GIANT DNA MOLECULE PER CHROMOSOME	133
"Lampbrush" Chromosomes in Vertebrate Oocytes	134
Viscoelastometric Evidence for Chromosome-Size DNA Molecules	134
Autoradiographic Evidence for Chromosome-Size DNA Molecules	136
PACKAGING THE GIANT DNA MOLECULES INTO CHROMOSOMES	136
Nucleosome Structure	137
The 300-Å Chromatin Fiber	138
"Scaffolds" Composed of Nonhistone Chromosomal Proteins	140
EUCHROMATIN AND HETEROCHROMATIN	143
REPETITIVE DNA AND SEQUENCE ORGANIZATION	143
Satellite DNAs	143
DNA Renaturation Kinetics	144
Interspersion of Single-Copy and Middle-Repetitive DNA Sequences	146
Function of Highly Repetitive DNA Sequences?	148
Telomere Structure	148
REPLICATION OF EUKARYOTIC CHROMOSOMES	149
The Cell Cycle	149
Semiconservative Replication of Eukaryotic Chromosomes	150
Multiple "Replicons" per Chromosome	150
Components of the Eukaryotic "Replication Apparatus"	153
SUMMARY	155
REFERENCES	155
PROBLEMS AND QUESTIONS	156

<b>Chapter 7</b>	
<b>Linkage, Crossing-over, and Chromosome Mapping</b>	<b>158</b>
LINKAGE	159
CROSSING-OVER	163
Cytological Basis of Crossing-Over	163
Crossing-Over Occurs in the Postreplication Tetrad Stage	165

CHROMOSOME MAPPING	167
Two-Factor Crosses	170
Maximum Frequency of Recombination Is 50 Percent	171
Three-Factor Crosses	174
Interference	177
Ordered Tetrad Data	178
Unordered Tetrad Data	183
Somatic-Cell Hybridization	183

#### MOLECULAR MECHANISM OF CROSSING-OVER 186

#### GENE CONVERSION 196

#### SUMMARY 199

#### REFERENCES 200

#### PROBLEMS AND QUESTIONS 200

### Chapter 8

#### Recombination in Bacteria 205

##### TRANSFORMATION 206

##### TRANSDUCTION 208

###### Generalized Transduction 209

###### Specialized Transduction 210

##### CONJUGATION 213

##### F-MEDIATED SEXDUCTION 219

##### RATIONALE OF FINE STRUCTURE MAPPING IN MEROZYGOTES 220

##### BREAKAGE AND REUNION AS THE MECHANISM OF RECOMBINATION IN BACTERIA 223

##### PLASMIDS AND EPISOMES 224

##### SUMMARY 227

##### REFERENCES 227

##### PROBLEMS AND QUESTIONS 228

### Chapter 9

#### Transposable Genetic Elements 230

##### GENETIC INSTABILITY AND THE DISCOVERY OF TRANSPOSABLE ELEMENTS 231

##### TRANSPOSABLE ELEMENTS IN BACTERIA 232

###### IS Elements 232

###### The Tn3 Family 233

###### Medical Significance of Bacterial Transposons 235

##### TRANSPOSABLE ELEMENTS IN EUKARYOTES 237

###### Yeast *TY* Elements 237

###### Maize Transposons 237

###### *Drosophila* Transposons 240

##### THE GENETIC AND EVOLUTIONARY SIGNIFICANCE OF TRANSPOSABLE ELEMENTS 243

###### Mutation and Chromosome Breakage 243

###### Use in Genetic Analysis 243

###### Evolutionary Issues 244

##### SUMMARY 245

##### REFERENCES 245

##### PROBLEMS AND QUESTIONS 246

### Chapter 10

#### Gene Expression 247

##### EVOLUTION OF THE ONE GENE-ONE POLYPEPTIDE CONCEPT 249

##### GENETIC CONTROL OF METABOLISM 251

##### PROTEIN SYNTHESIS 253

###### Transcription 255

###### Translation 258

###### Coupled Transcription and Translation in Prokaryotes 265

###### Transcription, RNA Processing and Transport, and Translation in Eukaryotes 265

###### Removal of Intron Sequences by RNA

###### Splicing 269

###### Three Distinct Types of RNA Splicing 270

###### Splicing tRNA Precursors: Unique Nuclease and Ligase 271

###### Autocatalytic Splicing of *Tetrahymena* rRNA Precursor 272

###### Pre-mRNA Splicing: snRNAs, snRNPs, and the Spliceosome 272

##### THE GENETIC CODE 275

###### Three Nucleotides per Codon 276

###### Deciphering the Code 278

###### Degeneracy and Wobble 279

###### Initiation and Termination Codons 281

###### Universality of the Code 281

###### Suppressor Mutations Producing tRNAs with Altered Codon Recognition 281

##### COLINEARITY OF GENE AND POLYPEPTIDE 282

##### SUMMARY 284

##### REFERENCES 285

##### PROBLEMS AND QUESTIONS 286

### Chapter 11

#### Mutation 288

##### SPONTANEOUS VERSUS INDUCED MUTATION 289

##### MUTATION: RANDOM RATHER THAN DIRECTED BY THE ENVIRONMENT 290

PHENOTYPIC EFFECTS OF MUTATIONS	291
Somatic and Germinal Mutations	294
Mutant Hemoglobins	295
Pleiotropy	296
Back-Mutations and Suppressor Mutations	297
THE MOLECULAR BASIS OF MUTATION	298
RADIATION-INDUCED MUTATION	301
Ionizing Radiation	301
Ultraviolet Radiation	304
DNA Repair Mechanisms	305
Xeroderma Pigmentosum	307
CHEMICALLY INDUCED MUTATION	308
Base Analogs	308
Nitrous Acid	310
Acridines	310
Alkylating and Hydroxylating Agents	311
CORRELATION BETWEEN MUTAGENICITY AND CARCINOGENICITY	312
MUTATION FREQUENCY	314
PRACTICAL APPLICATIONS OF MUTATIONS	314
Beneficial Mutations	314
High-Resolution Dissection of Biological Processes via Mutation Analysis	315
MUTATIONS AND HUMANS	316
SUMMARY	317
REFERENCES	317
PROBLEMS AND QUESTIONS	318

## Chapter 12

### Genetic Fine Structure: Classical Genetics 320

CLASSICAL VERSUS MOLECULAR CONCEPT OF THE GENE	321
EARLY EVIDENCE THAT THE GENE IS SUBDIVISIBLE	321
THE <i>CIS-TRANS</i> OR COMPLEMENTATION TEST FOR FUNCTIONAL ALLELISM	322
LIMITATIONS OF THE <i>CIS-TRANS</i> TEST	324
FINE STRUCTURE OF THE PHAGE T4 <i>rII</i> LOCUS	327
Deletion Mapping	332
Electron Microscope Heteroduplex Mapping	333
THE UNIT OF STRUCTURE IS THE SINGLE NUCLEOTIDE-PAIR	336
FINE STRUCTURE OF GENES AND "COMPLEX LOCI" IN EUKARYOTES	337
ONE GENE-ONE BAND IN <i>DROSOPHILA</i> SALIVARY GLAND CHROMOSOMES?	341

### GENES-WITHIN-GENES IN PHAGE $\Phi$ X174 342

SUMMARY	344
REFERENCES	345
PROBLEMS AND QUESTIONS	345

## Chapter 13

### Genetic Fine Structure: Gene Cloning and Sequencing 348

#### "RECOMBINANT DNA" AND "GENE CLONING" AS A TOOL FOR FINE STRUCTURE ANALYSIS 349

The <i>In Vitro</i> Synthesis of Recombinant DNA and Gene Cloning Techniques	349
Restriction Endonucleases	349
Cloning DNAs with Complementary Single-Stranded Ends	351
Cloning DNAs with Blunt Ends by Adding Complementary Tails	351
Cloning cDNAs Produced from Purified mRNAs	351
Screening a Genomic DNA Library by <i>In Situ</i> Colony Hybridization	351
Applications and Potential Hazards of Recombinant DNA and Gene Cloning Technology	354
NONCODING INTERVENING SEQUENCES OR INTRONS WITHIN EUKARYOTIC GENES	356
R-Loop Mapping	356
Mammalian Hemoglobin Genes	356
Eukaryotic Genes with Many Introns and With No Introns	358
Biological Significance of Introns?	359
MAPPING RESTRICTION ENZYME CLEAVAGE SITES OF CHROMOSOMES	359
NUCLEOTIDE SEQUENCES: THE ULTIMATE FINE STRUCTURE MAPS	364

ADVANCED CLONING VECTORS	367
Plasmids That Provide an Option to Package Single Strands in Viral Coats	368
<i>In Vitro</i> Transcription Vectors	372
Use of Linkers in Manipulating DNAs	373
MOLECULAR ANALYSIS OF GENES AND GENE-PRODUCTS	374
Analysis of DNAs by Southern Blot Hybridizations	376
Analysis of RNAs by Northern Blot Hybridizations	378
Analysis of Proteins by Western Blot Techniques	380
Amplification of DNAs by the Polymerase Chain Reaction	381
TAILORING GENES TO SUIT ONE'S NEEDS: <i>IN VITRO</i> SITE-SPECIFIC MUTAGENESIS	382

SUMMARY	386
REFERENCES	387
PROBLEMS AND QUESTIONS	387

## Chapter 14

### Regulation of Gene Expression in Prokaryotes 390

INDUCTION AND REPRESSION IN PROKARYOTES	392
The Operon Model	393
<i>lac</i> , an Inducible Operon	395
<i>trp</i> , a Repressible Operon	397
Positive Control of the <i>lac</i> Operon by CAP and Cyclic AMP	398
Complex Regulation of the <i>ara</i> Operon	400
Lambda Prophage Repression during Lysogeny	402
CONTROL OF THE <i>trp</i> OPERON BY ATTENUATION	403
FEEDBACK INHIBITION AND ALLOSTERIC ENZYMES	406
TEMPORAL SEQUENCES OF GENE EXPRESSION DURING PHAGE INFECTION	407
SUMMARY	407
REFERENCES	408
PROBLEMS AND QUESTIONS	408

## Chapter 15

### Regulation of Gene Expression and Development in Eukaryotes 410

CELLULAR DIFFERENTIATION IN HIGHER EUKARYOTES	411
GENETIC CONTROL OF DEVELOPMENT IN <i>DROSOPHILA</i>	413
Imaginal Discs and Determination	413
Homeotic Mutants	415
Transdetermination	417
Morphogenesis of a Fly: Sequential Anterior- Posterior Segmentation Events	417
A Temporally Ordered Cascade of Regulatory Genes	420
The <i>Bithorax</i> Complex	422
Role of the "Homeobox" Domain in Regulation of Transcription	422
Conserved "Homeobox" Sequences in Other Species	424
CAENORHABDITIS ELEGANS: A MODEL SYSTEM FOR THE STUDY OF DEVELOPMENT	424

### NEOCLASSICAL EXAMPLES OF DEVELOPMENTALLY REGULATED GENE EXPRESSION 425

Transcription on Lampbrush Chromosomes in Amphibian Oocytes	425
rRNA Gene Amplification in Amphibian Oocytes	426
GENE TRANSCRIPT POPULATIONS ARE DIVERGENT IN DIFFERENT CELL TYPES	426
MECHANISMS OF REGULATION OF TRANSCRIPTION IN HIGHER EUKARYOTES	427
Most Eukaryotic Transcription Units Are Monogenic	428
Enhancers and Silencers Modulate Transcription in Eukaryotes	428
Regulation of Levels of Transcription by DNA Methylation	430
Does Z-DNA Play a Regulatory Role?	432
Chromatin Structure: Nuclease Sensitive Sites Adjacent to Active Genes	434
HORMONAL CONTROL OF GENE EXPRESSION	435

Activation of Transcription by Steroid Hormones	435
Glucocorticoid Hormones Act via Enhancer Elements	437
Ecdysone and Chromosome "Puffs" in Flies	439
REGULATION BY ALTERNATE PATHWAYS OF TRANSCRIPT SPLICING	440
REGULATION OF COMPLEX CIRCUITS OF GENE EXPRESSION IN EUKARYOTES	441
SUMMARY	445
REFERENCES	445
PROBLEMS AND QUESTIONS	446

## Chapter 16

### Genetic Control of the Immune Response 448

COMPONENTS OF THE IMMUNE SYSTEM	449
VAST REPERTOIRE OF ANTIBODIES	449
Hypotheses: Genetic Basis of Antibody Diversity	449
Structure of Antibodies	450
ANTIBODY DIVERSITY: GENOME REARRANGEMENTS DURING B LYMPHOCYTE DIFFERENTIATION	451
Kappa Light Chains	452
Lambda Light Chains	452
Heavy Chains	452
Class Switching	454
ANTIBODY DIVERSITY: ALTERNATE PATHWAYS OF TRANSCRIPT SPLICING	454

SIGNAL SEQUENCES GOVERN GENOME REARRANGEMENTS	455
ANTIBODY DIVERSITY: VARIABLE JOINING SITES AND SOMATIC MUTATIONS	457
HOW MANY COMBINATIONS?	458
REGULATION OF TRANSCRIPTION: A TISSUE-SPECIFIC ENHANCER	458
CLONAL SELECTION	459
ALLELIC EXCLUSION	459
T CELL RECEPTOR VARIABILITY	460
MAJOR HISTOCOMPATIBILITY COMPLEX	461
SUMMARY	463
REFERENCES	464
PROBLEMS AND QUESTIONS	464

## Chapter 17

### Genetic Control of Cell Division: Oncogenes and Protooncogenes 465

REGULATION OF THE MITOTIC CELL CYCLE IN EUKARYOTES	466
INTERCELLULAR COMMUNICATION IN MULTI-CELLULAR EUKARYOTES	467
CANCER CELLS: LOSS OF CONTROL OF CELL DIVISION	468
TUMOR-INDUCING VIRUSES: VIRAL ONCOGENES	468
Life Cycle of Rous Sarcoma Virus	468
Diversity of Retroviral Oncogenes	470
Oncogene Products as Regulators of Cell Division	470
PROTOONCOGENES AND CELLULAR ONCOGENES	471
Homology with Viral Oncogenes	471
Transfection Experiments	472
Cellular Oncogenes Contain Introns—Their Viral Homologs Are Single Exons	473
Conservation of Protooncogenes during Evolution	473
Protooncogene Products: Key Regulators of Cell Division	474
<i>pjun</i> and <i>pfos</i> as Activators of Gene Transcription	475
Mutational Origin of <i>ras</i> Cellular Oncogenes	476
Translocation Breakpoints at Protooncogene Loci	478
Insertional Activation of Protooncogenes	479
Amplification of Protooncogenes in Cancer Cells	480
Origin of Viral Oncogenes	482

CANCER AS THE END PRODUCT OF A MULTISTEP PROCESS	484
SUMMARY	484
REFERENCES	485
PROBLEMS AND QUESTIONS	487

## Chapter 18

### Variations in Chromosome Structure 488

GIANT POLYTENE CHROMOSOMES IN DIPTERA	489
Features of Polytene Chromosomes	490
DEFICIENCIES	490
DUPLICATIONS	491
INVERSIONS	492
Inversions and Balanced Lethal Mutations	494
TRANSLOCATIONS	495
POSITION EFFECTS	498
CHROMOSOME ABERRATIONS AND EVOLUTION	499
HUMAN CHROMOSOME TECHNIQUES	499
CHROMOSOME ABERRATIONS IN HUMANS	499
BANDED CHROMOSOMES AND PHYLOGENY	506
Rhesus and African Green Monkey	507
SUMMARY	508
REFERENCES	509
PROBLEMS AND QUESTIONS	509

## Chapter 19

### Variations in Chromosome Number 511

TRISOMY IN HUMANS	512
Down Syndrome (47,+21)	512
Amniocentesis for Detecting Aneuploidy	515
Trisomy-13 and Trisomy-18	515
Turner Syndrome (45,X)	516
Klinefelter Syndrome (46,XXY)	519
Aneuploidy of X Chromosomes and Mental Deficiency	520
47,YYY and Behavior	520
CHROMOSOMAL MOSAICS	521
NONDISJUNCTION AND ANEUPLOIDY IN <i>DROSOPHILA</i>	521
ANEUPLOID SEGREGATION IN PLANTS	523
Tetrasomics and Nullisomics	523
EUPLOIDY	524
POLYPLOIDY IN ANIMALS	524

**POLYPLOIDY IN PLANTS 524**

Chromosome Doubling in Somatic and Germ Cells 524

Triploid Plants 525

Tetraploid Plants 526

**INDUCED POLYPLOIDY 527**

**EXPERIMENTAL PRODUCTION OF**

**POLYPLOIDS 528**

Sterile Hybrid Grass Made Fertile by Chromosome Doubling 528

**APPLICATIONS OF POLYPLOIDY 530**

New World Cotton 530

Primrose Hybridization 531

Disease Resistance in Tobacco 531

Polyloid Fruits, Flowers, and Wheat 531

**SUMMARY 532**

**REFERENCES 533**

**PROBLEMS AND QUESTIONS 533**

**Chapter 20**

**Extrachromosomal Inheritance 535**

**CRITERIA FOR EXTRANUCLEAR INHERITANCE 536**

**CYTOPLASMIC ORGANELLES AND SYMBIONTS 536**

DNA in Mitochondria 537

Organization of Mitochondrial Genomes 538

DNA in Plastids 539

Chloroplast DNA and Drug Resistance 541

Organization of Plastid Genomes 542

Symbiont Bacteria in *Paramecium* Cytoplasm 544

**PLASMID DNA AND TUMOR**

**TRANSFORMATION 547**

**CYTOPLASMIC MALE STERILITY IN PLANTS 547**

Male Sterility in a Cross-Pollinating Plant 547

Danger of Uniformity 548

**MATERNAL EFFECTS 548**

Maternal Effect in Snail Shell Coiling 549

Maternal Effect in *Drosophila* 549

**SUMMARY 550**

**REFERENCES 550**

**PROBLEMS AND QUESTIONS 551**

**Chapter 21**

**Quantitative Genetics 552**

**QUANTITATIVE TRAITS AND QUANTITATIVE GENETICS 553**

**DESCRIPTIVE STATISTICS 553**

The Mean 554

The Variance and Standard Deviation 554

Correlations 555

Additivity of Means and Variances 555

**THE MULTIPLE FACTOR HYPOTHESIS 556**

A Quantitative Model 556

Analysis of Variance 557

Broad-Sense Heritability 557

**PREDICTING PHENOTYPES 558**

Narrow-Sense Heritability 558

Estimating an Offspring's Phenotype 559

Predicting Responses to Artificial Selection 560

Practical Significance and Evolutionary Implications 562

**SUMMARY 564**

**REFERENCES 564**

**PROBLEMS AND QUESTIONS 564**

**Chapter 22**

**Population and Evolutionary Genetics 566**

**GENETIC VARIATION 567**

Allele Frequencies 568

**RANDOM MATING AND THE HARDY-WEINBERG METHOD 568**

Applications of the Hardy-Weinberg Method 569

**INBREEDING 570**

The Inbreeding Coefficient 570

Calculating  $F$  from Pedigrees 571

Genotype Frequencies under Inbreeding 572

Quantitative Effects of Inbreeding 572

**OUTBREEDING AND ASSORTATIVE MATING 572**

**CHANGES IN ALLELE FREQUENCIES 574**

Mutation 574

Migration 574

Selection 574

Selection Dynamics 575

Random Genetic Drift 578

Effects of Random Drift 578

**GENETIC EQUILIBRIUM 580**

Balancing Selection 580

Mutation-Selection Balance 581

Mutation-Drift Balance 582

**EVOLUTIONARY GENETICS 582**

The Synthetic Theory of Evolution 583

Evidence for Adaptive Evolution 584

Molecular Evolution 585

Evolution of Multigene Families 586

**SUMMARY 588**

**REFERENCES 588**

**PROBLEMS AND QUESTIONS 589**



**Chapter 23****Genetics of Behavior 591****GENETIC MECHANISMS 592**

Inheritance and Learning in Bees 594

**EXPERIMENTAL BEHAVIOR GENETICS 594***Escherichia coli* Chemotaxis 595*Drosophila* Behavior Genetics 596Response to Light and Gravity in *Drosophila* 596**GENETIC AND ENVIRONMENTAL INTERACTIONS IN DOGS 599**

Sociobiology 600

The Evolutionary Approach 601

**SUMMARY 601****REFERENCES 602****PROBLEMS AND QUESTIONS 602****Chapter 24****Genetic Engineering and the Future 604****DISSECTION OF BIOLOGICAL PROCESSES 605***Saccharomyces cerevisiae*, a Eukaryotic *Escherichia coli* 605*Drosophila*, P-Element Transformation 610*Arabidopsis thaliana*, the *Drosophila* of the Plant Kingdom 612**MICROORGANISMS 618**

Single-Cell Proteins 619

Industrial Chemicals 620

Proteins with Industrial Applications 620

Proteins with Medicinal Applications 621

Dairy Fermentations 623

Degradation of Pollutants 623

**PLANTS 624**

Regeneration of Plants from Protoplasts and Callus Tissue 624

The Ti Plasmid of *Agrobacterium tumefaciens* 625

Disarmed Ti Vectors 627

Chimeric Selectable Marker Genes 628

Binary Ti Vectors 630

Transformation by Co-cultivation of Tissue Explants with *Agrobacterium* 630

Direct Gene Transfer, Electroporation, and Microprojectile Guns 630

Herbicide-Tolerant Plants 631

Disease- and Insect-Resistant Varieties 632

"High-Lysine" Corn 634

Enhanced Nitrogen Utilization 635

**ANIMALS 637**

Microinjection of DNA into Fertilized Eggs 637

Retroviral Vectors 638

Gene Expression in Transgenic Mice 639

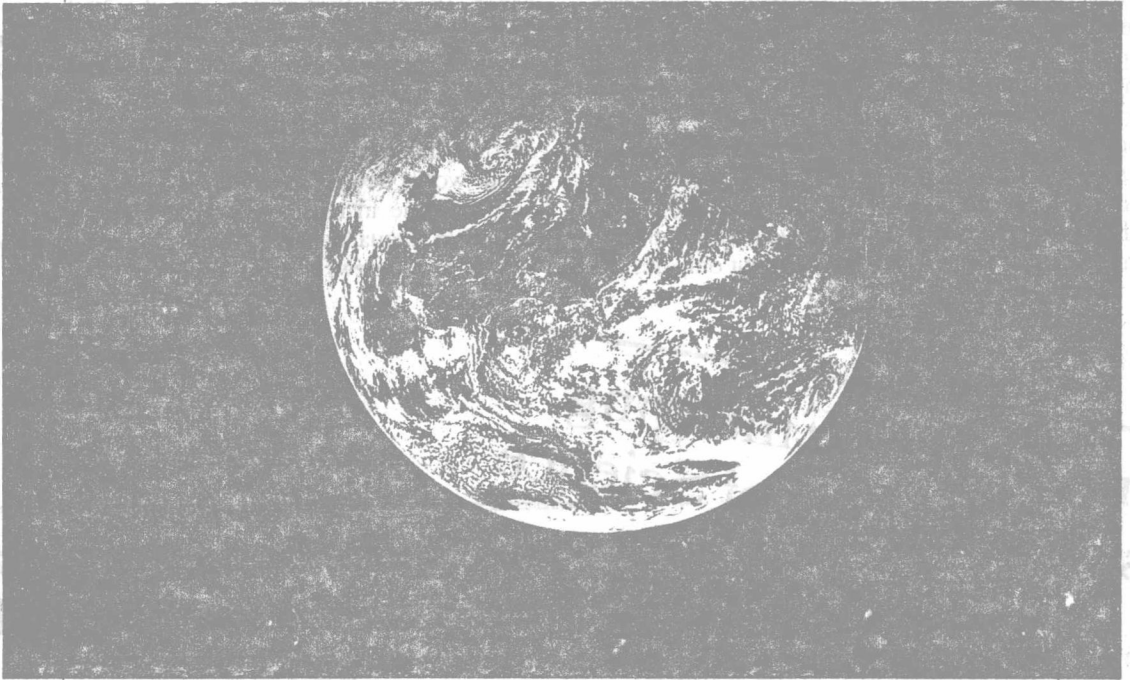
Increased Growth Rates via Amplified Growth Hormone Genes 640

Virus-Resistant Transgenic Chickens 642

Secretion of Valuable Proteins in Milk of Transgenic Animals 642

**"ANTI-SENSE RNA" STRATEGIES FOR REGULATING GENE EXPRESSION 643****SOMATIC-CELL GENE THERAPY TREATMENT OF HUMAN DISEASES 645****SUMMARY 647****REFERENCES 648****GLOSSARY G1****ANSWERS TO PROBLEMS A1****INDEX I1**

# Chapter 1



## Introduction

**The World is a Colossal Genetics Laboratory, 3**

**The Birth of Genetics, 15**

**Growth of a Science: From Mendel to Genetic**

**Engineering, 17**

**Summary, 18**

**References, 19**

(Photo above): View of the Earth from the Apollo 8 spacecraft.

**"G**ENE Studies Emerging as Key Engine of Science"—headline, *The New York Times*, September 6, 1988. The significance of this headline is not that it announces any new discovery. Geneticists have known for decades that genes play central roles in science and, in fact, in all aspects of the lives of all living organisms. The significance of this headline is that our society as a whole, or at least the more informed component of our society, now also recognizes the major roles that genes play in shaping our lives and the lives of all living creatures on planet Earth. Of course, such headlines have been commonplace in science journals for decades, but only during

the last few years have these headlines appeared frequently in the popular press, particularly the news media. For example, a quick scan of one major newspaper, *The New York Times*, over the 30-day period from September 6, 1988, to October 6, 1988, revealed three major headlines focusing on genetics; one, "Panel Backs Gene Transplant For Cancer Treatment Test," was on the front page (Fig. 1.1). In fact, as anyone who makes a significant effort to stay informed about world developments knows, similar headlines have become increasingly prevalent during the last decade in all quality newspapers as well as in the radio and television news media throughout the world.

## Gene Studies Emerging As Key Engine of Science

By HAROLD M. SCHMECK Jr.

**T**HE science of genetics is fast becoming what geneticists always knew it was: the central and most provocative science of life.

Toronto, president of the the Toronto meeting was meetings, which are he Four thousand scientist tries attended, twice t and four times the nur resented three decade:

## Panel Backs Gene Transplant For Cancer Treatment Test

By WARREN E. LEARY  
Special to The New York Times

WASHINGTON, Oct. 3 — An influential advisory committee recommended today that the Government for the first time approve the transplanting of foreign genes into human patients.

and the Food and Drug Administration. But the scientist and his principal co-worker on the cancer experiment, Dr. Steven Rosenberg, said they had indications from the remaining authorities

## DNA Pioneer to Tackle Biggest Gene Project Ever

By HAROLD M. SCHMECK Jr.

**D**R. James D. Watson, who is about to assume a key role in the largest biological research project ever contemplated, sees that effort as the redemption of a longstanding bargain between scientists and other citizens.

"I think it is imperative to keep our bargain with the people who have supported biomedical research," he said in an interview Friday. The project will immensely expand scientists' knowledge of human genetics. Directly and indirectly, that knowledge will be used to seek new ways of curing or coping with important diseases by teasing out the secrets of the genes.

Dr. Watson was co-discoverer in 1953 of the structure of DNA, the substance of the genes in all living things. In 1962, he shared a Nobel Prize for that work with Dr. Francis H. C. Crick, now at the Salk Institute in La Jolla, Calif., and Dr. Maurice H. F. Wilkins of Cambridge University in England. The discovery showed how DNA functions as the repository of all hereditary information and led to a revolution in biology that continues to accelerate and expand.

The new job that Dr. Watson begins this week in-



The New York Times/Michael Shavel

Dr. James D. Watson, who pioneered research into the structure of DNA and now heads an effort to map the human genome.

volves helping to organize a huge federally supported project to define all of the roughly 50,000 to 100,000 human genes. It is known as the genome project; a genome being the sum of all the genes of any organism. The project, if approved by Congress, is expected to take 15 years and to cost several billion dollars.

Dr. Watson has been appointed associate director of the National Institutes of Health for genome research, a newly created post to oversee the institutes' involvement with the genome project.

In his early years of research, Dr. Watson was known as a brash, witty and often abrasive enfant terrible. The new appointment seemingly completes his metamorphosis into a senior statesman of science.

"People know that I am interested in biology as a whole," he said. "Of course, I am also interested in human beings."

The project is a logical outcome of the discovery made in 1953, but until recently, the idea would have been derided as fantasy because it was so far beyond the technical capabilities of the science. Only a relatively few genes have been completely defined to date but the new project will try to define the chemical composition of them all. The effort is made possible now by a series of technical and

Continued on Page C78

Figure 1.1 Headlines in the *The New York Times* on September 6 and October 8, 1988, heralding some of the exciting recent developments in genetics. (Copyright 1988 by The

New York Times Company. Photograph by Michael Shavel, New York Times Pictures. Reprinted with permission.)