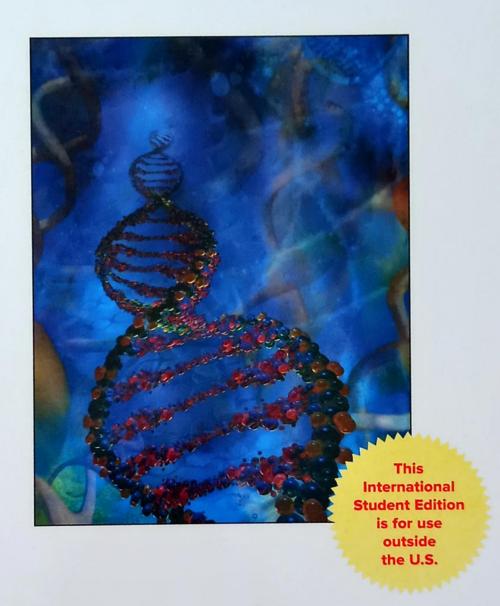
Ricki Lewis

Human Genetics

Concepts and Applications

Twelfth Edition







Human Genetics

Concepts and Applications

Ricki Lewis

Genetic Counselor CareNet Medical Group Schenectady, New York

Adjunct Assistant Professor of Medical Education Alden March Bioethics Institute Albany Medical College

Writer, Medscape Medical News Genetic Literacy Project

Blogger, Public Library of Science





HUMAN GENETICS

Published by McGraw-Hill Education, 2 Penn Plaza, New York, NY 10121. Copyright © 2018 by McGraw-Hill Education. All rights reserved. No part of this publication may be reproduced or distributed in any form or by any means, or stored in a database or retrieval system, without the prior written consent of McGraw-Hill Education, including, but not limited to, in any network or other electronic storage or transmission, or broadcast for distance learning.

Some ancillaries, including electronic and print components, may not be available to customers outside the United States.

Printed in the Philippines. Not for sale outside the Philippines.

1 2 3 4 5 6 7 8 9 PAB 20 20 19

ISBN 978-981-3157-84-2 MHID 981-3157-84-4

All credits appearing on page or at the end of the book are considered to be an extension of the copyright page.

The Internet addresses listed in the text were accurate at the time of publication. The inclusion of a website does not indicate an endorsement by the authors or McGraw-Hill Education, and McGraw-Hill Education does not guarantee the accuracy of the information presented at these sites.

Brief Contents



PART 1
Introduction 1

CHAPTER 1
What Is in a Human Genome? 1

CHAPTER 2 Cells 15

CHAPTER 3
Meiosis, Development, and Aging 40



Transmission

Genetics 66

CHAPTER 4 Single-Gene Inheritance 66

CHAPTER 5
Beyond Mendel's Laws 86

CHAPTER 6 Matters of Sex 106

CHAPTER 7
Multifactorial Traits 124

Genetics of Behavior 141



DNA and

Chromosomes 158

CHAPTER 9

DNA Structure and Replication 158

Gene Action: From DNA to Protein 175

Gene Expression and Epigenetics 193

Gene Mutation 205

CHAPTER 13 Chromosomes 229



PART 4

Population Genetics 256

CHAPTER 14
Constant Allele Frequencies and DNA Forensics 256

CHAPTER 15
Changing Allele Frequencies 272

CHAPTER 16
Human Ancestry and Evolution 294



P A R T 5

Immunity and

Cancer 319

CHAPTER 17
Genetics of Immunity 319

CHAPTER 18
Cancer Genetics and Genomics 344



P A R T 6

Genetic Technology 365

CHAPTER 19
DNA Technologies 365

CHAPTER 20
Genetic Testing and Treatment 383

CHAPTER 21
Reproductive Technologies 401

CHAPTER 22 Genomics 418

Contents

About the Author iii

Preface ix

Applying Human Genetics xiii

The Human Touch xiv

The Lewis Guided Learning System xv

PART 1 Introduction 1



CHAPTER 1

What Is in a Human

Genome? 1

- 1.1 Introducing Genes and Genomes 2
- 1.2 Levels of Genetics and Genomics 2
- 1.3 Applications of Genetics and Genomics 6
- 1.4 A Global Perspective on Genomes 10



CHAPTER 2

- 2.1 Introducing Cells 16
- 2.2 Cell Components 16
- 2.3 Cell Division and Death 27
- 2.4 Stem Cells 31
- 2.5 The Human Microbiome 36



CHAPTER 3

Meiosis, Development,

and Aging 40

- 3.1 The Reproductive System 41
- 3.2 Meiosis 42
- 3.3 Gametes Mature 46
- 3.4 Prenatal Development 49
- 3.5 Birth Defects 58
- 3.6 Maturation and Aging 60

PART 2 Transmission Genetics 66



CHAPTER

Single-Gene

Inheritance 66

- 4.1 Following the Inheritance of One Gene 67
- 4.2 Single-Gene Inheritance Is Rare 71

- 4.3 Following the Inheritance of More Than One Gene 75
- 4.4 Pedigree Analysis 78



Beyond Mendel's

Laws 86

- 5.1 When Gene Expression Appears to Alter Mendelian Ratios 87
- 5.2 Mitochondrial Genes 94
- 5.3 Linkage 97



Matters of Sex 106

- 6.1 Our Sexual Selves 107
- 6.2 Traits Inherited on Sex Chromosomes 111
- 6.3 Sex-Limited and Sex-Influenced Traits 116
- 6.4 X Inactivation 117
- 6.5 Parent-of-Origin Effects 118



CHAPTER 7
Multifactorial Traits 124

- 7.1 Genes and the Environment Mold Traits 125
- 7.2 Polygenic Traits Are Continuously Varying 127
- 7.3 Methods to Investigate Multifactorial
 Traits 129
- 7.4 A Closer Look: Body Weight 137



Genetics of Behavior 141

- 8.1 Genes and Behavior 142
- 8.2 Sleep 143
- 8.3 Intelligence and Intellectual Disability 144
- 8.4 Drug Addiction 145
- 8.5 Mood Disorders 148
- 8.6 Schizophrenia 149
- 8.7 Autism 152

PART 3 DNA and Chromosomes 158



CHAPTER 9

DNA Structure

and Replication 158

- 9.1 Experiments Identify and Describe the Genetic Material 159
- 9.2 DNA Structure 163
- 9.3 DNA Replication—Maintaining Genetic Information 167
- 9.4 Sequencing DNA 170



CHAPTER 10

Gene Action: From DNA

to Protein 175

- 10.1 The Importance of Proteins 176
- 10.2 Transcription Copies the Information in DNA 176
- 10.3 Translation of a Protein 182
- 10.4 Processing a Protein 187



CHAPTER 11

Gene Expression and Epigenetics 193

- 11.1 Gene Expression Through Time and Tissue 194
- 11.2 Control of Gene Expression 197
- 11.3 Maximizing Genetic Information 199
- 11.4 Most of the Human Genome Does Not **Encode Protein 201**



CHAPTER 12 Gene Mutation 205

- 12.1 The Nature of Gene Variants 206
- 12.2 A Closer Look at Two Mutations 208
- 12.3 Allelic Diseases 211

- 12.4 Causes of Mutation 212
- 12.5 Types of Mutations 215
- 12.6 The Importance of Position 219
- 12.7 DNA Repair 221



CHAPTER 13 Chromosomes 229

- 13.1 Portrait of a Chromosome 230
- 13.2 Detecting Chromosomes 232
- 13.3 Atypical Chromosome Number 237
- 13.4 Atypical Chromosome Structure 245
- 13.5 Uniparental Disomy—A Double Dose from One Parent 251

PART 4 Population Genetics 256



CHAPTER 14

Constant Allele

Frequencies and DNA Forensics 256

- 14.1 Population Genetics Underlies **Evolution 257**
- 14.2 Constant Allele Frequencies 258
- 14.3 Applying Hardy-Weinberg Equilibrium 260
- 14.4 DNA Profiling Uses Hardy-Weinberg Assumptions 261



CHAPTER 15

Changing Allele

Frequencies 272

15.1 Population Matters: Steel Syndrome in East Harlem 273

- 15.2 Nonrandom Mating 274
- 15.3 Migration 275
- 15.4 Genetic Drift 276
- 15.5 Mutation 280
- 15.6 Natural Selection 281
- 15.7 Eugenics 287



CHAPTER 16

Human Ancestry

and Evolution 294

- 16.1 Human Origins 295
- 16.2 Methods to Study Molecular Evolution 303
- 16.3 The Peopling of the Planet 307
- 16.4 What Makes Us Human? 312

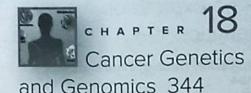
PART 5 Immunity and Cancer 319



Genetics of

Immunity 319

- 17.1 The Importance of Cell Surfaces 320
- 17.2 The Human Immune System 323
- 17.3 Abnormal Immunity 328
- 17.4 Altering Immunity 334
- 17.5 Using Genomics to Fight Infection 339



- 18.1 Cancer Is an Abnormal Growth That Invades and Spreads 345
- 18.2 Cancer at the Cellular Level 348
- 18.3 Cancer Genes and Genomes 352
- 18.4 Diagnosing and Treating Cancer 359

PART 6 Genetic Technology 365



DNA Technologies 365

- 19.1 Patenting DNA 366
- 19.2 Modifying DNA 367
- 19.3 Monitoring Gene Function 373
- 19.4 Gene Silencing and Genome Editing 375



Genetic Testing and

Treatment 383

- 20.1 Genetic Counseling 384
- 20.2 Genetic Testing 386
- 20.3 Treating Genetic Disease 393
- 20.4 CRISPR-Cas9 in Diagnosis and Treatment 397



Reproductive

Technologies 401

- 21.1 Savior Siblings and More 402
- 21.2 Infertility and Subfertility 403
- 21.3 Assisted Reproductive Technologies 406
- 21.4 Extra Embryos 413



Genomics 418

- 22.1 From Genetics to Genomics 419
- 22.2 Analysis of Human Genome Content 426
- 22.3 Personal Genome Sequencing 429

Glossary G-1 Index I-1



Additional McGraw-Hill Education International Editions are available in the following subjects:

Geology and Mineralogy Accounting

Industrial Arts and Vocational Education Agriculture

Management **Biological Sciences**

Management Information Systems **Business and Industrial Management**

Marketing Chemistry

Mathematics Chemistry and Chemical Engineering

Mechanical Engineering Civil Engineering

Computer Information Technology (CIT) Medicine

Meteorology Decision Science

Physics Economics

Political Science Education

Psychology **Electrical Engineering**

Sociology Electrical Engineering Technology

Electronics and Computer Science Tech and Trade

Finance Geography

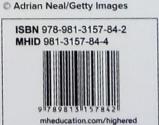
Some ancillaries, including electronic and print components, may not be available to customers outside the United States.

FOR SALE IN THE PHILIPPINES ONLY

Statistics







Cover Image:

