

## **TABLE OF CONTENTS**

About the Authors xi

Introduction xv

### **1 Exploring Online Genetics Sources 1**

Background 1

Human Genome Project 2

National Center for Biotechnology Information (NCBI) 2

UCSC Genome Browser 3

The Encyclopedia of DNA Elements (ENCODE) 3

Roadmap Epigenomic Project 5

References 5

Exercise Questions 6

Additional Exercise Questions 11

### **2 Observation of Human Inheritance 13**

Background 13

Mendelian Genetics 13

The Transmission of Hereditary Characteristics 16

Mendelian Disorders 19

References 20

Exercise Questions 21

Additional Exercise Questions 22

### **3 Reading, Understanding, and Constructing Human Pedigrees 25**

Background 25

Basic Pedigree Nomenclature 26

Modes of Inheritance 26

Autosomal Recessive Inheritance 28

Pedigrees Representing Autosomal Recessive Inheritance 28

Autosomal Dominant Inheritance 29

Pedigrees Representing Autosomal Dominant Inheritance	30
X-Linked Recessive Inheritance	30
Pedigrees Representing X-Linked Recessive Inheritance	31
X-Linked Dominant Inheritance	32
Pedigrees Representing X-Linked Dominant Inheritance	32
Y-Linked Inheritance	34
Pedigrees Representing Y-Linked Inheritance	34
Non-Mendelian Patterns of Inheritance	35
Confounding Factors in Pedigree Generation and Interpretation	36
References	36
Exercise Questions	38
Additional Exercise Questions	39

#### **4 Cytogenetics 43**

Background	43
Generation of Karyograms/Chromosome Banding	45
FISH, Fluorescent <i>In Situ</i> Hybridization	47
Comparative Genomic Hybridization (CGH) and Array-Based CGH Technology	48
Chromosomal Abnormalities	48
Numerical Chromosomal Abnormalities	49
Structural Chromosomal Abnormalities	50
References	51
Exercise Questions	52
Additional Exercise Questions	54

#### **5 Exploring DNA, RNA, and Protein Sequence Databases and Genome Browsers 55**

Background	55
General Biological Databases	57
RNA-Specific Databases	59

Protein-Specific Databases 61

Regulatory DNA Database 62

Genome Browsers 65

References 67

Exercise Questions 67

Additional Exercise Questions 68

## **6 Exploring Online Bioinformatics Tools 71**

Background 71

BLAST 71

ExPASy 72

Clustal Omega 74

Reactome 76

References 77

Exercise Questions 77

Additional Exercise Questions 79

## **7 Multifactorial Inheritance and Common Complex Diseases 81**

Background 81

Polygenic Complex Diseases 82

Investigating Complex Traits 82

Obesity as a Complex Disease 83

Diabetes Mellitus Type 2 as a Complex Disease 84

Coronary Artery Diseases as Complex Diseases 85

References 86

Exercise Questions 88

Additional Exercise Questions 88

## **8 Neurogenetics and Behavioral Genetics 91**

Background 91

Genetic Regulation of Neural Development 91

Structure and Function of the Nervous System 92

Understanding the Genetics of Neurological Disease 94

Examples of Neurogenetic Disorders 94

Huntington's Disease 94

Parkinson's Disease 95

Rett Syndrome 95

Behavioral Genetics 96

Genetics of Depression 97

Genetics of Drug Addiction 97

Genetics of Schizophrenia 98

References 99

Exercise Questions 100

Additional Exercise Questions 101

## **9 Cancer Genetics 103**

Background 103

Cell Cycle and Its Regulation 104

Oncogenes and Tumor Suppressor Genes 105

Carcinogens/Environmental Factors in Cancer 107

Breast Cancer 108

Role of Molecular Medicine in Breast Cancer Treatment 110

Recent Advances in Cancer Diagnosis and Treatment 111

References 112

Exercise Questions 113

Additional Exercise Questions 114

## **10 Genetic Counseling 115**

Background 115

References 117

Exercise Questions 117

Additional Exercise Questions 119

**11 Evolving Tools in Genome Editing: CRISPR-Cas 121**

Background 121

Mechanism of CRISPR-Cas9 System 123

Applications of CRISPR-Cas9 System 123

Generation of Animal Models and Engineering Cells 123

Somatic Genome Editing 124

Repair of Genetic Disorders 124

Functional Genomic Screening 124

Treatment of Infectious Diseases 125

Other Applications of CRISPR-Cas9 System 125

References 126

Acknowledgment 127

Exercise Questions 127

Additional Exercise Questions 128

Glossary 129

Index 139