# TABLE OF CONTENTS

**Preface**  
3

**Chapter 1** Genetics 101  
1.1 Cells, Genomes, DNA, and Genes  6  
1.2 Types of Genetic Disease  6  
1.3 Laws of Inheritance  7  
1.4 Genetic Variation  9

**Chapter 2** Diagnosis of a Genetic Disease  
11  
2.1 History and Physical Examination  12  
2.2 Red Flags for Genetic Disease  12  
2.3 Uses of Genetic Testing  13  
2.4 Types of Genetic Testing  13  
   2.4.1 Cytogenetic Testing  13  
   2.4.2 Biochemical Testing  14  
   2.4.3 Molecular Testing  14

**Chapter 3** Pedigree and Family History-taking  
15  
3.1 Importance of Family History  16  
3.2 How to Take a Family Medical History  17  
3.3 Pedigrees  17

**Chapter 4** Newborn Screening  
19  
4.1 Overview of Newborn Screening  20  
   4.1.1 Screening Procedure and Follow-up  20  
   4.1.2 Retesting  20  
   4.1.3 Clinical Evaluation and Diagnostic Testing  20  
   4.1.4 Treatment  20  
   4.1.5 Tests Performed  21  
4.2 Newborn Screening Programs  21  
4.3 Newborn Hearing Screening  22  
   4.3.1 Screening Procedure  22  
   4.3.2 Retesting  22  
   4.3.3 Treatment  22  
4.4 Newborn Hearing Screening Programs  23

**Chapter 5** Genetic counseling  
25  
5.1 Role of Genetic Counseling  26  
5.2 Process of Genetic Counseling  26  
5.3 Patient Education  27

**Chapter 6** Indications for a Genetic Referral  
29  
6.1 When to Refer to a Genetic Specialist  30  
   6.1.1 Family History  30  
   6.1.2 Delayed Growth and Development  30  
   6.1.3 Reproductive Issues  30

**Chapter 7** Psychological and Social Implications  
33  
7.1 Genetic Information and Other Medical Information  34  
7.2 A Lifetime of Affected Relationships  34
7.3 Impact of a Genetic Diagnosis 35
7.3.1 Patients 35
7.3.2 Parents 35
7.3.3 Family 35
7.3.4 Communities 36
7.4 Coping Mechanisms 36

Chapter 8 Ethical, Legal, and Social Issues 39
8.1 Description of Ethical, Legal, and Social Issues 40
8.1.1 Communicating Test Results 40
8.1.2 Direct-to-consumer Tests 40
8.1.3 Duty to Disclose 40
8.1.4 Genetic Discrimination 40
8.1.5 Informed Consent 41
8.1.6 Privacy 41
8.1.7 Psychosocial Impact 41
8.1.8 Reproductive Issues 41
8.1.9 Societal Values 42
8.1.10 Test Utility 42
8.1.11 Test Validity 42

Chapter 9 Patient Stories and Consumer Profiles 43
9.1 Inherited Breast & Ovarian Cancer 44
9.2 The Value of Newborn Screening 44
9.3 Hereditary Hemachromatosis 45
9.4 Type II Diabetes 46

Chapter 10 Genetics Resources and Services 47

Appendices 61
A. Basic Genetics Information 62
B. Family History is Important for Your Health 64
C. Family Health History Questionnaire 66
D. Healthcare Provider Card 68
E. Inheritance Patterns 70
F. Chromosomal Abnormalities 72
G. Genetic Testing 73
H. Prenatal Screening and Testing 75
I. Genetic Testing Methodologies 78
J. Newborn Screening 80
K. Birth Defects 81
L. Genetics and the Environment 82
M. Pharmacogenomics and Pharmacogenetics 84
N. Integrated Health Data Systems 86
O. Making Sense of Your Genes: A Guide to Genetic Counseling 87
P. Cultural Competency in Genetics 90
Q. National Coalition for Health Professional Education in Genetics (NCHPEG)—Principles of Genetics for Health Professionals 91
R. Centers for Disease Control and Prevention (CDC)—Genomic Competencies for All Public Health Professionals and Clinicians 98