

TABLE OF CONTENTS

PREFACE		3
CHAPTER 1	GENETICS 101	5
1.1	Cells, Genomes, DNA, and Genes	6
1.2	Major 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	The Importance of Newborn Screening Tests	20
4.2	Testing Procedure and Follow-up	20
4.3	Retesting	20
4.4	Tests Performed	21
4.5	Treatment	21
4.6	Newborn Screening Programs	21
4.7	Newborn Hearing Screening	22
4.8	Newborn Hearing Screening Programs	22
CHAPTER 5	GENETIC COUNSELING	25
5.1	Role of Genetic Counseling	26
5.2	Process of Genetic Counseling	27
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	Developmental Delay/Growth	30
6.1.3	Reproductive Issues	30
CHAPTER 7	PSYCHOLOGICAL AND SOCIAL IMPLICATIONS	33
7.1	Genetic Information vs. 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 ELSI Issues <i>40</i>	
8.1.1	Communicating Test Results <i>40</i>	
8.1.2	Direct-to-consumer Tests <i>40</i>	
8.1.3	Duty to Disclose <i>40</i>	
8.1.4	Genetic Discrimination <i>41</i>	
8.1.5	Informed Consent <i>41</i>	
8.1.6	Privacy <i>41</i>	
8.1.7	Psychosocial Impact <i>41</i>	
8.1.8	Reproductive Issues <i>42</i>	
8.1.9	Societal Values <i>42</i>	
8.1.10	Test Utility <i>42</i>	
8.1.11	Test Validity <i>42</i>	
CHAPTER 9	PATIENT STORIES AND CONSUMER PROFILES	43
9.1	Inherited Breast & Ovarian Cancer <i>44</i>	
9.2	The Value of Newborn Screening <i>44</i>	
9.3	Hereditary Hemochromatosis <i>45</i>	
9.4	Type II Diabetes <i>45</i>	
CHAPTER 10	NEW ENGLAND GENETICS RESOURCES & SERVICES	47
CHAPTER 11	CONSUMER FACT SHEETS	55
	Family History <i>56</i>	
	Basic Genetic Information <i>58</i>	
	Dominant and Recessive Genetic Diseases <i>59</i>	
	X-linked Genetic Diseases <i>60</i>	
	What is a Chromosome Abnormality? <i>61</i>	
	Understanding Genetic Testing <i>62</i>	
	Prenatal Diagnosis <i>63</i>	
	Birth Defects/Congenital Abnormalities <i>64</i>	
	Newborn Screening <i>65</i>	
	Genetic Counseling <i>66</i>	
APPENDIX		67
A.	Teratogens/Prenatal Substance Abuse <i>68</i>	
B.	Single-Gene Disorders <i>70</i>	
C.	Chromosomal Abnormalities <i>71</i>	
D.	Pharmacogenomics <i>72</i>	
E.	Cultural Competencies in Genetics <i>74</i>	
F.	NCHPEG Principles of Genetics for Health Professionals <i>75</i>	
G.	CDC Genomic Competencies for All Public Health Professionals and Clinicians <i>81</i>	