

Table of Contents

<i>Preface</i>	v
1. Scope of Medical Genetics	1
2. Skin.....	5
i. Icthyosis	5
ii. Psoriasis	9
iii. Multiple Neurofibromatosis	12
iv. Porphyrias	16
v. Blooms Syndrome.....	20
3. Skeletal System	23
i. Ankylosing Spondylitis.....	23
ii. Rheumatoid Arthritis.....	26
iii. Osteogenesis Imperfecta	29
4. Muscle	32
i. Muscular Dystrophies	32
a. Duchenne Muscular Dystrophy (DMD).....	32
b. Becker's Muscular Dystrophy (BMD)	34
ii. Myotonia	37

5. Eye.....	40
i. Glaucoma.....	40
ii. Cataract.....	43
6. Jaws.....	45
i. Cleft Lip and Palate.....	45
7. Ears.....	48
i. Deafness.....	48
8. Alimentary System.....	52
i. Pyloric Stenosis.....	52
ii. Cirrhosis of Liver.....	54
9. Respiratory System.....	56
i. Cystic Fibrosis.....	56
10. Cardio Vascular System.....	58
i. Congenital Heart Disease.....	58
ii. Hypertension.....	61
iii. Coronary Heart Disease.....	64
11. Central Nervous System.....	68
i. Neural Tube Defects.....	68
a. Spina Bifida.....	69

b. Anencephaly	71
ii. Hereditary Spastic Paraplegia	73
12. Kidney and Urinogenetal Tract	75
i. Cystinosis	75
ii. Polycystic Kidney Disease.....	77
13. Reproductive System.....	80
i. Polycystic Ovary Syndrome or Disease (PCOD/PCOS)	80
14. Endocrine System.....	84
i. Congenital Hypothyroidism.....	84
ii. Goiter	87
iii. Diabetes Mellitus	91
15. Cancer Genetics.....	96
i. Characteristics of Cancer Cells.....	96
ii. Chromosomes in Malignancies.....	102
iii. Cancer as a Genetic Disorder and Cancer in Families.....	108
iv. Inherited Versus Sporadic Cancers	120
v. Cell Cycle Regulation.....	123