

Introduction.

## **Chapter 1 Cytogenetic Abnormalities.**

Introduction to cytogenetic abnormalities.

Common aneuploidy – recurrence risks and counseling pitfalls.

Chromosomal mosaicism – prenatal diagnosis.

Chromosomal mosaicism – postnatal diagnosis.

Reciprocal translocations and structural abnormalities.

Robertsonian translocations.

Sex discrepancies.

## **Chapter 2 Introduction to Mendelian Disorders.**

### **Chapter 3 Autosomal Dominant Disorders.**

Features of autosomal dominant inheritance.

Polycystic kidney disease.

Hereditary non-polyposis colon cancer.

Huntington disease.

Marfan syndrome.

Retinoblastoma.

Tuberous sclerosis.

### **Chapter 4 Autosomal Recessive Disorders.**

Features of autosomal recessive inheritance.

Congenital adrenal hyperplasia.

Cystic fibrosis.

Spinal muscular atrophy.

Hemoglobinopathies.

Fanconi anemia.

Maple syrup urine disease.

### **Chapter 5 X-linked Disorders.**

Features of X-linked inheritance.

Duchenne muscular dystrophy.

Hunter syndrome.

Fragile X syndrome

Factor VIII deficiency.

X-linked adrenoleukodystrophy.

### **Chapter 6 Mitochondrial Inheritance.**

Features of mitochondrial inheritance.

Leigh syndrome.

### **Chapter 7 Multifactorial Inheritance.**

Features of multifactorial inheritance.

Multifactorial inheritance.

### **Chapter 8 Abnormal Ultrasound Findings.**

Recurrent hypotonia and polyhydramnios.

Holoprosencephaly.

Abnormalities of the digits.

Multicystic kidneys.

Omphalocele.

Recurrent fetal hydrops.

Nonmotile ciliopathies.

### **Chapter 9 Skeletal Dysplasias.**

Achondroplasia and hypochondroplasia.

Osteogenesis imperfecta type II.

Short rib polydactyly syndromes.

**Chapter 10 Imprinting Disorders.**

**Chapter 11 First and Second Trimester Screening Tests.**

**Chapter 12 Infertility.**

**Chapter 13 Family History.**

**Chapter 14 Consanguinity.**

**Chapter 15 Non-paternity.**

**Chapter 16 Fetal Infection.**

**Chapter 17 Teratogens.**

**Chapter 18 Autism.**

**Chapter 19 Glossary.**

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