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.