

PART ONE: BASIC CONCEPTS

1. Elements of Medical Cytogenetics
2. Chromosome Analysis
3. The Origins and Consequences of Chromosome Pathology
4. Deriving and Using a Risk Figure

PART TWO: PARENT OR CHILD WITH A CHROMOSOMAL ABNORMALITY

5. Autosomal Reciprocal Translocations
6. Sex Chromosome Translocations
7. Robertsonian Translocations
8. Insertions
9. Inversions
10. Complex Chromosomal Rearrangements
11. Autosomal Ring Chromosomes
12. Centromere Fissions, Complementary Isochromosomes, Telomeric Fusions, Balancing Supernumerary Chromosomes, Neocentromeres, Jumping Translocations, and Chromothripsis
13. Down Syndrome, Other Full Aneuploidies, Polyploidy, and the Influence of Parental Age
14. Autosomal Structural Rearrangements: Deletions and Duplications
15. Sex Chromosome Aneuploidy and Structural Rearrangement
16. Chromosome Instability Syndromes

PART THREE: CHROMOSOME VARIANTS

17. Normal Chromosomal Variation
18. Copy Number Variants

PART FOUR: DISORDERS ASSOCIATED WITH ABERRANT GENOMIC IMPRINTING

19. Uniparental Disomy and Disorders of Imprinting

PART FIVE: REPRODUCTIVE CYTOGENETICS

20. Reproductive Failure
21. Prenatal Testing Procedures
22. Chromosome Abnormalities Detected at Prenatal Diagnosis
23. Preimplantation Genetic Diagnosis

PART SIX: DISORDERS OF SEX DEVELOPMENT

24. Chromosomal Disorders of Sex Development

PART SEVEN: PHENOTYPES

25. Chromosomal Phenotypes

PART EIGHT: NOXIOUS AGENTS

26. Gonadal Cytogenetic Damage from Exposure to Extrinsic Agents

PART NINE: ETHICS

27. Ethical Issues

APPENDIXES

A. Ideograms of Human Chromosomes

B. Cytogenetic Nomenclature

C. Penetrance Data for Certain Copy Number Variants