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Autosomal Recessive InheritanceX-Linked Inheritance; Using Pedigrees to Study Human Genetic Disorders; from the human genetics files Calculating Mendelian Frequencies; Detection and Estimation of Genetic Linkage in Humans; The Logarithm of the Likelihood Ratio Method of Linkage Analysis: LOD Score; Key Terms; Summary; References; Review Questions; chapter 4 The Molecular Biology of the Gene; Properties of Genetic Material; Structure of DNA; DNA Replication; Decoding Genetic Information: RNA and Protein; Translation; Regulation of mRNA Transcription
from the human genetics files Hemoglobinopathies and Thalassemias: An Abundance of MutationsNucleotide Sequence Alteration: Mutation; Mutations of Structural Genes; Nomenclature for Mutations; Dominant Mutations and Genetic Disorders; Key Terms; Summary; References; Review Questions; chapter 5 Recombinant DNA Technology; Restriction Endonucleases; Cloning Vectors; Plasmid Cloning Vector pUC19; Screening DNA Constructs by DNA Hybridization; In Situ Hybridization; Chemical Synthesis of DNA; Sequencing DNA; Polymerase Chain Reaction; Human-Rodent Somatic Cell Hybrids; Human DNA Libraries Genomic LibrariesChromosome DNA Libraries; from the human genetics files Multicolor Karyotyping: Coloring Chromosomes; Region-Specific Chromosome Libraries; Constructing a cDNA Library; Key Terms; Summary; References; Review Questions; chapter 6 Genetic and Physical Mapping of the Human Genome; Genetic Mapping of Human Chromosomes; Genetic Polymorphism; Restriction Fragment Length Polymorphism; Short Tandem Repeat Polymorphism; Mapping of a Genetic Disease Locus to a Chromosome Location; Multilocus Mapping of Human Chromosomes; Inserting a Disease Gene into a Linkage Map; Homozygosity Mapping
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Sommario/riassunto

An Introduction to Human Molecular GeneticsSecond EditionJack J. PasternakThe Second Edition of this internationally acclaimed text expands its coverage of the molecular genetics of inherited human diseases with the latest research findings and discoveries. Using a unique, systems-based approach, the text offers readers a thorough explanation of the gene discovery process and how defective genes are linked to inherited disease states in major organ and tissue systems. All the latest developments in functional genomics, proteomics, and microarray technology have been thoroughly inco