

1. Record Nr.	UNINA9910813517203321
Autore	Pasternak Jack J
Titolo	An introduction to human molecular genetics : mechanisms of inherited diseases // Jack J. Pasternak
Pubbl/distr/stampa	Hoboken, N.J. : Wiley-Liss, c2005
ISBN	9786610275861 9781280275869 1280275863 9780470357866 047035786X 9780471719175 047171917X 9780471719182 0471719188
Edizione	[2nd ed.]
Descrizione fisica	1 online resource (660 p.)
Disciplina	616/042
Soggetti	Human molecular genetics Genetic disorders
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di contenuto	AN INTRODUCTION TO Human Molecular Genetics; Contents; Preface; Preface to the First Edition; chapter 1 Understanding Human Disease; Human Genetic Disease; Human Genetics from 1900 to 1957; Eugenics: Genetics Misinterpreted; The Molecularization of Genetics; Genes and Phenotypes; from the human genetics files OMIM: An Important Online Source of Information About Human Genetic Disorders; Key Terms; Summary; References; Review Questions; chapter 2 The Genetic System: Chromosomes; Human Chromosomes; Maintaining the Chromosome Number; Cell Division Cycle: The Mitotic Process; The Meiotic Process Characterizing Human ChromosomesChromosome Abnormalities; Whole Chromosome Changes: Aneuploidy; Chromosome Structural Changes; from the human genetics files Determining the Phases of the Cell Cycle; Key Terms; Summary; References; Review Questions; chapter 3 The Genetic System: Mendel's Laws of Inheritance and Genetic

Linkage; Dominance, Recessiveness, and Segregation; Independent Assortment; Genetic Linkage; Constructing Genetic Maps; Three-Point Cross; Chi-Square Distribution: Testing for Significance; Multiple Alleles; Human Genetics; Autosomal Dominant Inheritance; Autosomal Recessive Inheritance; X-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 Mutations; Nucleotide 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 Libraries; Chromosome 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; Linkage Disequilibrium Mapping

---

#### Sommario/riassunto

An Introduction to Human Molecular Genetics, Second Edition  
Jack J. Pasternak  
The 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 incorporated.

---