



# An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases

By Jack J. Pasternak

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## An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases By Jack J. Pasternak

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 into the text.

The first part of the text introduces readers to the fundamentals of cytogenetics and Mendelian genetics. Next, techniques and strategies for gene manipulation, mapping, and isolation are examined. Readers will particularly appreciate the text's exceptionally thorough and clear explanation of genetic mapping. The final part features unique coverage of the molecular genetics of distinct biological systems, covering muscle, neurological, eye, cancer, and mitochondrial disorders. Throughout the text, helpful figures and diagrams illustrate and clarify complex material.

Readers familiar with the first edition will recognize the text's same lucid and engaging style, and will find a wealth of new and expanded material that brings them fully up to date with a current understanding of the field, including:

- \* New chapters on complex genetic disorders, genomic imprinting, and human population genetics
- \* Expanded and fully revised section on clinical genetics, covering diagnostic testing, molecular screening, and various treatments

This text is targeted at upper-level undergraduate students, graduate students, and medical students. It is also an excellent reference for researchers and physicians who need a clinically relevant reference for the molecular genetics of inherited human diseases.

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### Editorial Review

#### Review

"With over 600 pages, this book provides students and professionals in life sciences, pharmacy and biochemistry with all they need to know about molecular biotechnology." (NewsRx, 30 September 2011)

"...very readable...I would recommend this text to students studying human molecular genetics as well as those involved in their instruction." (American Journal of Medical Genetics, June 15, 2006)

"...tailor-made to address...[the] need for a textbook...more focused on the nature and characterization of common, classical genetic diseases...an excellent book that will be applicable to a wide range of students." (*The Quarterly Review of Biology*, June 2006)

"An excellent course book or text to read for updating the established investigator." (*American Journal of Therapeutics*, March/April 2006)

"This book would make a worthy addition to any geneticist's library." (*Clinical Chemistry*, February 2006)

"The abundance of clearly presented information renders this textbook a chef d'oeuvre." (*New England Journal of Medicine*, January 5, 2006)

#### From the Publisher

Engaging writing style and numerous illustrations make complex concepts accessible to all students. Chapter outlines, lists of key terms, extensive summaries of key concepts, and review questions are provided for each chapter.

Up-to-the-minute research and numerous references cited throughout.

"From the Human Genetics Files" sections in each chapter present historical and innovative applications and contributions of Human Molecular Genetics (like the use of DNA analysis to identify the remains of the Russian Imperial family).

Full-color insert illustrates the latest cytogenetic imaging techniques (FISH, SKY).

#### From the Back Cover

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## **Users Review**

### **From reader reviews:**

#### **Deborah Allen:**

This An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases book is absolutely not ordinary book, you have it then the world is in your hands. The benefit you have by reading this book is definitely information inside this reserve incredible fresh, you will get information which is getting deeper a person read a lot of information you will get. This particular An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases without we realize teach the one who examining it become critical in imagining and analyzing. Don't possibly be worry An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases can bring once you are and not make your handbag space or bookshelves' come to be full because you can have it inside your lovely laptop even telephone. This An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases having good arrangement in word as well as layout, so you will not feel uninterested in reading.

#### **Timothy Bennington:**

The experience that you get from An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases may be the more deep you rooting the information that hide within the words the more you get thinking about reading it. It does not mean that this book is hard to know but An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases giving you enjoyment feeling of reading. The article writer conveys their point in specific way that can be understood through anyone who read this because the author of this publication is well-known enough. This kind of book also makes your personal vocabulary increase well. Making it easy to understand then can go along, both in printed or e-book style are available. We suggest you for having this kind of An Introduction to Human Molecular Genetics: Mechanisms of Inherited Diseases instantly.

#### **Ariane Swanson:**

Exactly why? Because this An Introduction to Human Molecular Genetics: Mechanisms of Inherited

Diseases is an unordinary book that the inside of the e-book waiting for you to snap the idea but latter it will jolt you with the secret the item inside. Reading this book close to it was fantastic author who also write the book in such wonderful way makes the content within easier to understand, entertaining approach but still convey the meaning completely. So , it is good for you because of not hesitating having this anymore or you going to regret it. This book will give you a lot of benefits than the other book get such as help improving your ability and your critical thinking means. So , still want to hold off having that book? If I ended up you I will go to the e-book store hurriedly.

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