

# Chapter 14 The Human Genome

## Section Review 1 Answer Key

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*Chapter 14  
The Human  
Genome  
Section Review  
1 Answer Key*      2019-12-15

### **ARTHUR BRENDEN**

Advances in Animal Genomics Academic Press  
Advances in Animal Genomics provides an outstanding collection of integrated strategies involving traditional and modern - omics (structural, functional, comparative and epigenomics) approaches and genomics-assisted breeding methods which animal biotechnologists can utilize to dissect and decode the molecular and gene regulatory networks involved in the complex quantitative yield and stress tolerance traits in livestock. Written by international experts on

animal genomics, this book explores the recent advances in high-throughput, next-generation whole genome and transcriptome sequencing, array-based genotyping, and modern bioinformatics approaches which have enabled to produce huge genomic and transcriptomic resources globally on a genome-wide scale. This book is an important resource for researchers, students, educators and professionals in agriculture, veterinary and biotechnology sciences that enables them to solve problems regarding sustainable development with the help of current innovative biotechnologies. Integrates basic and advanced concepts of

animal biotechnology and presents future developments Describes current high-throughput next-generation whole genome and transcriptome sequencing, array-based genotyping, and modern bioinformatics approaches for sustainable livestock production Illustrates integrated strategies to dissect and decode the molecular and gene regulatory networks involved in complex quantitative yield and stress tolerance traits in livestock Ensures readers will gain a strong grasp of biotechnology for sustainable livestock production with its well-illustrated discussion Science, Ethics, and Governance Elsevier Health Sciences

This text clearly explains the key principles needed by medical and health sciences students, from the basis of molecular genetics, to clinical applications used in the treatment of both rare and common conditions. A newly expanded Part 1, *Basic Principles of Human Genetics*, focuses on introducing the reader to key concepts such as Mendelian principles, DNA replication and gene expression. Part 2, *Genetics and Genomics in Medical Practice*, uses case scenarios to help you engage with current genetic practice. Now featuring full-color diagrams, *Human Genetics and Genomics* has been rigorously updated to reflect today's genetics teaching, and includes updated discussion of genetic risk assessment, single gene disorders and therapeutics. Key learning features include: Clinical snapshots to help relate science to practice 'Hot topics' boxes that focus on the latest developments in testing, assessment and treatment 'Ethical issues' boxes to prompt further thought and discussion on the implications of genetic developments 'Sources of information' boxes to

assist with the practicalities of clinical research and information provision. *What Genes Do, How They Malfunction, and Ways to Repair Damage* Academic Press Significant advances in our knowledge of genetics were made during the twentieth century but in the most recent decades, genetic research has dramatically increased its impact throughout society. Genetic issues are now playing a large role in health and public policy, and new knowledge in this field will continue to have significant implications for individuals and society. Written for the non-majors human genetics course, *Human Genetics, 3E* will increase the genetics knowledge of students who are learning about human genetics for the first time. This thorough revision of the best-selling *Human Genome, 2E* includes entirely new chapters on forensics, stem cell biology, bioinformatics, and societal/ethical issues associated with the field. New special features boxes make connections between human genetics and human health and disease. Carefully crafted pedagogy includes

chapter-opening case studies that set the stage for each chapter; concept statements interspersed throughout the chapter that keep first-time students focused on key concepts; and end-of-chapter questions and critical thinking activities. This new edition will contribute to creating a genetically literate student population that understands basic biological research, understands elements of the personal and health implications of genetics, and participates effectively in public policy issues involving genetic information. Includes topical material on forensics, disease studies, and the human genome project to engage non-specialist students Full, 4-color illustration program enhances and reinforces key concepts and themes Uniform organization of chapters includes interest boxes that focus on human health and disease, chapter-opening case studies, and concept statements to engage non-specialist readers **The -Omics Revolution and Its Impact on Human Reproductive Medicine** CRC Press Concepts of Biology is designed for the single-semester introduction to

biology course for non-science majors, which for many students is their only college-level science course. As such, this course represents an important opportunity for students to develop the necessary knowledge, tools, and skills to make informed decisions as they continue with their lives. Rather than being mired down with facts and vocabulary, the typical non-science major student needs information presented in a way that is easy to read and understand. Even more importantly, the content should be meaningful. Students do much better when they understand why biology is relevant to their everyday lives. For these reasons, Concepts of Biology is grounded on an evolutionary basis and includes exciting features that highlight careers in the biological sciences and everyday applications of the concepts at hand. We also strive to show the interconnectedness of topics within this extremely broad discipline. In order to meet the needs of today's instructors and students, we maintain the overall organization and coverage found in most syllabi for this course. A

strength of Concepts of Biology is that instructors can customize the book, adapting it to the approach that works best in their classroom. Concepts of Biology also includes an innovative art program that incorporates critical thinking and clicker questions to help students understand--and apply--key concepts. [Genome Engineering via CRISPR-Cas9 System](#) Human Genome Epidemiology A Scientific Foundation for Using Genetic Information to Improve Health and Prevent Disease The #1 NEW YORK TIMES Bestseller The basis for the PBS Ken Burns Documentary The Gene: An Intimate History From the Pulitzer Prize-winning author of The Emperor of All Maladies—a fascinating history of the gene and “a magisterial account of how human minds have laboriously, ingeniously picked apart what makes us tick” (Elle). “Sid Mukherjee has the uncanny ability to bring together science, history, and the future in a way that is understandable and riveting, guiding us through both time and the mystery of life itself.” —Ken Burns “Dr. Siddhartha Mukherjee

dazzled readers with his Pulitzer Prize-winning *The Emperor of All Maladies* in 2010. That achievement was evidently just a warm-up for his virtuoso performance in *The Gene: An Intimate History*, in which he braids science, history, and memoir into an epic with all the range and biblical thunder of *Paradise Lost*” (The New York Times). In this biography Mukherjee brings to life the quest to understand human heredity and its surprising influence on our lives, personalities, identities, fates, and choices. “Mukherjee expresses abstract intellectual ideas through emotional stories...[and] swaddles his medical rigor with rhapsodic tenderness, surprising vulnerability, and occasional flashes of pure poetry” (The Washington Post). Throughout, the story of Mukherjee’s own family—with its tragic and bewildering history of mental illness—reminds us of the questions that hang over our ability to translate the science of genetics from the laboratory to the real world. In riveting and dramatic prose, he describes the centuries of research and experimentation—from

Aristotle and Pythagoras to Mendel and Darwin, from Boveri and Morgan to Crick, Watson and Franklin, all the way through the revolutionary twenty-first century innovators who mapped the human genome. “A fascinating and often sobering history of how humans came to understand the roles of genes in making us who we are—and what our manipulation of those genes might mean for our future” (Milwaukee Journal-Sentinel), *The Gene* is the revelatory and magisterial history of a scientific idea coming to life, the most crucial science of our time, intimately explained by a master. “The Gene is a book we all should read” (USA TODAY).

*Diagnostic Molecular Biology* Oxford University Press

Every new copy includes access to the student companion website Updated throughout to reflect the latest discoveries in this fast-paced field, *Essential Genetics: A Genomics Perspective*, Sixth Edition, provides an accessible, student-friendly introduction to modern genetics. Designed for the shorter, less comprehensive course,

the Sixth Edition presents carefully chosen topics that provide a solid foundation to the basic understanding of gene mutation, expression, and regulation. It goes on to discuss the development and progression of genetics as a field of study within a societal and historical context. The Sixth Edition includes new learning objectives within each chapter which helps students identify what they should know as a result of their studying and highlights the skills they should acquire through various practice problems. What's new in the Sixth Edition? Chapter 1 includes a new section on the origin of life Chapter 2 includes a revised discussion of the complementation test and how it is used to determine whether two mutations have defects in the same gene Chapter 3 incorporates new data showing that the folding of interphase chromatin into chromosome territories has the form of a fractal globule. It also includes a new section on progenitor cells and embryonic stem cells Chapter 4 includes a new section discussing how copy-number variation in human amylase evolved in response to increased

dietary starch as well as the latest on hotspots of recombination Chapter 5 is updated with the latest information on hazards of polycarbonate food containers. It also includes a new section on the genetics of schizophrenia and autism spectrum disorder Chapter 6 includes a revised section on restriction mapping and also discusses the newest massively parallel DNA sequencing technologies that can yield the equivalent of 200 human genomes' worth of DNA sequence in a single sequencing run Chapter 7 has been updated with a shortened and streamlined discussion of recombination in bacteriophage Chapter 8 includes new discoveries concerning the mechanisms of intrinsic transcriptional termination as well as rho-dependent termination Chapter 9 is updated with a new section on stochastic effects on gene expression and an expanded discussion of the lactose operon. There is also a revised discussion of galactose gene regulation in yeast, as well as new sections on lon noncoding RNAs Chapter 10 includes new

sections on ancient DNA sequences of the Neandertal and Denisovan genomes Chapter 11 examines master control genes in development Chapter 12 includes a new section on the repair of double-stranded breaks in DNA by nonhomologous end joining or template-directed gap repair Chapter 13 has been extensively revised with the latest data on cancer. Chapter 14 includes a new section on the detection of natural selection, as well as a new section on conservation genetics Key Features of Essential Genetics, Sixth Edition: New Learning Objectives within each *How Genotype and Gene Interactions Affect Behavior* John Wiley & Sons

This comprehensive account of the human herpesviruses provides an encyclopedic overview of their basic virology and clinical manifestations. This group of viruses includes human simplex type 1 and 2, Epstein-Barr virus, Kaposi's Sarcoma-associated herpesvirus, cytomegalovirus, HHV6A, 6B and 7, and varicella-zoster virus. The viral diseases and cancers they cause are significant and often recurrent. Their prevalence in the

developed world accounts for a major burden of disease, and as a result there is a great deal of research into the pathophysiology of infection and immunobiology. Another important area covered within this volume concerns antiviral therapy and the development of vaccines. All these aspects are covered in depth, both scientifically and in terms of clinical guidelines for patient care. The text is illustrated generously throughout and is fully referenced to the latest research and developments. Issues of Ethics, Law, Regulation and Communication Academic Press

Recent advances in genomic and omics analysis have triggered a revolution affecting nearly every field of medicine, including reproductive medicine, obstetrics, gynecology, andrology, and infertility treatment. Reproductomics: The -Omics Revolution and Its Impact on Human Reproductive Medicine demonstrates how various omics technologies are already aiding fertility specialists and clinicians in characterizing patients, counseling couples

towards pregnancy success, informing embryo selection, and supporting many other positive outcomes. A diverse range of chapters from international experts examine the complex relationship between genomics, transcriptomics, proteomics, and metabolomics and their role in human reproduction, identifying molecular factors of clinical significance. With this book Editors Jaime Gosálvez and José A. Horcajadas have provided researchers and clinicians with a strong foundation for a new era of personalized reproductive medicine. Thoroughly discusses how genomics and other omics approaches aid clinicians in various areas of reproductive medicine Identifies specific genomic and molecular factors of translational value in treating infertility and analyzing patient data Features chapter contributions by leading international experts **Genomics I** Cambridge University Press RNA-based Regulation in Human Health and Disease offers an in-depth exploration of RNA mediated genome regulation at different

hierarchies. Beginning with multitude of canonical and non-canonical RNA populations, especially noncoding RNA in human physiology and evolution, further sections examine the various classes of RNAs (from small to large noncoding and extracellular RNAs), functional categories of RNA regulation (RNA-binding proteins, alternative splicing, RNA editing, antisense transcripts and RNA G-quadruplexes), dynamic aspects of RNA regulation modulating physiological homeostasis (aging), role of RNA beyond humans, tools and technologies for RNA research (wet lab and computational) and future prospects for RNA-based diagnostics and therapeutics. One of the core strengths of the book includes spectrum of disease-specific chapters from experts in the field highlighting RNA-based regulation in metabolic & neurodegenerative disorders, cancer, inflammatory disease, viral and bacterial infections. We hope the book helps researchers, students and clinicians appreciate the role of RNA-based regulation in genome regulation, aiding the development of useful

biomarkers for prognosis, diagnosis, and novel RNA-based therapeutics. Comprehensive information of non-canonical RNA-based genome regulation modulating human health and disease Defines RNA classes with special emphasis on unexplored world of noncoding RNA at different hierarchies Disease specific role of RNA - causal, prognostic, diagnostic and therapeutic Features contributions from leading experts in the field The Autobiography of a Species in 23 Chapters Academic Press Genes, Brain Function, and Behavior offers a concise description of the nervous system that processes sensory input and initiates motor movements. It reviews how behaviors are defined and measured, and how experts decide when a behavior is perturbed and in need of treatment. Behavioral disorders that are clearly related to a defect in a specific gene are reviewed, and the challenges of understanding complex traits such as intelligence, autism and schizophrenia that involve numerous genes and environmental factors are explored. New methods of altering genes

offer hope for treating or even preventing difficulties that arise in our genes. This book explains what genes are, what they do in the nervous system, and how this impacts both brain function and behavior. Presents essential background, facts, and terminology about genes, brain function, and behavior Builds clear explanations on this solid foundation while minimizing technical jargon Explores in depth several single-gene and chromosomal neurological disorders Derives lessons from these clear examples and highlights key lessons in boxes Examines the intricacies of complex traits that involve multiple genetic and environmental factors by applying lessons from simpler disorders Explains diagnosis and definition Includes a companion website with Powerpoint slides and images for each chapter for instructors and links to resources

**Bioinformatics for Geneticists** Academic Press Cytogenomics demonstrates that chromosomes are crucial in understanding the human genome and that new high-throughput



approaches are central to advancing cytogenetics in the 21st century. After an introduction to (molecular) cytogenetics, being the basic of all cytogenomic research, this book highlights the strengths and newfound advantages of cytogenomic research methods and technologies, enabling researchers to jump-start their own projects and more effectively gather and interpret chromosomal data. Methods discussed include banding and molecular cytogenetics, molecular combing, molecular karyotyping, next-generation sequencing, epigenetic study approaches, optical mapping/karyomapping, and CRISPR-cas9 applications for cytogenomics. The book's second half demonstrates recent applications of cytogenomic techniques, such as characterizing 3D chromosome structure across different tissue types and insights into multilayer organization of chromosomes, role of repetitive elements and noncoding RNAs in human genome, studies in topologically associated domains, interchromosomal interactions, and

chromoanagenesis. This book is an important reference source for researchers, students, basic and translational scientists, and clinicians in the areas of human genetics, genomics, reproductive medicine, gynecology, obstetrics, internal medicine, oncology, bioinformatics, medical genetics, and prenatal testing, as well as genetic counselors, clinical laboratory geneticists, bioethicists, and fertility specialists. Offers applied approaches empowering a new generation of cytogenomic research using a balanced combination of classical and advanced technologies Provides a framework for interpreting chromosome structure and how this affects the functioning of the genome in health and disease Features chapter contributions from international leaders in the field  
Using Genetic Information to Improve Health and Prevent Disease Academic Press  
 A unique exploration of the principles and methods underlying the Human Genome Project and modern molecular genetics and biotechnology-from

two top researchers In Genomics, Charles R. Cantor, former director of the Human Genome Project, and Cassandra L. Smith give the first integral overview of the strategies and technologies behind the Human Genome Project and the field of molecular genetics and biotechnology. Written with a range of readers in mind-from chemists and biologists to computer scientists and engineers-the book begins with a review of the basic properties of DNA and the chromosomes that package it in cells. The authors describe the three main techniques used in DNA analysis-hybridization, polymerase chain reaction, and electrophoresis-and present a complete exploration of DNA mapping in its many different forms. By explaining both the theoretical principles and practical foundations of modern molecular genetics to a wide audience, the book brings the scientific community closer to the ultimate goal of understanding the biological function of DNA. Genomics features:  
 Topical organization within chapters for easy reference A discussion of

the developing methods of sequencing, such as sequencing by hybridization (SBH) in which data is read through words instead of letters. Detailed explanations and critical evaluations of the many different types of DNA maps that can be generated— including cytogenetic and restriction maps as well as interspecies cell hybrids. Informed predictions for the future of DNA sequencing.

*Biology, Therapy, and Immunoprophylaxis*

CreateSpace

Get a quick, expert overview of the fast-changing field of perinatal genetics with this concise, practical resource. Drs. Mary Norton, Jeffrey A. Kuller, Lorraine Dugoff, and George Saade fully cover the clinically relevant topics that are key to providers who care for pregnant women and couples contemplating pregnancy. It's an ideal resource for Ob/Gyn physicians, maternal-fetal medicine specialists, and clinical geneticists, as well as midwives, nurse practitioners, and other obstetric providers. Provides a comprehensive review of basic principles of medical genetics and genetic counseling,

molecular genetics, cytogenetics, prenatal screening options, chromosomal microarray analysis, whole exome sequencing, prenatal ultrasound, diagnostic testing, and more. Contains a chapter on fetal treatment of genetic disorders. Consolidates today's available information and experience in this important area into one convenient resource.

**Human Genes and Genomes** National Academies Press

In the 1960's and 1970's, personality and mental illness were conceptualized in an intertwined psychodynamic model. Biological psychiatry for many un-weaved that model and took mental illness for psychiatry and left personality to psychology. This book brings personality back into biological psychiatry, not merely in the form of personality disorder but as part of a new intertwined molecular genetic model of personality and mental disorder. This is the beginning of a new conceptual paradigm!! This breakthrough volume marks the beginning of a new era, an era made possible by the

electrifying pace of discovery and innovation in the field of molecular genetics. In fact, several types of genome maps have already been completed, and today's experts confidently predict that we will have a smooth version of the sequencing of the human genome -- which contains some 3 billion base pairs. Such astounding progress helped fuel the development of this remarkable volume, the first ever to discuss the brand-new -- and often controversial -- field of molecular genetics and the human personality. Questioning, critical, and strong on methodological principles, this volume reflects the point of view of its 35 distinguished contributors -- all pioneers in this burgeoning field and themselves world-class theoreticians, empiricists, clinicians, developmentalists, and statisticians. For students of psychopathology and others bold enough to hold in abeyance their understandable misgivings about the conjunction of "molecular genetics" and "human personality," this work offers an authoritative and up-to-date introduction to the molecular genetics of



human personality. The book, with its wealth of facts, conjectures, hopes, and misgivings, begins with a preface by world-renowned researcher and author Irving Gottesman. The authors masterfully guide us through Chapter 1, principles and methods; Chapter 4, animal models for personality; and Chapter 11, human intelligence as a model for personality, laying the groundwork for our appreciation of the remaining empirical findings of human personality qua personality. Many chapters (6, 7, 9, 11, and 13) emphasize the neurodevelopmental and ontogenetic aspects of personality, with a major emphasis on the receptors and transporters for the neurotransmitters dopamine and serotonin. Though these neurotransmitters are a rational starting point now, the future undoubtedly will bring many other candidate genes that today cannot even be imagined, given our ignorance of the genes involved in the prenatal development of the central nervous system. Chapter 3 provides an integrative overview of the broad

autism phenotype, and as such will be of special interest to child psychiatrists. Chapters 5, 8, and 10 offer enlightening information on drug and alcohol abuse. Chapter 14 discusses variations in sexuality. Adding balance and mature perspectives on how all the chapters complement and sometimes challenge one another are Chapter 2, written by a major figure in the renaissance of the relevance to psychopathology of both genetics and personality; Chapters 15-17, informed critical appraisals citing concerns and cautions about premature applications of this information in the policy arena; and Chapter 18, a judicious contemplation by the editors themselves of this promising -- and, to some, alarming -- field. Clear and meticulously researched, this eminently satisfying work is written to introduce the subject to postgraduate students just beginning to develop their research skills, to interested psychiatric practitioners, and to informed laypersons with some scientific background. Human Herpesviruses  
John Wiley & Sons  
The human genome is a

linear sequence of roughly 3 billion bases and information regarding this genome is accumulating at an astonishing rate. Inspired by these advances, *The Human Genome in Health and Disease: A Story of Four Letters* explores the intimate link between sequence information and biological function. A range of sequence-based functional units of the genome are discussed and illustrated with inherited disorders and cancer. In addition, the book considers valuable medical applications related to human genome sequencing, such as gene therapy methods and the identification of causative mutations in rare genetic disorders. The primary audiences of the book are students of genetics, biology, medicine, molecular biology and bioinformatics. Richly illustrated with review questions provided for each chapter, the book helps students without previous studies of genetics and molecular biology. It may also be of benefit for advanced non-academics, which in the era of personal genomics, want to learn more about their genome. Key selling features: Molecular sequence perspective,

explaining the relationship between DNA sequence motifs and biological function Aids in understanding the functional impact of mutations and genetic variants Material presented at basic level, making it accessible to students without previous studies of genetics and molecular biology Richly illustrated with questions provided to each chapter

### **Fundamentals of Forensic DNA Typing**

Academic Press

Heritable human genome editing - making changes to the genetic material of eggs, sperm, or any cells that lead to their development, including the cells of early embryos, and establishing a pregnancy - raises not only scientific and medical considerations but also a host of ethical, moral, and societal issues. Human embryos whose genomes have been edited should not be used to create a pregnancy until it is established that precise genomic changes can be made reliably and without introducing undesired changes - criteria that have not yet been met, says Heritable Human Genome Editing. From an international commission of the U.S. National Academy of Medicine,

U.S. National Academy of Sciences, and the U.K.'s Royal Society, the report considers potential benefits, harms, and uncertainties associated with genome editing technologies and defines a translational pathway from rigorous preclinical research to initial clinical uses, should a country decide to permit such uses. The report specifies stringent preclinical and clinical requirements for establishing safety and efficacy, and for undertaking long-term monitoring of outcomes. Extensive national and international dialogue is needed before any country decides whether to permit clinical use of this technology, according to the report, which identifies essential elements of national and international scientific governance and oversight.

### **A Scientific Foundation for Using Genetic Information to Improve Health and Prevent Disease**

American Academic Press  
Scientific Frontiers in Developmental Toxicology and Risk Assessment reviews advances made during the last 10-15 years in fields such as developmental biology, molecular biology, and

genetics. It describes a novel approach for how these advances might be used in combination with existing methodologies to further the understanding of mechanisms of developmental toxicity, to improve the assessment of chemicals for their ability to cause developmental toxicity, and to improve risk assessment for developmental defects. For example, based on the recent advances, even the smallest, simplest laboratory animals such as the fruit fly, roundworm, and zebrafish might be able to serve as developmental toxicological models for human biological systems. Use of such organisms might allow for rapid and inexpensive testing of large numbers of chemicals for their potential to cause developmental toxicity; presently, there are little or no developmental toxicity data available for the majority of natural and manufactured chemicals in use. This new approach to developmental toxicology and risk assessment will require simultaneous research on several fronts by experts from multiple scientific disciplines, including developmental

toxicologists, developmental biologists, geneticists, epidemiologists, and biostatisticians. Academic Press

*Our Genes, Our Choices: How Genotype and Gene Interactions Affect Behavior* - First Prize winner of the 2013 BMA Medical Book Award for Basic and Clinical Sciences - explains how the complexity of human behavior, including concepts of free will, derives from a relatively small number of genes, which direct neurodevelopmental sequence. Are people free to make choices, or do genes determine behavior? Paradoxically, the answer to both questions is "yes," because of neurogenetic individuality, a new theory with profound implications. Author David Goldman uses judicial, political, medical, and ethical examples to illustrate that this lifelong process is guided by individual genotype, molecular and physiologic principles, as well as by randomness and environmental exposures, a combination of factors that we choose and do not choose. Written in an authoritative yet accessible style, the book

includes practical descriptions of the function of DNA, discusses the scientific and historical bases of genethics, and introduces topics of epigenetics and the predictive power of behavioral genetics. First Prize winner of the 2013 BMA Medical Book Award for Basic and Clinical Sciences Poses and resolves challenges to moral responsibility raised by modern genetics and neuroscience Analyzes the neurogenetic origins of human behavior and free will Written by one of the world's most influential neurogeneticists, founder of the Laboratory of Neurogenetics at the National Institutes of Health

*Nutrition and Genomics* Jones & Bartlett Learning

Ancestral DNA, Human Origins, and Migrations describes the genesis of humans in Africa and the subsequent story of how our species migrated to every corner of the globe. Different phases of this journey are presented in an integrative format with information from a number of disciplines, including population genetics, evolution, anthropology, archaeology, climatology, linguistics, art, music,

folklore and history. This unique approach weaves a story that has synergistic impact in the clarity and level of understanding that will appeal to those researching, studying, and interested in population genetics, evolutionary biology, human migrations, and the beginnings of our species. Integrates research and information from the fields of genetics, evolution, anthropology, archaeology, climatology, linguistics, art, music, folklore and history, among others Presents the content in an entertaining and synergistic style to facilitate a deep understanding of human population genetics Informs on the origins and recent evolution of our species in an approachable manner

*Pan-genomics: Applications, Challenges, and Future Prospects* John Wiley & Sons

A thought-provoking exploration of deleterious mutations in the human genome and their effects on human health and wellbeing Despite all of the elaborate mechanisms that a cell employs to handle its DNA with the utmost care, a

newborn human carries about 100 new mutations, originated in their parents, about 10 of which are deleterious. A mutation replacing just one of the more than three billion nucleotides in the human genome may lead to synthesis of a dysfunctional protein, and this can be inconsistent with life or cause a tragic disease. Several percent of even young people suffer from diseases that are caused, exclusively or primarily, by pre-existing and new mutations in their genomes, including both a wide variety of genetically simple Mendelian diseases and diverse complex diseases such as birth anomalies, diabetes, and schizophrenia. Milder, but still substantial, negative effects of mutations are even more pervasive. As of now, we possess no means of reducing the rate at which mutations

appear spontaneously. However, the recent flood of genomic data made possible by next-generation methods of DNA sequencing, enabled scientists to explore the impacts of deleterious mutations on humans with previously unattainable precision and begin to develop approaches to managing them. Written by a leading researcher in the field of evolutionary genetics, *Crumbling Genome* reviews the current state of knowledge about deleterious mutations and their effects on humans for those in the biological sciences and medicine, as well as for readers with only a general scientific literacy and an interest in human genetics. Provides an extensive introduction to the fundamentals of evolutionary genetics with an emphasis on mutation and selection. Discusses

the effects of pre-existing and new mutations on human genotypes and phenotypes. Provides a comprehensive review of the current state of knowledge in the field and considers crucial unsolved problems. Explores key ethical, scientific, and social issues likely to become relevant in the near future as the modification of human germline genotypes becomes technically feasible. *Crumbling Genome* is must-reading for students and professionals in human genetics, genomics, bioinformatics, evolutionary biology, and biological anthropology. It is certain to have great appeal among all those with an interest in the links between genetics and evolution and how they are likely to influence the future of human health, medicine, and society.