

Learning Objectives After this lesson, you will be able to identify disorders that are caused by chromosomal changes and explain the effects of these changes. You will also be able to identify disorders that are caused by single-gene mutations and explain the effects of these mutations. You will also be able to identify disorders that are caused by multifactorial inheritance and explain the effects of these mutations.

| Disorder | Genetic Cause | Effect |
|----------------------------|--|---|
| Down Syndrome (Trisomy 21) | Trisomy 21: Having three copies of chromosome 21 instead of the usual two. | Physical and mental retardation, characteristic facial features, and a higher risk of heart disease and leukemia. |
| Phenylketonuria (PKU) | Autosomal recessive mutation in the PAH gene, leading to a deficiency in the enzyme phenylalanine hydroxylase. | Intellectual disability and other health problems if not treated with a low-phenylalanine diet. |
| Sickle Cell Anemia | Autosomal recessive mutation in the HBB gene, leading to a change in the structure of hemoglobin. | Anemia, pain, and an increased risk of complications such as stroke and heart disease. |
| Tay-Sachs Disease | Autosomal recessive mutation in the HEXA gene, leading to a deficiency in the enzyme hexosaminidase A. | Progressive neurological damage, blindness, and death in early childhood. |

Lesson Summary

Chromosomal Disorders

Chromosomal disorders are the result of changes in the number or structure of chromosomes. These changes can be inherited or acquired. Some chromosomal disorders are caused by a change in the number of chromosomes (aneuploidy), while others are caused by a change in the structure of a chromosome (structural chromosomal disorders).

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Human Genetic Disorders Answer Key Chapter 13

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Human Genetic Disorders Answer Key Chapter 13:

Human Genes and Genomes Leon E. Rosenberg, Diane Drobnis Rosenberg, 2012-05-18 In the nearly 60 years since Watson and Crick proposed the double helical structure of DNA the molecule of heredity waves of discoveries have made genetics the most thrilling field in the sciences The study of genes and genomics today explores all aspects of the life with relevance in the lab in the doctor's office in the courtroom and even in social relationships In this helpful guidebook one of the most respected and accomplished human geneticists of our time communicates the importance of genes and genomics studies in all aspects of life With the use of core concepts and the integration of extensive references this book provides students and professionals alike with the most in depth view of the current state of the science and its relevance across disciplines Bridges the gap between basic human genetic understanding and one of the most promising avenues for advances in the diagnosis prevention and treatment of human disease Includes the latest information on diagnostic testing population screening predicting disease susceptibility pharmacogenomics and more Explores ethical legal regulatory and economic aspects of genomics in medicine Integrates historical classical genetics approach with the latest discoveries in structural and functional genomics

The Human Genome in Health and Disease Tore Samuelsson, 2019-02-07 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

50 Critical Cancer Answers Francisco Contreras, Daniel Kennedy, 2013-06-18 Cancer it's a menacing word and when we hear it from our own doctor it can be terrifying But there's hope In this practical comprehensive field manual from seasoned cancer fighters and renowned clinicians Francisco Contreras MD and Daniel Kennedy MC you will grab hold of 50 tangible tips plans and prescriptive measures for tackling cancer and finding renewed health 50 Critical Cancer Answers provides the essential information a person needs to create a personal action plan to deal with cancer effectively Each of the 50 short easy to digest chapters

includes a concise explanation of the most effective and sought after cancer treatment in the realms of traditional medicine natural medicine emotional support and spiritual care A succinct commentary is provided to help the reader understand potential benefits and if it is a realistic treatment option or not Each chapter includes An interview with an expert in the field of the chapter s topic A smartphone scannable QR code linking the reader to You Tube video containing author commentary on the relevant topic 5 tips from cancer survivors all 50 cancer survivors have received treatment from the authors Oasis of Hope cancer centers Don t let a cancer diagnosis define your circumstances Instead develop a plan to identify attack and beat cancer

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Human Biological Diversity Daniel E. Brown, 2015-11-17 This text is intended for the sophomore level course in human variation human biology taught in anthropology departments It may also serve as a supplementary text in introductory physical anthropology courses In addition to covering the standard topics for the course it features contemporary topics in human biology such as the Human Genome Project genetic engineering the effects of stress obesity and pollution

Burton's Microbiology for the Health Sciences, Enhanced Edition Paul G. Engelkirk, Janet Duben-Engelkirk, Robert C. Fader, 2020-05-07 Emphasizing the relevance of microbiology to a career in the health professions Burton's Microbiology for the Health Sciences provides the vital microbiology information you need to protect yourself and your patients from infectious diseases

Medical Genetics: First South Asia Edition - E-Book Lynn B. Jorde, John C. Carey, Michael J. Bamshad, 2017-01-20 Popular for its highly visual clinical approach Medical Genetics First South Asia Edition delivers an accessible yet thorough understanding of this active and fast changing field Key updates in this new edition cover the latest developments which are integrated with clinical practice to emphasize the central principles and how they apply to practice Photographs illustrations and tables along with boxes containing patient family vignettes demonstrate clinical relevance and enhance visual impact of the material for easier and more effective learning and retention Mini summaries study questions suggested reading and a detailed glossary supplement and reinforce what you learn from the text More than 230 photographs illustrations and tables along with patient family vignettes clarify difficult concepts and demonstrate clinical significance Clinical Commentary Boxes help demonstrate how the hard science of genetics has real applications to everyday patient problems and prepare you for problem based integrated courses

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