

**Chapter 14 The Human Genome****Making Karyotypes****Introduction**

Several human genetic disorders are caused by extra, missing, or damaged chromosomes. In order to study these disorders, cells from a person are grown with a chemical that stops cell division at the metaphase stage. During metaphase, a chromosome exists as two chromatids attached at the centromere.

The cells are stained to reveal banding patterns and placed on glass slides. The chromosomes are observed under the microscope, where they are counted, checked for abnormalities, and photographed. The photograph is then enlarged, and the images of the chromosomes are individually cut out. The chromosomes are identified and arranged in homologous pairs. The arrangement of homologous pairs is called a karyotype. In this investigation, you will use a sketch of chromosomes to make a karyotype. You will also examine the karyotype to determine the presence of any chromosomal abnormalities.

**Problem**

How can chromosomes be observed?

**Pre-Lab Discussion**

Read the entire investigation. Then work with a partner to answer the following questions.

1. What clues to the presence of certain genetic disorders can be seen in a karyotype?

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2. Why might a laboratory worker attempting to diagnose a genetic disorder prefer to work with photographs of chromosomes rather than the chromosomes themselves?

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3. Why would it be much more difficult to construct a karyotype of unstained chromosomes?

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4. Which pair of chromosomes can contain two very different chromosomes and still be considered normal? Explain your answer.

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5. How do autosomes differ from sex chromosomes?

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# **Making Karyotypes Chapter 14 The Human Genome**

**E. Albert Reece, MD, PhD, MBA, John C.  
Hobbins**



## **Making Karyotypes Chapter 14 The Human Genome:**

**Prentice Hall Miller Levine Biology Laboratory Manual a for Students Second Edition 2004** Kenneth Raymond Miller, Joseph S. Levine, Prentice-Hall Staff, 2003-02 Authors Kenneth Miller and Joseph Levine continue to set the standard for clear accessible writing and up to date content that engages student interest Prentice Hall Biology utilizes a student friendly approach that provides a powerful framework for connecting the key concepts a biology Students explore concepts through engaging narrative frequent use of analogies familiar examples and clear and instructional graphics Whether using the text alone or in tandem with exceptional ancillaries and technology teachers can meet the needs of every student at every learning level *Miller Levine Biology 1e Lab Manual a (Average Advanced) Student Edition 2002c* Prentice Hall Direct Education Staff, 2001-04 One program that ensures success for all students **Stem Cells in Human Reproduction** Carlos Simon, Antonio Pellicer, 2006-12-05 This book gives an overview of the revolutionary advances in stem cell science that may potentially impact human reproductive medicine The contents cover the production and regeneration of female and male germ cells trophoblasts and endometrium from human embryonic and adult stem cells New developments in hESC derivation that will impact clinic **Flow Cytogenetics**, 2012-12-02 This is the first book to be devoted entirely to the application and development of flow techniques in cytogenetics It provides comprehensive information on the use of flow cytometry and sorting for chromosome classification and purification Cytogenetics and molecular biologists will find this book an invaluable reference source Practical details for the preparation and analysis of chromosomes using flow cytometry Flow karyotyping for sensitive rapid analysis of chromosome normality and the detection of aberrant chromosomes Flow sorting as a source of chromosome specific DNA for gene mapping and recombinant DNA libraries Construction and current status of chromosome specific recombinant DNA libraries **Ssg- Human Biology 6E Student Study Guide** Chiras, 2008-02 Human Biology Sixth Edition provides students with a clear and concise introduction to the general concepts of mammalian biology and human structure and function With its unique focus on health and homeostasis Human Biology enhances students understanding of their own health needs and presents the scientific background necessary for students to think critically about biological information they encounter in the media The completely revised content and exceptional new art and photos provide students with a more user friendly text while excellent learning tools maximize comprehension of material **ICRF Handbook of Genome Analysis** N. S. Spurr, S. Bryant, Bryan Young, 2009-06-03 The combined power of genetic analysis and recombinant DNA technology to analyse entire genomes has moved biomedical research into a new and revolutionary phase The complete sequencing and mapping of the human genome as well as the genomes of other model organisms will be the basis for our future understanding of human disease and will allow us to answer fundamental questions about development and evolution T The new ICRF Handbook of Genome Analysis is the essential guide to the enormous range of techniques available to the researcher for both the genetic and physical mapping of the genome as well as the sequencing and analysis

of DNA It is both a protocol manual and a comprehensive information resource Written by international experts each chapter presents a state of the art review of a methodology Methods are fully described and evaluated their advantages and disadvantages discussed and their suitability for different investigations considered Step by step protocols including computer analyses are given for 123 essential experimental procedures Troubleshooting sections discuss possible reasons for failure and offer remedies The primary focus is on human genetics and the benefits of an understanding of the genome for the diagnosis and treatment of human disease The book also considers the current state of progress in the analysis of genomes of many model organisms including plants A major part of the work provides detail on Internet resources as well as basic data on human and other genomes including mapped disease genes and mouse knockouts Covers not only the human genome in relation to cancers and other human diseases but also the genomes of all important model organisms Contains 123 easy to follow protocols for essential experimental procedures Reviews a vast range of other information resources including journals and the Internet provides an invaluable listing of suppliers of laboratory materials Has been written by international experts from their own practical experience Is mandated by the Imperial Cancer Research Fund a leader in research in this field Has a sturdy spiral binding within a hardback case for ease of use in the lab

### **Genetic Disorders and the Fetus**

Aubrey Milunsky, Jeff M. Milunsky, 2021-07-06 Explore the latest edition of the definitive resource on prenatal genetic diagnosis In the newly revised eighth edition of Genetic Disorders and the Fetus authors and acclaimed medical doctors Aubrey and Jeff Milunsky deliver a thorough and comprehensive reference perfect for academicians students in post graduate specialization courses and working medical professionals This book incorporates the knowledge wisdom perspectives and recommendations from a renowned team of contributing authors drawing upon their extensive experience in prenatal genetic diagnosis to present the definitive reference work used routinely around the world In addition to fundamental information on established prenatal diagnosis and exhaustively referenced coverage of new techniques you ll find new chapters on preconception genetic counselling preimplantation genetic diagnosis advances in fetal imaging and gene therapy Genetic Disorders and the Fetus is authored by a global team of internationally recognized contributors all of whom are leading voices in the field The eighth edition also contains A thorough discussion of the public policy and ethics of embryo editing including mitochondrial replacement treatment and gene patents prenatal diagnosis and polygenic disease risk prediction An exploration of preimplantation genetic diagnosis pharmacogenetics and prenatal diagnosis and whole genome sequencing A treatment of genetic disorders and pharmacologic therapy including spinal muscular atrophy and fragile X syndrome A discussion of legal issues including the fetus as plaintiff and the increasing liability of physicians due to advances in genetics Perfect for obstetricians clinical geneticists molecular and biochemical geneticists and pediatricians Genetic Disorders and the Fetus will also earn a place in the libraries of neonatologists genetics counsellors ethicists radiologists and professionals working in public policy and health departments

The AGT Cytogenetics Laboratory Manual

Marilyn S. Arsham, Margaret J. Barch, Helen J. Lawce, 2017-04-24 Cytogenetics is the study of chromosome morphology structure pathology function and behavior The field has evolved to embrace molecular cytogenetic changes now termed cytogenomics Cytogeneticists utilize an assortment of procedures to investigate the full complement of chromosomes and or a targeted region within a specific chromosome in metaphase or interphase Tools include routine analysis of G banded chromosomes specialized stains that address specific chromosomal structures and molecular probes such as fluorescence in situ hybridization FISH and chromosome microarray analysis which employ a variety of methods to highlight a region as small as a single specific genetic sequence under investigation The AGT Cytogenetics Laboratory Manual Fourth Edition offers a comprehensive description of the diagnostic tests offered by the clinical laboratory and explains the science behind them One of the most valuable assets is its rich compilation of laboratory tested protocols currently being used in leading laboratories along with practical advice for nearly every area of interest to cytogeneticists In addition to covering essential topics that have been the backbone of cytogenetics for over 60 years such as the basic components of a cell use of a microscope human tissue processing for cytogenetic analysis prenatal constitutional and neoplastic laboratory safety and the mechanisms behind chromosome rearrangement and aneuploidy this edition introduces new and expanded chapters by experts in the field Some of these new topics include a unique collection of chromosome heteromorphisms clinical examples of genomic imprinting an example driven overview of chromosomal microarray mathematics specifically geared for the cytogeneticist usage of ISCN s cytogenetic language to describe chromosome changes tips for laboratory management examples of laboratory information systems a collection of internet and library resources and a special chapter on animal chromosomes for the research and zoo cytogeneticist The range of topics is thus broad yet comprehensive offering the student a resource that teaches the procedures performed in the cytogenetics laboratory environment and the laboratory professional with a peer reviewed reference that explores the basis of each of these procedures This makes it a useful resource for researchers clinicians and lab professionals as well as students in a university or medical school setting

**Biology** Eric Strauss, Marylin Lisowski, 2000 *Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics* Reed E. Pyeritz, Bruce R. Korf, Wayne W. Grody, 2021-11-02 Selected for Doody s Core Titles 2024 in Clinical Genetics Emery and Rimoin s Principles and Practice of Medical Genetics and Genomics Perinatal and Reproductive Genetics Seventh Edition includes the latest information on seminal topics such as prenatal diagnosis genome and exome sequencing public health genetics genetic counseling and management and treatment strategies in this growing field The book is ideal for medical students residents physicians and researchers involved in the care of patients with genetic conditions This comprehensive yet practical resource emphasizes theory and research fundamentals related to applications of medical genetics across the full spectrum of inherited disorders and applications to medicine more broadly Chapters from leading international researchers and clinicians focus on topics ranging from single gene testing to whole genome sequencing whole

exome sequencing gene therapy genome editing approaches FDA regulations on genomic testing and therapeutics and ethical aspects of employing genomic technologies Fully revised and up to date this new edition introduces genetic researchers students and healthcare professionals to genomic technologies testing and therapeutic applications Examines key topics and developing methods within genomic testing and therapeutics including single gene testing whole genome and whole exome sequencing gene therapy and genome editing variant Interpretation and classification and ethical aspects of applying genomic technologies Includes color images that support the identification concept illustration and method of processing Features contributions by leading international researchers and practitioners of medical genetics Provides a robust companion website that offers further teaching tools and links to outside resources and articles to stay up to date on the latest developments in the field Swaiman's Pediatric Neurology - E-Book Stephen Ashwal, Phillip L. Pearl, 2025-05-09 Selected for 2025 Doody's Core Titles in Pediatrics and with Essential Purchase designation in Neurology For fifty years experienced clinicians and physicians in training have relied on Swaiman's cornerstone text as their 1 source for authoritative guidance in pediatric neurology Swaiman's Pediatric Neurology Principles and Practice Seventh Edition continues this tradition of excellence under the expert editorial direction of Drs Stephen Ashwal and Phillip L Pearl along with a team of key leaders in the field who serve as associate and section editors in their areas of expertise Thorough revisions including new chapters new videos new editors and expanded content bring you up to date with this dynamic field Contains new sections on global child neurology and environment and brain development and a greatly expanded section on neurogenetics in addition to new chapters on autoimmune epilepsies immune mediated movement disorders and more Offers expanded online content including additional figures tables and text as well as new personal introductory videos by many chapter authors Covers new emerging or controversial topics such as COVID 19 teleneurology environment and brain development immune mediated disorders of the nervous system functional neurological disorders in children nonverbal learning disorders and the pharmacological and future genetic treatment of neurodevelopmental disabilities Provides authoritative coverage of perinatal acquired and congenital disorders neurodevelopmental disabilities extensive sections on pediatric epilepsy and movement disorders nonepileptiform paroxysmal disorders and disorders of sleep Features nearly 3 000 line drawings photographs tables and boxes that highlight the text clarify key concepts and make it easy to find information quickly **Genetics** Arthur P. Mange, Elaine Johansen Mange, 1980 *Genetics* Peter J. Russell, 1998 Containing updated information on molecular genetics Peter J Russell's text emphasises a problem solving approach that helps students to develop and apply their critical thinking and analysis skills **Biosocial Genetics** Gerald James Stine, 1977 *Medical Genetics at a Glance* Dorian J. Pritchard, Bruce R. Korf, 2013-07-08 Medical Genetics at a Glance covers the core scientific principles necessary for an understanding of medical genetics and its clinical applications while also considering the social implications of genetic disorders This third edition has been fully updated to include the latest

developments in the field covering the most common genetic anomalies their diagnosis and management in clear concise and revision friendly sections to complement any health science course Medical Genetics at a Glance now has a completely revised structure to make its content even more accessible Other features include Three new chapters on Gene Identification The Biology of Cancer and Genomic Approaches to Cancer A much extended treatment of Biochemical Genetics A completely revised chapter on The Cell Cycle explaining principles of biochemistry and genetics which are fundamental to understanding cancer causation Two new chapters on Cardiac Developmental Pathology An extended Case Studies section Providing a broad understanding of one of the most rapidly progressing topics in medicine Medical Genetics at a Glance is perfect for students of medicine molecular biology genetics and genetic counselling and is a previous winner of a BMA Award

Clinical Obstetrics E. Albert Reece, MD, PhD, MBA, John C. Hobbins, 2008-04-15 Highly Commended in the Obstetrics and Gynaecology category at the British Medical Association Book Awards 2008 The third edition of Clinical Obstetrics The Fetus and Mother is unique in that it gives in depth attention to the two patients fetus and mother giving special coverage to each patient Clinical Obstetrics The Fetus and Mother covers the biology pathology and clinical management of disorders affecting the fetus and the mother with illustrations highlighting essential diagnostic features and treatment procedures The book has been extensively revised and in some cases reorganized for clarity and flow This book is fully up to date with special chapters on first trimester prenatal screening 3 D and 4 D ultrasound medical and surgical fetal therapy foetal stem cell therapy prenatal diagnosis and fetal developmental biology benign tumors in pregnancy and extensive discussion of each of the main areas of maternal diseases The book has been made more succinct yet heavily annotated and illustrated giving greater clinical emphasis enabling fast decisions at the point of care all chapters now have key summary points making it easy to retrieve information In addition Clinical Obstetrics The Fetus and Mother includes a new searchable CD of the text including over 100 colour illustrations and a Q A book allowing you to test your learning Formerly known as Medicine of the Fetus and Mother and highly successful in two previous editions published by Lippincott

**Childhood Leukemias** Ching-Hon Pui, 2012-06-21 New insights into the molecular biology of childhood leukemias have stimulated numerous advances in diagnostic methods strategies for risk assessment and the development of novel therapy for genetic subtypes of the diseases Fully revised and updated this new edition of Childhood Leukemias provides the most comprehensive clinically oriented and authoritative reference dedicated to these diseases Beginning with an overview of history cell biology and pathology subsequent chapters review approaches in the evaluation and management of specific leukemias new therapeutic development and the unique pharmacodynamics and pharmacogenetics of individual patients New chapters include epigenetics of leukemias leukemias in patients with Down syndrome and leukemia in adolescents and young adults The final section covers the complications associated with the disease or its treatment and supportive care during and after treatment Authored by leading experts this is a must have for any physician or investigator who deals with

leukemias in childhood     The Molecular and Genetic Basis of Neurologic and Psychiatric Disease Roger N. Rosenberg, 2008  
Completely updated for its Fourth Edition this book is the most comprehensive current review of the molecular and genetic basis of neurologic and psychiatric diseases More than 120 leading experts provide a fresh new assessment of recent molecular genetic and genomic advances offer new insights into disease pathogenesis describe the newest available therapies and explore promising areas of therapeutic development This edition features an updated section on psychiatric disease and expanded updated chapters on human genomics gene therapy and ethical issues Six new chapters cover congenital myasthenic syndromes hereditary spastic paraplegia ion channel disorders the phakomatoses beta galactosidase deficiency and prion diseases A Neurologic Gene Map describes the chromosome locus of all the genetic diseases and their gene product where known The fully searchable online text will be available on a companion Website [www.rosenbergneuroandpsychdisease.com](http://www.rosenbergneuroandpsychdisease.com)     Instructor's Resource Manual for Starr and Taggart's Biology Larry G. Sellers, 1998

**Clinical Laboratory Immunology** Connie R. Mahon, Diane G. Tice, 2006 This unique resource is the first covering molecular diagnostic technology that is specifically geared to the needs of those in clinical laboratory science or medical technology This book covers molecular diagnostic technology and the multidisciplinary clinical applications of this technology Topics include immunology infectious and autoimmune diseases clinical applications of the flow of cytometry organ transplantation molecular methods and more Clinical Laboratory Science Medical Technology students



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