

Karyotypes

Pre-Lab Discussion

Several human genetic disorders are caused by additional, missing, or damaged chromosomes. One way of studying genetic disorders is to observe the chromosomes themselves. In order to do this, cells from a person are grown in a laboratory. After the cells have reproduced a few times, they are treated with a chemical that stops cell division at the metaphase stage. During metaphase, the chromosomes are at the best length for identification.

The cells are treated further, stained, and then 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 chromosomes are individually cut out. The chromosomes are identified and arranged in homologous pairs. Homologous chromosomes are identical, or matching, chromosomes. 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 genetic defects.

Part A: Analyzing a Karyotype

1. Observe the *normal* karyotype in Figure 1. Notice that the two sex chromosomes, pair numbers 23, do not look alike. They are different because this karyotype is of a male, and a male has an X and a Y chromosome.
2. Identify the centromere in each pair of chromosomes. The centromere is the area where each chromosome narrows.

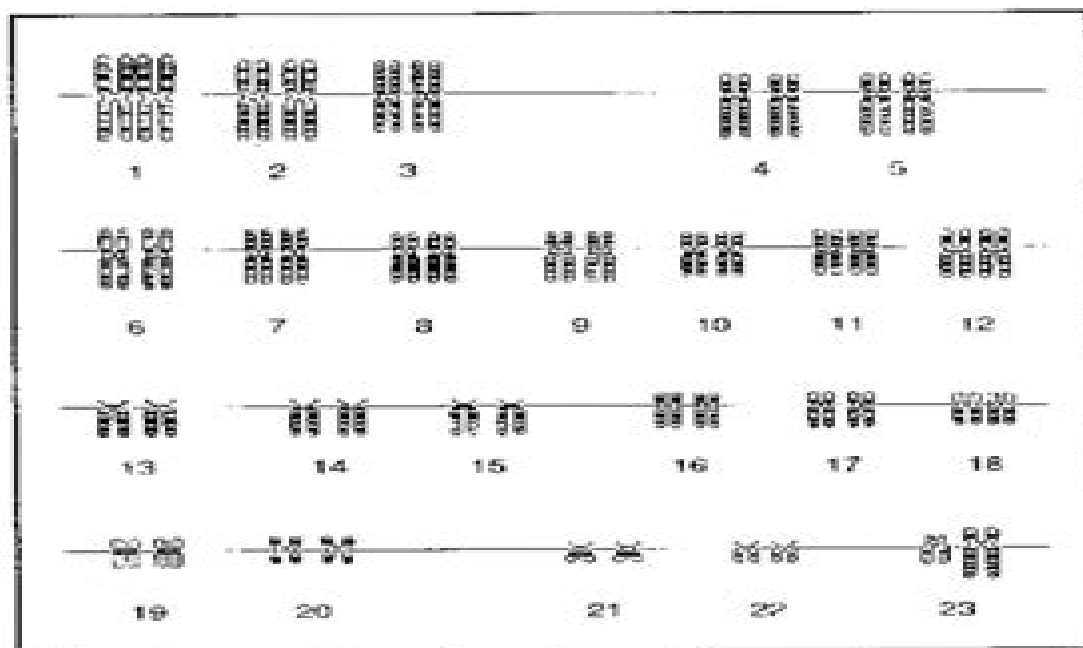


Figure 1

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**Douglas T. Carrell, C. Matthew
Peterson**



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Cases in Laboratory Genetics and Genomics (LGG) Practice Xia Li, 2023-06-05 Cases in Laboratory Genetics and Genomics LGG Practice instructs readers in the lab based diagnosis of genetic conditions including inborn and acquired disorders using cytogenetics and molecular genetics technologies This entirely case based book covers a wide range of genetic cases from prenatal to postnatal and oncology genetic disorders which lab professionals and geneticists encounter daily in the diagnostic field Each disorder discussed includes a section on clinical background clinical indication tests ordered laboratory tests performed test results results with interpretations future testing and recommendations and references The book will help lab professionals understand and navigate clinical cases using an integrative approach and thoroughly understand the methodologies and interpretations involved in high complexity genetic testing Presents clinical cases illustrating the complexity of the genetic abnormalities and successful diagnoses Discusses the technologies best suited to detect DNA mutations copy number variations and chromosome or RNA translocations Aids lab professionals in ensuring tests ordered are optimal for clinical indications Prepares trainees for the American Board of Medical Genetics and Genomics ABMGG LGG course and exam

Handbook of Autopsy Practice Jurgen Ludwig, 2002-07-18 The second edition of Handbook of Autopsy Practice appeared in 1979 under the title Current Methods of Autopsy Practice W B Saunders Company that edition was out of print in the early 1980s Now over 20 years later it appeared timely to thoroughly update the material in a third edition by adding what we have learned in the meantime and eliminating text that has become obsolete There is an acute need for a complete and readily accessible resource for autopsy work because few pathologists still specialize in autopsy practice and as a consequence expertise in autopsy technology and autopsy pathology has declined Our colleagues in the forensic field have remained the only large group of autopsy practitioners For most other pathologists the economic situation time constraints and the steadily decreasing autopsy rates have made a career in autopsy pathology unattractive This state of affairs is perpetuated by a lack of interest among many of our young colleagues partly because the teaching of autopsy pathology and autopsy techniques during most residencies is insufficient Numerous articles have been moaned this situation but the trend I fear is irreversible Still autopsies will be requested particularly in complex and difficult situations where the questions remaining after the death of the patient might challenge even experienced autopsy pathologists Under these circumstances this Handbook of Autopsy Practice should meet a particular need by providing the prosecutor with a source of information when it is most required in the autopsy room

Ordinary Genomes Karen-Sue Taussig, 2009-09-23 Explores the mutually constructive relationship between increasing scientific knowledge of human genetics and cultural identity through a case study of the development and reception of genomics in the Netherlands

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

Reproductive Endocrinology and Infertility Douglas T. Carrell,C. Matthew Peterson,2010-03-23 Management of the modern reproductive endocrinology and infertility clinic has become very complex In addition to the medical and scientific aspects it is crucial that the modern director be aware of of incongruent fields such as marketing accounting management and regulatory issues Reproductive Endocrinology and Infertility Integrating Modern Clinical and Laboratory Practice was developed to assist the practicing reproductive endocrinologist and or laboratory director by providing an overview of relevant scientific medical and management issues in a single volume Experts in all pertinent areas present concise practical evidence based summaries of relevant topics producing a key resource for physicians and scientists engaged in this exciting field of medicine As novel technologies continue to amplify Reproductive Endocrinology and Infertility Integrating Modern Clinical and Laboratory Practice offers insight into development and imparts extra confidence to practitioners in handling the many demands presented by their work

Creasy and Resnik's Maternal-Fetal Medicine: Principles and Practice E-Book Robert Resnik,Robert K. Creasy,Jay D. Iams,Charles J. Lockwood,Thomas Moore,Michael F Greene,2008-11-25 In your practice you require advanced knowledge of the obstetrical medical genetic and surgical complications of pregnancy and their effects on the mother and fetus With both basic science and clinical information six new chapters and an updated color design you need look no further than the 6th edition of this long time best seller Includes both basic science and clinical information to give you comprehensive knowledge of the biology of pregnancy Acts as an excellent resource for OB GYNs studying for their

Maternal Fetal Medicine boards and for practitioners who need quick access to practical information Provides an updated and focused reference list to keep you up to date on the standards of care in maternal fetal medicine today Keeps you current with a new section Disorders at the Maternal Fetal Interface and 6 new chapters Biology of Parturition Developmental Origins of Health and Disease Intrapartum Assessment of Fetal Health Pathogenesis of Pre term Birth Maternal and Fetal Infectious Disorders and Benign Gynecological Conditions of Pregnancy Features over 50% new authorship with increased focus on international perspectives Includes the following hot topics in Maternal Fetal Medicine o Biology of Parturition o Fetal Growth o Prenatal Genetic Screening and Diagnosis o Fetal Cardiac Malformations and Arrhythmias o Thyroid Disease and Pregnancy o Management of Depression and Psychoses during Pregnancy and the Puerperium Focuses on evidence based medicine the current best practice in MFM for diagnosing and treating high risk pregnancies Includes new illustrations and an updated color design

Chestnut's Obstetric Anesthesia: Principles and Practice E-Book David H. Chestnut, Cynthia A Wong, Lawrence C Tsen, Warwick D Ngan Kee, Yaakov Beilin, Jill Mhyre, 2014-02-28 From basic science to various anesthesia techniques to complications the meticulously updated fifth edition of Chestnut's Obstetric Anesthesia Principles and Practice covers all you need to know about obstetric anesthesia An editorial team of leading authorities presents the latest on anesthesia techniques for labor and delivery and medical disorders that occur during pregnancy New chapters and rewritten versions of key chapters cover topics such as psychiatric disorders in the pregnant patient neurologic disorders and critical care of obstetric patients It is an invaluable comprehensive reference textbook for specialists in obstetric anesthesiology and obstetricians as well as anesthesiology and obstetric residents This book also serves as a clear user friendly guide for both anesthesiologists and obstetricians who are in clinical practice Consult this title on your favorite e reader conduct rapid searches and adjust font sizes for optimal readability Get all the accuracy expertise and dependability you could ask for from the most important names in the fields of obstetric anesthesia and maternal fetal medicine Master the current best practices you need to know for treating the fetus and the mother as separate patients each with distinct needs Search and retain difficult concepts easily with the help of key point summaries in each chapter Stay current on the latest advancements and developments with sweeping updates and new chapters on topics such as patient safety and team approach transthoracic echocardiography and noninvasive measurement of cardiac output in obstetric patients psychiatric disorders during pregnancy neurologic injuries and more Prevent and plan for potential complications associated with the advancing age of pregnant women An extensive state of the art discussion of critical care of obstetric patients equips you to address any special considerations for this increasing segment Know exactly how to proceed An abundance of tables and boxes illustrate the step by step management of a full range of clinical scenarios Choose the best drugs available while adhering to the most recent guidelines for obstetric anesthesia

Handbook of Autopsy Practice Brenda L.

Waters, 2010-05-19 Handbook of Autopsy Practice Fourth Edition is divided into three parts Part I contains six new chapters

in which the reader will find an assortment of tools that will increase the value of the autopsy Included in the section are valuable resources and tools such as a sample next of kin letter a quality assurance worksheet new discourse on the dissection procedure which is accompanied by a worksheet and template for the gross description There is also a new detailed discussion of the safe handling of sharps complete with photographs and the reader will also find the updated requirements of the Eye Bank of America and the U S Food and Drug Administration for ocular tissue transplantation Part II has been updated with new diseases and recent references added Practicing autopsy pathologists residents and students are invited to review this alphabetical listing of disorders before each autopsy in order to re acquaint themselves with what they might encounter Part III provides a series of tables providing organ weights and body measurements for fetuses children and adults Handbook of Autopsy Practice Fourth Edition is an essential resource for clinicians pathologists residents and students who strive to hone their trade and increase the value of the autopsies they perform **Medical Cytogenetics** Hon Fong L. Mark, 2000-04-11 The only monograph on cytogenetics for the pathologist this up to the minute reference text contains the most up to date research findings on many important topics in medical genetics notably FISH fluorescent in situ hybridation based molecular cytogenetic technologies and spectral karyotyping An excellent resource for cytogeneticists prepar

Equine Reproductive Procedures John Dascanio, Patrick McCue, 2021-02-17 Dieses vollst ndig aktualisierte Referenzwerk erl utert die Techniken der Theriogenologie bei Pferden Die 2 Auflage von Equine Reproductive Procedures ist ein vollst ndig aktualisiertes Praktikerbuch f r Pferdetier rzte Studenten der Veterin rmedizin und Fachtier rzte f r Theriogenologie Es beschreibt Schritt f r Schritt die Verfahren der Theriogenologie bei Pferden und vermittelt die notwendigen klinischen Kenntnisse Die Themen behandeln das Reproduktionsmanagement bei Pferden u a Diagnoseverfahren und therapeutische Prozeduren bei Stuten Hengsten und Fohlen Die verschiedenen Verfahren werden klar beschrieben und anhand von Fotos verdeutlicht Eine Liste der f r das jeweilige Verfahren notwendigen Instrumente und Hilfsmittel und ein Referenzverzeichnis runden die Publikation ab Enth lt Schritt f r Schritt Beschreibungen der verschiedenen Verfahren der Theriogenologie bei Pferden Pr sentiert als wertvolles Praktikerbuch 39 neue Verfahren die in der 1 Auflage nicht enthalten sind Verfahren die in der Erstauflage erl utert wurden wurden aktualisiert Studenten der Veterin rmedizin die sich mit der Reproduktion bei Pferden besch ftigen und Fachtier rzte f r Theriogenologie erhalten mit diesem Praxisratgeber alles Wissenswerte in einer Publikation

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