

Sex Chromosomes Aneuploidy

Understanding Genetics Genetic Alliance 2009 The purpose of this manual is to provide an educational genetics resource for individuals, families, and health professionals in the New York - Mid-Atlantic region and increase awareness of specialty care in genetics. The manual begins with a basic introduction to genetics concepts, followed by a description of the different types and applications of genetic tests. It also provides information about diagnosis of genetic disease, family history, newborn screening, and genetic counseling. Resources are included to assist in patient care, patient and professional education, and identification of specialty genetics services within the New York - Mid-Atlantic region. At the end of each section, a list of references is provided for additional information. Appendices can be copied for reference and offered to patients. These take-home resources are critical to helping both providers and patients understand some of the basic concepts and applications of genetics and genomics.

Living with Klinefelter Syndrome, Trisomy X, and 47, Xyy: A Guide for Families and Individuals Affected by X and Y Chromosome Variations Virginia Isaacs Cover Msw 2012-03 This comprehensive guide to X and Y chromosome aneuploidy is written in lay language for affected individuals and their families, providing an authoritative volume that explains X and Y chromosome variations in clear and accurate terms. These surprisingly common genetic conditions, affecting 1 in 500 individuals, include Klinefelter syndrome, Trisomy X and 47, XYY. This guide provides a lifespan approach to the three trisomy conditions, as well as their less common variations involving 48 and 49 chromosomes. Readers are provided clear explanations of the genetics involved, diagnosis and disclosure issues, development from infancy through early adulthood, potential health and fertility concerns, and educational and psychosocial considerations. The text is illustrated with actual quotations from those who live with the disorders, and provides not only descriptions of potential concerns, but also strategies for successfully addressing the challenges that may develop.

Sex and Cardiac Electrophysiology Marek Malik 2020-07-11 Sex and Cardiac Electrophysiology: Differences in Cardiac Electrical Disorders Between Men and Women is a comprehensive investigation into all aspects of sex differences in cardiac electrophysiology. As there are substantial differences between female and male patients in physiology, pathology triggering factors, disease progression, clinical approaches and treatment outcome, this book provides a comprehensive examination. In cardiology, the differences between women and men are more recognized, hence this title summarizes these important differences, providing the essential information needed for clinical specialists and researchers involved in the design and implementation of clinical studies. Explores topics ranging from the physiologic differences between women and men to the differences in clinical handling of arrhythmic disorders between female and male patients Provides sex differences in cardiac electrophysiology in separate chapters Covers the sex differences of cardiac electrical disorders, providing insights beyond cardiac metabolic syndrome, hypertension, atherogenesis and heart failure

Human Development and Disease Antonia Alisia Dominguez 2014 1 -- 2% of all human fetal conceptions result in Turner syndrome -- the complete or partial loss of the second sex chromosome (45, X karyotype). 90% of Turner cases terminate in spontaneous abortion during the first two trimesters; however, those who survive have a wide array of clinical characteristics. Infertility is very common in Turner syndrome women, making a link between the X chromosome and causes of infertility. Induced pluripotent stem cells (iPSCs) offer a means of obtaining insight on the genetic requirements of germ cell development. I have used iPSCs to characterize XIST, a non-coding RNA that initiates X chromosome inactivation (XCI), and shown that newly reprogrammed female iPSCs reflect the developmental state of the preimplantation embryo following embryonic genome activation. From the generation of iPSCs from four Turner syndrome, one premature ovarian failure and a control female, I demonstrated that two X

chromosomes are not necessary for reprogramming to pluripotency. Single cell analysis of genes that escape XCI within control, Turner syndrome and Triple X iPSCs revealed minimal expression differences when compared to a female human embryonic stem cell (hESC) line. Next, I investigated the ability of X chromosome aneuploidy and control iPSCs to form germ cells by differentiating iPSCs and analyzing single cells and populations throughout a differentiation protocol using BMP4/8 and Retinoic acid. An increase in germ cell associated genes was not observed, but rather a loss. Finally, xenotransplantation was used to assess germ cell formation across different X chromosome aneuploidy states. All lines were capable of forming early germ cells in vivo, independent of their X chromosome composition. These results demonstrate that two complete X chromosomes are not necessary for the formation of early germ cells, and X aneuploidy iPSCs are amenable for the study for germ cell formation.

Aneuploidy Vicki Dellarco 2012-12-06 The "Symposium on Aneuploidy: Etiology and Mechanisms" was held at the Carnegie Institution of Washington Auditorium from March 25-29, 1985. This Symposium developed as a consequence of the concern of the Environmental Protection Agency with the support of the National Institute of Environmental Health Sciences about human exposure to environmental agents that cause aneuploidy. The program was chosen to explore what is currently known about the underlying causes, the origins, and the extent of the problem of human aneuploidy, and whether exposure to environmental agents is associated with an increased incidence of aneuploidy in humans. Basic research findings in the area of mitosis and meiosis were presented and related to possible mechanisms of how aneuploidy may be produced spontaneously and chemically. A survey of data regarding the chemical induction of aneuploidy in experimental organisms was presented. Outstanding scientists from different fields were invited to cover a broad perspective of aneuploidy from the molecular aspects to the human situation. We hope that the publication of the proceedings will share the enthusiasm of the meeting and its scientific content. The topic of aneuploidy has received little attention and it is the purpose of this Volume to establish a scientific basis for assessing health risks posed by environmental exposures to aneuploidy-inducing chemicals. Vicki L. DeJlarco Peter E. Voytek Alexander Hollaender vii ACKNOWLEDGEMENT The Editors of the proceedings of the "Symposium on Aneuploidy" wish to acknowledge the support of Dr. Elizabeth L.

Human Interphase Chromosomes Yuri B. Yurov 2013-04-03 This title will focus on the study of human interphase chromosomes and its relation to health and disease. Orchestrated organization and human genome function in interphase nuclei at the chromosomal level have been repeatedly shown to play a significant role in a variety of basic biological processes involved in realization and inheritance of genetic information within and between species. Current biomedical sciences of post-genomic era refocus basic and applied studies of interphase nuclei genetics and genomics with special attention to interphase chromosome behavior in health and disease. Additionally, related processes are a target of studies elucidating the role of interphase chromosome behavior during development, chromosome/DNA replication, DNA repair etc. Studies of interphase nuclei have an appreciable impact on different areas of biomedical sciences such as cell biology, neurobiology, cancer research, developmental biology, epigenetics, cytogenetics, and medical genetics, as a whole. Moreover, development of innovative and emergent technologies to analyze interphase nuclei are closely associated with application of these techniques in clinical, diagnostic and research practice to solve reproductive problems (including infertility and spontaneous abortions), to investigate congenital malformations (including those produced by aneuploidy and other chromosome abnormalities); genetic diseases (including cardiac, immune, neurological and psychiatric diseases), and cancer. This title will serve as a source of new valuable information and promising ideas for a wide audience of professionals in biomedicine including researchers, scientists, and healthcare professionals in human genetics, cytogenetics, and developmental biology.

Modeling Human Aneuploidy Kara Elaine Koehler 1997

Exploring the Biological Contributions to Human Health Institute of Medicine 2001-07-02 It's obvious why only men develop prostate cancer and why

only women get ovarian cancer. But it is not obvious why women are more likely to recover language ability after a stroke than men or why women are more apt to develop autoimmune diseases such as lupus. Sex differences in health throughout the lifespan have been documented. Exploring the Biological Contributions to Human Health begins to snap the pieces of the puzzle into place so that this knowledge can be used to improve health for both sexes. From behavior and cognition to metabolism and response to chemicals and infectious organisms, this book explores the health impact of sex (being male or female, according to reproductive organs and chromosomes) and gender (one's sense of self as male or female in society). Exploring the Biological Contributions to Human Health discusses basic biochemical differences in the cells of males and females and health variability between the sexes from conception throughout life. The book identifies key research needs and opportunities and addresses barriers to research. Exploring the Biological Contributions to Human Health will be important to health policy makers, basic, applied, and clinical researchers, educators, providers, and journalists-while being very accessible to interested lay readers.

Human Chromosomes Edward Hugh Rawlinson Ford 1973

Prospective Studies on Children with Sex Chromosome Aneuploidy Shirley G. Ratcliffe 1986 Overview of behavioral development in individuals with neonatally identified X and Y aneup.

Sex Chromosomes and Micronuclei in Human Lymphocytes John Christopher Hando 1996

Clinical Cytogenetics, An Issue of Clinics in Laboratory Medicine - E-Book Caroline Astbury 2011-12-28 This issue of Clinics in Laboratory Medicine, Guest Edited by Caroline Astbury, PhD, FACMG, will focus on Cytogenetics, with topics including: Chronic lymphocytic leukemia; Acute lymphocytic leukemia; Acute myelogenous leukemia; Chronic myelogenous leukemia; Plasma cell myeloma; Lymphomas; Solid tumors; Myelodysplastic syndromes; SNP arrays in clinical practice; Prenatal arrays; FISH (including Paraffin-embedded (PET) FISH); New and old microdeletion and microduplication syndromes; Sex chromosome and sex chromosome abnormalities; Autosomal aneuploidy; Microarray-CGH interpretation and Genomic Integrity; Structural chromosome rearrangements and complex chromosome rearrangements; and UPD/imprinting.

Chromosome Abnormalities and Genetic Counseling R.J. MKinlay Gardner 2011-11-11 Advances in cytogenetics continue to crop up in wonderful ways, and we know exponentially more about chromosomes now than mere decades ago. Likewise, the necessary skills in offering genetic counseling continue to evolve. This new edition of Chromosome Abnormalities in Genetic Counseling offers a practical, up-to-date guide for the genetic counselor to marshal cytogenetic data and analysis clearly and effectively to families.

Heritable Human Genome Editing The Royal Society 2021-01-16 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.

Sex Chromosomes—Advances in Research and Application: 2013 Edition 2013-06-21 *Sex Chromosomes—Advances in Research and Application: 2013 Edition* is a ScholarlyBrief™ that delivers timely, authoritative, comprehensive, and specialized information about Y Chromosome in a concise format. The editors have built *Sex Chromosomes—Advances in Research and Application: 2013 Edition* on the vast information databases of ScholarlyNews.™ You can expect the information about Y Chromosome in this book to be deeper than what you can access anywhere else, as well as consistently reliable, authoritative, informed, and relevant. The content of *Sex Chromosomes—Advances in Research and Application: 2013 Edition* has been produced by the world's leading scientists, engineers, analysts, research institutions, and companies. All of the content is from peer-reviewed sources, and all of it is written, assembled, and edited by the editors at ScholarlyEditions™ and available exclusively from us. You now have a source you can cite with authority, confidence, and credibility. More information is available at <http://www.ScholarlyEditions.com/>.

Human Reproductive and Prenatal Genetics Peter C.K. Leung 2023-03-22 *Human Reproductive and Prenatal Genetics, Second Edition* provides application-driven coverage of key topics in human reproductive and prenatal genetics, including genetic control underlying the development of the reproductive tracts and gametogenesis, the genetics of fertilization and implantation, the genetic basis of female and male infertility, as well as genetic and epigenetic aspects of assisted reproduction. Also examined are the genetics and epigenetics of the placenta in normal and abnormal pregnancy, preimplantation genetic diagnosis and screening, and cutting-edge advances in noninvasive prenatal screening, prenatal genetic counseling, and bioethical and medicolegal aspects of relevance in the lab and clinic. This new edition has been fully revised to address new and evolving technologies in human reproductive genetics, with new chapters added on chromatin landscapes and sex determination, genetic alterations of placental development and preeclampsia, metabolism and inflammation in PCOS, pre-implantational genetic testing, maternal genetic disorders, bioethics, and future applications. Features chapter contributions from leading international scientists and clinicians Provides in-depth coverage of key topics in human reproductive and prenatal genetics, including genetic controls, fertilization, placental development, embryo implantation, in vitro culture of the human embryo for the study of post-implantation development, and more Identifies how researchers and clinicians can implement the latest genetic, epigenetic, and -omics-based approaches Includes all new chapters on evolving technologies and recent genetic discoveries of relevance to reproductive medicine

Genetic Disorders and the Fetus Aubrey Milunsky 2012-12-06 About 21 years ago prenatal diagnosis became part of the physician's diagnostic armamentarium against genetic defects. My first monograph in 1973 (*The Prenatal Diagnosis of Hereditary Disorders*) critically assessed early progress and enunciated basic principles in the systematic approach to prenatal genetic diagnosis. Six years later and under the current title, a subsequent volume provided the first major reference source on this subject. The present second (effectively third) edition, which was urged in view of the excellent reception of the two earlier volumes, reflects the remarkable growth of this new discipline and points to significant and exciting future developments. Notwithstanding these advances, the use of the new tools and techniques for the benefit of at-risk parents has taken many more years than most anticipated. Key factors have been the lack of teaching of human genetics in medical schools in the preceding decades and the difficulty of educating practicing physicians in a new scientific discipline. Even today the teaching of genetics in medical schools leaves much to be desired and this will further delay the introduction of newer genetic advances to the bedside.

Preventive Health Care for Children with Genetic Conditions Golder Wilson 2006-05-11 An invaluable, structured approach to the preventive care of children with congenital disorders.

Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling R.J. McKinlay Gardner 2018-02-06 Even as classic cytogenetics has given way to molecular karyotyping, and as new deletion and duplication syndromes are identified almost every day, the fundamental role of the

genetics clinic remains mostly unchanged. Genetic counselors and medical geneticists explain the "unexplainable," helping families understand why abnormalities occur and whether they're likely to occur again. *Chromosome Abnormalities and Genetic Counseling* is the genetics professional's definitive guide to navigating both chromosome disorders and the clinical questions of the families they impact. Combining a primer on these disorders with the most current approach to their best clinical approaches, this classic text is more than just a reference; it is a guide to how to think about these disorders, even as our technical understanding of them continues to evolve. Completely updated and still infused with the warmth and voice that have made it essential reading for professionals across medical genetics, this edition of *Chromosome Abnormalities and Genetic Counseling* represents a leap forward in clinical understanding and communication. It is, as ever, essential reading for the field.

Chromosome Abnormalities and Genetic Counseling R. J. McKinlay Gardner 2003-08-28 Chromosomal abnormalities can cause disability in children, and reproductive difficulty in parents. Many parents and couples seek genetic counseling in order to learn why they, or a relative, may have had a child with a particular collection of medical problems and/or intellectual disability. There may have been a history of multiple miscarriage, or infertility. They may want to know the outlook for a pregnancy, and what the risks might be. These and other questions concerning chromosome abnormalities are addressed in this standard text, which will be of interest to genetic counselors, medical geneticists, pediatricians and obstetricians, infertility specialists, and laboratory cytogeneticists. This third edition has been thoroughly updated, and is richly illustrated and fully referenced. New chapters have been written on preimplantation diagnosis and on reproductive risks due to environmental agents. The practical applications of recent advances in molecular cytogenetics are noted. The book will give counselors the information that will enable them to help concerned parents accommodate to their particular "chromosomal situation", and to determine what may be, for them, the best course of action.

Genetics: Questions and Problems John Kuspira 1973 Mitosis and meiosis. Lige (Chromosome) cycles of eukaryotes. Monohybrid inheritance. Dihybrid and multihybrid inheritance. Probability. Gene interaction. Lethal genes. Multiple alleles. Sex determination and sex differentiation. Sex linkage. Sex-influenced and sex-limited inheritance. Linkage and crossing-over. Chromosome mapping. Extranuclear inheritance and related phenomena. Recombination in bacteria. Recombination in viruses. Genotype, environment, and phenotype. Pleiotropism, penetrance, expressivity, and phenocopies. Euploidy: haploidy and polyploidy. Aneuploidy. Chromosome aberrations. Balanced lethal systems and *Oenothera* Cytogenetics. Gene mutation. Chemical nature and structure of genes and chromosomes. The gene: genetics of gross and fine structure and interallelic. Biochemical genetics. Protein synthesis. Coding, collinearity, and suppressors. Development and regulation. Inbreeding, outbreeding, and heterosis. Population genetics. The genetics of race and species formation.

The Y Chromosome: Clinical aspects of Y chromosome abnormalities 1985

An Impaired DNA Damage Response Alters Neural Aneuploidy McConnell Michael James 2004

Chromosome Segregation and Meiotic Defects in Carriers of Chromosomal Abnormalities 2006 Male carriers of chromosomal abnormalities (CA) are more frequent in the infertile population. These men have higher levels of sperm aneuploidy due to the aberrant segregation of the chromosomes involved in the abnormality. The presence of a CA may also influence the segregation of other chromosomes, in a process known as in interchromosomal effect (ICE). The behaviour of the CA during meiosis may account for the infertility observed in this population. We studied chromosome segregation, ICE and meiotic defects in a variety of CA. In carriers of CA, we determined the segregation patterns of chromosomes involved in the abnormality. With the exception of the carriers of mosaic aneuploidy, we found significantly increased frequencies of unbalanced chromosome complements. We observed ICE in six of twelve carriers, which were confined largely to the acrocentric chromosomes 13 and 21. We compared the frequency of chromosome imbalance in CA carriers with infertile, but karyotypically normal, and found higher levels of sperm

aneuploidy than CA carriers or controls. We observed synapsis and recombination of homologous chromosomes in carriers of chromosomal abnormalities, as well infertile and fertile men. We observed reduced recombination in two of the carriers of CA and in three of the infertile men. Increased synaptic errors were observed in all carriers of CA and in four of the infertile men. We noted an increased proportion of cells lacking sex chromosome recombination in all of the CA carriers. We studied chromosome-specific recombination patterns on chromosomes 13, 18 and 21 and compared those results with levels of aneuploidy in the sperm but observed no relationship. We studied the recombination and sex chromosome association, of the involved chromosomes, in the three carriers of CA, and observed decreased recombination on the involved chromosomes and frequent association between the chromosome abnormality and the sex chromosomes. We report the use of a novel technique.

The Biology of Reproduction Giuseppe Fusco 2019-10-10 A look into the phenomena of sex and reproduction in all organisms, taking an innovative, unified and comprehensive approach.

Chromosomes Today A.T. Sumner 2012-12-06 When the late Professor C. D. Darlington founded what developed into the International Chromosome Conferences in Oxford in 1964, he was concerned that scientists who worked on different aspects of chromosomes, or who studied them in different ways, should have the opportunity of "discussing the fundamental problems of chromosomes with one another". The fact that well over 300 scientists with a wide variety of interests came to Edinburgh in August 1992 for the 11th International Chromosome Conference shows that there is still the same need, and also the desire among chromosomologists to have such discussions. The present volume contains almost all the invited contributions, and attests to the diversity of approaches and applications in chromosomal studies. A few years ago it may have seemed to some that chromosome studies were being superseded by molecular biology, but the molecular biologists have now realized that they need to know about chromosomes, and indeed an important, if ill-defined discipline of 'molecular cytogenetics' has grown up in recent years. We are pleased that in planning the Conference and this book, so much of the work presented is at the interface between cytogenetics and molecular biology. This will surely continue in the future, as boundaries between disciplines are largely artificial, and each has much to learn from the others.

Neurogenetics 2018-01-08 Genetic methodologies are having a significant impact on the study of neurological and psychiatric disorders. Using genetic science, researchers have identified over 200 genes that cause or contribute to neurological disorders. Still an evolving field of study, defining the relationship between genes and neurological and psychiatric disorders is evolving rapidly and expected to grow in scope as more disorders are linked to specific genetic markers. Part I covers basic genetic concepts and recurring biological themes, and begins the discussion of movement disorders and neurodevelopmental disorders, leading the way for Part II to cover a combination of neurological, neuromuscular, cerebrovascular, and psychiatric disorders. This volume in the Handbook of Clinical Neurology will provide a comprehensive introduction and reference on neurogenetics for the clinical practitioner and the research neurologist. Presents a comprehensive coverage of neurogenetics Details the latest science and impact on our understanding of neurological psychiatric disorders Provides a focused reference for clinical practitioners and the neuroscience/neurogenetics research community

THE IMPACT OF SEX CHROMOSOME COMPOSITION ON GENE EXPRESSION AND REGULATORY DIMORPHISMS IN MOUSE

EMBRYONIC STEM CELLS Rachael Jane Werner 2020 The variation between males and females constitutes the largest phenotypic dimorphism in any given species and, in humans, contributes to differences seen in the risk, incidence and response to treatment for a number of diseases. A primary point of divergence is driven by variation in the sex hormone composition between males and females. However, we hypothesize that the sex chromosome composition alone drives dramatic events to which subsequent hormonal exposure either ameliorates or potentiates these differences. Methods to study early development are often limited to in vitro systems or in model organisms, such as in mouse. Human embryonic stem (ES) cell

lines are available but are of limited access, are of high passages, and require more stringent growth conditions. Direct comparisons between male and female lines are extremely difficult, due to the abundance of multiple X chromosome statuses as well as non-random X chromosome inactivation. In the absence of more refined culture methods for human ES cells, we opted to use mouse ES cell lines derived in-house as this would enable us to determine allelic expression patterns as well as more easily maintain pluripotency and random X chromosome inactivation with established culture parameters. As such, to test our hypothesis, we derived an extensive panel of low passage mouse embryonic stem (ES) cell lines from reciprocal crosses between the C57BL/6 and CAST/EiJ mice. In total, I had access to over 20 unique mouse ES lines, including two of which show aneuploidy with loss of one of the sex chromosomes. The addition of these two lines to the experimental design grants us the ability to tease apart the individual contributions of the X and the Y chromosome in early development. Additionally, sex chromosome aneuploidies have yet to be evaluated in terms of their effects on the epigenome as well as their influence on directed differentiation in vitro. To set the foundation for our studies, we first performed a series of RNA-seq analyses in which we expanded the number and variation of sex-specific differences from previous reports using microarray. We then interrogated the contribution from each of the sex chromosome complements on gene ontology. Additionally, we identified and validated sex-specific alternative splicing events, for which there is very limited reporting. With an emphasis on genome-wide regulatory patterns, we then performed an unbiased weighted gene co-expression network analysis (WGCNA) for which we identified a key sex-specific expression module. The main driver of this module was the gene encoding Prdm14, a pivotal transcription factor involved in pluripotency. Luciferase assays with a known Prdm14-responsive enhancer showed higher expression when transfected into female than in male ES cells. Because Prdm14 is more abundant in female ES cells, this suggests that the dosage of this transcription factor is a key factor of its capacity to activate gene expression. This is the first ever documented sex-specific differential enhancer activity and further underscores the need to not only evaluate expression but functionality of the protein product within biological systems. Prdm14 has the dual capability of promoting and repressing transcription depending on its binding partners. Evaluation of histone modifications overlapping with known Prdm14 binding motifs in the promoters of the co-expressed genes revealed a unique signature between the male and female mouse ES lines. Based on our analyses, we hypothesize that the higher Prdm14 abundance in XX ES cells can activate gene expression even if genes harbor a repressive histone modification in their promoters. We propose that the lower abundance of Prdm14 in male ES cells can only activate genes that do not exhibit repressive histone modifications. With this information in hand, we then performed a directed differentiation assay to the cardiomyocyte lineage. From these experiments, we identified an XX-specific impairment to differentiate without chromosome loss. Additionally, the 39,X lines exhibited dysregulation of cardiac-related genes, potentially correlating with the defects seen in Turner syndrome patients. Overall these findings help to expand upon an underrepresented field in the basic sciences, namely the underlying contributions of the sex chromosome complement on gene expression and regulatory dimorphisms.

X & Y Chromosomal Variations Carole A. Samango-Sprouse 2016-10-11 This is the first book on X and Y chromosomal disorders to address these common but rarely diagnosed conditions. This book seeks to present the latest in research and clinical care addressing neuroimaging, the interaction between hormones, brain development, and neurodevelopmental progression. This book will primarily focus on 47, XXY (Klinefelter syndrome, or KS), 47, XYY (Jacobs' syndrome), and 47, XXX (Triple X). More variant disorders such as 48, XXXX, 48, XXXY and 49, XXXXY will be discussed. Topics of interest include neurological functioning, neuroimaging, social language, and the evolving perspectives of these XY chromosomal disorders. The effects of testosterone supplementation in males with 47, XXY will also be examined.

Polyploidy and Genome Evolution Pamela S. Soltis 2012-10-03 Polyploidy - whole-genome duplication (WGD) - is a fundamental driver of biodiversity with significant consequences for genome structure, organization, and evolution. Once considered a speciation process common only in

plants, polyploidy is now recognized to have played a major role in the structure, gene content, and evolution of most eukaryotic genomes. In fact, the diversity of eukaryotes seems closely tied to multiple WGDs. Polyploidy generates new genomic interactions - initially resulting in "genomic and transcriptomic shock" - that must be resolved in a new polyploid lineage. This process essentially acts as a "reset" button, resulting in genomic changes that may ultimately promote adaptive speciation. This book brings together for the first time the conceptual and theoretical underpinnings of polyploid genome evolution with syntheses of the patterns and processes of genome evolution in diverse polyploid groups. Because polyploidy is most common and best studied in plants, the book emphasizes plant models, but recent studies of vertebrates and fungi are providing fresh perspectives on factors that allow polyploid speciation and shape polyploid genomes. The emerging paradigm is that polyploidy - through alterations in genome structure and gene regulation - generates genetic and phenotypic novelty that manifests itself at the chromosomal, physiological, and organismal levels, with long-term ecological and evolutionary consequences.

Genetic Disorders, Syndromology and Prenatal Diagnosis T.V.N. Persaud 2012-12-06 Birth defects have assumed an importance even greater now than in the past because infant mortality rates attributed to congenital anomalies have declined far less than those for other causes of death, such as infectious and nutritional diseases. As many as 50 % of all pregnancies terminate as miscarriages, and in the majority of cases this is the result of faulty intrauterine development. Major congenital malformations are present in at least 2 % of all liveborn infants, and 22 % of all stillbirths and infant deaths are associated with severe congenital anomalies. Not surprisingly, there has been a great proliferation of research into the problems of developmental abnormalities over the past few decades. This series, *Advances in the Study of Birth Defects*, was conceived in order to provide a comprehensive focal source of up-to-date information for physicians concerned with the health of the unborn child and for research workers in the fields of fetal medicine and birth defects. The first four volumes featured recent experimental work on selected areas of high priority and intensive investigation, including mechanisms of teratogenesis, teratological evaluation, molecular and cellular aspects of abnormal development, and neural and behavioural teratology. It seems logical and timely that the clinical aspects should now be presented. Accordingly, leading experts were invited to review a broad range of common problems from the standpoint of embryology, aetiology, clinical manifestations, diagnosis and management. This volume deals with genetic disorders and prenatal diagnosis.

Sex Chromosome Abnormalities And Human Behavior Daniel B Berch 2019-07-11 This volume is based on a symposium, "Cognitive and Psychosocial Dysfunctions Associated with Sex Chromosome Abnormalities," presented at the 1986 Annual Meeting of the American Association for the Advancement of Science. It contains reports from individual research groups and a psychological study.

Human Chromosomes Eeva Therman 2012-12-06 "This book provides an introduction to human cytogenetics. It is also suitable for use as a text in a general cytogenetics course, since the basic features of chromosome structure and behavior are shared by all eukaryotes. Because my own background includes plant and animal cytogenetics, many of the examples are taken from organisms other than the human. Since the book is written from a cytogeneticist's point of view, human syndromes are described only as illustrations of the effects of abnormal chromosome constitutions on the phenotype. The selection of the phenomena to be discussed and of the photographs to illustrate them is, in many cases, subjective and arbitrary and is naturally influenced by my interests and the work done in our laboratory. " The above paragraph from the Preface of the first edition of this book also fits the present edition. However, so much has happened in five years in cytogenetics that-apart from a couple of pages here and there-the whole book has been rewritten and nine new chapters have been added. The system used in the first edition to cite, whenever possible, the latest and/or the most comprehensive review rather than the original publications has been followed here also. Not only would complete literature citations increase the size of the book too much, but many readers have expressed satisfaction with the referencing method used here.

Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling R. J. M. Gardner 2018 Preceded by Chromosome abnormalities and genetic counseling / R.J. McKinlay Gardner, Grant R. Sutherland, Lisa G. Shaffer. c2012.

MRCOG Part One Alison Fiander 2016-10-13 A fully updated and illustrated handbook providing comprehensive coverage of all curriculum areas covered by the MRCOG Part 1 examination.

Noninvasive Prenatal Testing (NIPT) Lieve Page-Christiaens 2018-08-19 Since its introduction in 2012, cell-free (cf) DNA based Non-Invasive Prenatal Testing (NIPT) has been employed to test for fetal chromosome abnormalities, and gene mutations that lead to a variety of genetic conditions, by millions of pregnant women, in more than 90 countries worldwide. With Noninvasive Prenatal Testing (NIPT): Applied Genomics in Prenatal Screening and Diagnosis, Dr Lieve Page-Christiaens and Dr Hanns-Georg Klein have compiled the first authoritative volume on cfDNA NIPT methods and their clinical implementation. Provides a thorough, practical examination of the history of NIPT, NIPT laboratory techniques and bioinformatics, NIPT screening and diagnostics for a wide range of disorders and birth defects Presents leading, international experts who discuss the application of NIPT in early screening for common aneuploidies, fetal chromosome anomalies, autosomal trisomies, fetal blood group typing, and maternal constitutional and acquired copy number variants Includes full color imagery that enhances concept illustration, along with detailed descriptions of the benefits (and limitations) of NIPT Offers clinicians, researchers, genetic counselors and reproductive specialists of all kinds the required background information, methodologies and essential patient counseling techniques

Sex Chromosomes Ursula Mittwoch 2014-06-28 Sex Chromosomes focuses on the study of sex chromosomes, including human chromosomal abnormalities, behavior and characteristics of chromosomes, and cell division. The book first offers information on the chromosomal basis of sex determination, as well as development of the cell theory, mitosis, fertilization, meiosis, and discovery of sex chromosomes. The publication also ponders on the mitosis, meiosis, and formation of gametes. Discussions focus on the special characteristics of sex chromosomes, abnormalities of cell division, and sexual differentiation. The manuscript reviews sex chromosomes in plants, Drosophila, and Lepidoptera. The book also examines sex-chromosome mechanisms that differ the classic type; sex chromosomes in fishes, amphibia, reptiles, and birds; and sex chromosomes in man. Discussions focus on normal human sex chromosomes, Turner's syndrome, Klinefelter's syndrome, true hermaphrodites, testicular feminization, and pseudohermaphrodites. Sex chromosomes in mammals other than man, including monotremata, marsupialia, insectivora, rodentia, and carnivora, are discussed. The publication is a dependable reference for readers interested in the study of sex chromosomes.

Aneuploidy in Health and Disease Zuzana Storchova 2012-05-16 Aneuploidy means any karyotype that is not euploid, anything that stands outside the norm. Two particular characteristics make the research of aneuploidy challenging. First, it is often hard to distinguish what is a cause and what is a consequence. Secondly, aneuploidy is often associated with a persistent defect in maintenance of genome stability. Thus, working with aneuploid, unstable cells means analyzing an ever changing creature and capturing the features that persist. In the book *Aneuploidy in Health and Disease* we summarize the recent advances in understanding the causes and consequences of aneuploidy and its link to human pathologies.

Sex Chromosomes: Advances in Research and Application: 2011 Edition 2012-01-09 Sex Chromosomes: Advances in Research and Application: 2011 Edition is a ScholarlyBrief™ that delivers timely, authoritative, comprehensive, and specialized information about Sex Chromosomes in a concise format. The editors have built Sex Chromosomes: Advances in Research and Application: 2011 Edition on the vast information databases of ScholarlyNews.™ You can expect the information about Sex Chromosomes in this eBook to be deeper than what you can access anywhere else, as well as consistently reliable, authoritative, informed, and relevant. The content of Sex Chromosomes: Advances in Research and Application: 2011 Edition has been produced by the world's leading scientists, engineers, analysts, research institutions, and companies. All of the content is from peer-

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A Sex Chromosome Survey of Hyperactive Children William Paul Karduck 1969 The documented sex chromosome include aneuploidy of both the X and Y sex chromosomes. Males with additional X chromosomes are chromatin positive and can be detected by the nuclear chromatin analysis. An additional Y chromosome in males can be detected only by karyotype analysis. Cytological studies have shown that in comparison to the general population, there is an increased incidence of X and Y chromosome aneuploidy among mental defectives and criminals. Since incarcerated individuals and hyperactive children have in common certain aggressive and antisocial behavioral traits, this study set out to determine whether a sex chromosome aneuploidy existed in these children. The present study screened hyperactive children, patients from St. Louis Children's Hospital, for sex chromosome aneuploidy. Two cytological approaches were employed. In one, cells from the buccal mucosa were assayed for X chromosome aneuploidy using sex chromatin as the index. Analyses were done on 96 hyperactive children: 14 females and 82 males. No abnormalities were detected. The second cytological approach involved chromosome analyses of cells from peripheral blood. Chromosomes were analyzed on 23 hyperactive children: 3 females and 20 males. Microscopic and karyotypic analyses revealed normal chromosome constitution for both the females and males. In addition, the buccal mucosal cells of 20 of these children were assayed for sex chromatin and confirmed the karyotype results.

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