

Sex Chromosomes Results

Chromosome Biology as a Key to Understand Disease Mechanisms, Genome Architecture and Evolution Anja Weise 2021-04-16 This topic has been realized, and is in collaboration with Dr. Constanze Pentzold, Post Doctoral Researcher at the Institute of Human Genetics, University Hospital Jena.

Turner Syndrome Theola Stubby 2021-03-31 Turner syndrome, a condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure of the ovaries to develop, and heart defects. This book is storytelling about a girl who found out she has a form of Turner syndrome when she was 16. It was difficult to see how anything positive was going to come out of the new journey she was faced with. The journey has not always been easy but, through everything, she has become a stronger person and learned a lot.

Molecular Mechanisms Underlying Sexual Differentiation of the Brain and Brain Behavior Tuck Cheong Ngun 2012 The brains of males and females are different anatomically and chemically. There are also sex differences in neurological disease, cognition and behavior that are presumed to be downstream consequences. Two main factors have been implicated in sexual differentiation of the brain: gonadal hormones and direct genetic effects. Here, we explore the role of sex chromosomes in the brain and behavior and the molecular mechanisms mediating the effects of these factors. We investigated the contribution of sex chromosomes to sex differences in brain and behavior by studying a novel mouse model of Klinefelter Syndrome (KS) termed the Sex Chromosome Trisomy (SCT) model. KS is characterized by the presence of an additional X chromosome in men. We investigated the extent of feminization in XXY male mice. We found that partner preference in XXY males is feminized and that these differences are likely due to interactions of the additional X chromosome with the Y. We also found that expression of a small but highly significant proportion of genes is feminized in the bed nucleus of the stria terminalis/preoptic area (BNST/POA) of XXY males, which represent strong candidates for dissecting the molecular pathways responsible for KS-specific phenotypes. We also investigated whether DNA methylation could be one of the molecular mechanisms that mediate the long-lasting, irreversible effects of perinatal testosterone in the BNST/POA. Using a genome-wide approach, we found that methylation at 45 genes was affected three days after the exposure. This number ballooned to 740 in adult animals. There was also a shift to a more masculine pattern of DNA methylation during adulthood in females that had seen perinatal testosterone. These results strongly suggest that perinatal testosterone confers an initial imprint that is amplified over postnatal development. We also observed sex differences in methylation at numerous genes. The interplay between gonadal hormones and sex chromosomes is a complex one. Collectively, our results provide further support for the theory of direct genetic effects in brain sexual differentiation and suggest that DNA methylation may be one mechanism that mediates not only the effects of gonadal hormones but also direct genetic effects.

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.

Spectrum of Sex Minoru Tanaka 2022-11-07 This book introduces cutting-edge studies on the spectrum of sex. The sex spectrum can be understood as an interwoven mechanism sustaining graded phenotypes between the two sexes. The book overviews three elements that develop the sex spectrum: genetics, the endocrine system, and the environment. Part I discusses the genetic regulation during sex determination, which often results in a mixture of two sexes or sex reversal. The evolutionary aspects of the genetic determinants are also discussed. Part II presents the involvement of endocrine regulation in the sex spectrum, which covers a broad range of phenotypic events, including sexual behavior and metabolism. Interestingly,

sex hormones can also act as sex determinants. Finally, Part III shows that intrinsic factors, such as sex-determining genes and sex hormones, are not the only factors in sex development. The environment surrounding organisms, such as symbiosis and metabolism, act on the sex as critical factors, generating the sex spectrum. Determination and development of the two sexes have been a topic of great interest and a long-standing issue in biology. The book updates the conventional view that biological sex is fixed after birth and sets new perspectives for understanding sex as a spectrum manifested in multiple phenomena. Each chapter contributed by leading experts explains the sex spectrum in various organisms and their underlying mechanisms from the latest ongoing studies. The book provides a valuable resource for not only experts in developmental biology, physiology, and medical science, but also non-scientists and anyone interested in the topic.

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.

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.

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.

Sex-linked Inheritance in Drosophila Thomas Hunt Morgan 2022-08-10 The following book was written by Thomas Hunt Morgan and Calvin Bridges, and made the former world-famous. It was in the studies covered in the following publication that Morgan discovered that genes are carried on chromosomes and are the mechanical basis of heredity. These discoveries formed the basis of the modern science of genetics; and he would later win the Nobel Prize in Physiology or Medicine in 1933 for his findings.

Molecular Genetics of Sex Determination Stephen S. Wachtel 2014-07-23 In this era of accelerated discovery and prolific output, Molecular Genetics of Sex Determination keeps readers abreast of this field's fast-moving biology. Its chapters were completed by experts in each area only months before publication. The text is organized into two parts. First, it reviews the basic biology of sex determination and summarizes groundbreaking work in mouse, marsupial, and *Drosophila* systems. Second, it covers current human genetics, clinical studies, and the syndromes of abnormal sex differentiation. With chapters by preeminent reproductive biologists, this is a capital work. Ohno's law is described by Ohno; the Lyon

hypothesis, by Lyon; Sinclair tells how he cloned the testis-determining gene; and so on. *Molecular Genetics of Sex Determination* is authoritative, comprehensive, and current. It is prime reading for geneticists, developmental biologists, graduate students in these and related fields, clinical researchers, physicians, and medical students. Reviews the genetics of sex determination in 19 up-to-date chapters Features research on sex chromosomes and sex-determining genes Includes abnormalities of sex determination and clinical genetics Written by scientists who pioneered work in this field

Your Easy Way to Chromosomes Sana Nimer Abu Shihab 2012-03 These days, hardly a week goes by in the media, without mention of a remarkable advancement in the field of genetics. Cytogenetics is a branch of genetics that is concerned with the study of the structure and function of the chromosomes and their role in heredity. Every individual inherits a pair of chromosomes from each of his parents. Each cell in our body has 46 chromosomes each. Chromosomes carry genetic information in the form of genes. The genes within the chromosomes have a powerful impact on our health, either directly through chromosomal or single gene disorders or by influencing our susceptibility to disease. Cytogenetic study is performed in order to diagnose certain genetic disorders such as; congenital birth defects, mental retardation, growth and developmental delay, defects of sexual development, ambiguous genitalia, congenital defects, abnormal facial features, infertility, multiple miscarriages, amenorrhea, autism, malignancies and hematological disorders, early embryonic death, and gene mutations among others. These can be identified by chromosomal analysis and molecular cytogenetic techniques such as Fluorescent in Situ Hybridization (FISH) and Microarray, which have enormously expanded in recent years.

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 an 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 techniq.

Facts about Down Syndrome for Women Over 35 National Institute of Child Health and Human Development (U.S.) 1979

Genetics of Sex Differentiation Ursula Mittwoch 2012-12-02 *Genetics of Sex Differentiation* intends to help readers understand the genetic basis of sex differentiation. The book focuses on explaining how the sex chromosomes affect the process of sex differentiation by influencing the rates at which cells divide. The book is composed of seven chapters. It provides overviews of classical genetics and structure of cells. It also explains the

chromosomal basis of sex determination and sex determination using *Drosophila*. Polygenetics and continuous and quasicontinuous variations are also discussed. The book also discusses sex factors, determination, and disorders. Moreover, it explains the heterochromatin, embryological basis of sex differentiation, and triploidy and autosomal effects. In addition, it discusses the relationship of genes, chromosomes, growth, and sex. The book is an excellent "bedside book" for students in biology, specifically in genetics and developmental biology. Lecturers and professionals in biology and genetics will also find this book invaluable for their practice.

The Danish Cytogenetic Central Register Johannes Nielsen 1980

Human Cytogenetics John L. Hamerton 2013-10-22 Human Cytogenetics: Clinical Cytogenetics, Volume II presents the general theoretical principles and clinical aspects of cytogenetics, a branch of genetics that deals specifically with the study of the chromosomes. The volume focuses on the clinical cytogenetics of human. It discusses the sex chromosomes and their abnormalities and the abnormalities of sexual development and differentiation; mechanism of sex determination in mammals; major autosomal abnormalities found in human populations; and chromosome abnormalities in relation to human pregnancy wastage and chromosome changes in neoplasia. The book will be a great reference book for geneticists, cytogeneticists, pathologists, clinicians, and medical students.

A Handbook of Clinical Genetics J. S. Fitzsimmons 2013-10-22 A Handbook of Clinical Genetics focuses on clinical genetics and the growing demand for genetic counseling. This book begins by introducing issues regarding changes in morbidity and mortality; fall in birth rate; advances in technology and treatment; and complex social changes. Other topics covered include genetic and environmental factors in disease; the genetic code; pedigree information; inheritance patterns; genetic counseling; prenatal diagnosis of genetic disease; special problems; and ethical issues and future developments. The last portion of this text is devoted to a glossary of unfamiliar medical terms, list of recommended books for further research and study, and appendices consist of a case on genetic counseling for Down's syndrome. This handbook is suitable for nurses, medical students, and doctors needing an introduction to clinical genetics.

Down Syndrome: From Understanding the Neurobiology to Therapy 2012-10-16 Down syndrome (DS) is the most common example of neurogenetic aneuploid disorder leading to mental retardation. In most cases, DS results from an extra copy of chromosome 21 (HSA21) producing deregulated gene expression in brain that gives rise to subnormal intellectual functioning. The topic of this volume is of broad interest for the neuroscience community, because it tackles the concept of neurogenomics, that is, how the genome as a whole contributes to a neurodevelopmental cognitive disorders, such as DS, and thus to the development, structure and function of the nervous system. This volume of Progress in Brain Research discusses comparative genomics, gene expression atlases of the brain, network genetics, engineered mouse models and applications to human and mouse behavioral and cognitive phenotypes. It brings together scientists of diverse backgrounds, by facilitating the integration of research directed at different levels of biological organization, and by highlighting translational research and the application of the existing scientific knowledge to develop improved DS treatments and cures. Leading authors review the state-of-the-art in their field of investigation and provide their views and perspectives for future research. Chapters are extensively referenced to provide readers with a comprehensive list of resources on the topics covered. All chapters include comprehensive background information and are written in a clear form that is also accessible to the non-specialist.

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.

Splitting the Sexes Aivars Cirulis 2022 The evolution of gonochorism from hermaphroditism can be gradual by increasing investment in one sex role while decreasing in the other, or rapid through the fixation of sex-role sterility mutations, eventually leading to the evolution of sex chromosomes. It is expected that the transition will involve a temporary state of gynodioecy or androdioecy as the mutations are not expected to take place at the same time. If the first mutation is a dominant female-sterility mutation, later accompanied by a recessive male sterility mutation, then an XY sex chromosome system evolves, while the opposite combination of mutations will result in a ZW system. Later on sexually antagonistic (SA) genes can be linked to the newly established sex-determining regions on the sex chromosomes. This is followed by recombination arrest in the region, so that the inheritance pattern is sex-limited for all these sex-specific genes. However, the lack of recombination leads to degeneration of the genetic content on the sexlimited chromosomes, since recombination is important for repairing mutations. Nevertheless, recombination arrest does not necessarily mean a dead-end for the sex-limited chromosomes. As our understanding of the very early stages of sex chromosome evolution is mainly based on theory and comparative evidence, we developed a system which we hoped would make it possible to observe in real time what happens after the acquisition of a new sex-determining gene. We used a previously established green fluorescent protein (GFP) line of the simultaneous hermaphrodite *Macrostomum lignano*. We used the GFP locus as a dominant sterility mutation, which is inherited in a Mendelian fashion. By allowing the GFP allele to be inherited only through sperm, we created male-limited selection lines (resembling the early stages in XY chromosome evolution), and by allowing the GFP allele to be inherited only through egg cells, we created female-limited selection lines (resembling the early stages in ZW chromosome evolution). We also created control lines, where the inheritance pattern was equally mixed. After tens of generations, we investigated how these lines have responded on the level of the genome, the transcriptome, and the phenotype. We sequenced genomes and analysed changes in SNP frequency and structural variant (SV) distribution in pairwise comparisons to see changes across the genome, but particularly on the scaffold where the GFP is located. We also sequenced transcriptomes and performed pairwise comparisons to detect differentially expressed genes, and analysed significant GO terms and KEGG pathways to see how the gene regulation has changed. Besides genomic analyses, we also looked at how mating behaviour (copulation frequency and duration, as well as probability of post-copulatory sucking behaviour) and sexual anatomy (gonad size and morphology of the male copulatory organ called stylet) has changed. We observed that the female-selected lines seemed to have responded the most at the genomic level. For example, the number of significantly differentially expressed transcripts was largest between the female-selected lines and the control lines. These changes seemed to involve downregulation of testes-biased genes. In addition, we observed the highest number of SVs in the female-selected lines, which could be related to changes in recombination rate. In contrast, the male-selected lines seemed to have responded the most at the phenotypic level, since we observed a decrease in the ovary size and body size in the male-selected lines, as well as behavioural changes that may be related to changes in the ejaculate. Both sex-specific selection regimes showed evidence of

alterations in the shape of the stylet. Based on these results, we can conclude that our worms have indeed responded to the sex-limited selection in a way that is generally consistent with our expectations from other young sex chromosome systems. The evidence of a decrease in the testes function in the female-selected lines resembles adaptation towards gynodioecy, and the evidence of a decrease in the ovary size in the male-selected lines resembles adaptation towards androdioecy.

Accurate Map[s] of Cumberland, Westmorland, and Lancashire [...] Peter Crosthwaite 1809

The Chromosome Disorders Gordon Howard Valentine 1975

Females Are Mosaics Barbara Migeon 2007-03-15 Women can be described as genetic mosaics because they have two distinctly different types of cells throughout their bodies. Unlike males, who have one X chromosome (inherited from their mother), females have two X chromosomes in every cell (one from each parent). The fathers copy works in some cells, while the mothers copy works in others. These two X chromosomes often function differently, especially if one carries a defective gene. Much has been written about the Y chromosome and its role in inducing maleness. This will be the first book about the X chromosome as a key to female development and the role of X-related factors in the etiology of sex differences in human disease. Barbara Migeon, from the renowned McKusick-Nathan Institute at Johns Hopkins, is a major figure in clinical genetics and is eminently qualified to write this book, and she writes clearly and effectively. She describes both the underlying molecular mechanisms and the remarkable genetic consequences of X inactivation and its role in determining the biological concepts characteristic of women. *Females are Mosaics* will be valuable to geneticists, biologists, and all health professionals interested in women's health.

Sex Chromosomes Franz Schrader 1928

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.

Genetics of Sex Determination R.S. Verma 1996-04-23 The Genetical Theory of Natural Selection by R.A. Fisher (1930) dictated that sexual dimorphisms may depend upon a single medelian factor. This could be true for some species but his suggestion could not take off the ground as gender in *Drosophila* is determined by the number of X chromosomes. Technical advances in molecular biology have revived the initial thinking of Fisher and dictate that TDF or SRY genes in humans or Tdy in mice are sex determining genes. The fortuitous findings of XX males and XY female, which are generally termed sex reversal phenomenon, are quite bewildering traits that have caused much amazement concerning the pairing mechanism(s) of the pseudoautosomal regions of human X and Y chromosomes at meiosis. These findings have opened new avenues to explore further the genetic basis of sex determination at the single gene level. The aim of the fourth volume, titled *Genetics of Sex Determination* is to reflect

on the latest advances and future investigative directions, encompassing 10 chapters. Commissioned several distinguished scientists, all pre-eminent authorities in each field to shed their thoughts concisely but epitomise their chapters with an extended bibliography. Obviously, during the past 60 years, the metric advances are voluminous and to cover every account of genes, chromosomes, and sex in a single volume format would be a herculean task. Therefore, a few specific topics are chosen, which may be of great interest to scientists and clinicians. The seasoned scientists who love to inquire about the role of genes in sex determination should find the original work of these notable contributors very enlightening. This volume is intended for advanced students who want to keep abreast as well as for those who indulge in the search for genes of sex determination.

Facts about Mongolism for Women Over 35 1973

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.

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.

Evolution of Transcriptional Inactivation on Sex Chromosomes in Birds and Mammals Alexandra Maxine Livernois 2012 Differentiated sex chromosomes (XY or ZW) result in a 2:1 gene dosage imbalance between the X or Z and the autosomes, in the heterogametic sex, because genes have been lost from the usually small and heterochromatic sex-specific element (Y or W). This gene loss also results in a 2:1 dosage difference between the sexes for unpartnered genes on the X or Z. Such dosage imbalances may be compensated for by various strategies in different taxa. In this thesis I examine dosage compensation of the X chromosomes in a monotreme mammal, and the Z chromosome of a representative bird.

Sex Chromosome Evolution in Blow Flies Anne Amarila Andere 2020 Chromosomal mechanisms of sex determination vary greatly in phylogenetically closely related species, indicative of rapid evolutionary rates. Sex chromosome karyotypes are generally conserved within families;

however, many species have derived sex chromosome configurations. Insects display a plethora of sex chromosome systems due to rapid diversification caused by changes in evolutionary processes within and between species. A good example of such a system are insects in the blow fly family Calliphoridae. While cytogenetic studies observe that the karyotype in blow flies is highly conserved (five pairs of autosomal chromosomes and one pair sex chromosome), there is variation in sex determining mechanisms and sex chromosome structure within closely related species in blow flies. The evolutionary history of sex chromosomes in blow fly species have not been fully explored. Therefore, the objective of this research was to characterize the sex chromosome structures in four species of blow flies and investigate the selective forces which have played a role in shaping the diverse sex chromosome system observed in blow flies. The blow fly species used in this study are *Phormia regina*, *Lucilia cuprina*, *Chrysomya rufifacies* and *Chrysomya albiceps*. *Phormia regina*, and *Lucilia cuprina* have a heteromorphic sex chromosome system and are amphogenic (females produce both male and female offspring in equal ratio). In contrast, *Chrysomya rufifacies* and *Chrysomya albiceps*, have a homomorphic sex chromosome system, are monogenic (females produce unisexual progeny), have two types of females (arrhenogenic females - male producers and thelygenic females - female producers), and sex of the offspring is determined by the maternal genotype. To accomplish these tasks, a total of nine male and female individual draft genomes for each of the four species (including three individual draft genomes of *Chrysomya rufifacies* - male, and the two females) were sequenced and assembled providing genomic data to explore sex chromosome evolution in blow flies. Whole genome analysis was utilized to characterize and identify putative sex chromosomal sequences of the four blow fly species. Genomic evidence confirmed the presence of genetically differentiated sex chromosomes in *P. regina* and *L. cuprina*; and genetically undifferentiated sex chromosomes in *C. rufifacies* and *C. albiceps*. Furthermore, comparative analysis of the ancestral Dipteran sex chromosome (Muller element F in *Drosophila*) was determined to be X-linked in *P. regina* and *L. cuprina* contributing to sex chromosome differentiation but not sex-linked in *C. rufifacies* and *C. albiceps*. Evolutionary pressures are often quantified by the ratio of substitution rates at non-synonymous (dN) and synonymous (dS) sites. Substitution rate ratio analysis (dN/dS) of homologous genes indicated a weaker purifying selection may have contributed to the loss of sex-linked genes in Muller element F genes of the undifferentiated sex chromosome as compared to the differentiated sex chromosome system. Overall, the results presented herein greatly expands our knowledge in sex chromosome evolution within blow flies and will reinforce the study of sex chromosome evolution in other species with diverse sex chromosome systems.

The X in Sex David Bainbridge 2004-09-30 A tiny scrap of genetic information determines our sex; it also consigns many of us to a life of disease, directs or disrupts the everyday working of our bodies, and forces women to live as genetic chimeras. The culprit--so necessary and yet the source of such upheaval--is the X chromosome, and this is its story. An enlightening and entertaining tour of the cultural and natural history of this intriguing member of the genome, *The X in Sex* traces the journey toward our current understanding of the nature of X. From its chance discovery in the nineteenth century to the promise and implications of ongoing research, David Bainbridge shows how the X evolved and where it and its counterpart Y are going, how it helps assign developing human babies their sex--and maybe even their sexuality--and how it affects our lives in infinitely complex and subtle ways. X offers cures for disease, challenges our cultural, ethical, and scientific assumptions about maleness and femaleness, and has even reshaped our views of human evolution and human nature.

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.

Textbook of Human Reproductive Genetics Karen Sermon 2014-04-10 What happens with our genome and epigenome in the first fundamental days of our development? How can this be analysed? What do we need to know when faced with patients' questions about their own infertility, or how to prevent the birth of affected children? For the first time, this book brings together both scientists' and clinicians' viewpoints on human reproductive genetics, making for a more comprehensive discussion of interest to ART professionals and developmental biologists. With worldwide leaders in this burgeoning field guiding the reader through from the basics to the most exciting recent discoveries, this book presents the wider picture of how reproductive medicine and biology links with genetics. The editors also address the new challenges raised in how to treat and counsel patients at fertility and genetic clinics, as well as eliciting vivid bioethical debates. This book brings together genetics, reproductive biology and medicine for practitioners and geneticists.

Gender Differences: Their Ontogeny and Significance Christopher Ounsted 1972

Chromosomes of Man Hans Zellweger 1977

Genes and Mechanisms in Vertebrate Sex Determination Gerd Scherer 2013-04-17 Following an opening chapter by the late Susumu Ohno on paralogues of sex-determining genes, the five best-studied genes essential for early mammalian gonadal development are portrayed in detail: SF-1 and WT1 and their roles in early events in gonadal development, SRY and SOX9 in testis determination, and the anti-testis gene DAX-1. Subsequent chapters look at the roles of these genes in sex determination in marsupial mammals, birds, reptiles, amphibians and fish, and review the different sex-determining mechanisms, genetic and environmental, that operate in these different vertebrate classes. Two insights emerge: one, that the same basic set of genes appears to operate during early gonadal development in all vertebrates, despite the differences in mechanisms; the other, that sex determination in vertebrates results from a complex network of regulatory interactions and not from a simple hierarchical cascade of gene actions.

Cytogenetic Abnormalities Susan Mahler Zneimer 2014-09-22 Cytogenetics is the study of the structure and function of chromosomes in relation to phenotypic expression. Chromosomal abnormalities underlie the development of a wide variety of diseases and disorders ranging from Down syndrome to cancer, and are of widespread interest in both basic and clinical research. Cytogenetic Abnormalities: Chromosomal, FISH, and Microarray-Based Clinical Reporting is a practical guide that describes cytogenetic abnormalities, their clinical implications and how best to report and communicate laboratory findings in research and clinical settings. The text first examines chromosomal, FISH, and microarray-based analyses in constitutional disorders. Using these same methodologies, the book's focus shifts to acquired abnormalities in cancers. Both sections provide illustrative examples of cytogenetic abnormalities and how to communicate these findings in standardized laboratory reports. Providing both a wealth of cytogenetic information, as well as practical guidance on how best to communicate findings to fellow research and medical professionals, Cytogenetic Abnormalities will be an essential resource for cytogeneticists, laboratory personnel, clinicians, research scientists, and students in the field. A guide to interpreting and reporting cytogenetic laboratory results involved in constitutional disorders and cancers Guides the reader on implementing the International System for Human Cytogenetic Nomenclature in written reports Provides information to allow scientists and medical professionals to fully understand and communicate cytogenetic abnormalities Describes a wide array of cytogenetic abnormalities observed in the

laboratory Divided into user-friendly sections devoted to methodologies and implications of specific diseases

Chromosome Abnormalities and Genetic Counseling R. J. M. Gardner 1989 Chromosomal defects are one of the most common causes of genetic disorders and are responsible for a large proportion of miscarriages. As a result, more and more parents are becoming interested in genetic counseling in order to learn about the risks of reproductive failure, or to understand why they had a child with a particular defect, whether it will happen again, and what might be done to prevent it. This book addresses these and other questions concerning chromosome abnormalities. Although the book is written primarily for genetic counselors and laboratory workers in cytogenetics, it will be of interest to all physicians concerned with genetic counseling. Each chapter discusses a different type of defect, such as translocations, duplications, insertions, deletions, and non-disjunction. The basic biological theory underlying the problem is discussed and in-depth information is provided on the reproductive risks of chromosomally abnormal individuals and of normal parents who have a child with a chromosome abnormality. The book will help concerned parents determine an appropriate course of action, including prenatal diagnosis, and help counselors to communicate the information necessary for responsible decision-making.

The Genomic Basis of the Response to Female-limited X-chromosome Evolution Yesbol Manat 2021 In this thesis, I attempted to achieve a better understanding of the nature of X-linked polymorphic loci using a female-limited X chromosome (FLX) experimental evolution in *Drosophila melanogaster*. I expected that expressing the evolved X chromosome will result in an increase in female fitness and a decrease in male fitness. I first investigated the effect of an experimentally evolved female-limited X chromosome on male reproductive traits (Paper I). Secondly, I examined how the genome-wide expression pattern responds to the presence of the evolved X chromosome (Paper II), and then I analysed the changes in allele frequencies across the genome (Paper III). Finally, I attempted to study the changes in genetic variances and covariances in sexually homologous traits in response to FLX evolution, as well as change in the cross-sex genetic correlations for these sexually homologous traits (Paper IV). Contrary to the initial expectation, I found evidence of trade-offs between various components of male reproductive success rather than an overall decline (Paper I). However, I identified a more 'feminized' gene expression profile as the result of FLX evolution (Paper II) and found evidence of adaptation in the methodological control treatment which was a necessary part of the experimental design. The analysis of differences in allele frequencies between selection regimes showed no evidence of overrepresentation of SA loci on the X chromosome, but these results suggest an interesting avenue for future study of sexual conflict over sensory ability (Paper III). Finally, I found evidence of a breakdown in the intersexual genetic correlation for locomotory activity in FLX populations compared to control populations (Paper IV). These results indicate that the X chromosome may not possess as many SA mutations as previously thought, and they are by nature difficult to study in a species with old, already highly degenerated sex chromosomes. However, the results presented here highlight the importance of sex-specific selection pressures in shaping the genetic architecture of many traits.

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