

# Sex Chromosomes Trisomy

*Genetic Disorders of Human Sexual Development* - Leonard Pinsky 1999

There have been many advances in understanding mammalian sex determination and differentiation during the last decade. Using these advances to elucidate clinical conditions of abnormal sexual development, the authors of this book bring together expertise in molecular endocrinology, molecular genetics, and dysmorphology. This is not a conventional textbook. It is seldom dogmatic, frequently presents alternatives, highlights speculation, raises questions, and attempts to provide answers. This book will be welcomed by medical geneticists, genetic counselors, endocrinologists, gynecologists, urologists, and students who need fully-referenced information about the genetic aspects of human sexual maldevelopment in order to better manage their patients and their patients' families.

**Trisomy 21** - G.R. Burgio 2013-03-07

With contributions by numerous experts

**Human Afflictions and Chromosomal Aberrations** - Raymond Turpin 2013-10-22

Modern Trends in Physiological Sciences, Volume 32: Human Afflictions and Chromosomal Aberrations presents the study of the links between chromosome aberrations and physical and mental congenital anomalies. This book discusses the possibilities of human cytogenetic research as well as its difficulties. Organized into 15 chapters, this volume begins with an overview of the development of human chromosome investigations. This text then explains the methods for studying human chromosomes, which can be applied without controlling the atmosphere of the incubator. Other chapters describe the structural features of the normal human karyotype. This book discusses as well the early appearance of a chromosome aberration that produces a change in the hereditary patrimony manifest in a constitutional disorder of the individual. The final chapter deals with the biochemical effects that correspond to numerical or structural

anomalies in chromosome 21. This book is a valuable resource for genetecists, cytogeneticists, physicians, and clinical researchers.

**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. *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,XYX. 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.

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.

**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.

**Chromosomal Aberrations in Ophthalmology** - Jules François 1975

**Sex Chromosomes and Testosterone: A Novel Mouse Model Sheds Light on the Etiology of Klinefelter Syndrome** - Shayna Marie Williams-Burris 2016

Klinefelter syndrome (KS) occurs when boys are born with a second X chromosome so their cells are XXY instead of XY. Men with KS have low testosterone, infertility, obesity and metabolic dysfunction, and some conditions that typically affect women such as osteoporosis, lupus, and breast cancer. Notably, they exhibit cognitive differences such as speech delay in early childhood and difficulty with executive functions as adults. Historically, KS symptoms were

thought to be primarily due to low testosterone levels, yet testosterone treatment does not cure KS. Therefore, we hypothesize that direct genetic effects of the second X chromosome play an essential role. Previously, it was impossible to separate the influence of genes vs. hormones, but our lab has established the novel Sex Chromosome Trisomy (SCT) mouse model, which can be used to distinguish the two factors. The SCT model produces mice with XY and XXY chromosomes that are either male or female. XXY males have low testosterone so differences between the male groups may be due either genetic or hormonal differences. However, both female groups have low testosterone, so differences in females provide strong evidence for direct genetic effects. We find that XXY mice have an obesity phenotype like that seen in human KS, and that sex chromosomes influence this trait, though the genetic effect may be facilitated by the presence of hormones. The mice also show an osteoporosis phenotype like humans that we find to be influenced by hormones only. Finally, a preliminary study in another mouse model of KS, the XY\* model, show XXY\* mice may have executive functions deficit like those seen in humans. Next steps should test the executive functioning in the SCT model, and to also measure pup vocalizations as a model of speech delay. Further research should expand the use of the model to gene expression studies to identify which genes are responsible for direct genetic effects in SCT mice. Ideally the SCT model will be increasingly characterized and lead to a better understanding of the genetic factors at play in KS, potentially leading to better treatment options for KS patients.

*ABNORMAL KARYOTYPES* - Sana Nimer Abu Shihab 2013-10

In my first book (*Your Easy Way To Chromosomes*), the main topic was about the human chromosomes, their structures, abnormalities, syndromes, and chromosome analysis. In this book I focused on abnormal karyotypes and how chromosomal abnormalities happen. A karyotype is a picture of a person's chromosomes from body cells (blood, hair, or any other tissue), photographing them through a microscope and arranging them in pairs, ordered by size and position of centromere for

chromosomes of the same size. Karyotype test (alternative names are Chromosome Analysis, Chromosomal Analysis) plays a role in: diagnosis genetic diseases which are related to chromosomal abnormalities, diagnosis some birth defects, and provides clinical utility in the diagnosis and treatment of hematologic malignancies. On the other hand some genetic abnormalities cannot be detected by karyotype analysis such as microdeletions. Karyotype helps clinical cytogeneticist to identify abnormalities by: Counting the number of chromosomes and looking for extra chromosome such as in trisomy 21 or missing chromosome in a karyotype such as in Turner syndrome. Looking for changes in chromosome structure such as chromosomal deletions, duplications, translocations, insertions, inversions and other chromosomal abnormalities. Writing a book related to your field shows your passion and commitment to your job. Sana Nimer sananimer1@gmail.com sananimer1@hotmail.com

**A Glossary of Genetics and Cytogenetics** - R. Rieger 2013-04-17

The past two decades have witnessed a truly phenomenal growth and expansion in our knowledge of the principles and mechanisms of inheritance. Molecular and microbial genetics, for all purposes non-existent at the outset of this period, have developed and flourished to the extent of becoming major branches of genetics from which the most exciting and edifying concepts of gene function and structure have been derived. Similarly, man, heretofore a genetic curiosity, has become in his own right a genetic organism of first rank importance. It is, therefore, not without reason that accompanying the rapid proliferation of genetic knowledge, a parallel increase has occurred in the technical nomenclature and terminology special to the field of genetics and often special to specific branches of genetics. In preparing this glossary of ca. 2500 entries, we have attempted to compile and collate the terminology from seemingly unrelated, widely separated branches of genetics - classical and molecular; microbial and human; cytogenetics and population genetics. We have not been content merely to collect terms and definitions much as is found in a dictionary. Rather our aim has been to provide material suitable and usable both for students

and research workers. Accordingly, depending upon our evaluation, some terms have simply been defined, others have been described at some length even to the extent of providing experimental data.

*Chromosomal Variation in Man* - Digamber S. Borgaonkar 1977

Over 1500 entries to literature (mostly English-language journal articles). Sources were Current contents, various genetics journals, *Excerpta medica*, and *Index medicus*. Entries arranged under sections titled Structural variations and anomalies, Numerical anomalies, and Chromosome breakage syndromes. Author, selected syndrome index.

**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.

*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.

Understanding the Effects of Sex Chromosomes and Sex Hormones on Sex Differences - Qinze Arthur Zhang 2021

Biological factors that determine sex and drive sexual differentiation have long been established in humans and other mammalian species. Yet there is a limited understanding of the mechanisms by which these factors give rise to sex-biased outcomes in health and disease. Growing evidence for sex differences in preclinical and clinical studies has led to research funding regulations that require clinical studies and drug trials to account for sex as a biological variable. This new wave of interest in sex differences is focused on three aspects: First, what factors show quantitative sex differences at the molecular and cellular levels, and how are they regulated, over the course of a lifetime, by the classic sex-biasing factors, i.e., genes encoded on the sex chromosomes and sex steroid hormones secreted by gonads. Second, can we utilize knowledge of sex differences (e.g., sex-biased gene expression) gained from studying model animals to generate and test hypotheses regarding these sex-driven variables in a multitude of biological functions of interest. Third, can we translate basic knowledge of sex differences into new therapeutic targets/strategies that alleviate sex-biased outcomes in common diseases, developing sex-specific treatments or extending sex-biased protection from one sex to the other. In this thesis, I will first review the fundamental causes

of sex differences and the classical strategies used to study sex-biased phenotypes. Next, I will introduce existing mice models (i.e., the "Four-Core-Genotype", XY\* and Sex Chromosome Trisomy models) that aid in delineating the effects of sex hormones and sex chromosomes on sex differences. Finally, I will highlight the insights gained from recent studies using these mouse models and discuss the need for a "systems approach" in future studies of sex differences. (276 words.)

**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.

**Understanding Down Syndrome** - Cliff Cunningham 1996

Using positive, readable language, this book helps parents understand Down syndrome  
The Chromosome Disorders - Gordon Howard Valentine 1975

**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.

**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.

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