

# Sex Chromosomes Abnormalities

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.

**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.

**Chromosome Abnormalities in Domestic Animals as Causes of Disorders of Sex Development Or Impaired Fertility** - Izabela Szczerbal 2016

Cytogenetic evaluation is an important step in the diagnosis of infertile or sterile animals. Moreover, the analysis of sex chromosomes is crucial for a proper classification of disorders of sex development (DSD). For many years, chromosome studies mainly addressed the livestock species, while recently, increasing interest in such analysis in companion animals is observed. New molecular and cytogenetic tools and techniques have given opportunities for a precise identification of chromosome mutations. Among them, fluorescence in situ hybridization, besides chromosome banding, has become a gold standard. In this chapter, recent advances in the cytogenetic diagnosis of cattle, pigs, horses, dogs and cats are presented.

*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.

**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.  
*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.  
*Abnormalities of the Sex Chromosome Complement in Man* - Medical Research Council (Great Britain) 1964

*The Chromosome Disorders* - Gordon Howard Valentine 1969

*Gender and Genes* - Klasien Horstman 2013

This Yearbook of Women's History (Jaarboek voor Vrouwengeschiedenis)

is dedicated to Gender and Genes. Intruding upon our everyday lives, the world of DNA, genes and genomics has become a challenging field of research, both clinical and biomedical as well as socio-cultural. It is also a challenging topic for a Yearbook which traditionally focuses on women and gender from a historical point of view. Gender issues are part and parcel of genes and genomics in scientific research and socio-cultural discourses and representations. Current literature on genes and genomics does not abound in analyses of biomedical and socio-cultural realms where gender aspects are played out and exchanged. This Yearbook may thus contribute to a field of analysis which contextualizes history from the viewpoint of current biotechnological developments. This volume contains articles on medical cases (reproductive testing and the case of the sex chromosomes, and framing cancer risk in women and men), cultural representations, a portrait of female scientist Rosalind Franklin and interviews with feminist science philosophers Katarina Karkazis and Donna Dickenson.

**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.

**The Y Chromosome: Clinical aspects of Y chromosome abnormalities** - 1985

**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.

*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.

*Chromosomal Abnormalities* - 1972\*

**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 Chromosomes** - Cynthia N. Weingarten 2009

Sex chromosomes determine the sex of an organism. A human somatic cell has two sex chromosomes: XY in male and XX in female. A human germ cell has one sex chromosome: X or Y in a sperm and X in an egg. When an X-sperm is combined with an egg, the resulting zygote (fertilised egg) will contain two X chromosomes. A person developed from the XX-zygote will have the characteristics of a female. Combination of a Y-sperm and an egg will produce a male. Usually, a woman has two X chromosomes (XX) and a man one X and one Y (XY). However, both male and female characteristics can sometimes be found in one individual, and it is possible to have XY women and XX men. Analysis of such individuals has revealed some of the molecules involved in sex determination, including one called SRY, which is important for testis formation. SRY (which stands for sex-determining region Y gene) is found on the Y chromosome. In the cell, it binds to other DNA and in doing so distorts it dramatically out of shape. This alters the properties of the

DNA and likely alters the expression of a number of genes, leading to testis formation. Most XX men who lack a Y chromosome do still have a copy of the SRY gene on one of their X chromosomes (moved there by chromosomal translocation). This copy accounts for their maleness. However, because the remainder of the Y chromosome is missing they frequently do not develop secondary sexual characteristics in the usual way. This book presents the latest research in the field from around the world.

**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.

**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.

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

**Human Chromosomal Abnormalities: Introduction to Common Gross Defects of Human Chromosomes** -

Dennis O'Neil presents an online tutorial on the common defects of human chromosomes. The tutorial offers an overview, information about detection, and descriptions of common, autosomal, and sex chromosome abnormalities. O'Neil provides access to learning activities, a glossary, links to additional resources. and illustrations of sex organs and diagnostic tests.

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