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Cytogenetic and Genome Research

publishes high quality original reports and reviews covering all aspects of chromosomes, genes and genomes in man, animals and plants.

Categories of publications in conventional issues Original Articles are reports on Human, Animal and Plant Cytogenetics Cancer Cytogenetics Molecular Cytogenetics Gene Mapping Cloning and Sequencing Gene Characterization Comparative Gene Mapping Somatic Cell Genetics Genes and Diseases Developmental Genetics Epigenetics. Other categories closely related to the above topics could be considered by contacting the editors.

Reviews covering a timely topic by experts in the field are either invited by the Editors or may be submitted for consideration.

Short Reports must present results of sufficient importance to justify accelerated acceptance. They should not be longer than 4 printed pages, including figures, tables and references.

Human Cytogenetics Case Reports are for de novo observations, particularly such which apply the use of HGMP (Human Genome Mapping Project) resources to the detailed characterization of chromosome rearrangements thereby further highlighting the nature of the abnormalities being described.

Cytogenetics of Rare or Endangered Species with straightforward karyotype description of animals or plants should not be longer than 3 printed pages, including figures, tables and references.

Chromosome and Gene Workshops or Reports compile data that have been collected for single chromosomes, genes or genomes, hereditary diseases and gene-phenotype correlations in man, animals and plants. Illustrations and tables may be included.

Collaborative and Interactive Research Projects are contributions from investigators who are in need of research materials, or need the assistance of colleagues with specialized expertise, or who have data that is inadequate for a full report but which could be published when combined with data of others.

Commentaries are for observations, opinions, and comments outside the realm of conventional scientific papers. Original data, illustrations and tables may be included.

Single topic issues The journal has a tradition in publishing a series of themed issues. For these volumes we are soliciting the assistance of one or more expert investigators to act as Guest Editors in the area that is particularly interesting and/or one in which there is a need for a thorough overview. The Guest Editors invite top researchers to contribute original research reports or reviews of a topic that is in their main area of interest. These manuscripts are peer reviewed the same way as the papers received for the conventional issues. Proposals clearly outlining a theme and nominating potential Guest Editors are welcome.

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Arrangement Title page: The first page should give the full names of the authors and their affiliations, the full postal address, telephone and fax numbers, as well as the e-mail address of the corresponding author.

Abstracts should be provided for all Original Articles, Reviews, Human Cytogenetics Case Reports, and Cytogenetics of Rare or Endangered Species.

Figures: Digital illustrations should be sharp with good contrast and color rendition. Resolution for all graphics should be at least 300 dots per inch. We request that all illustrations be in a common format such as .jpg (.jpeg .jpgcs .tif .tiff .eps .ps .png).

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Guidelines set forth by the HUGO Gene Nomenclature Committee are available at www.genenames.org/guidelines.html New symbols and names for genes can be requested electronically through the online gene symbol request form at http://www.genenames.org/cgi-bin/hgnc_request.pl

Animal gene symbols: Authors submitting material on mouse and rat genetics should obtain correct genetic nomenclature before publication.

Lois Maltais MGD Nomenclature Coordinator The Jackson Laboratory 600 Main Street Bar Harbour, ME 04609, USA Tel.: +1 (207) 288 6429 Fax: +1 (207) 288 6132 E-Mail: nomen@informatics.jax.org MGD home page: www.informatics.jax.org Guidelines set forth by the International Committee on Standardized Genetic Nomenclature for Mice are available at www.informatics.jax.org/mgihome/nomen/table.shtml New symbols and names for genes can be requested electronically through the on-line symbol registry form at www.informatics.jax.org.

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20 short talks will be selected among the abstracts submitted to the meeting.
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Prenatal screening by cell-free (cf) DNA in maternal plasma has opened a new landscape for fetal medicine. Large clinical series have demonstrated that cfDNA analysis of maternal blood can achieve trisomy 21 detection rate of 99% for false-positive rate below 0.1%, which is a great improvement upon the current screening strategies. However, there is a need to define the optimal way of combining cfDNA testing with other first trimester exams. Likewise, several commercial cfDNA tests have rapidly become available, but their main features are not always readily comprehensible for clinicians. In this special issue, we combine a practical overview from a clinical perspective with meta-analysis and theoretical models that help explain the impact of introducing cfDNA testing, along with a number of important recent research contributions. This issue offers clinicians involved in fetal medicine up-to-date and useful information about the current state of cfDNA testing and how to use it in their daily practice.

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Exploring vertebrate sexual differentiation

Sexual Differentiation of Vertebrate Reproductive Organs

Editors
Marilyn B. Renfree
Humphrey H.-C. Yao

This special topic issue on Sexual Differentiation of Vertebrate Reproductive Organs continues and complements the story that was first taken up in a previously published issue dealing with the Control of Gonadal Development. This new publication focuses on the transition from sex determination to sexual differentiation. Due to the fact that most work has been conducted on mice, it is not surprising that many generalisations have developed, even though only some of these apply to all mammals. This special topic issue aims to shed some light on these generalisations. It covers the different mechanisms of sex determination in mice, reptiles and birds. Further insights are provided on the somatic lineages, gonadal differentiation and disorders of sexual development in mammals, fishes and humans. The last few chapters focus on the development and molecular control of the duct system and the development of external genitalia in mammals and also discuss how these evolved in reptiles.

This publication is recommended for anyone who would like to know more about what directs male and female sexual dimorphisms at molecular, cellular, developmental, physiological and endocrinological levels.

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An in-depth compendium of current knowledge of chromosome dynamics and arrangement in bacteria and archaea

Bacterial Chromosomal Segregation

Editor
Peter L. Graumann

All cellular life depends on the accurate duplication and partitioning of the genome. It has become clear in recent years that the chromosomes of many bacterial species show a well-defined arrangement, and their layout, integrity and segregation is supported by a variety of conserved proteins. The architecture of the chromosome also has a large impact on global transcription, and proteins involved in the topology of the DNA play major roles in chromosome segregation and compaction. In this special issue, the molecular biological principles of these fundamental processes are described in several model and non-model bacteria.

Providing an in-depth compendium of our current knowledge of chromosome dynamics and arrangement in bacteria and archaea, this publication is a must-read for any researcher studying or teaching bacterial genetics or physiology.

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Editor: Graumann, P.L. (Marburg)
142 p., 34 fig., 12 in color, soft cover, 2015
CHF 45.00 / EUR 38.00 / USD 53.00 (soft cover)/
CHF 54.00 / EUR 46.00 / USD 64.00 (online)
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Cutting-edge advances in the embryology, epidemiology, genetics, diagnosis, imaging, and therapy of congenital heart disease

Congenital Heart Disease

Molecular Genetics, Principles of Diagnosis and Treatment

Editors
Maximilian Muenke
Paul S. Kruszka
Craig A. Sable
John W. Belmont

This cutting-edge book encompasses the latest advances in the embryology, epidemiology, genetics, diagnosis, imaging, and therapy of congenital heart disease. The international cast of authors are leaders in their fields and have combined their talents to produce a unique and expert perspective on congenital heart disease. The work is of interest to pediatricians, internal medicine specialists, medical geneticists, both pediatric and adult cardiologists, embryologists, imaging physicians, and cardiac surgeons. This book arrives at a very exciting time as new genetic, imaging, and therapeutic developments are changing the field of congenital heart disease. It takes the reader on a journey that begins with a historical overview of congenital cardiovascular anomalies and ends with developments in stem cell and tissue engineering. In between these chapters are destinations that include cardiac embryogenetics, epidemiology, genetic syndromes associated with cardiovascular anomalies, single-gene disorders, cardiac imaging, surgical and interventional therapies, and ethical considerations. Congenital Heart Disease is an invaluable reference. In short, it provides important pearls of wisdom to create a comprehensive reference for all physicians involved with congenital heart disease.

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