European Human Genetics Conference Eshg 2017

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European Human Genetics Conference 2000 SICS Editore
Biobanking, i.e. storage of biological samples or data emerging from such samples for diagnostic, therapeutic or research purposes, has been going on for decades. However, it is only since the mid 1990s that these activities have become the subject of considerable public attention, concern and debate. This shift in climate is due to several factors. The purpose of this book is to investigate some of the ethical, legal and social challenges raised by research biobanking in its different modern forms and formats. The issues raised by research biobanking in its modern form can be divided into four main clusters: how biological materials are entered into the bank; research biobanks as institutions; under what conditions researchers can access materials in the bank, and problems concerning ownership of biological materials and of intellectual property arising from such materials; and how the information is collected and stored, e.g. access-rights, disclosure, confidentiality, data security and data protection.
Human Reproductive Genetics CRC Press
With advances in personalised medicine, the field of medical law is being challenged and transformed. The nature of the doctor-patient relationship is shifting as patients simultaneously become consumers. The regulation of emerging technologies is being thrown into question, and we face new challenges in the context of global pandemics. This volume identifies significant questions and issues underlying the philosophy of medical law. It brings together leading philosophers, legal theorists, and medical specialists to discuss these questions in two parts. The first part deals with key foundational theories, and the second addresses a variety of topical issues, including euthanasia, abortion, and medical privacy. The wide range of perspectives and topics on offer provide a vital introduction to the philosophical underpinnings of medical law.
Science, Ethics, and Governance RCOG
This volume contains several analyses of health rights issues related to children. The various chapters provide an overview of this captivating area and may be of special interest to lawyers, health care professionals, ethicists, psychologists, judicial institutions, policy makers, interest groups, students and all others who are concerned with the children’s rights perspective on health care.
32nd Annual Meeting of the ESHG : Amsterdam, the Netherlands, Saturday-Tuesday, 27 May-30 May 2000 : Final Programme and Abstracts Springer
Infertility affects more than one in ten couples worldwide and is related to highly heterogeneous pathologies sometimes only discernible in the germ line. Its complex etiology often, but not always, includes genetic factors besides anatomical defects, immunological interference, and environmental aspects. Nearly 30% of infertility cases are probably caused only by genetic defects. Thereby experimental animal knockout models convincingly show that infertility can be caused by single or multiple gene defects. Translating those basic research findings into clinical studies is challenging, leaving genetic causes for the vast majority of infertility patients unexplained. Nevertheless, a large number of candidate genes have been revealed

by sophisticated molecular methods. This book provides a comprehensive overview on the subject of infertility written by the leading authorities in this field. It covers topics including basic biological, cytological, and molecular studies, as well as common and uncommon syndromes. It is a must-read for human geneticists, endocrinologists, epidemiologists, zoologists, and counsellors in human genetics, infertility, and assisted reproduction.
The 2019 MDPI Writing Prize Academic Press
The 2016 edition of the International System for Human Cytogenomic Nomenclature (ISCN 2016) offers standard nomenclature that is used to describe any genomic rearrangement identified by techniques ranging from karyotyping to FISH, microarray, various region specific assays, and DNA sequencing. Suggestions from the international cytogenetics community have been reviewed by the Standing Committee, an international group of experts, nominated by their peers. This updated edition offers: * many new examples, particularly for microarray and region specific assays * trackable changes in the main text compared to the previous edition for easier identification * a nomenclature standard to facilitate the description of chromosome rearrangements characterized by DNA sequencing developed through collaboration between the Human Genome Variation Society (HGVS) and ISCN to accommodate the increased use of sequencing technologies in the characterization of chromosomal abnormalities The ISCN 2016 is an indispensable reference volume for human cytogeneticists, molecular geneticists, technicians, and students for the interpretation and communication of human cytogenetic and molecular cytogenomic nomenclature. After a long collaboration with Cytogenetic and Genome Research, ISCN is now again a part of this leading journal on chromosome and genome research, combining the day-to-day business with the latest findings.
Rare Diseases Epidemiology: Update and Overview Emerald Group Publishing
The majority of patients with myopathies have an inherited disease. Symptoms alone are not usually enough to diagnose myopathy, but they warrant further neurological examinations that are performed in larger hospitals and in special outpatient clinics for neuromuscular disorders.
Genetic Counseling Research Karger Medical and Scientific Publishers
Genome editing is a powerful new tool for making precise alterations to an organism's genetic material. Recent scientific advances have made genome editing more efficient, precise, and flexible than ever before. These advances have spurred an explosion of interest from around the globe in the possible ways in which genome editing can improve human health. The speed at which these technologies are being developed and applied has led many policymakers and stakeholders to express concern about whether appropriate systems are in place to govern these technologies and how and when the public should be engaged in these decisions. Human Genome Editing considers important questions about the human application of genome editing including: balancing potential benefits with unintended risks, governing the use of genome editing, incorporating societal values into clinical applications and policy decisions, and respecting the inevitable differences across nations and cultures that will shape how and whether to use these new technologies. This report proposes criteria for heritable germline editing, provides conclusions on the crucial need for public education and engagement, and presents 7 general principles for the governance of human genome editing.
Precision Public Health Encyclopaedia Britannica
This remarkable publication focuses on the importance of genetics in mental retardation, investigating the extent to which molecular diagnostic capability and the understanding of genetic causes have improved over recent years. As a result, clinical evaluation and diagnostic laboratory practice are now undergoing an unprecedented period of change.In a single volume, a unique combination of key individuals and world-class clinical, diagnostic and research-based experts share specialized, state-of the-art knowledge in this field. The parents' perspective lies behind chapters dealing with issues such as:- Classification nomenclature- Well-known syndromes- How modern technologies have resulted in newly identified syndromes- How genome architecture can influence disease- Guidelines for clinical evaluation- Valuable database resources for clinical, diagnostic and research departments- Challenges involved in data interpretation and determining clinical relevance- Genetic overlaps with autism and schizophrenia- Processes of health service implementationGenetics of Mental Retardation is an invaluable resource for researchers and students with an active interest in the field. Furthermore, consultants and trainees in clinical genetics and pediatrics, and researchers working in clinical genetics laboratories will benefit from these reviews.
CRISPR-Cas Systems Academic Press
Molecular Diagnostics, Third Edition, focuses on the technologies and applications that professionals need to work in, develop, and manage a clinical diagnostic laboratory. Each chapter contains an expert introduction to each subject that is next to technical details and many applications for molecular genetic testing that can be found in comprehensive reference lists at the end of each chapter. Contents are divided into three parts, technologies, application of those technologies, and related issues. The first part is dedicated to the battery of the most widely used molecular pathology techniques. New chapters have been added, including the various new technologies involved in next-generation sequencing (mutation detection, gene expression, etc.), mass spectrometry, and protein-specific methodologies. All revised chapters have been completely updated, to include not only technology innovations, but also novel diagnostic applications. As with previous editions, each of the chapters in this section includes a brief description of the technique followed by examples from the area of expertise from the selected contributor. The second part of the book attempts to integrate previously analyzed technologies into the different aspects of molecular diagnostics, such as identification of genetically modified organisms, stem cells, pharmacogenomics, modern forensic science, molecular microbiology, and genetic diagnosis. Part three focuses on various everyday issues in a diagnostic laboratory, from genetic counseling and related ethical and psychological issues, to safety and quality management. Presents a comprehensive account of all new technologies and applications used in clinical diagnostic laboratories Explores a wide range of molecular-based tests that are available to assess DNA variation and changes in gene expression Offers clear translational presentations by the top molecular pathologists, clinical chemists, and molecular geneticists in the field
Understanding Disease Genetics Using Genomic Approaches Routledge
CRISPR/Cas is a recently described defense system that protects bacteria and archaea against invasion by mobile genetic elements such as viruses and plasmids. A wide spectrum of distinct CRISPR/Cas systems has been identified in at least half of the available prokaryotic genomes. On-going structural and functional analyses have resulted in a far greater insight into the functions and possible applications of these systems, although many secrets remain to be discovered. In this book, experts summarize the state of the art in this exciting field.
The Ethics of Research Biobanking Demos Medical Publishing
This book, written by a leading geneticist, examines the ethical and social issues raised by the genetic testing of children. The opinions of geneticists, ethicists and affected families are all included to give a balanced view of this controversial field. Issues covered include confidentiality, potential abuses of genetic information (eg the use of test results by insurance companies) and the value of predictive genetic testing. The aim of the book is to improve awareness of the complexity of the issues raised and provide suggestions as to how the discussions must develop - it therefore raises new questions as well as answering those that already exist.
Genetic Testing and Gene Therapy European Human Genetics Conference 200032nd Annual Meeting of the ESHG.European Human Genetics Conference 200032nd Annual Meeting of the ESHG : Amsterdam, the Netherlands, Saturday-Tuesday, 27 May-30 May 2000 : Final Programme and AbstractsHistory of Human GeneticsAspects of Its Development and Global Perspectives
Analogies play a fundamental role in science. To understand how and why, at a given moment, a certain analogy was used, one has to know the specific, historical circumstances under which the new idea was developed. This historical background is never presented in scientific articles and quite rarely in books. For the general reader, the undergraduate or graduate student who learns the subject for the first time, but also for the practitioner who looks for inspiration or who wants to understand what his colleague working in another field does, these historical circumstances can be fascinating and useful. This book discusses a series of analogy effects in subatomic physics, the prediction and theory of which the author has contributed to in the last 50 years. These phenomena are presented at a level accessible to the non-specialist, without formulae but with emphasis on the personal and historical background: memoirs of meetings, discussions and correspondence with collaborators and colleagues. As such, besides its scientific aspects, the book constitutes an absorbing witness account of a holocaust survivor who subsequently illegally crossed the Iron Curtain to escape communist persecution.
European Human Genetics Conference 2000 Oxford University Press (UK)
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.
Returning Individual Research Results to Participants Karger Medical and Scientific Publishers
The introduction and widespread implementation of newborn bloodspot screening (NBS) for cystic fibrosis (CF) has offered earlier diagnosis and better outcomes for children with CF in many countries of the world. It represents a paradigm shift in the diagnostic pathway for these families. In contrast to a clinical diagnosis, infants are now referred for diagnostic testing after a positive NBS result. The introduction of NBS has enabled the provision of early appropriate treatment to prevent the manifestations of the disease. In the near future, early diagnosis will facilitate the prompt use of new CFTR modulator therapies that correct the basic underlying molecular defect. NBS for CF has been a global success but continues to raise questions with many varied approaches and the development of new technologies, in particular the ability to undertake extensive gene examination. Which is the best protocol to achieve high sensitivity and specificity, and how to evaluate and manage infants with inconclusive diagnosis are all subjects of ongoing discussion. It is also open to question: what is the best approach to informing and counselling the parents about a positive or inconclusive NBS result? These questions are not easy to answer and require a balanced solution that reflects the local health care system and may appropriately result in different answers around the globe. The articles in this book try to answer these questions and give an overview of the current state of knowledge in NBS for CF.
Inherited Metabolic Epilepsies Garland Science
"Human DNA: Law and Policy" provides the first international debate on a topic of universal concern. No book has brought together such a diverse range of multidisciplinary ethical and legal expertise on the highly controversial issues surrounding the use, storage, exchange and sale of the very stuff' of which we are made - human genetic material. Testing of human genetic material involves a variety of samples (pathological samples, newborn screening samples, samples leftover' after testing, and research samples), shared around the world. This places consent issues on an individual, familial, and societal level. The comparative and international perspectives presented reveal the transnational nature of genetic studies. This book focuses on the issues of DNA sampling and testing, consent and confidentiality, banking policies, genetic epidemiology and diversity. Since financial and technological pressures are inextricably linked to human genetics research, commercialization and patents are also examined. Academic researchers, policy makers and industry will benefit from the learned papers and reports of the discussion, which is rich in diversity of opinion, controversial in the diversity of policy and approaches presented, anchored on scientific facts and yet sensitive to cultural, political and economic differences.
Guidance for a New Research Paradigm Pergamon
This book presents applications of bioinformatics tools that experimental research scientists use in "daily practice." Its interdisciplinary approach combines computational and experimental methods to solve scientific problems. The book begins with reviews of computational methods for protein sequence-structure-function analysis, followed by methods that use experimental data obtained in the laboratory to improve functional predictions.
RNA-mediated Adaptive Immunity in Bacteria and Archaea National Academies Press
Precision Public Health is a new and rapidly evolving field, that examines the application of new technologies to public health policy and practice. It

draws on a broad range of disciplines including genomics, spatial data, data linkage, epidemiology, health informatics, big data, predictive analytics and communications. The hope is that these new technologies will strengthen preventive health, improve access to health care, and reach disadvantaged populations in all areas of the world. But what are the downsides and what are the risks, and how can we ensure the benefits flow to those population groups most in need, rather than simply to those individuals who can afford to pay? This is the first collection of theoretical frameworks, analyses of empirical data, and case studies to be assembled on this topic, published to stimulate debate and promote collaborative work.

Summary of a Workshop Academic Press

This volume focuses on critical issues surrounding the intersection of genetics, health, and society. It provides a critical examination of sociological and biomedical approaches to genomics, including strengths and limitations of each perspective.

The Genetic Testing of Children Springer Science & Business Media

The 2019 MPDI Writing Prize invited early stage researchers who are not native English speakers to write on the subject of "how research should be evaluated and how researchers should be rewarded". Six prizes were awarded, however there were many more entries. This book collates many of those entries and contains inspiring, thought-provoking and original viewpoints of open science through the eyes of those conducting research on a daily basis.

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Genomic and Personalized Medicine Academic Press

Cytogenomics demonstrates that chromosomes are crucial in understanding the human genome and that new high-throughput approaches are central to advancing cytogenetics in the 21st century. After an introduction to (molecular) cytogenetics, being the basic of all cytogenomic research, this book highlights the strengths and newfound advantages of cytogenomic research methods and technologies, enabling researchers to jump-start their own projects and more effectively gather and interpret chromosomal data. Methods discussed include banding and molecular cytogenetics, molecular combing, molecular karyotyping, next-generation sequencing, epigenetic study approaches, optical mapping/karyomapping, and CRISPR-cas9 applications for cytogenomics. The book’s second half demonstrates recent applications of cytogenomic techniques, such as characterizing 3D chromosome structure across different tissue types and insights into multilayer organization of chromosomes, role of repetitive elements and noncoding RNAs in human genome, studies in topologically associated domains, interchromosomal interactions, and chromoanagenesis. This book is an important reference source for researchers, students, basic and translational scientists, and clinicians in the areas of human genetics, genomics, reproductive medicine, gynecology, obstetrics, internal medicine, oncology, bioinformatics, medical genetics, and prenatal testing, as well as genetic counselors, clinical laboratory geneticists, bioethicists, and fertility specialists. Offers applied approaches empowering a new generation of cytogenomic research using a balanced combination of classical and advanced technologies Provides a framework for interpreting chromosome structure and how this affects the functioning of the genome in health and disease Features chapter contributions from international leaders in the field