Public Health Genomics
Despite the fact that the prevalence of obesity in early childhood has been stable and is no longer increasing in many developed and industrialized countries, the incidence of both obesity and full-blown metabolic syndrome in children and adolescents is still very high. Obesity is a major disease burden in all societies and needs to be prevented early in life. New approaches are eagerly sought and absolutely necessary.

This book presents a comprehensive and state-of-the-art summary of current and new knowledge in this critical field. Crucial issues such as nutrition and genetics are described in detail. In addition, new ideas such as e-health and the consequences of urban living conditions are explored. Last but not least, modern treatment concepts and prevention even at an early age are competently discussed.

Offering a valuable update on new developments in obesity research and the treatment in children and adolescents, this book is essential reading for all pediatricians and health-care professionals who look after young patients on a regular basis.
Public Health Genomics

Founded 1998 as ‘Community Genetics’ by Leo ten Kate (1998–2008)
Continued by B.M. Knoppers (2009–2011), M. Gwinn (2012–2013) and
A.M. Brand as ‘Public Health Genomics’.

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• Reviews
• Short Communications
• Policy Statements

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Chlamydial Infection: A Clinical and Public Health Perspective

Editor: Carolyn M. Black

Chlamydiae are obligate intracellular bacteria that cause one of the most common sexually transmitted infectious diseases in the world. The infection disproportionately impacts women and the highest prevalence of infection is found in adolescents. Most chlamydial infections are asymptomatic. Untreated infections are sources of further spread of infection and can lead to serious consequences including pelvic inflammatory disease, infertility and chronic pelvic pain. Chlamydial infections also increase a person’s susceptibility to HIV and other STDs. Featuring contributions by internationally recognized experts in epidemiology, infectious disease research and chlamydial biology, this book provides up-to-date reviews from a clinical and public health perspective on chlamydia epidemiology and control programs, genomics and pathogenicity, diagnosis, treatment, host immune responses, and the latest on the search for an effective vaccine. Also included are chapters on the impact of chlamydial infection on specific populations such as the lesbian, gay, bisexual and transgender community, and an update on the outbreak in Europe of the invasive chlamydial infection, lymphogranuloma venereum or LGV. This comprehensive publication is intended for clinicians, public health workers and scientists with interest in sexually transmitted diseases, medical microbiology, infectious diseases and clinical research.
Research of Rare and Orphan Diseases

RE(ACT), International Congress on Research of Rare and Orphan Diseases

Editor
Olivier Menzel

The medical community does not see rare diseases as a field of priority. In fact, poor characterization of the natural causes, pathologies and low numbers of cases make diagnosis difficult, often resulting in a real ordeal for patients and their families. This special issue of Molecular Syndromology presents six selected contributions of the First International Congress on Research of Rare and Orphan Diseases which brought together courageous researchers working on rare and orphan diseases. It shows the growing research interest in rare and orphan diseases, the new experimental approaches, such as next generation sequencing and therapeutic possibilities. Topics outlined in this issue are:

- Current debates on the emerging technical, medical and ethical issues
- Potential optimum use of the available technology
- Molecular genetics of Charcot-Marie-Tooth diseases
- Amelogenesis imperfecta – a clinically and genetically heterogeneous group of inherited defects of enamel formation
- Smith-Lemli-Opitz syndrome – an autosomal recessive disorder characterized by multiple congenital abnormalities and mental retardation
- Strategies for therapeutic suppression of nonsense mutations in inherited metabolic diseases

Furthermore, the involvement of patient organizations in the development of orphan drugs for rare diseases is emphasized. This volume is essential reading for all researchers in the fields of human genetics and syndromology and everyone interested in the study of rare diseases.

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Consanguinity and Genomics

Guest Editors

Giovanni Romeo

Alan Bittles

When researching the genetics of consanguineous marriages, the medical and scientific community continues to pay attention mainly to rare autosomal recessive diseases. But from an overall population and health perspective, consanguinity is a much more extensive and complex topic that is influenced by major social, economic, and demographic factors. Genome analysis is, however, the principal unifying theme in this special topic issue since next-generation sequencing has become increasingly indispensable for profiling populations and diagnosing diseases, and thus is central to investigations into the biological and health outcomes arising as a consequence of consanguinity. With some 1.1 billion people living in countries, mainly in North and Sub-Saharan Africa, the Middle East, and West, Central and South Asia, where 20–50% of marriages are between close kin, consanguinity is a topic of major contemporary significance. Written by experts in the field, this thematically diverse collection of papers provides new and exciting insights into the rationale for and outcomes of consanguineous marriage.

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Understanding disease by focusing on genome-mediated somatic cell evolution

Genetic Heterogeneity and Human Diseases

Editor
Henry Heng

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The contribution of karyotype heterogeneity to human diseases is a crucial but overlooked issue. Recent genomic research has revealed high levels of genetic/epigenetic heterogeneity, in particular karyotypic heterogeneity and somatic mosaicism associated with many complex but common human diseases. These important findings challenge the current gene-based concept of many common diseases. To frame this new emerging field, this publication presents pertinent examples linking karyotype heterogeneity to diseases and identifying it in the general population. Specifically, a few key topics essential to understanding karyotypic heterogeneity are discussed, including genomic instability, non-clonal chromosome aberrations, previously unreported/ignored types of chromosome aberrations, cell death heterogeneity and somatic mosaicism. These subjects are discussed with an emphasis on determining the biological implications of genomic heterogeneity and synthesizing these implications into the frameworks of systems biology and genome theory.
Over the past decade, there has been unprecedented progress in our understanding of the genetic basis of type 2 diabetes driven largely by technological advances which have facilitated genome-wide studies. Bringing together comprehensive synopses written by leaders in the field, this book discusses the recent progress in defining the genetic basis for type 2 diabetes and related traits.

The opening section focuses on the impact of genome-wide association studies on our understanding of the genetic landscape of type 2 diabetes, whilst later chapters explore how variants affecting relevant metabolic traits influence diabetes susceptibility. The impact of next-generation sequencing on gene discovery efforts in both type 2 diabetes and mendelian forms of diabetes and insulin resistance is covered in detail. Particular attention is also given to efforts to increase our understanding of sequence annotation in diabetes-relevant tissues and how this can be harnessed to leverage molecular mechanisms for genetic association signals. Finally, efforts to translate this improved knowledge into both molecular mechanisms and clinical care are discussed.

Providing a comprehensive overview of the current knowledge of the genetic basis for type 2 diabetes and related traits, this book is essential reading for researchers with an interest in the etiology of diabetes and human genetics.
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A One-Page Summary Report of Genome Sequencing for the Healthy Adult