# Prenatal Diagnosis and Reproductive Genetics

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# **Prenatal Diagnosis And Reproductive Genetics**

Sean Kehoe,Lyn Chitty,Tessa Homfray

# **Prenatal Diagnosis And Reproductive Genetics:**

Women and Prenatal Testing Karen H. Rothenberg, Elizabeth Jean Thomson, 1994 For pregnant women in the 1990s technological developments have ushered in new and expanding reproductive genetic testing options Some herald these procedures as advances providing women with previously unavailable information about their pregnancies Others contend that with this surge of information come increasing and perhaps unwarranted obligations while some women have greater knowledge about their pregnancies they also face far more complex decisions and a greater pressure to do as much as is technologically possible to ensure the birth of a healthy child This book focuses on the major women s issues surrounding the development and application of reproductive genetic testing Although much has been written about the biological safety and efficacy of these technologies few publications have addressed their psychological sociocultural ethical legal and political impact on women and their experience of pregnancy The first of three sections provides the contextual framework in which the debate should be analyzed The second section sets forth the philosophical foundations and complex ethical and legal questions that need to be addressed and the final section delineates a variety of perspectives on the psychological and sociocultural issues raised by reproductive genetic testing These fourteen essays on the cutting edge of the debate are essential reading for anyone interested in women s studies human genetics health law and bioethics and prenatal care providers BOOK JACKET Title Summary field provided by Blackwell North America Inc All Rights Reserved

**Reproductive Genetics** Sean Kehoe, Lyn Chitty, Tessa Homfray, 2009-11-01 Great progress has been made in genetics in recent years with significant consequences for the field of reproductive genetics. The ability to identify genetic defects before implantation to diagnose fetal abnormalities and to introduce screening programmes means that genetic testing now has a major role in preventive medicine The 57th RCOG Study Group brought together a range of experts to examine the many clinical ethical and societal issues around reproductive genetics This book presents the findings of the Study Group covering a range of disorders advances in treatments and techniques as well as the importance of educating the public and the potential ethical dilemmas that may arise as a result of new scientific findings and new methodologies Born Well: Prenatal Genetics and the Future of Having Children Megan A. Allyse, Marsha Michie, 2021-11-24 This book brings together an international collection of experts in reproductive ethics law disability studies and medicine to explore the challenging future of reproduction and children From the medical to the social and from the financial to the legal the authors explore the expanding impact of reproductive genetics on our society New advances in genetic technologies are revolutionizing the practice of reproductive medicine We have expanded our ability to detect genetic changes in embryos and fetuses in ways that potentially allow to identify treat or prevent a growing range of diseases The development of gene editing technologies raises questions about the possibility of removing disease causing variants from embryos before pregnancy implantation The growing sophistication of prenatal genomic sequencing offers us glimpses into the whole genome of the developing fetus And the increasingly sophisticated science of gene matching allows us greater and greater foreknowledge of how the genomes of two individuals will combine in a future child This is an indispensable book on the newest developments in bioethics caused by the sciences Chapter 5 is available open access under a Creative Commons Attribution 4 0 International License via link springer com \*\*Reproductive Genetics\*, Gender and the Body\* Elizabeth Ettorre, 2013-04-15 The new reproductive technologies are currently hot topics in medical sociology Our book explores the complexities of genetic technologies with special reference to biomedical prenatal practices It includes material from interviews with doctors lawyers and midwives in Greece The Netherlands Finland and England It is written by a leading figure in the field from a feminist perspective

Reproductive Genetics, an Issue of Obstetrics and Gynecology Clinics Lorraine Dugoff, 2018-03-06 This issue provides a timely update for for the ob gyn on genetics in reproductive medicine Dr Dugoff has created an issue with the goals of providing the most currently clinical information on gentic screening and prenatal genetics Top authors have written reviews on the following topics Genetic counseling overview for the ob gyn Cell free DNA screening for an euploidy Cell free DNA screening for single gene disorders The use of microarray in prenatal diagnosis Whole exome sequencing Applications in prenatal diagnosis Screening for an euploidy in multiple gestations The challenges and available options Expanded carrier screening Ethnicity based carrier screening overview Prenatal genetic diagnosis and prenatal genetic screening Ethical issues in prenatal genetics Ultrasound findings and associated genetic syndromes Hereditary cancers in gynecology and What physicians should know about genetic testing screening and risk reduction Readers will come away with the knowledge they need to diagnose treat and manage patients based on the most current evidence and data Prenatal Diagnosis & **Reproductive Genetics** Jeffrey A. Kuller, Nancy Chescheir, Robert C. Cefalo, 1996 An innovative text in question answer format this text includes medical and surgical fetal treatment preimplantation embryo analysis preimplantation genetics stem cell transplantation fetal cells in maternal circulation embryology and teratology Technologies for Prenatal Diagnosis and Assessment of Genetic Disorders Fan Jin, Yueqiu Tan, Evica Rajcan-Separovic, Peter C. K. Leung, 2020-06-22 Birth defects are one of the major public health concerns in the world as they cause approximately 20% of infant deaths Genetic disorders including chromosome abnormalities and single gene disorders are the most common causes of birth defects for which there is no efficient treatment Prenatal genetic screening and diagnosis allow early identification of affected conceptuses and facilitates reproduction planning or counseling Molecular technologies have developed rapidly in recent years and have been widely used in screening and diagnosis of genetic disorders at all stages of prenatal development e g pre implantation embryonic and fetal However their performance still needs to be validated and assessed as the balance between their advantages and disadvantages need to be discussed With the ability to detect copy number variations CNVs polyploidy uniparental disomy and maternal cell contamination SNP based chromosomal microarray analysis CMA is showing the unique importance in diagnosing chromosomal abnormalities. The interpretation of CNVs remains a challenge however

ultrasound and biochemical screening improve the diagnosis of fetal chromosomal abnormalities Whole exome sequencing WES and whole genome sequencing WGS play increasingly significant roles in prenatal and carrier screening for genetic disorders NGS based non invasive prenatal screening NIPS is now widely used for detecting common autosomal aneuploidies and has shown the potential of detecting microdeletions and microduplications However further investigations of the sensitivity and accuracy are required and large scale data is necessary to evaluate the performance and clinical applications of current and new methods Recently reports of application of newer technologies in prenatal setting became available Examples include third generation sequencing reading the nucleotide sequences at the single molecule level digital PCR used for direct quantification of DNA and cell based NIPT In the followed listed papers the authors showed their successful experiences in identifying novel mutation detecting low level mosaicism or de novo mutations limited in germline cells investigating the association of the CNVs with specific phenotypic alterations by using WES CMA digital PCR and some other new developed molecular techniques More interesting the authors also presented a report about the evaluation of diagnostic yield in fetal WES which suggested a new tendency to apply WES or WGS directly for prenatal diagnosis We believed that the efficiency of scanning causative mutations and prenatal or preimplantion genetic diagnosis for genetic disorders will further improved based on the technologies of whole genomic sequencing with further improved output and resolution New techniques such as quick WES for the newborn in intensive care unit direct WGS for prenatal diagnosis and non invasive test for fetal monogenic disorders will become available in the near future **Prenatal Diagnostic Testing for Genetic Disorders** Gian Carlo Di Renzo, 2023-07-21 This comprehensive volume covers all aspects of the revolution in prenatal diagnosis brought about by the introduction of non invasive prenatal testing NIPT which primarily relies on the detection of free fetal DNA circulating in maternal blood from the early stages of pregnancy The book explores the potential of NIPT to provide full genome screening of the fetus and identify many common or rare disorders. The counseling process as well as the limitations and pitfalls of various techniques used to perform NIPT are described evaluated and critically discussed by renowned international experts The book also compares the new technology with more conventional tests preimplantation diagnosis and the invasive procedures currently in use This book will be a valuable resource for gynecologists obstetricians geneticists maternal fetal medicine specialists pathologists neonatologists reproductive medicine specialists midwives and Reproductive Genetics Sean Kehoe, Lyn Chitty, Tessa Homfray, 2009-11 anyone interested in prenatal genetic diagnosis This book presents the findings of the RCOG Study Group findings on genetics underlying reproductive function Health Care Ethics John F. Monagle, David C. Thomasma, 2005 Provides expert help you need to make difficult bio ethical decisions covering a broad range of current and future health care issues as well as institutional and social issues applicable to multiple disciplines and settings

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