



Prenatal Diagnosis and Reproductive Genetics

Kuller MD, Jeffrey A.

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Prenatal Diagnosis And Reproductive Genetics

**Aleksandra Jezela-Stanek, Sheela
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Hu**

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 *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](https://www.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

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

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 genetic 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 aneuploidy Cell free DNA screening for single gene disorders The use of microarray in prenatal diagnosis Whole exome sequencing Applications in prenatal diagnosis Screening for aneuploidy 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 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 anyone interested in prenatal genetic diagnosis *Human Reproductive Genetics* Juan A. Garcia-Velasco, Emre Seli, 2020-04-19 Human Reproductive Genetics Emerging Technologies and Clinical Applications presents a great reference for clinicians and researchers in reproductive medicine Part I includes a brief background of genetics and epigenetics probability of disease and the different techniques that are being used today for analysis and genetic counseling Part II focuses on the analysis of the embryo current controversies and future concepts Part III comprises different clinical scenarios that clinicians frequently face in practice The increasing amount of genetic tests available and the growing information that patients handle makes this section a relevant part of the fertility treatment discussion Finally Part IV concludes with the psychological aspects of genetic counseling and the role of counselor and bioethics in human reproduction Provides an essential reference for clinicians involved in reproductive medicine Builds

foundational knowledge on new genetic tests coming into the clinical scenario for physicians involved with patients

Assembles critically evaluated chapters that cover basic concepts of genetics and epigenetics and the techniques involved including preimplantation genetic testing controversies and more **Human Reproductive and Prenatal Genetics** Peter C.K. Leung, Jie Qiao, 2018-08-28 Human Reproductive and Prenatal Genetics presents the latest material from a detailed molecular cellular and translational perspective Considering its timeliness and potential international impact this all inclusive and authoritative work is ideal for researchers students and clinicians worldwide Currently there are no comprehensive books covering the field of human reproductive and prenatal genetics As such this book aims to be among the largest and most useful references available Named a Highly Commended book in the Basic and Clinical Sciences by the British Medical Association Features chapter contributions from leading international scientists and clinicians Provides in depth coverage of key topics in human reproductive and prenatal genetics including genetic controls fertilization and implantation in vitro culture of the human embryo for the study of post implantation development and more Identifies how researchers and clinicians can implement the latest genetic epigenetic and omics based approaches **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 **Reproductive Genetics** Sean Kehoe, Lyn Chitty, Tessa Homfray, 2009-11 This book presents the findings of the RCOG Study Group findings on genetics underlying reproductive function 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 preimplantation 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

Reproductive Genetics, Gender and the Body

Elizabeth Ettorre,2013-04-15 This book is all about reproductive genetics a sociological concept developed to define the use of DNA based technologies in the medical management and supervision of reproduction and pregnant women In a searching analysis Elizabeth Ettorre uncovers the hidden social processes involved in the development of these technologies Focussing on prenatal screening she explores how the key concepts of gender and the body are intertwined with the process of building genetic knowledge and some of the unintended consequences for women These include the injection of biology into social relationships and the development of a gendered discourse of shame and stigmatisation in which the perfect body becomes idealised and new conceptions of disability are shaped It becomes clear that the modernist tradition of scientific disinterestedness is being replaced by a new ethic the making of moral judgements by scientists Reproductive Genetics Gender and the Body draws on interviews with European medical legal and nursing professionals and raises important issues around the gendered female body the site of genetic capital It challenges professional and scholar alike to grapple with and think through their responsibilities in this complex field where the competing issues have yet to be resolved

Prenatal Testing and Disability Rights Erik Parens,Adrienne Asch,2000-09-28 As prenatal tests proliferate the medical and broader communities perceive that such testing is a logical extension of good prenatal care it helps parents have healthy babies But prenatal tests have been criticized by the disability rights community which contends that advances in science should be directed at improving their lives not preventing them Used primarily to decide to abort a fetus that would have been born with mental or physical impairments prenatal tests arguably reinforce discrimination against and misconceptions about people with disabilities In these essays people on both sides of the issue engage in an honest and occasionally painful debate about prenatal testing and selective abortion The contributors include both people who live with and people who theorize

about disabilities scholars from the social sciences and humanities medical geneticists genetic counselors physicians and lawyers Although the essayists don't arrive at a consensus over the disability community's objections to prenatal testing and its consequences they do offer recommendations for ameliorating some of the problems associated with the practice

Textbook of Human Reproductive Genetics Stéphane Viville, Karen D. Sermon, 2023-01-05 This book combines genetics reproductive biology and medicine for an integrative view of the emerging specialism of reproductive genetics

The Evolution of Medical Genetics Peter Harper, 2019-10-08 This informative new book presents an accessible account of the development of medical genetics over the past 70 years one of the most important areas of 20th and now 21st century science and medicine Based largely on the author's personal involvement and career as a leader in the field over the last half century both in the UK and internationally it draws on his interest and involvement in documenting the history of medical genetics Underpinning the content is a unique series of 100 recorded interviews undertaken by the author with key older workers in the field the majority British providing invaluable information going back to the very beginnings of human and medical genetics Focusing principally on medically relevant areas of genetics rather than the underlying basic science and technological aspects the book offers a fascinating insight for those working and training in the field of clinical or laboratory aspects of medical genetics genomics and allied areas it will also be of interest to historians of science and medicine and to workers in the social sciences who are increasingly attracted by the social and ethical challenges posed by modern medical genetics and genomics

Emery's Elements of Medical Genetics E-Book Peter D Turnpenny, Sian Ellard, 2016-11-30 Everything a student needs to know about medical genetics is here in the 15th edition of this award winning textbook Thoroughly updated and revised throughout to map a fast moving area the 15th edition continues Emery's enviable reputation for successfully balancing up to dateness in a rapidly developing field with a strong basis in practical clinical genetics for medical students With MCQs and Case Based Review Questions end of chapter summaries it is the essential tool for this complex but foundational topic for all medical undergraduates as well as postgraduates seeking to improve their understanding and knowledge Divided into three restructured sections to make the book easier to use for a variety of readers Scientific Basis of Human Genetics Genetics in Medicine and Genomic Medicine Clinical Genetics Counselling and Ethics Interactive self assessment questions Case based questions Online hyperlinks to important genetics websites and clinical databases Update of clinical figures to include more full colour images An extensive glossary of terms Full colour art to visualise the appearance of genetic disorders and assist with the understanding of complex genetic structures Explore the social ethical and counselling issues surrounding the study and treatment of genetic disorders Elements boxes at the end of each chapter summarizing the basics at a glance

Foundations of Perinatal Genetic Counseling Amber Mathiesen, Kali Roy, 2018-04-30 An essential new text for genetic counseling's most sought after skills Foundations of Perinatal Genetic Counseling is a practical introduction to the concepts and skills in genetic counseling with

clients before and during pregnancy Authored by genetic counselors at the forefront of contemporary perinatal practice this all in one reference provides an accessible yet comprehensive overview of the basics of pregnancy including assisted reproductive technologies and high risk pregnancy management preimplantation and prenatal genetic screening and diagnosis the structure and goals of a genetic counseling appointment common clinical scenarios and best practice approaches Distilling the most pertinent information for new learners and practicing counselors Foundations of Perinatal Genetic Counseling is an essential companion for both classroom and clinic Perinatal genetic counselors will find themselves returning to this unique resource long after their training has come to an end **Assessing Genetic Risks** Institute of Medicine,Committee on Assessing Genetic Risks,1994-01-01 Raising hopes for disease treatment and prevention but also the specter of discrimination and designer genes genetic testing is potentially one of the most socially explosive developments of our time This book presents a current assessment of this rapidly evolving field offering principles for actions and research and recommendations on key issues in genetic testing and screening Advantages of early genetic knowledge are balanced with issues associated with such knowledge availability of treatment privacy and discrimination personal decision making public health objectives cost and more Among the important issues covered Quality control in genetic testing Appropriate roles for public agencies private health practitioners and laboratories Value neutral education and counseling for persons considering testing Use of test results in insurance employment and other settings **Cumulated Index Medicus** ,1993

Prenatal genetic screening, diagnosis and treatment Aleksandra Jezela-Stanek,Sheela Nampoothiri,Przemysław Kosinski,Ting Hu,2023-09-06

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