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Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Primary prevention of genetic disorders and place of preimplantation diagnosis -- Approaches to preimplantation diagnosis -- Obtaining Biopsy Material -- Polar Body Diagnosis -- Pre-embryonic Genetic Diagnosis (PEGD) -- Preconception Testing for Paternally Derived Mutations by Sperm Duplication -- Development of artificial human gametes in vitro -- Embryo Biopsy -- Single cell genetic analysis -- DNA Analysis -- FISH Analysis -- Microarray analysis -- Preimplation diagnosis for single gene disorders -- Autosomal Recessive Diseases -- Autosomal Dominant Disorders -- X-linked disorders -- Homozygous or Double Heterozygous Recessive Conditions -- Conditions with No Available Direct Mutation Testing -- De Novo Mutations -- Late-onset disorders with genetic predisposition -- Inherited Predisposition to Cancer -- Alzheimer Disease -- Inherited Cardiac Diseases -- Blood Group Incompatibility -- Congenital Malformations -- Currarino Triad -- Dynamic Mutations -- Overall Experience of PGD for Mendelian Disorders -- PGD for HLA typing -- Fanconi Anemia -- World’s First PGD for HLA Typing -- Thalassemia -- Immunodeficiencies -- Preimplantation HLA Matching without PGD -- Limitations and future prospect of PGD for HLA Typing -- Practical Implications of PGD for HLA Typing -- Preimplantation diagnosis for chromosomal disorders -- First Polar Body Morphological Grading as

Possible Potential Means for Preselecting Viable Oocytes -- Aneuploidy in human oocytes -- Testing for both meiosis I and meiosis II errors required for PGD of aneuploidies -- Inconsistency between Aneuploidy Types Predicted by PB1 and Detected by Cleavage Stage Testing -- Complex Errors and Aneuloidy Rescue in Female Meiosis -- Chromosome Specific Meiotic Error Origin and Its Impact on Embryo Viability -- Mitotic Errors in Cleaving Embryos in Relation to Meiosis Errors -- PCR-based aneuploidy testing in cleaving embryos -- Practical Relevance of Autosomal Monosomy Detection -- Uniparental Disomies -- Impact of PB Testing in Detection and Avoidance of Aneuploid Embryos for Transfer -- Chromosomal rearrangements -- Polar Boby Approach -- Blastomere Nuclear Conversion by Fusion with Mouse Oocytes -- Chemical Conversion Method -- Clinical outcome of preimplantation genetic diagnosis -- Safety of PGD -- Diagnostic Accuracy of PGD -- Reproductive outcome of PGD -- Controversy in Assessing Clinical Outcome of Preimplantation Aneuploidy Testing -- Reproductive Outcome Before and after PGD in Same Couples -- Possible Impact of Aneuploidy Origin -- Possible Impact of 24 Chromosome Aneuploidy Testing -- Preimplantation diagnosis and establishment of disease and individual specific human embryonic stem cell lines -- Sources for Establishing Human Embryonic Stem Cell Lines -- Human Embryonic Stem Cell Lines with Genetic Disorders -- Human Embryonic Stem Cell Lines with Chromosomal Disorders -- Genetic Disease Specific Human Embryonic Stem Cell Lines -- Development of individual specific hESC lines -- Human Embryonic Stem Cell Lines Resistant to HIV -- Progress in Study of Disease specific hESC Lines -- Social, ethical and legal aspects.

Sommario/riassunto

Although treatment remains the major goal in the control of genetic disease, this is not yet a reality for most inherited conditions. In the absence of radical treatment, preimplantation genetic diagnosis (PGD) offers the answer to the control of these inherited conditions by predicting reproductive outcome. PGD is now entering its third decade as an established procedure for genetic and assisted reproduction practices, with new and exciting developments changing the whole concept of prevention of congenital disorders. The availability of practical experience from tens of thousands of PGD cases makes it necessary to update the current information on its accuracy, reliability and safety. The Second Edition of this successful book updates the progress in prevention of genetic disorders to demonstrate the important place of PGD in primary preventive measures and its increasing role in providing the whole range of reproduction options to couples at risk. In addition, it provides an extensive review of the most recent developments within the field of PGD including, PGD for expanding indications, such as de novo mutations, cancers, inherited cardiac diseases and combined PGD for single gene disorders, HLA typing and 24 chromosome testing in patients of advance reproductive age. This practical book is vital for all practitioners within the field of fertility, reproductive medicine and medical genetics. It will also be useful for those responsible for planning and organizing PGD services and provides a working manual for the establishment and performance of PGD in the framework of IVF and genetic practices. .