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Nota di contenuto	Preface -- 1. Introduction -- 2. A brief history of non-invasive prenatal diagnosis and its forecast -- Part 1. Clinical Genetics -- 3. The Nexus Between Chromosomal Abnormalities and Single Gene Disorders -- 4. Clinical implications of chromosomal polymorphisms in congenital disorders -- 5. Placental genetics. Fetus-placental discrepancies: Challenges in prenatal genetic diagnosis -- 6. Underpinnings of the Conundrum Between Genetic Screening and Testing -- 7. Epidemiology of birth defects in twins -- 8. Screening of aneuploidies in twin pregnancies -- Part 2. Non Invasive Diagnosis -- 9. Congenital Anomalies: the Role of Ultrasound -- 10. Customary complications and screening techniques of early pregnancy -- 11. First trimester screening for common and rare chromosomal abnormalities as well as for major defects – which tests should be combined? -- 12. The Technology of Cell Free Fetal DNA-based NIPT -- 13. The technologies: comparisons on efficiency, reliability and costs -- 14. Pre and Post Test Counseling -- 15. CfDNA testing in IVF pregnancies -- 16. “RATs” – Rare autosomal trisomies and their relevance in cfDNA testing -- 17. Rapid Detection of Fetal Mendelian Disorders: Thalassemia and Sickle Cell Syndrome -- 18. Noninvasive Antenatal Screening for Fetal RHD in RhD Negative Women to Guide Targeted Anti-D Prophylaxis -- 19. Genome Wide Cell Free Fetal DNA-based Prenatal Testing: Limits and Perspectives -- Part 3. Clinical setting and trends -- 20. Developing

and delivering a clinical service for the non-invasive prenatal diagnosis of monogenic conditions -- 21. Counseling in a changing world of genetics -- 22. Maternal Secondary Genomic Findings Detected By Fetal Genetic Testing -- 23. Prenatal genome-wide sequencing for the investigation of fetal structural anomalies - is there a role for non-invasive prenatal diagnosis? -- 24. Cross-cultural Perspectives on Noninvasive Prenatal Testing -- 25. International Guidelines for implementation of NIPT -- 26. Overview of Preimplantation Genetic Diagnosis (PGD): Historical Perspective and Future Directions.

Sommario/riassunto

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.
