1. Record Nr. UNINA9910583398303321

Titolo	Noninvasive prenatal testing (NIPT) [[electronic resource]] : applied genomics in prenatal screening and diagnosis / / edited by Lieve Page-Christiaens, Hanns-Georg Klein
Pubbl/distr/stampa	London : , : Academic Press, , [2018] ©2018
ISBN	0-12-814190-5
Descrizione fisica	1 online resource (xxvii, 377 pages) : illustrations
Disciplina	52
Soggetti	Prenatal diagnosis Diagnosis, Noninvasive Genetic Testing Prenatal Diagnosis DNA - blood
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Section 1: Cell-freeDNA (CfDNA): overview and technology. Fetal DNA in maternal plasma: an amazing two decades -- Understanding the basics of next generation sequencing in the context of cell-free DNA based NIPT -- The technology and bioinformatics of cell-free DNA based NIPT -- Section 2: CfDNA in clinical practice. Prenatal screening for common aneuploidies before and after the introduction of cell-free DNA based NIPT -- Why cell-free DNA based NIPT for fetal chromosome anomalies is not diagnostic -- The role of cell-free DNA based NIPT in twin pregnancy -- Genomewide testing for autosomal trisomies and copy number variations -- Non-invasive fetal blood group typing -- Noninvasive prenatal diagnosis (NIPD) of monogenic disorders -- Maternal constitutional and acquired copy number variations (CNVs) -- Section 3: Clinical integration. Best practices for integrating cell-free DNA based NIPT into clinical practice -- Quality assurance and standardization of cell-free DNA based NIPT laboratory procedures -- Decisional support for expectant parents -- Cell-free DNA based NIPT and society -- Ethics of cell-free DNA based NIPT for

	sex chromosome aneuploidies and sex determination -- Cost-effectiveness of cell-free DNA based NIPT: summary of evidence and challenges -- Section 4: The future. Exome sequencing in the evaluation of the fetus with structural anomalies -- Cell-based NIPT: a promising path for prenatal diagnosis -- Maternal circulating nucleic acids as a marker of placental health -- Prenatal treatment of genetic diseases in the unborn
Sommario/riassunto	"Since its introduction in 2012, cell-free (cf) DNA based Non-Invasive Prenatal Testing (NIPT) has been employed to test for fetal chromosome abnormalities, and gene mutations that lead to a variety of genetic conditions, by millions of pregnant women, in more than 90 countries worldwide. With Noninvasive Prenatal Testing (NIPT): Applied Genomics in Prenatal Screening and Diagnosis, Dr Lieve Page-Christiaens and Dr Hanns-Georg Klein have compiled the first authoritative volume on cfDNA NIPT methods and their clinical implementation"--Publisher's description.