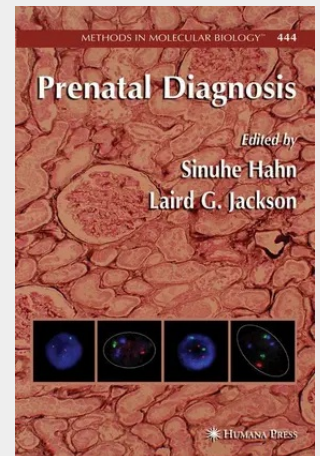


Prenatal Diagnosis

With molecular biology now allowing greater accuracy in prenatal diagnosis given amounts of fetal material as small as single cells, a major current focus has grown in the development of rapid, cost-effective diagnoses. This book has a totally unique focus on novel non-invasive approaches for prenatal diagnosis. Top experts provide cutting edge applications for the rapid assessment of fetal aneuploidies and Mendelian disorders on fetal material gained by invasive approaches, as well as procedures being validated for routine, non-invasive clinical analysis of cell free fetal DNA. Following the Methods in Molecular Biology™ series format, the chapters feature step-by-step laboratory protocols, lists of the necessary materials, and tips on troubleshooting and avoiding known pitfalls. Thorough and state-of-the-art, this is an ideal volume for researchers and molecular biologists invested in ever-growing field of prenatal medicine.

Molecular biology has transformed prenatal diagnosis because it permits an accurate diagnosis to be made from very small quantities of fetal material, even single cells. Although the latter permits the analysis of preimplantation embryos and, perhaps in the future, the analysis of fetal cells enriched from maternal blood, a major current focus is to facilitate rapid, cost-effective diagnoses. In this manner, the use of fluorescence in situ hybridization (FISH) or polymerase chain reaction (PCR)-based approaches on uncultured amniocytes or chorionic villi already permits a rapid assessment of the most common fetal aneuploidies (chromosomes 13, 18, 21, X, and Y) to be obtained within 24 h, thereby obviating the need for a 2-week culture period previously required for a traditional karyotype. Although the accuracy of karyotypic analysis is greatly enhanced by methodologies such as spectral karyotyping (SKY) or comparative genomic hybridization (CGH), the advent of high-density nucleotide arrays (chips) facilitates rapid assessment of the fetal genotype for a large number of mutations of frequent Mendelian disorders, e. g. , cystic fibrosis, the hemoglobinopathies, and Tay-Sachs syndrome. A further focus is the development of noninvasive, and hence, risk-free alternatives for prenatal diagnosis that no longer rely on invasive procedures such as amniocentesis or chorionic villus sampling. The most successful of these approaches is the analysis of placentally derived cell-free DNA in maternal blood.



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