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Non-invasive prenatal screening: A 20-year experience in Italy

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ABSTRACT

Over the past two decades, there has been a rapid evolution in prenatal screening for fetal chromosome abnormalities. Initially, testing was focused on the identification of affected pregnancies in either the first, or, the second trimester (e.g. the Combined test or the triple test). This was replaced by sequential modalities (e.g. contingent screening) that have enhanced detection while reducing the need for invasive testing. More recently, the introduction of technologies based on cell-free DNA (cfDNA) in maternal plasma and enrichment of fetal cells in maternal circulation have further refined the concept of sequential screening. In this review, we document our experience with serum and ultrasound-based contingent screening where we were able to achieve a detection rate of 96.8%, a false-positive rate of 2.8% and an odds of being affected given a positive result of 1:11. We also describe our initial experience with a novel sequential protocol that includes the analysis of fetal cells in maternal blood.

Methods for enrichment for fetal cells cfDNA and cfDNA technologies offer the possibility of greater sensitivity and specificity as well as expansion in the scope of genetic disorders detectable. As costs decline, these technologies will become increasingly used as primary screening tools. In the meantime, sequential use offers a practical approach to maximizing the benefits of prenatal testing.

Contents

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