Trisomy 21 screening utilizing cell-free fetal DNA from maternal blood becomes commercially available in the US

MaterniT21(tm) was launched in the United States as a blood test for trisomy 21. This test, which is currently marketed for women with singleton gestations at >10 weeks at high risk for Down syndrome, utilizes cell-free fetal DNA in maternal circulation. The results become available in about 10 days. The International Society for Prenatal Diagnosis (ISPD) issued a rapid response position statement supporting the use of this test as an advanced screening test or women with singleton pregnancies considered at high risk for trisomy 21. The ISPD recommends that positive results be confirmed by invasive testing.

The test was validated in a multicenter study of high risk singleton pregnancies, defined as the presence of advanced maternal age, an ultrasound finding suggestive of aneuploidy, positive serum screening or a personal or family history of Down syndrome. In the validation study, the performance characteristics of the MaterniT21(tm) test included sensitivity 98.6%, specificity 99.8%, false negative rate of 1.4% and false positive rate of 0.2%. Testing was technically successful in 99.2% of the samples. The test has not been validated in low risk women, multiple gestations, or in pregnancies with trisomy 21 mosaicism, yet studies are underway to validate use in low-risk pregnancies.