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NIPT: cost-effective, first-line screening for all pregnant women

Background

Before 2011, prenatal aneuploidy screening options for trisomy 21 (T21) included measurement of serum markers and/or sonographic evaluation of the fetus.[1] These tests could also report a risk for trisomy 18.[1] The introduction of cell-free DNA (cfDNA)-based noninvasive prenatal testing (NIPT) created a new screening option and facilitated screening for a greater range of fetal aneuploidies (trisomies 21, 18, 13, and certain sex chromosome aneuploidies).[3] NIPT is now endorsed as a screening option for all pregnant women.[1,4,5] Although NIPT is more expensive than serum screening, it is actually cost-effective.

Finding the most cost-effective solution

While NIPT is an endorsed screening option,[1,4,5] professional societies recommend that diagnostic testing be done following any positive or failed screening test for confirmation.[6,7] Although these invasive diagnostic tests are necessary to confirm results, they're expensive.[8-10] Therefore, false positive rates (FPR), technical failure rates, and the costs associated with invasive confirmatory procedures need to be considered in cost modeling. Compared with a trisomy 21 FPR of around 5% with conventional screening approaches,[11-13] NIPT has an FPR of around 0.1%.[14]

Studies found that using NIPT as a first-line screen becomes cost effective at a price of $619–744,[8-10]* with most of this value derived from screening for trisomy 21.[8]

0.1% FPR with NIPT

Reduction in need for confirmatory invasive procedures

Cost-effective at a price of $619-744

The Verifi™ Prenatal Test. Maximize cost effectiveness with the lowest failure rate

Of all the available NIPTs, the Verifi Prenatal Test offers the lowest reported technical failure rate,[15-19] substantially reducing additional costs associated with technical failures.[20] The failure rate of 0.1% is 10-fold less than that of other NIPTs on the market.

100,000 samples 1:500 incidence T21	200 pregnancies with T21	99,800 unaffected pregnancies	Potential number of unaffected invasive procedures	Potential costs of invasive procedures on unaffected pregnancies
	**In-lab test**			
	Verifi Prenatal Test[15] > 199 detected	< 0.1% false positive + 0.1% assay failure →	200 invasive	$0.2M
	Harmony Prenatal Test[19] > 188 detected	< 0.1% false positive + 6.0% assay failure →	6082 invasive	$3.3M
	Test send out			
	MaterniT21 PLUS[17] > 197 detected	< 0.1% false positive + 1.3% assay failure →	1396 invasive	$1.5M
	Panorama Prenatal Screen[18] > 192 detected	< 0.1% false positive + 3.8% assay failure →	3888 invasive	$4.1M
	Serum screen[11,13,21] > 188 detected	~5% false positive + 0% assay failure →	4990 invasive	$5.3M

NIPT and serum screening: Impact of false positive rates and test failures on the number and cost of invasive procedures for unaffected pregnancies.† Theoretical example of the number of invasive procedures, and the associated total cost, for serum screening and for commercial NIPTs currently available in the US. Based on published cost estimates for invasive testing[8-10] and published failures rates.[15-19]‡

NIPT is cost effective for use in the general pregnancy population

NIPT provides cost-effective, making it a viable option for first-line screening in the general pregnancy population. Ultimately, to maximize cost effectiveness in the general pregnancy population, the Verifi Prenatal Test is the NIPT of choice.

* These studies modeled the annual US pregnancy population that undergoes prenatal screening, and determined at what NIPT price point first-line screening by NIPT was cost effective compared with traditional screening options (measurement of serum markers with or without sonographic evaluation of the fetus). Modeling took into consideration: the detection rates and false positive rates of the two screening options; costs of traditional screening, diagnostic testing, and affected births; current clinical practices in terms of screening uptake and termination rates. The per-patient cost-effective price of NIPT reflects the total costs incurred by payers for the screening population divided by the number of patients being screened.[8-10]

† Affected pregnancies with a screening test failure were excluded from the number of detected T21.

‡ Assay failure rate for the Harmony test is based on next-generation sequencing studies and may not be consistent with actual test results achieved using the array-based Harmony test currently in use (published clinical experience data not available).

The Verifi™ Prenatal test was developed by, and its performance characteristics were determined by Verinata Health, Inc. (VHI), a wholly owned subsidiary of Illumina, Inc. The VHI laboratory is CAP-accredited and certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing. It has not been cleared or approved by the U.S. Food and Drug Administration.

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