



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

BACKGROUND

Prior to 2011, prenatal aneuploidy screening options for trisomy 21 included measurement of serum markers and/or sonographic evaluation of the fetus.^{1,2} 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 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, as shown below.

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 confirmation procedures need to be considered in cost modelling. Compared with a trisomy 21 FPR of around 5% with conventional screening approaches,¹¹⁻¹⁴ NIPT has a FPR of around 0.1%.¹⁵

THE ILLUMISCREEN® PRENATAL TEST. MAXIMISE COST-EFFECTIVENESS WITH THE LOWEST FAILURE RATE.

Of all the NIPTs, the Illumiscreen Prenatal Test offers the lowest reported technical failure rate,¹⁶⁻²⁰ substantially reducing additional costs associated with technical failures.²¹ 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
	Illumiscreen Prenatal Test ¹⁶ > 199 detected	< 0.1% false positive + 0.1% assay failure →	200 invasive
	MaterniT21 PLUS ¹⁸ > 199 detected	< 0.1% false positive + 1.3% assay failure →	1396 invasive
	Harmony Prenatal Test ²⁰ > 199 detected	< 0.1% false positive + 3.0% assay failure →	3091 invasive
	Panorama Prenatal Screen ¹⁹ > 199 detected	< 0.1% false positive + 3.8% assay failure →	3888 invasive
	Serum screen ^{11,14,22} > 199 detected	~0.1% false positive + 0% assay failure →	4990 invasive

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 testing⁸⁻¹⁰ and published failures rates.^{16-20†}

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* 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).

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