ILLUMI SCREEN

TTAGCGRACTCCC
TTTAACCUTTAAAEG
AAGGCCACCACGGCAAA
AAGGAAACCTCGG

Don't settle for failure

The Illumiscreen prenatal test uses proven NGS technology to provide accurate NIPT results with the lowest failure rate

WHAT IS TEST FAILURE?

For noninvasive prenatal testing (NIPT), test failure indicates that no call for chromosomal status can be made. This is an important factor in the reliability and clinical utility of NIPT. NIPT test failure rates vary significantly based on the test. Using whole-genome NGS, the Illumiscreen prenatal test from Illumina achieves the lowest test failure rate in NIPT (Figure 1).

THE IMPACT OF TEST FAILURE?

As test failure is really an inconclusive result, it leads to increased anxiety on the part of the patient and the physician, and it can potentially lead to an increased number of follow-up invasive procedures to obtain information. Although ordering a second blood draw to repeat NIPT is an option, there are no guarantees that repeated NIPT test will provide a definitive result. In fact, as many as 65% of patients who receive a no-call result on their first draw fail to receive a conclusive result, even after factoring in repeat attempts.[1,*]

An additional concern related to test failure rate is that methods that do not use whole-genome sequencing (WGS) have higher test failure rates. According to the Society for Maternal-Fetal Medicine (SMFM), "women with failed cfDNA tests are at an increased risk for aneuploidy, and therefore need careful counselling about further testing, including the offer of diagnostic testing."[2] With a lower test failure rate, whole-genome NGS-based assays are more likely to detect these aneuploidies the first time.

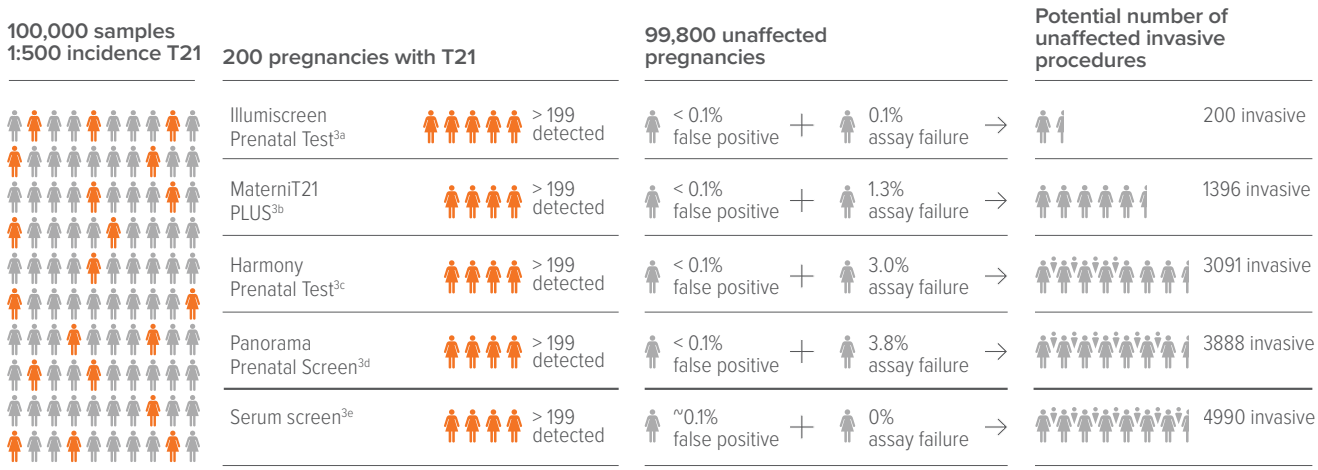

Figure 1: Test Failures May Lead to Invasive Procedures.[†]—Theoretical example of the number of invasive procedures requested due to NIPT failure and false positive rates of the assays. Failure rates include assay failures and samples rejected due to low fetal fraction. Assay failure rate for the Harmony test is based on NGS studies and may not be consistent with actual test results achieved using the array-based Harmony Test currently in use.[3a-3e]

* This 65% includes test failures from redraws and patients that either chose not to submit a second sample or are ineligible for a redraw due to specific features that prevent resolution with SNP-based NIPT (ie, large regions exhibiting loss of heterozygosity [LOH]).

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

To learn more about NIPT using the Illumiscreen prenatal test, visit **illumiscreen.co.nz**

References

1. Dar P, Curnow KJ, Gross SJ, et al. Clinical experience and follow-up with large scale single-nucleotide polymorphism-based non-invasive prenatal aneuploidy testing. *Am J Obstet Gynecol*. 2014;211(5):527.e521-517.
1. Cell-Free DNA Screening - Publications. Society for Maternal-Fetal Medicine. https://www.smfm.org/publications/193-cell-free-dna-screening. Accessed June 5, 2015.

3a. Taneja PA, Snyder HL, de Feo E, et al. Noninvasive prenatal testing in the general obstetric population: clinical performance and counselling considerations in over 85,000 cases. *Prenat Diagn*. 2015;doi:10.1002/pd.4766.

3b. Mccullough RM, Almasri EA, Guan X, et al. Noninvasive prenatal chromosomal aneuploidy testing—clinical experience: 100,000 clinical samples. PLoS One. 2014;9:e109173.

3c. Norton ME, Jacobsson B, Swamy GK, et al. Cell-free DNA analysis for noninvasive examination of trisomy. *N Engl J Med*. 2015;doi: 10.1056/NEJMoa1407349.

3d. Pergament E, Cuckle H, Zimmermann B, et al. Single-nucleotide polymorphism-based noninvasive prenatal screening in a high-risk and low-risk cohort. *Obstet Gynecol*. 2014;124(2 Pt 1):210-218.

3e. Nicolaides KH. Screening for fetal aneuploidies at 11 to 13 weeks. *Prenat Diagn.* 2011;31(1):7-15. Wald NJ, Rodeck C, Hackshaw AK, et al. SURUSS in perspective. *Semin Perinatol*. 2005;29(4):225-235.

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