Don’t settle for failure

The verifi® 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 chromosomal status can be made. This is an important factor in the reliability and clinical utility of NIPT. NIPT failure rates vary significantly based on the type of test used. Using whole-genome next-generation sequencing (NGS), the verifi 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 can lead 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 will provide a result. In fact, as many as 65% of patients who receive a test failure result on their first draw fail to receive a conclusive result, even after factoring in repeat attempts.1,4

Whole-genome sequencing (WGS) methods for NIPT have lower test failure rates than other targeted methods. 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 counseling 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.4,7

<table>
<thead>
<tr>
<th>100,000 samples 1:500 incidence T21</th>
<th>200 pregnancies with T21</th>
<th>99,800 unaffected pregnancies</th>
<th>Potential number of unaffected invasive procedures</th>
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</thead>
<tbody>
<tr>
<td>verifi Prenatal Test2</td>
<td>&gt; 199 detected</td>
<td>&lt; 0.1% false positive + 0.1% assay failure → 200 invasive</td>
<td></td>
</tr>
<tr>
<td>MaterniT21 PLUS®</td>
<td>&gt; 197 detected</td>
<td>&lt; 0.1% false positive + 1.3% assay failure → 1396 invasive</td>
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</tr>
<tr>
<td>Harmony Prenatal Test®</td>
<td>&gt; 194 detected</td>
<td>&lt; 0.1% false positive + 3.0% assay failure → 3091 invasive</td>
<td></td>
</tr>
<tr>
<td>Panorama Prenatal Screen®</td>
<td>&gt; 192 detected</td>
<td>&lt; 0.1% false positive + 3.8% assay failure → 3888 invasive</td>
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<tr>
<td>Serum screen®</td>
<td>&gt; 188 detected</td>
<td>&lt; 5% false positive + 0% assay failure → 4990 invasive</td>
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To learn more about NIPT using the verifi Prenatal Test, visit www.illumina.com/verifi.
References

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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