### Screening Options

<table>
<thead>
<tr>
<th></th>
<th>First Trimester Screening</th>
<th>Second Trimester Screening</th>
<th>Non-Invasive Prenatal Testing</th>
<th>Chromosome Analysis</th>
<th>Prenatal Microarray</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Markers/ Technology</strong></td>
<td>NT, PAPP-A, hCG</td>
<td>First Trimester: NT, PAPP-A, hCG</td>
<td>SequentialScreen®</td>
<td>Random analysis of circulating cell-free DNA (cfDNA) in maternal blood, Massively Parallel Sequencing (MPS)</td>
<td>Chorionic Villi Sampling</td>
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<td>Second Trimester: NT, PAPP-A, AFP, hCG, uE3, inhibin</td>
<td>IntegratedScreen®</td>
<td>G-banded karyotyping/ chromosome analysis</td>
<td>Amniocentesis</td>
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<tr>
<td></td>
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<td>First Trimester: NT, PAPP-A, hCG</td>
<td>Non-Invasive</td>
<td>Aneuploidy of chromosomes, large deletions/duplications, chromosome translocations, inversions, triploidy and tetraploidy</td>
<td>Reveal® SNP Microarray Prenatal</td>
</tr>
<tr>
<td></td>
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<td>Second Trimester: NT, PAPP-A, AFP, hCG, uE3, inhibin</td>
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<td>Aneuploidy of chromosomes, large deletions/duplications, chromosome translocations, inversions, triploidy and tetraploidy</td>
<td>Non-Invasive</td>
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<td>First Trimester: NT, PAPP-A, hCG</td>
<td>Non-Invasive</td>
<td>Aneuploidy of all unbalanced chromosomes, large deletions/duplications, chromosome translocations and inversions. Also, small (submicroscopic) deletions/duplications, provides detection of uniparental disomy of any chromosome, homozygosity as it pertains to identity by descent</td>
<td>Non-Invasive</td>
</tr>
<tr>
<td>What conditions does it detect?</td>
<td>Trisomy 21, 18</td>
<td>Trisomy 21, 18, Open Neural Tube Defects (ONTD)</td>
<td>Trisomy 21, 18, Open Neural Tube Defects (ONTD)</td>
<td>Trisomy 21, 18, Sex chromosome aneuploidy (Monosomy X, XXX, XXY, XY, XXY)</td>
<td>Trisomy 21, 18, Sex chromosome aneuploidy (Monosomy X, XXX, XXY, XY, XXY)</td>
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<td></td>
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<td>Aneuploidy of chromosomes, large deletions/duplications, chromosome translocations, inversions, triploidy and tetraploidy</td>
</tr>
<tr>
<td>What conditions are not detected?</td>
<td>All other aneuploidies and microdeletions/duplications, ONTD</td>
<td>All other aneuploidies and microdeletions/duplications, ONTD</td>
<td>All other aneuploidies and microdeletions/duplications, ONTD</td>
<td>All other aneuploidies and microdeletions/duplications, ONTD</td>
<td>Small microdeletions/duplications, ONTD</td>
</tr>
</tbody>
</table>
| Detection rates for the conditions it detects | T21 - 83.0%1 T18 - 80.0%2 | T21 - 90.4%3 T18 - 90.0%2 | T21 - 92.0%1 T18 - 90.0%2 | T21 - 99.15%4 T18 - 98.15%4 T13 - 98.15%4 MX - 95.0%5 XX - 97.6%, XY - 99.1%5 XXX/XXX/XXY - limited clinical data1 | ≥99% for aneuploidies2
| False positive rates (FPRs) for the conditions it detects | T21 - 5.0%1 T18 - <0.2%2 | T21 - 3.7%3 T18 - <0.2%2 | T21 - 5.0%1 T18 - <0.2%2 | T21 - 0.06%4 T18 - 0.1%4 T13 - 0.05%4 MX - 1.0%5 XX - 0.8%, XY - 1.1%5 XXX/XXX/XXY - limited clinical data1 | -0%2
| Gestational age when performed | 10w 3d - 13w 6d | 10w 3d - 13w 6d | 10w 3d - 13w 6d | ≥10 weeks | ≥10 weeks for CVS
| Turnaround time | 2-3 days | 2-3 days | 2-3 days | 5-7 days | ≥15 weeks | ≥15 weeks for amnio

### Diagnostic Options

- **Chorionic Villi Sampling**
  - G-banded karyotyping/ chromosome analysis

- **Amniocentesis**
  - G-banded karyotyping/ chromosome analysis

- **Reveal® SNP Microarray Prenatal**
  - Whole genome SNP-based copy-number microarray and analysis targeting 2.695 million copy number and allele-specific genome sites

- **Integrated Genetics 800-848-4436**

- **www.integratedgenetics.com**

### The Right Test at the Right Time

- **Markers/Technology**
  - NT, PAPP-A, hCG
  - First Trimester:
    - NT, PAPP-A, hCG
  - Second Trimester:
    - NT, PAPP-A, AFP, hCG, uE3, inhibin

- **What conditions does it detect?**
  - Trisomy 21, 18
  - Open Neural Tube Defects (ONTD)

- **What conditions are not detected?**
  - All other aneuploidies and microdeletions/duplications, ONTD

- **Detection rates for the conditions it detects**
  - T21 - 83.0%1 T18 - 80.0%2
  - T21 - 90.4%3 T18 - 90.0%2
  - T21 - 92.0%1 T18 - 90.0%2
  - T21 - 99.15%4 T18 - 98.15%4 T13 - 98.15%4 MX - 95.0%5 XX - 97.6%, XY - 99.1%5 XXX/XXX/XXY - limited clinical data1

- **False positive rates (FPRs) for the conditions it detects**
  - T21 - 5.0%1 T18 - <0.2%2
  - T21 - 3.7%3 T18 - <0.2%2
  - T21 - 5.0%1 T18 - <0.2%2
  - T21 - 0.06%4 T18 - 0.1%4 T13 - 0.05%4 MX - 1.0%5 XX - 0.8%, XY - 1.1%5 XXX/XXX/XXY - limited clinical data1

- **Gestational age when performed**
  - 10w 3d - 13w 6d
  - 10w 3d - 13w 6d

- **Turnaround time**
  - 2-3 days
  - 2-3 days

- **The Right Test at the Right Time**

- **Integrated Genetics 800-848-4436**

- **www.integratedgenetics.com**
Professional Society Recommendations and the Integrated Genetics Difference

<table>
<thead>
<tr>
<th>Maternal Serum Screening</th>
<th>Non-Invasive Prenatal Testing</th>
<th>Cytogenetics and Microarrays</th>
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<tbody>
<tr>
<td>• The American College of Obstetricians and Gynecologists (ACOG) recommendations indicate that &quot;a strategy that incorporates both first- and second-trimester screening should be offered to women who seek prenatal care in the first trimester&quot;.6</td>
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<tr>
<td>• &quot;...trisomies 13, 18, and 21 comprise a smaller proportion of the chromosome abnormalities found in the general obstetric population. Traditional serum analyte screening methods allow for higher detection rates of these other chromosome abnormalities as well as the risk of other adverse pregnancy outcomes.&quot;7</td>
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<td>An ACOG and Society of Maternal Fetal Medicine (SMFM) Committee Opinion:7</td>
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<td>• Recognized the tremendous potential of cell-free DNA (cfDNA) testing as a screening tool for fetal aneuploidy.</td>
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<tr>
<td>• Recommended genetic counseling prior to offering cfDNA screening and a discussion of the risks, benefits and potential alternative testing.</td>
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<tr>
<td>ACOG and SMFM recognize :</td>
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<tr>
<td>• Regardless of age and in concert with genetic counseling... &quot;patients with a fetus with one or more major structural abnormalities identified on ultrasonographic examination and who are undergoing invasive prenatal diagnosis... [chromosomal microarray] replaces the need for fetal karyotype.&quot;8</td>
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<tr>
<td>• &quot;In patients with a structurally normal fetus undergoing invasive prenatal diagnostic testing, either fetal karyotyping or a chromosomal microarray analysis can be performed.&quot;8</td>
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</tbody>
</table>

### Integrated Genetics, an experienced partner you and your patients can trust

- Over 25 years of genetic testing expertise.
- A complete menu of screening and diagnostic tests focused on women’s reproductive health.
- Largest national commercial genetic counseling network with over 140 genetic counselors.
- Extensive managed care contracts helping patients maximize their benefits.

Integrated Genetics – a leading provider of reproductive genetic testing services, driven by its commitment to physicians and patients. Learn more by visiting [www.integratedgenetics.com](http://www.integratedgenetics.com) or calling Integrated Genetics Client Services at 800-848-4436.

### Test Numbers and Names

<table>
<thead>
<tr>
<th>Test Number</th>
<th>Test Name</th>
</tr>
</thead>
<tbody>
<tr>
<td>315</td>
<td>FirstScreen®</td>
</tr>
<tr>
<td>335/336</td>
<td>SequentialScreenSM</td>
</tr>
<tr>
<td>302</td>
<td>IntegratedScreenSM</td>
</tr>
<tr>
<td>550746</td>
<td>informaSeq®</td>
</tr>
<tr>
<td>550757</td>
<td>informaSeq® with Y Analysis</td>
</tr>
<tr>
<td>510988</td>
<td>informaSeq® with XY Analysis</td>
</tr>
<tr>
<td>110</td>
<td>Chorionic Villi Sampling (CVS) Chromosome Analysis</td>
</tr>
<tr>
<td>100</td>
<td>Amniotic Fluid Chromosome Analysis</td>
</tr>
<tr>
<td>477</td>
<td>Reveal® SNP Microarray Prenatal</td>
</tr>
</tbody>
</table>

### Test Numbers and Names

- [FirstScreen®](http://www.integratedgenetics.com/)
- [SequentialScreenSM](http://www.integratedgenetics.com/)
- [IntegratedScreenSM](http://www.integratedgenetics.com/)
- [informaSeq®](http://www.integratedgenetics.com/)
- [informaSeq® with Y Analysis](http://www.integratedgenetics.com/)
- [informaSeq® with XY Analysis](http://www.integratedgenetics.com/)
- [Chorionic Villi Sampling (CVS) Chromosome Analysis](http://www.integratedgenetics.com/)
- [Amniotic Fluid Chromosome Analysis](http://www.integratedgenetics.com/)
- [Reveal® SNP Microarray Prenatal](http://www.integratedgenetics.com/)

**REFERENCES**


informaSeq® Prenatal Test is Powered by Illumina® sequencing technology.