Prenatal Testing Services
Amniotic Fluid | Chorionic Villi Sampling

CHROMOSOME ANALYSIS
• Detects extra or missing whole chromosomes
  ◦ Down syndrome
  ◦ Trisomy 13
  ◦ Trisomy 18
  ◦ Triploidy
  ◦ Turner syndrome
  ◦ Other sex chromosome aneuploidies
• Detects structural aberrations
  ◦ Large-scale deletions and duplications
  ◦ Balanced and unbalanced rearrangements

FLUORESCENCE IN SITU HYBRIDIZATION (FISH)
• Useful for targeted assessment of various chromosomal abnormalities
  ◦ Common aneuploidies (13, 18, 21, X, and Y)
  ◦ Familial chromosomal abnormalities
  ◦ Microdeletion or microduplication syndromes
• Results for STAT indications communicated to referring health care provider within 24 hours of specimen receipt

MICROARRAY ANALYSIS
• Detects submicroscopic copy number changes (microdeletions and microduplications) in targeted regions throughout the genome

INDICATIONS FOR TESTING

CHROMOSOME ANALYSIS with the option of FISH
♦ Advanced maternal age
♦ Abnormal ultrasound findings
♦ Abnormal NIPT, 1st trimester, 2nd trimester, or combined maternal serum screening result
♦ Family history of genetic or chromosomal anomalies

PRENATAL MICROARRAY
♦ Abnormal ultrasound findings
♦ Abnormal screening test (NIPT) for microdeletions
♦ Family history of genetic or chromosomal anomalies
♦ History of pregnancy loss
Feelings of guilt and blame are common in women experiencing a loss; these symptoms are exacerbated for some women by misconceptions about the cause of their pregnancy loss.

At least 10-15% of all recognized pregnancies are miscarried. Chromosome abnormalities cause more than 50% of first trimester losses and 15% of second trimester losses.1

In most instances, identification of a chromosomal cause for pregnancy loss allows calculation of accurate recurrence risks; families and providers use risks to determine appropriate fertility and pregnancy management.

Women with normal genetic test results may require further clinical evaluation, which may include additional laboratory studies, prior to their next pregnancy.

Testing Options

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<tr>
<th>Patient History</th>
<th>Our Recommended Testing</th>
<th>Rationale</th>
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<tbody>
<tr>
<td>Woman with a history of recurrent pregnancy loss currently not pregnant</td>
<td>Chromosome Analysis for woman and her partner</td>
<td>Chromosome Analysis identifies a chromosomal rearrangement, such as a balanced translocation, in 5-8% of individuals with recurrent loss.2,3</td>
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<tr>
<td>Woman with a history of recurrent pregnancy loss currently experiencing a miscarriage</td>
<td>Pregnancy Loss Microarray performed on fetal tissue</td>
<td>Pregnancy Loss Microarray offers an increased detection rate and greater likelihood of obtaining results as compared to traditional chromosome analysis.4</td>
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<tr>
<td>Woman currently experiencing a stillbirth</td>
<td>Pregnancy Loss Microarray performed on fetal tissue</td>
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<tr>
<td>Woman without a history of recurrent pregnancy loss currently experiencing a miscarriage</td>
<td>Pregnancy Loss Microarray performed on fetal tissue</td>
<td>Testing may be appropriate for women experiencing their first or second loss, particularly when the family or pregnancy history is suspicious for a chromosomal cause of pregnancy loss.</td>
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