## Specimen Types and Sample Requirements

### Prenatal Analysis

<table>
<thead>
<tr>
<th>Test Specimen Type</th>
<th>Specimen Requirement</th>
</tr>
</thead>
<tbody>
<tr>
<td>CombiSNP™ Array Prenatal Direct†</td>
<td>Amniotic fluid or Chorionic villi</td>
</tr>
<tr>
<td>Direct† Amniotic fluid or Chorionic villi</td>
<td>15-20 mL amniotic fluid or 5-10 mg cleaned chorionic villi</td>
</tr>
<tr>
<td>Karyotype Analysis</td>
<td>Amniotic fluid Chorionic villi</td>
</tr>
<tr>
<td>Amniotic fluid or 25-30 mL with reflex to microarray or 5-10 mg cleaned chorionic villi</td>
<td></td>
</tr>
<tr>
<td>Amniotic Fluid AFP/AChE</td>
<td>Amniotic fluid Chorionic villi</td>
</tr>
<tr>
<td>Amniotic fluid or 5 mL amniotic fluid or 5 mg cleaned chorionic villi</td>
<td></td>
</tr>
<tr>
<td>Family Studies</td>
<td>Peripheral blood</td>
</tr>
<tr>
<td>Contact our Genetic Counseling Services Team: 949.255.0921</td>
<td></td>
</tr>
<tr>
<td>Maternal Cell Contamination (MCC) Studies</td>
<td>Peripheral blood</td>
</tr>
<tr>
<td>4 mL blood in an EDTA (purple top) tube</td>
<td></td>
</tr>
<tr>
<td>Pregnancy Loss Analysis</td>
<td>Specimen Type</td>
</tr>
<tr>
<td>CombiSNP™ Array for Pregnancy Loss</td>
<td>Fresh tissue in RPMI with antibiotics*</td>
</tr>
<tr>
<td>CombiSNP™ Array for Pregnancy Loss</td>
<td>Formalin-fixed paraffin-embedded (FFPE) tissue block or unstained slides</td>
</tr>
<tr>
<td>Maternal Cell Contamination (MCC) Studies</td>
<td>Peripheral blood</td>
</tr>
<tr>
<td>Parental Karyotype Analysis</td>
<td>Peripheral blood</td>
</tr>
</tbody>
</table>

### Preimplantation Genetic Testing

<table>
<thead>
<tr>
<th>Test Specimen Type</th>
<th>Specimen Requirement</th>
</tr>
</thead>
<tbody>
<tr>
<td>CombiPGD™ for Single Gene Disorders</td>
<td>Cells from embryo biopsy</td>
</tr>
<tr>
<td>CombiPGD™ for Chromosome Translocations</td>
<td>Cells from embryo biopsy</td>
</tr>
<tr>
<td>CombiPGS™</td>
<td>Cells from embryo biopsy</td>
</tr>
<tr>
<td>Parental Karyotype Analysis</td>
<td>Peripheral blood</td>
</tr>
</tbody>
</table>

### Pediatric Analysis

<table>
<thead>
<tr>
<th>Test Specimen Type</th>
<th>Specimen Requirement</th>
</tr>
</thead>
<tbody>
<tr>
<td>CombiSNP™ Array for Pediatric Analysis</td>
<td>Peripheral blood</td>
</tr>
<tr>
<td>Buccal swab †</td>
<td>Use ORAcollect OC-100 kit (follow enclosed directions)</td>
</tr>
<tr>
<td>Fragile X Analysis</td>
<td>Peripheral blood</td>
</tr>
<tr>
<td>Karyotype Analysis</td>
<td>Peripheral blood</td>
</tr>
</tbody>
</table>

* CombiMatrix provides this media at no charge.
† Direct array can only be performed if a sufficient number of fetal cells are present.
‡ CombiFISH must be ordered in conjunction with either a microarray analysis or karyotype analysis.
§ If results are positive, a follow up peripheral blood sample is required for confirmatory FISH analysis.

CombiMatrix offers comprehensive result interpretation services and ready access to our medical team and board-certified genetic counselors.

For more information, please contact your local CombiMatrix representative or Client Services at 949.255.0920.
Prenatal Analysis

**CombiSNP™ Array Direct Analysis**
Performed on uncultured amniocytes or chorionic villi to minimize result turn-around time, this high resolution SNP microarray detects numeric chromosome abnormalities, unbalanced structural rearrangements, microdeletion/duplication syndromes, and long contiguous segments of homologous material, which can indicate shared ancestry or uniparental disomy (UPD). SNP analysis also enables detection of trisomy, mosaicism and maternal cell contamination.

7-8 days*

**CombiSNP™ Array for Prenatal Analysis**
Performed on cultured amniocytes or chorionic villi, this high resolution SNP microarray is typically performed as a reflex test following a normal karyotype. The CombiSNP™ Array detects numeric chromosome abnormalities, unbalanced structural rearrangements, microdeletion/duplication syndromes, and long contiguous segments of homology, which can indicate shared ancestry or uniparental disomy (UPD). SNP analysis also enables detection of trisomy, mosaicism and maternal cell contamination.

7-8 days*

**Karyotype Analysis**
G-banded karyotyping for the detection of numeric chromosome abnormalities, balanced and unbalanced chromosome rearrangements, and polyplody. (Ammiotic and CVS)

10-12 days

**Amniotic Fluid AFP/AChE Analysis**
Analyte assay for detection of open neural tube defects and abdominal wall defects.

3 days

**CombiFISH™ Analysis**
Rapid interphase FISH assay performed on uncultured amniocytes or chorionic villi for detecting of aneuploidy of chromosomes 13, 18, 21, X and Y.

24-48 hrs†

**Family Studies**
Parental and family studies may be performed using either customized FISH analysis or microarray, depending on the size and nature of the familial variant.

7-8 days

**Maternal Cell Contamination (MCC) Studies**
Molecular assay to detect the presence of maternal cells in the fetal sample, which may obscure fetal results. Often ordered following a normal female karyotype when microarray is not desired, or as a follow-on if microarray results suggest a possible mosaic sex chromosome abnormality.

17 days†

Pregnancy Loss Analysis

**CombiSNP™ Array for Reflex Request**
Performed on fresh tissue, FFPE or unstained slides

10-12 days

**Maternal Cell Contamination (MCC) Studies**
Molecular assay to detect the presence of maternal cells in the fetal sample, which may obscure fetal results. Typically ordered as a follow-on test if CombiSNP™ Array results suggest a possible mosaic sex chromosome abnormality.

17 days†

**Parental Karyotype Analysis**
G-banded karyotyping for the detection of balanced chromosomal rearrangements.

14 days

Preimplantation Genetic Testing

**CombiPGD™ for Single Gene Disorders**
Performed on cells from human embryos during the course of in vitro fertilization, this test determines whether or not an embryo is affected with a specific single gene disorder.

12-14 days

**CombiPGD™ for Chromosome Translocations**
Performed on cells from human embryos during the course of in vitro fertilization, this test determines whether or not an embryo is affected with a specific segmental aneuploidy due to the presence of a balanced structural chromosomal abnormality in one of the parents.

12-14 days

**CombiPGS™ Analysis**
Performed on cells from embryos during the course of in vitro fertilization (IVF), CombiPGS screens for whole and segmental chromosomal deletions and duplications which can lead to IVF failure, miscarriages and live births with chromosomal abnormalities.

24 hrs (Day 3 biopsy)

3-4 days (Day 5/6 biopsy & freeze)

**Parental Karyotype Analysis**
G-banded karyotyping for the detection of balanced chromosomal rearrangements.

14 days

Pediatric Analysis

**CombiSNP™ Array for Pediatric Analysis**
High resolution SNP microarray detects numeric chromosome abnormalities, unbalanced structural rearrangements, microdeletion/duplication syndromes, and long contiguous segments of homology, which can indicate shared ancestry or uniparental disomy (UPD). The design of the CombiSNP™ array is based on the International Collaboration for Clinical Genomics (ICCG) and ACMG consensus recommendations, along with additional content targeted to autism spectrum disorders.

10-12 days

**Fragile X Analysis**
Molecular assay to identify Fragile X and FMR1-related disorders.

10 days

**Karyotype Analysis**
G-banded karyotyping for the detection of numeric chromosome abnormalities as well as balanced and unbalanced chromosome rearrangements.

12 days

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* From date of reflex request.
† Samples received on Friday or Saturday will be reported on Monday.
‡ If sufficient sample is received.

CombiMatrix is pleased to offer the following specialized genetic testing services.

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