

## Specimen Types and Sample Requirements

### Prenatal Analysis

Test	Specimen Type	Specimen Requirement
CombiSNP™ Array Prenatal Direct†	Amniotic fluid Chorionic villi	15-20 mL amniotic fluid or 5-10 mg cleaned chorionic villi
CombiSNP™ Array for Prenatal Analysis	Cultured amniocytes Cultured villi	One confluent T-25 flask
Karyotype Analysis	Amniotic fluid Chorionic villi	15 mL amniotic fluid or 25-30 mL with reflex to microarray or 5-10 mg cleaned chorionic villi
Amniotic Fluid AFP/AChE	Amniotic fluid	2 mL amniotic fluid
CombiFISH™ ‡	Amniotic fluid Chorionic villi	5 mL amniotic fluid or 5 mg cleaned chorionic villi
Family Studies	Peripheral blood	Contact our Genetic Counseling Services Team: 949.255.0921
Maternal Cell Contamination (MCC) Studies	Maternal peripheral blood	4 mL blood in an EDTA (purple top) tube

### Pregnancy Loss Analysis

Test	Specimen Type	Specimen Requirement
CombiSNP™ Array for Pregnancy Loss	Fresh tissue in RPMI with antibiotics*	15-20 mg fetal or placental tissue placed in RPMI in a 50 mL tube
CombiSNP™ Array for Pregnancy Loss	Formalin-fixed paraffin-embedded (FFPE) tissue block or unstained slides	1 FFPE block with >1 cm <sup>3</sup> of fetal tissue or 10 unstained slides with 7 micron-thick sections plus one H&E stained slide
Maternal Cell Contamination (MCC) Studies	Peripheral blood	4 mL blood in an EDTA (purple top) tube
Parental Karyotype Analysis	Peripheral blood	4 mL blood in a NaHep (green top) tube

### Preimplantation Genetic Testing

Test	Specimen Type	Specimen Requirement
CombiPGD™ for Single Gene Disorders	Cells from embryo biopsy	1 cell (blastomere) from a Day 3 embryo or 5-10 trophectoderm cells from a Day 5/6 embryo
CombiPGD™ for Chromosome Translocations	Cells from embryo biopsy	1 cell (blastomere) from a Day 3 embryo or 5-10 trophectoderm cells from a Day 5/6 embryo
CombiPGS™	Cells from embryo biopsy	1 cell (blastomere) from a Day 3 embryo or 5-10 trophectoderm cells from a Day 5/6 embryo
Parental Karyotype Analysis	Peripheral blood	4 mL blood in a NaHep (green top) tube

### Pediatric Analysis

Test	Specimen Type	Specimen Requirement
CombiSNP™ Array for Pediatric Analysis	Peripheral blood	4 mL blood in an EDTA (purple top) tube and 4 mL NaHep (green top) tube (NICU/PICU patients, minimum 1-2 mL per tube)
	Buccal swab §	Use ORAcollect OC-100 kit (follow enclosed directions)
Fragile X Analysis	Peripheral blood	4 mL blood in an EDTA (purple top) tube
Karyotype Analysis	Peripheral blood	4 mL blood in a NaHep (green top) tube

\* 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](tel:949.255.0920).

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

## Prenatal Analysis

Test	Description	Avg. TAT
CombiSNP™ Array Direct Analysis	Performed on <i>uncultured</i> 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 homozygosity, which can indicate shared ancestry or uniparental disomy (UPD). SNP analysis also enables detection of triploidy, mosaicism and maternal cell contamination.	7-8 days*
CombiSNP™ Array for Prenatal Analysis	Performed on <i>cultured</i> 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 stretches of homozygosity, which can indicate shared ancestry or uniparental disomy (UPD). SNP analysis also enables detection of triploidy, 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 polyploidy. (Amniotic and CVS)	10-12 days
Amniotic Fluid AFP/ACHe	Analyte assay for detection of open neural tube defects and abdominal wall defects.	3 days
CombiFISH™	Rapid interphase FISH assay performed on <i>uncultured</i> 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

Test	Description	Avg. TAT
CombiSNP™ Array Fresh tissue, FFPE or unstained slides	Performed on fresh tissue, this high resolution SNP microarray detects triploidy, numeric chromosome abnormalities, unbalanced structural rearrangements, microdeletion / duplication syndromes, long contiguous segments of homozygosity, which can indicate shared ancestry or uniparental disomy (UPD) and maternal cell contamination.	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

Test	Description	Avg. TAT
CombiPGD™ for Single Gene Disorders	Performed on cells from human embryos during the course of <i>in vitro</i> fertilization, this test determines whether or not an embryo that is at risk for a familial single gene disorder is affected.	12-14 days
CombiPGD™ for Chromosome Translocations	Performed on cells from human embryos during the course of <i>in vitro</i> 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™	Performed on cells from embryos during the course of <i>in vitro</i> 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

Test	Description	Avg. TAT
CombiSNP™ Array for Pediatric Analysis	High resolution SNP microarray detects numeric chromosomal abnormalities, unbalanced structural rearrangements, microdeletion/duplication syndromes, and long contiguous segments of homozygosity, which can indicate common 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

\* From date of reflex request.

† Samples received on Friday or Saturday will be reported on Monday.

‡ If sufficient sample is received.



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