CombiMatrix

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^3 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.

CombiMatrix is pleased to offer the following specialized genetic testing services

Prenatal Analysis

Test	Description	Avg.TAT
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 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 *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 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 *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

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 *in vitro* 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 *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™	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

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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Lit.No. 04282016