Revealing Answers to Complex Questions

Reveal® SNP Microarray is a high density copy number array that detects genomic imbalance related to congenital anomalies, ultrasound abnormalities, miscarriage, and stillbirth.

www.integratedgenetics.com
American College of Obstetricians and Gynecologists (ACOG) and Society of Maternal-Fetal Medicine (SMFM) Recommendations for chromosomal microarray analysis include:\(^1\)

Regardless of age and in concert with genetic counseling…“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.”

- In patients with a structurally normal fetus undergoing invasive prenatal diagnostic testing, either fetal karyotyping or a chromosomal microarray analysis can be performed...
- In cases of intrauterine fetal demise or stillbirth when further cytogenetic analysis is desired, chromosomal microarray analysis on fetal tissue…“is recommended because of its increased likelihood of obtaining results and improved detection of causative abnormalities.”

Over 70,000 SNP Arrays Analyzed!

- Enhanced detection of chromosomal abnormalities
- Detects copy neutral changes, such as uniparental disomy (UPD) and consanguinity
- Detects small abnormalities that are typically undetectable by routine karyotype
  - More than 750,000 SNP probes provide both genotyping and copy number analysis
  - More than 1.9 million region specific copy number probes
- Whole genome coverage helps resolve marker chromosome origin and identify unbalanced rearrangements undetected by routine cytogenetics
- An extensive database* of abnormalities provides an exceptional reference to support evidence-based interpretation

A Recent Study has Shown:

“Microarray analysis provided additional clinically relevant information in 1.7% of pregnancies with standard indications for prenatal diagnosis (such as advanced maternal age and positive aneuploid screening result) and in 6.0% of cases with an anomaly on ultrasonography. These data indicate a benefit to chromosomal microarray analysis as a standard part of prenatal testing, bearing in mind that, as with karyotyping, the detection of variants of uncertain clinical significance present a challenge for counseling and cause anxiety.”\(^2\)

*Database of over 70,000 samples run at LabCorp’s Center for Molecular Biology and Pathology
Clinical Indications:
- As a first line test for patients with a fetus with abnormal anatomic findings, who are undergoing invasive diagnostic testing
- For clarification of an abnormal fetal karyotype requiring further characterization (e.g. identification of a marker chromosome)
- For an individual undergoing invasive diagnostic prenatal testing
- In cases of intrauterine fetal demise or stillbirth

Reveal\textsuperscript{®} SNP Microarray Product Offerings
- Reveal\textsuperscript{®} SNP Microarray Prenatal
- Full Chromosome analysis with reflex to Reveal\textsuperscript{®}
- Reveal\textsuperscript{®} SNP Microarray and Abbreviated Chromosomal Analysis

Results Reported for:
- Deletions larger than 1 Mb, duplications larger than 2 Mb
- Deletions or duplications as small as 50 Kb for genes with established clinical significance
- Detection of UPD and consanguinity
- Susceptibility genes are reported when they are associated with a clinical syndrome that has a clear phenotype

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<th>Specimen Requirements\textsuperscript{†}</th>
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*Direct testing available for women over 15 weeks gestation. MCC strongly recommended (7 ml whole blood in lavender top EDTA or yellow top ACD tube)
†Additional fluid or CVS specimen maybe be necessary if additional tests are ordered

A completed Clinical Questionnaire for Reveal\textsuperscript{®} SNP Microarray – Prenatal must be completed, and is available on our website or by calling 800-345-GENE (4363).
Experience you can trust. From your comprehensive laboratory partner.

Providing over 25 years of genetic testing expertise from board certified Geneticists

**Broad test menu**

- Integrated Genetics offers a full menu of diagnostic services including appropriate microarray, cytogenetics, FISH, and molecular genetics testing

**Comprehensive Service offerings**

- Genetics experts are readily accessible to answer questions
- Extensive managed care contracts, helping patients maximize their benefits
- Largest commercial genetic counseling network with over 140 genetic counselors
- Over 1,700 patient service centers providing an extensive patient service network

To learn more about prenatal diagnosis of chromosome abnormalities, please visit www.integratedgenetics.com or call 800-345-GENE (4363).

**REFERENCES**