Reveal®

SNP Microarray

PRENATAL

Reveal answers to complex diagnostic questions

Integrated GENETICS

LabCorp Specialty Testing Group

SNP microarray is a first-line test for all women, regardless of age, who undergo prenatal diagnostic testing, according to ACOG guidelines[1]

Where some genetic testing companies require patients to go elsewhere for diagnostic testing, Integrated Genetics offers continuity of care.

Whether you need diagnostic testing for routine prenatal diagnosis, confirmation of positive NIPT results, pregnancy loss, ultrasound anomalies, confirmation of complex rearrangements, interrogation of apparently "balanced" translocations and characterization of marker chromosomes , Reveal® Prenatal SNP microarray is an advanced genetic test that delivers results.

When a clinical phenotype does not fit a specific syndrome, microarray analysis can identify the etiology. Reveal Prenatal will identify and characterize abnormalities currently detected by routine cytogenetics (with the exception of balanced chromosome rearrangements) as well as genetic changes that may be missed by standard karyotype.[2]

Advantages of Reveal

- **Combine SNP microarray with Insight® (FISH)** for rapid answers and valuable insights during your patient's pregnancy
- **Detects genomic imbalances** associated with congenital abnormalities, intellectual disabilities, miscarriage and stillbirth[1]

REVEAL PRENATAL - FEATURES	
>230,000 arrays analyzed*	Extensive experience and database of abnormalities from specimens tested over time providing an exceptional resource to support interpretation
Low variants of uncertain significance (VUS) rate	Low VUS rates of 1-2% (where the vast majority are familial[3]) due to extensive variant database and use of conservative reporting cutoffs in genes of unknown function
High success rate	Greater than a 99.5% success rate[3]
Flexible test options	Various FISH, chromosome analysis, molecular, and microarray testing combinations to expedite results and help eliminate unnecessary testing
One microarray utilized for all sample types	One microarray platform for prenatal and pediatric testing, providing a robust database for interpretation and enabling reanalysis of data after delivery using pediatric cut-offs without the need for an additional specimen

*Prenatal, postnatal, products of conception

SERVICES	
Access to genetic experts	Network of lab genetic counselors, medical geneticists and lab directors available to support your results interpretation and patient management protocols
1900 Patient Service Centers	Convenient specimen draw sites nationwide for patients
>400 managed care plans	Extensive in-network coverage and expanded financial services to increase patient access
~140 genetic counselors on staff, plus online videos and resources	Largest national commercial network of genetic counselors to help inform and support patients

Detects copy neutral changes associated with increased risk for autosomal recessive conditions, uniparental disomy (UPD)[4], identity by descent (IBD) and molar genotypes[5]

Whole genome coverage helps resolve marker chromosome origin and identify unbalanced rearrangements undetectable by routine cytogenetics[1,6]

Ability to analyze DNA from most tissues, including paraffin-embedded tissue or tissue that cannot be cultured

Why choose Integrated Genetics for your SNP microarrays?

Years of experience	Access to experts
35+ years experience performing genetic testing **11+** years experience in microarray testing **300+** combined years cytogenetics experience for array laboratory directors	**~140** genetic counselors nation-wide: **~100** dedicated to direct patient care **15+** lab directors involved in microarray analysis

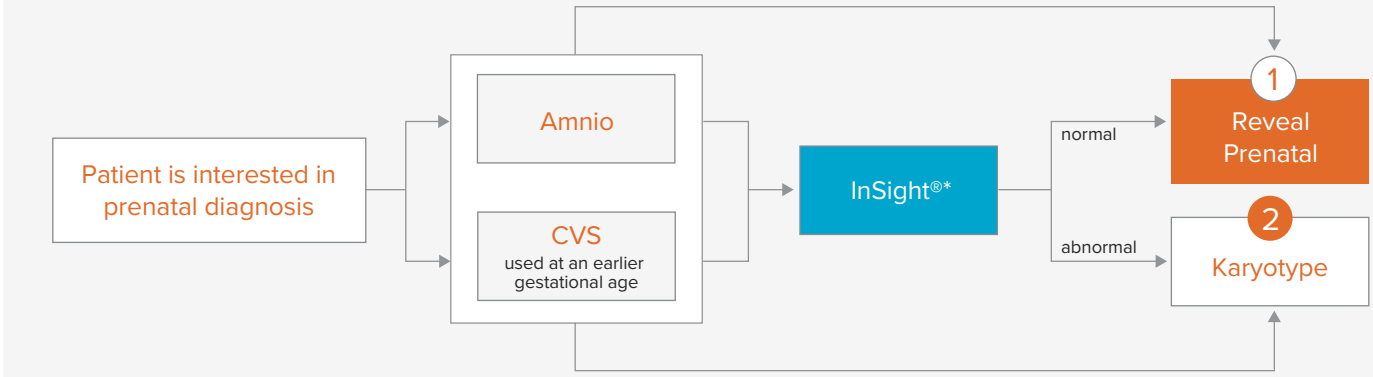
Reveal SNP Microarray is clinically validated, highly reliable and supported by society guidelines

In a study involving 50,000 prenatal microarray patients, it was determined SNP microarray was an effective technology and first-line test for prenatal diagnosis of patients. Microarray analysis detected cytogenetic abnormalities (when karyotype was normal) for both advanced maternal age (1.7%) and abnormal ultrasound findings (~4.3%).[3] This was even greater when the ultrasound revealed a major ultrasound abnormality (~9.2%).[3]

Table below demonstrates the additional yield for microarray after normal karyotype.[3]

	PATHOGENIC	IBD	UPD	TOTAL
Major	9.2%	2.9%	0.8%	12.9%
Major - Heart	8.1%	2.5%	0.3%	10.9%
Multiple Anomalies	4.3%	4.8%	0.1%	9.2%
Multiple Anomalies - Heart	8.2%	3.8%	0.8%	12.8%
Nuchal Translucency	2.5%	3.0%	0.3%	5.8%
Diaphragmatic Hernia	6.2%	2.1%	0.5%	8.8%
Holoprosencephaly	6.9%	2.5%	2.9%	12.3%

Example of a Reveal Prenatal ordering pathway



***Combination reflex testing available**

InSight: Fluorescence in situ Hybridization (FISH), rapid aneuploidy analysis (specific for chromosomes 13, 18, 21, X, Y), with reflex to:

1. high resolution SNP microarray, if results are normal
2. standard chromosome analysis, if results are abnormal

We can help determine your patient's prior-authorization simply by calling 855-488-8750 or emailing the Reveal Clinical Questionnaire to PriorAuth@LabCorp.com

Specimen requirements

	DIRECT TESTING
Specimen quantity	Amnio: 15-20 mL (15-17 weeks); 10 mL (≥ 17 weeks) CVS: 15-20 mg (2) confluent T-25 flasks for testing on cultured cells Special studies may call for additional specimen. Contact the lab to discuss requirements.
Expected turnaround time*	7-10 days If cell culture required, additional 1-2 weeks needed

*Turnaround time is calculated from the time the specimen arrives at the lab in North Carolina and varies if tissue culture is required

A completed clinical questionnaire for Reveal Prenatal SNP Microarray should accompany the specimen to aid in interpretation and is available on our website or by calling Client Services at 800-345-GENE (4363).

In 2018, 75% of patients paid $0 for Reveal Prenatal.

*Based on managed care claim data in 2018. For 48% of patients, insurance paid in full and 27% of patients had no patient responsibility (non-covered and coverage-related).[7]

Specifications

Probe density	• 2.6 million+ copy number and allele-specific genomic markers
	  • 1.9 million+ structural probes to detect copy number variants
	  • 750,000+ SNP probes to detect copy neutral changes, triploidy and molar genotypes
	  • 100% ISCA constitutional gene and X chromosome coverage

Specimen types	• Direct amnio or chorionic villi • Cultured chorionic villi or amniocytes
Reporting cut-offs for copy-number changes	• ≥25 kb for genes with established clinical significance • Deletions: ≥1 Mb with at least one OMIM gene • Duplications: ≥2 Mb with at least one OMIM gene
Identification of copy neutral changes	• IBD, UPD, regions of homozygosity associated with recessive disease risk
Exon analysis	• Specific genes may be analyzed at an exon level upon request
Susceptibility genes	• Reported when associated with a clinical syndrome that has a clear phenotype
Family studies policy	• Prenatal testing with appropriate technologies is performed at no charge when required to interpret the fetal result

Toll-free (within the US)
800.345.GENE (4363)

www.integratedgenetics.com

askCMBP@labcorp.com
Fax: 919.361.7700

Center for Molecular Biology and Pathology (CMBP)
1904 TW Alexander Drive
Research Triangle Park, NC 27709

View short videos on genetic testing:
www.integratedgenetics.com/videos

Integrated GENETICS
LabCorp Specialty Testing Group

Test codes

Integrated Genetics client	LabCorp client
477 Reveal SNP Microarray Prenatal	510200 Reveal SNP Microarray (Direct)–Prenatal
	510100 Reveal SNP Microarray–Prenatal (culture)
105 InSight® (Interphase FISH for Rapid Detection of Numerical Abnormalities of Chromosomes 13, 18, 21, X & Y)	052104 Reveal Chromosome Analysis, Amniotic Fluid With Reflex to SNP Microarray
	511590 Reveal Chromosome Five-cell Count Plus Microarray, Amniotic Fluid
100 Cytogenetics Amniotic Fluid Chromosome Analysis	511555 Reveal Chromosome Five-cell Count Plus Microarray, CVS
	511535 Reveal Chromosome Five-cell Count plus Microarray, Whole Blood
110 Cytogenetics Chorionic Villi Sampling (CVS) Chromosome Analysis	511033 Reveal Chromosome Analysis, Chorionic Villi Biopsy With Reflex to SNP Microarray
*Indicate reflex to chromosome analysis or microarray on test requisition form	511625 Fluorescence in situ Hybridization (FISH), Prenatal Aneuploid Evaluation, Chorionic Villus Sampling With Reflex to Microarray or Chromosome Analysis
	511966 InSight: Fluorescence in situ Hybridization (FISH), Prenatal Aneuploid Evaluation, Amniotic Fluid With Reflex to Microarray or Chromosome Analysis

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3. The impact of microarray studies on prenatal diagnosis: analysis of 50,000 prenatal samples and 15,000 POCs. Schwartz, Stuart; Burnside, Rachel; Gadi, Inder; etc. Presented at The 10th Prenatal Diagnosis: Ultrasounds/Fetal ECHO, Genetics & MFM/OB Conference in Philadelphia. May 2018.
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6. Savage Melissa, Mourad Mirella, Wapner Ronald. Evolving Applications of Microarray Analysis in Prenatal Diagnosis. *Curr Opin Obstet Gynecol.* 2011; 23(2):103-108. doi: 10.1097/GCO.0b013e32834457c7.
7. Internal LabCorp billing data (2018).

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