GENETICS & WOMEN'S HEALTH

Reveal® SNP Microarray Prenatal

Reveal answers to complex diagnostic questions

labcorp

Some genetic testing companies require patients to go elsewhere for diagnostic testing, in contrast, Labcorp 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 structural abnormalities and intellectual disabilities[1,2]
- **Detects copy neutral changes** associated with increased risk for autosomal recessive conditions, uniparental disomy (UPD)[5], identity by descent (IBD) and molar genotypes[6]
- **Whole genome coverage** helps resolve marker chromosome origin and identify unbalanced rearrangements undetectable by routine cytogenetics[1,7]
- **Ability to analyze DNA** from most tissues, including paraffin-embedded tissue or tissue that cannot be cultured

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

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

Reveal Prenatal - Features	
>284,000 arrays analyzed*	Extensive experience and database of abnormalities from specimens tested over time providing an exceptional resource to support interpretation
Low rate of variants of uncertain significance (VUS)	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[4]
Flexible test options	Various FISH, chromosome analysis, molecular, and microarray testing combinations to provide comprehensive results as well as to expedite results and reduce unnecessary testing
Reanalysis after delivery using pediatric cut-offs	One microarray platform for prenatal and pediatric testing, providing a robust database for interpretation. An additional benefit allows for a prenatal array to be recalculated using pediatric cutoffs without requiring an additional sample from the newborn. This allows the opportunity for faster results without additional cost to the patient

*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 needs
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

Why choose Integrated Genetics for your SNP microarrays?

Years of experience

- **35+** years experience performing genetic testing
- **11+** years experience in microarray testing
- **300+** combined years cytogenetics experience for array laboratory directors

Access to experts

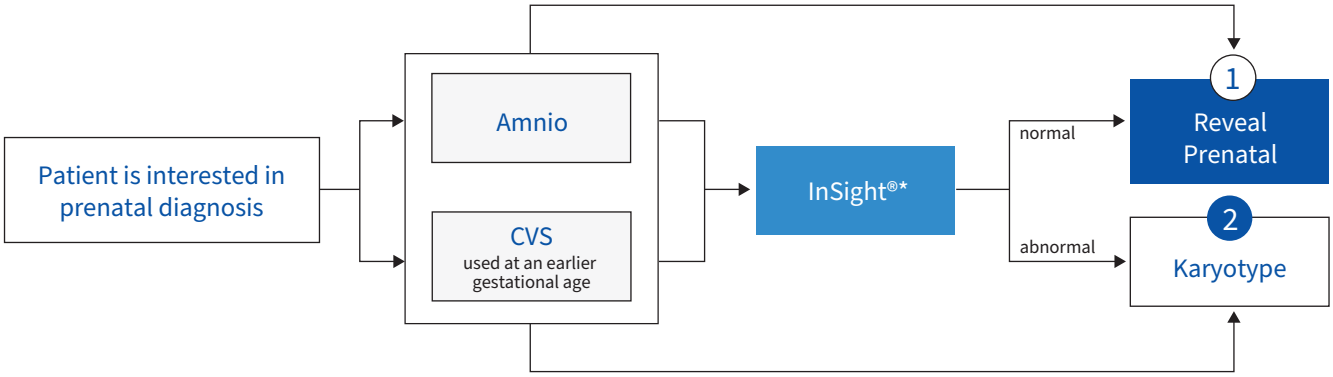
- **~140** genetic counselors nation-wide:
- **~100** dedicated to direct patient care
- **15+** lab directors involved in microarray analysis

In a study involving over 60,000 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 (1.8%) and abnormal ultrasound findings (5%).[3] This was even greater when the ultrasound revealed a major ultrasound abnormality (8.1-8.2%).[3]

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

	Pathogenic	IBD	UPD	Total
Major	4.90%	2.90%	0.40%	8.20%
Major - Heart	8.10%	2.40%	0.30%	10.80%
Multiple Anomalies	6.10%	5.70%	1.20%	13%
Multiple Anomalies - Heart	8.20%	3.50%	1.00%	12.70%
Nuchal Translucency	3.50%	4.40%	0.30%	8.20%
Diaphragmatic Hernia	6.20%	2.10%	0.50%	8.80%
Holoprosencephaly	6.90%	2.50%	2.90%	12.30%

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 suggest a common trisomy

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 Or (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 2020, 71% of patients paid $0 for Reveal Prenatal.

*Based on managed care claim data in 2020. For 36% of patients, insurance paid in full and 35% of patients had no patient responsibility (non-covered and coverage-related).[8]

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
Identification of 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 • Ability to identify triploidy
Identification of copy neutral changes	• IBD, UPD, complete molar pregnancy, and 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

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

Continuity of care, pioneering science, professional service

Labcorp delivers continuity of care for your patients, from carrier screening to noninvasive prenatal testing (NIPT, also known as cfDNA testing) to diagnostic testing.

We provide the scientific expertise you need, and the customer experience patients want.

Results reporting
Samples have a turnaround time of ~21-24 days from the date of pickup of a specimen for testing to when the result is released.

Extensive managed care contracts
Help patients maximize their benefits.

Convenient blood draws
We have a nationwide network of patient service centers, allowing for convenient access to sample collection. Visit **Labcorp.com** to find your nearest location.

Genetic counseling
Labcorp offers a national network of genetic counselors to help inform and support your patients. Patients can schedule an appointment online at **integratedgenetics.com/genetic-counseling**. Genetic education patient videos are available at **integratedgenetics.com/videos** to help patients learn about basic genetics concepts, navigate available testing options, and provide information to assist in understanding their results.

References
1. American College of Obstetricians and Gynecologists. Prenatal diagnostic testing for genetic disorders. Practice Bulletin No. 162 (2016). *Obstet Gynecol*. doi: 10.1097/AOG.0000000000001405.
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3. Schwartz, S. Prenatal SNP microarray analysis of over 60,000 patients: implications, importance and intriguing findings. Poster presented at: ACMG Annual Clinical Genetics Meeting; 2020 March 17-21; San Antonio, TX.
4. 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.
5. American College of Obstetricians and Gynecologists. The Use of Chromosomal Microarray Analysis ion Prenatal Diagnosis. Committee Opinion No. 581 (2013). *Obstet Gynecol*. doi: 10.1097/01.AOG.0000438962.16108.d1.
6. Xie Yingjun, Pei Xiaojuan, Dong Yu, Wu Huiqun, Shi Huijuan. Single Nucleotide Polymorphism-based Microarray Analysis for the Diagnosis of Hydatidiform Moles. *Mol Med Rep*. 2016; 14(1):137-144. doi: 10.3892/mmr.2016.5211.
7. 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.
8. Internal LabCorp billing data (2019).

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