Reveal®

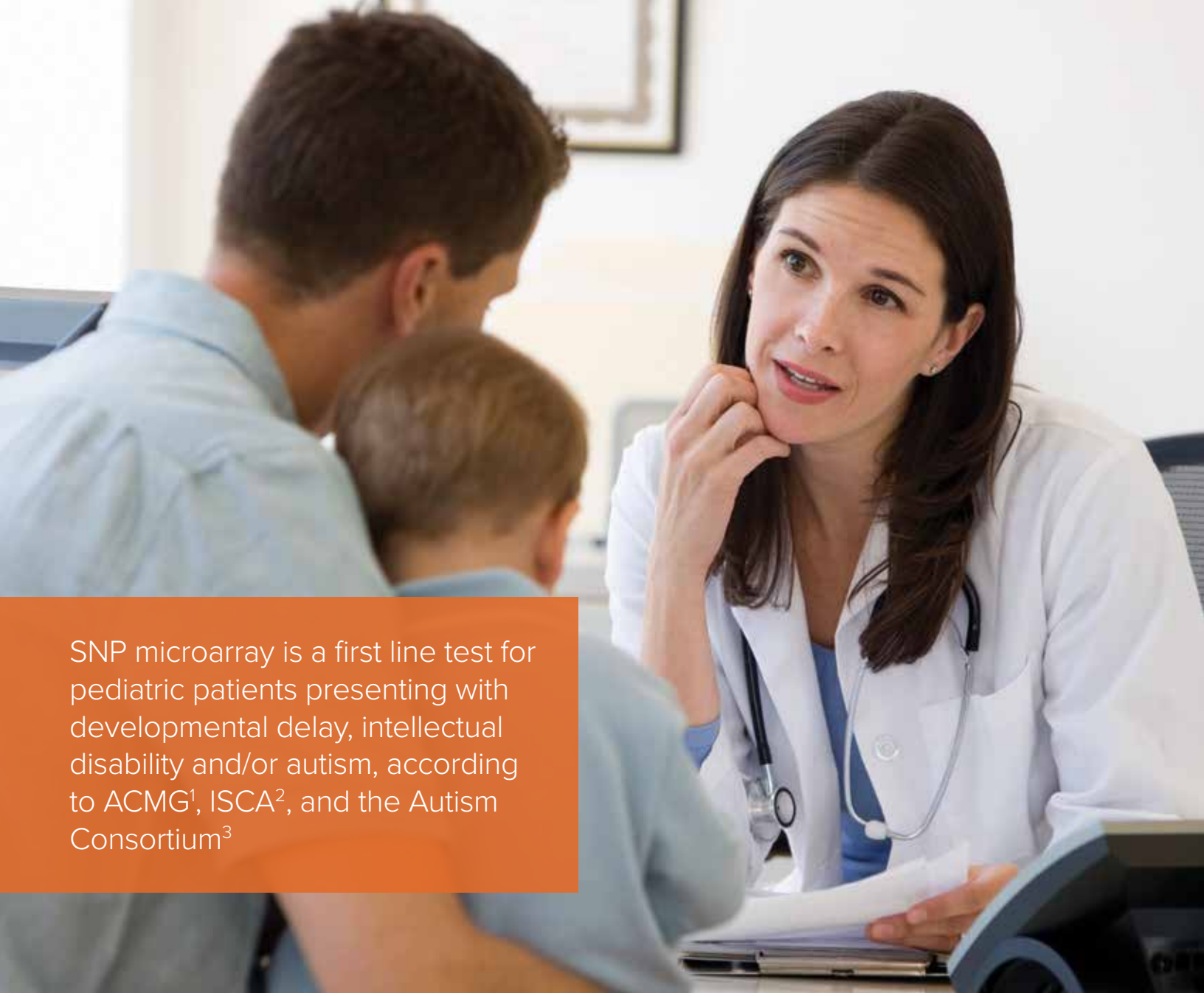
SNP Microarray

PEDIATRIC

Revealing answers early to help shorten the journey to diagnosis

Integrated GENETICS

LabCorp Specialty Testing Group



SNP microarray is a first line test for pediatric patients presenting with developmental delay, intellectual disability and/or autism, according to ACMG[1], ISCA[2], and the Autism Consortium[3]

Integrated Genetics, a LabCorp specialty testing group, offers a robust test menu and over 35 years of diagnostic experience to meet your pediatric genetic testing needs.

Whether a suspected genetic abnormality requires standard karyotyping, fluorescence in situ hybridization (FISH), single gene or panel testing, metabolic testing, microarray or whole exome sequencing, Integrated Genetics offers breadth and depth of diagnostic services.

In some patients, indications for testing may be apparent early in life, but in some cases, the phenotype evolves over time, and making a diagnosis becomes more challenging. In these cases, it may take years to uncover a definitive diagnosis, resulting in a costly and often frustrating journey for providers, patients and parents alike. Reveal Pediatric SNP Microarray provides a high resolution, whole genome analysis to identify copy number and copy neutral abnormalities to help provide diagnostic insights.

Reveal Pediatric can be performed via a cord blood sample or buccal swab in a neonate, or via a buccal swab or venous sample on an older child. Early diagnosis supports appropriate management and the provision of a realistic prognosis.

REVEAL PEDIATRIC - FEATURES	
>230,000 arrays analyzed	Extensive experience and database of abnormalities from specimens tested over time providing an exceptional resource to support interpretation
Flexible specimen type	Ability to analyze blood, as well as a buccal swab sample, to eliminate the stress of collecting a blood sample on a young child
High success rate	> 99.95% success rate on both blood and buccal samples providing highly reliable results
Low rate of variants of uncertain significance (VUS)	Low VUS rates of ~7.4% due to comprehensive variant database
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 a new sample

SERVICES	
Access to genetic experts	In-house 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	Broad in-network coverage and patient-friendly financial programs to increase patient access to genetic testing

Benefits of Reveal Pediatric

Easy-to-understand reports provide clinically relevant interpretation support

Whole genome coverage helps resolve marker chromosome origin and identify unbalanced rearrangements undetectable by routine cytogenetics[1,5,6]

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

Reanalysis of a prenatal sample using pediatric cut-offs for deeper interpretation, without requiring a new sample or incurring additional cost

Why choose Integrated Genetics for your SNP microarrays?

Depth of experience	Access to experts
35+ years experience performing genetic testing	**~140** genetic counselors nation-wide
11+ years experience in microarray testing	**~40** dedicated to clinician support
300+ combined years cytogenetics experience for array laboratory directors	**15+** lab directors involved in microarray analysis

Reveal Pediatric is a clinically validated, highly reliable test, supported by professional societies.

In a study involving over 21,000 patients referred for the investigation of developmental delay/intellectual disability, multiple congenital anomalies and/or autism spectrum disorder, the diagnostic yield of SNP microarray was 12.2%, about 10% more than standard karyotype alone.[2] In a separate study of over 36,000 patients with developmental delay/intellectual disability, SNP microarray identified a pathogenic finding in 19% of patents.[7]

When a clinical phenotype does not fit a specific syndrome, microarray analysis may identify the etiology. Clinical indications for Reveal Pediatric include[1,2,8]

- Multiple anomalies not specific to a well-defined syndrome
- Nonsyndromic developmental delay and/or intellectual disability
- Autism spectrum disorders
- Dysmorphic facial features
- Abnormal phenotype with apparently balanced translocation or marker chromosome
- Child that resembles a particular phenotype, but is not a perfect fit

1 in 6

~1 in 6 children has a developmental delay from mild speech delay to serious intellectual disability[9]

1 in 68

~1 in 68 children are diagnosed with autism spectrum disorder[10]

Specimen requirements

	DIRECT TESTING	
Specimen quantity	**Blood** • Children: 2-5 mL (less for newborns) • Infants: 2 mL • Blood should be collected in a Sodium Heparin (green top), EDTA (lavender top) or ACD-A (yellow top) tube *Special studies may call for an additional specimen. Contact the lab to discuss requirements.	**Buccal swab** • Minimum of 2 buccal swabs required • Buccal swab collection kit contains instructions for the use of a buccal swab
Expected turnaround time*	11-14 days	

*Turnaround time is calculated from the time the specimen arrives at the Center for Molecular Biology and Pathology (CMBP) in North Carolina.

A completed questionnaire for Reveal Pediatric SNP Microarray should accompany the specimen to aid in interpretation. The form is available on our website, or by calling 800-345-GENE (4363), or through your Integrated Genetics sales representative.

Integrated Genetics is contracted with 400+ managed care plans and regularly bills third party payers for testing reimbursement. This could help minimize or eliminate your patient's expense.

In 2018, 69% of patients paid $0 for Reveal Pediatric.

*Based on managed care claim data in 2018. For 52% of patients, insurance paid in full and 17% of patients had no patient responsibility (non-covered and coverage-related).[11]

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 number and copy neutral changes, triploidy and IBD • 100% ISCA constitutional gene and X chromosome coverage
Specimen types	• Whole blood • Buccal swab
Reporting cut-offs for copy-number changes	• ≥25 kb for genes with established clinical significance • Deletions: ≥ 200 kb with at least one OMIM gene • Duplications: ≥500 kb with at least one OMIM gene
Identification of copy neutral changes	• Regions of homozygosity suggestive of uniparental disomy or identity by descent, associated with an increased risk for recessive disease
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	• Familial testing using appropriate technologies is performed at no charge when required to interpret the proband's result

Toll-free (within the US)
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View short videos on genetic testing:
www.integratedgenetics.com/videos

Integrated GENETICS
LabCorp Specialty Testing Group

Test codes

Integrated Genetics client	LabCorp client
476 Reveal® SNP Microarray Pediatric	510002 SNP Microarray - Pediatric (Reveal®)
	052045 Chromosome Analysis with Reflex to SNP Microarray - Pediatric (Reveal®)
120 Cytogenetics Blood Chromosome Analysis	511535 Chromosome Five-Cell Count Plus Microarray (Reveal®) Whole Blood

*Indicate reflex to chromosome analysis

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