WOMEN'S HEATH AND GENETICS

Reveal® SNP Microarray Pediatric

Revealing answers early to help shorten the journey to diagnosis

labcorp

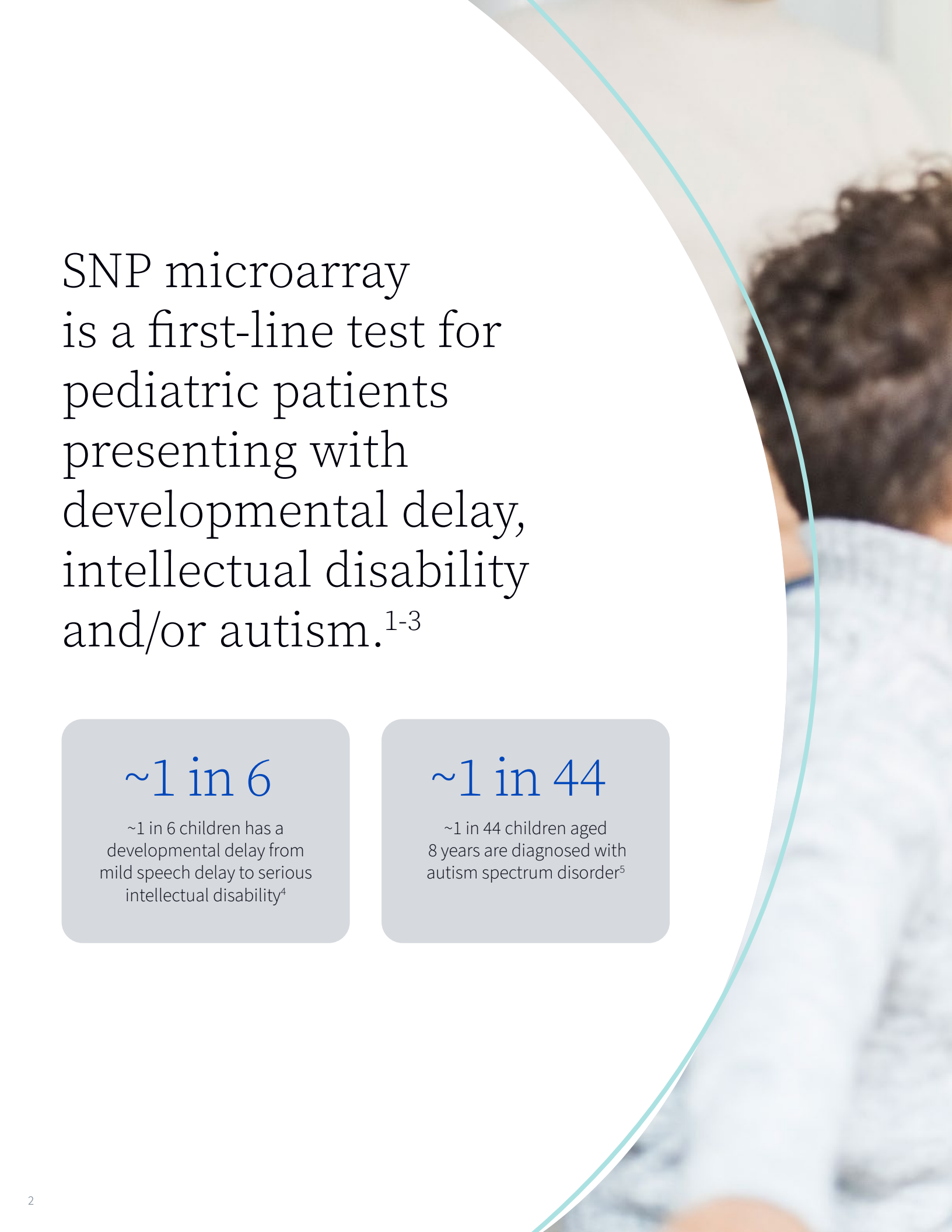
SNP microarray is a first-line test for pediatric patients presenting with developmental delay, intellectual disability and/or autism.[1-3]

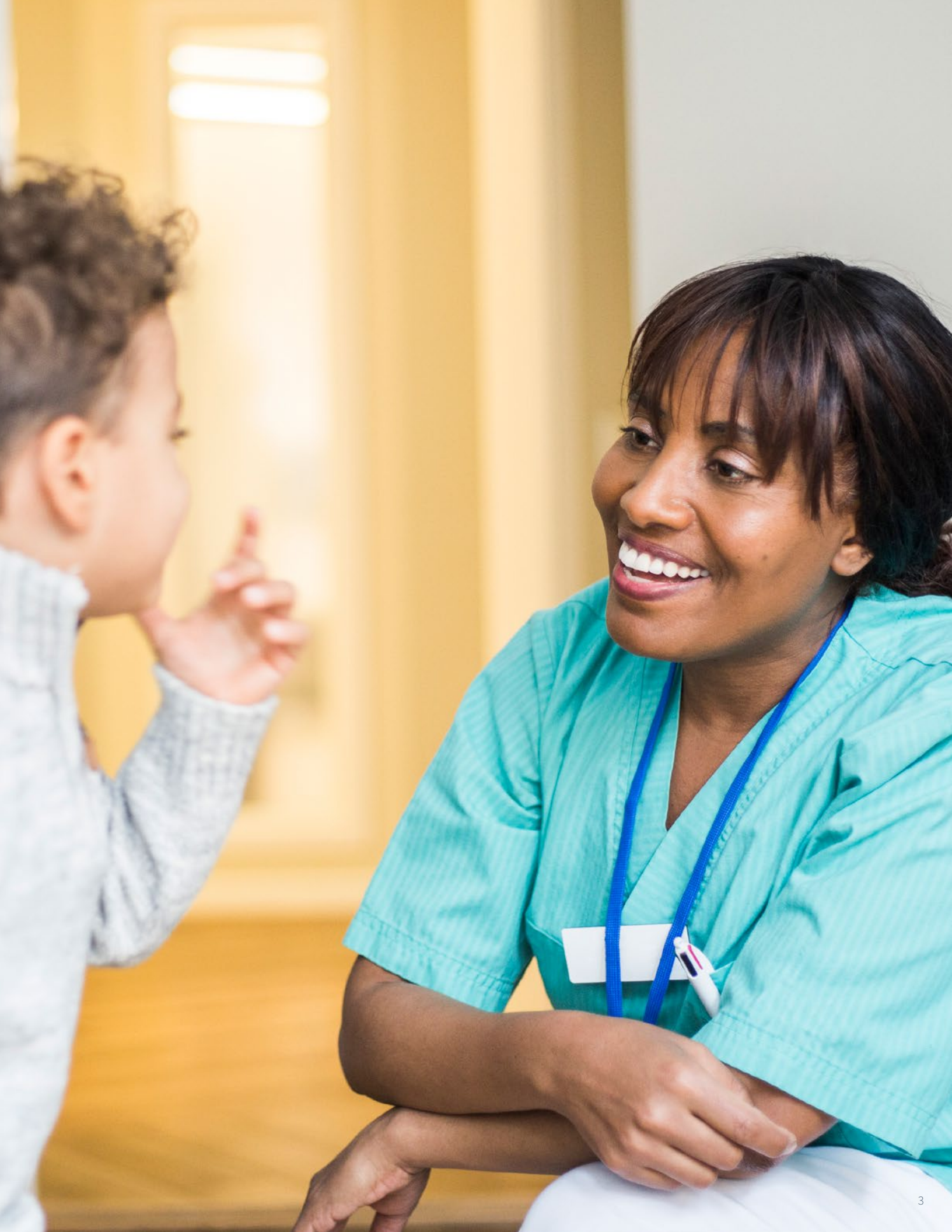
~1 in 6

~1 in 6 children has a developmental delay from mild speech delay to serious intellectual disability[4]

~1 in 44

~1 in 44 children aged 8 years are diagnosed with autism spectrum disorder[5]





Labcorp offers a robust test menu and over 35 years of diagnostic experience to meet your pediatric genetic testing needs

Labcorp offers breadth and depth of diagnostic services 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.

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

Prenatal, postnatal, products of conception

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
2,000+ 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[1,6,7]
- Detects copy neutral changes which may be associated with increased risk for autosomal recessive conditions, uniparental disomy (UPD) and identity by descent (IBD)
- Reanalysis of a prenatal sample using pediatric cut-offs for deeper interpretation, without requiring a new sample or incurring additional cost

Why choose Labcorp for your SNP microarrays?

Depth 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 nationwide
- **~40** genetic counselors dedicated to clinician support
- **15+** lab directors involved in microarray analysis

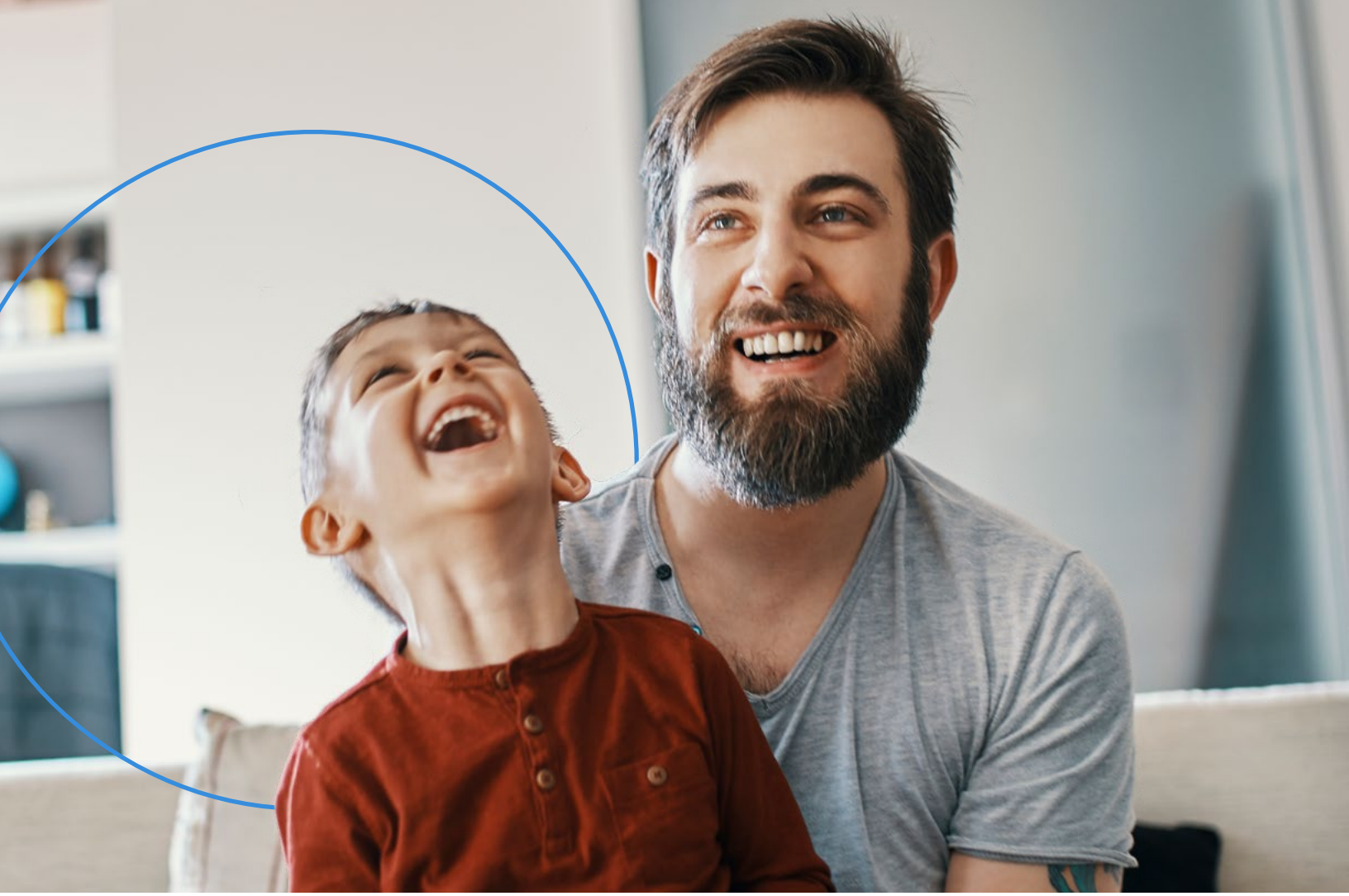
Reveal Pediatric is a clinically validated, highly reliable test, supported by professional societies

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.[8]

When a clinical phenotype does not fit a specific syndrome, microarray analysis may identify the etiology.

Clinical indications for Reveal Pediatric include[1-3]:

- 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 with symptoms that resemble, but are not a perfect fit for, a particular phenotype



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	**Buccal swab** • Minimum of 2 buccal swabs required • Buccal swab collection kit contains instructions for the use of a buccal swab *Special studies may call for an additional specimen. Contact the lab to discuss requirements.
Expected turnaround time*	14–17 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 Labcorp sales representative.

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

In 2022, 62% of patients paid $0 for Reveal Pediatric.

*Based on managed care claim data in 2022. For 48% of patients, insurance paid in full and 14% of patients had no patient responsibility (non-covered and coverage-related).[9]

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

Women's Health and Genetics client	Labcorp client
476 Reveal® SNP Microarray Pediatric	510002 SNP Microarray — Pediatric (Reveal®)
120 Cytogenetics Blood Chromosome Analysis	052045 Chromosome Analysis with Reflex to SNP Microarray — Pediatric (Reveal®)
	511535 Chromosome Five-Cell Count Plus Microarray (Reveal®) Whole Blood

References

1. Manning M, Hudgins L; Professional Practice and Guidelines Committee. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities *Genet Med*. 2010;12(11):742-745. doi:10.1097/GIM.0b013e3181f8baad
2. Miller DT, Adam MP, Aradhya S, et al. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. *Am J Hum Genet*. 2010;86(5):749-764. doi:10.1016/j.ajhg.2010.04.006
3. Shen Y, Dies KA, Holm IA, et al. Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics*. 2010;125(4):e727-e735. doi:10.1542/peds.2009-1684
4. Zablotsky B, Black LI, Maenner MJ, et al. Prevalence and Trends of Developmental Disabilities among Children in the United States: 2009-2017. *Pediatrics*. 2019;144(4):e20190811. doi:10.1542/peds.2019-0811.
5. Maenner MJ, Shaw KA, Bakian AV, et al. Prevalence and Characteristics of Autism Spectrum Disorder Among Children Aged 8 Years — Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2018. *MMWR Surveill Summ*. 2021;70(No. SS-11):1–16.
6. Savage MS, Mourad MJ, Wapner RJ. Evolving Applications of Microarray Analysis in Prenatal Diagnosis. *Curr Opin Obstet Gynecol*. 2011; 23(2):103-108. doi: 10.1097/GCO.0b013e32834457c7.
7. American College of Obstetricians and Gynecologists. Prenatal diagnostic testing for genetic disorders. Practice Bulletin No. 162 (2016). *Obstet Gynecol*. doi: 10.1097/AOG.0000000000001405.
8. Hochstenbach R, van Binsbergen E, Engelen J, et al. Array analysis and karyotyping: workflow consequences based on a retrospective study of 36,325 patients with idiopathic developmental delay in the Netherlands. *Eur J Med Genet*. 2009;52:161–169.
9. Internal Labcorp billing data (2022).

Call Us
Toll-free (within the US) at
800-345-GENE (4363)

Follow Us
@LabcorpWomensHealth
@labcorp

Visit Us
womenshealth.labcorp.com

Educational Videos
View short videos on genetic testing: womenshealth.labcorp.com/videos

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

labcorp