

Revealing Answers to Complex Questions

Reveal[®] SNP Microarray is a high density copy number array which enhances the detection of all chromosome abnormalities. It can also detect copy neutral changes such as uniparental disomy (UPD) and consanguinity.

Over 60,000 SNP Arrays Analyzed

- Enhanced detection of chromosomal abnormalities.
- Detects copy neutral changes, such as uniparental disomy (UPD) and consanguinity which helps identify candidate genes for recessive disorders.
- 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.
- More sensitive than routine cytogenetics in detecting small clinically-significant changes to identify:
 - Autism spectrum disorders¹
 - Genome imbalance in developmentally delayed children²

Clinical Indications Reveal may be Performed for:

- Individuals with non-syndromic congenital anomalies, dysmorphic features, developmental delay, mental retardation, intellectual disability, and/or autism spectrum disorders (ASD).
- Individuals with any of the above when previous chromosome analysis was normal.
- Detects small abnormalities that are typically undetectable by routine karyotype.
- Phenotypically symptomatic individuals with apparently balanced chromosome rearrangements or unidentified marker chromosomes.

American College of Medical Geneticists (ACMG) Practice Guidelines include:³

- "CMA [Cytogenetic microarray] testing for [copy number variation] CNV is recommended as a first-line test in the initial postnatal evaluation of individuals with the following:
 - Multiple anomalies not specific to a well-delineated genetic syndrome.
 - Apparently non-syndromic DD/ID [developmental delay/intellectual disability].
 - Autism spectrum disorders."
- "Appropriate follow up is recommended in cases of chromosome imbalance identified by CMA, to include cytogenetic/FISH studies of the patient, parental evaluation, and clinical genetic evaluation and counseling."

The Autism Consortium in Pediatrics 2010⁴

- "...our results suggest that CMA [Chromosomal microarray] with whole-genome coverage should be adopted as the national standard of care for genetic testing among patients with autism spectrum disorders."

* Database of over 60,000 samples run at LabCorp's Center for Molecular Biology and Pathology.

Pediatric



Reveal® SNP Microarray Product Offerings:

- Reveal® SNP Microarray Pediatric.
- Full Chromosome Analysis with reflex to Reveal®.
- Reveal® SNP Microarray and Abbreviated Chromosomal Analysis.

Specimen Requirements	
Blood	Buccal Swab†
<ul style="list-style-type: none">■ 1 tube of whole blood in a Na Heparin (green top) tube preferred (yellow or lavender top tubes also accepted)■ Children and Adults: 4 ml■ Infants: 2 ml <p>To reflex from a chromosome analysis, two tubes of blood are preferred and minimize turn around time</p>	<ul style="list-style-type: none">■ Buccal swab collection kit contains instructions for the use of a buccal swab■ Minimum of 2 buccal swabs required <p>†Buccal swab samples available for Reveal® SNP Microarray only</p>
A completed Clinical Questionnaire for Reveal® SNP Microarray – Pediatric must be completed, and is available on our website or by calling 800-345-GENE (4363).	

Rely on Integrated Genetics for all of your Microarray and Cytogenetic Testing Needs

- Cutting edge technology platform provides enriched gene coverage and enhanced detection of mosaicism.
- An extensive database* of abnormalities detected by high resolution microarray provides an exceptional reference to support interpretation.
- Easy-to-understand reports provide clinically relevant interpretations.

The Integrated Genetics Differences

- Integrated Genetics' comprehensive diagnostic services provide added convenience for your patient-appropriate microarray, cytogenetics, FISH, and molecular genetics testing needs.
- Genetics experts are readily accessible to answer questions.
- Integrated Genetics is contracted with over 400 health plans to help minimize patient out-of-pocket costs.
- Integrated Genetics is a leader in genetic testing with more than 150 genetic counselors and over 1,700 patient service centers.



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

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REFERENCES

- 1) Weiss, LA et al. Association between microdeletion and microduplication at 16p11.2 and autism. *N Engl J Med.* 2008; 358(7):667-675.
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- 3) Manning, M. and Hudgins, L. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. *Genet Med* 2010; 12(11):742-5.
- 4) Shen, Y et al. Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics* 2010;125:e727-e735; originally published online Mar 15, 2010;DOI: 10.1542/peds.2009-1684.



LabCorp Specialty Testing Group

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