Reveal[SM] SNP Microarray

Integrated GENETICS
LabCorp Specialty Testing Group

Revealing Answers to Complex Questions

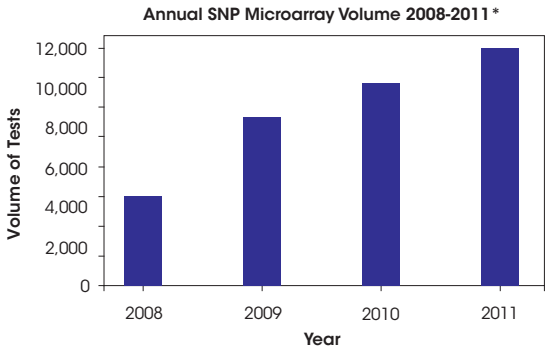
Reveal[SM] SNP Microarray is a high density copy number array which enhances the detection of all chromosome abnormalities. It can also detect copy neutral changes such as uniparental disomy (UPD) and consanguinity. Both of these are associated with an increase in risk for autosomal recessive conditions, and UPD is also associated with imprinting disorders.

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.
- 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.
- Contracted with close to 700 health plans to help minimize patient out-of-pocket costs.

Integrated Genetics Comprehensive Testing Services

Over 30,000 SNP arrays analyzed since 2008.*



American College of Medical Geneticists (ACMG) Practice Guidelines include:[1]

- *"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."*

* Database of over 30,000 samples run at LabCorp's Center for Molecular Biology and Pathology.

Clinical Indications:

- 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
- Phenotypically symptomatic individuals with apparently balanced chromosome rearrangements or unidentified marker chromosomes

> *"…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."*
> The Autism Consortium in *Pediatrics* 2010[2]

Reveal[SM] SNP Microarray Product Specifications and Advantages:

- More than 2.6 million copy and allele specific genomic sites
  - Probe median marker spacing in the International Standards for Cytogenomic Arrays (ISCA) genes ~384 bp
  - More than 750,000 SNP probes provide both genotyping and copy number analysis
  - More than 1.9 million region specific copy number probes
  - 100% ISCA constitutional gene and X chromosome coverage
- Highly sensitive in identifying extremely small genomic variations—especially important in detecting:
  - Autism spectrum disorders[3]
  - Genome imbalance in developmentally delayed children[4]
- Detection of UPD and consanguinity which helps identify candidate recessive disorders

Specimen Requirements:

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

To reflex from a chromosome analysis, two tubes of blood are preferred and minimize turn around time.

A completed Clinical Questionnaire for Reveal[SM] SNP Microarray - Pediatric **must** be completed, and is available at www.integratedgenetics.com or by calling 800-345-GENE (4363).

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

REFERENCES

1) 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.
2) Shen, Y et al. Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics* 2010; 125:e727-35.
3) Weiss, LA et al. Association between microdeletion and microduplication at 16p11.2 and autism. *N Engl J Med.* 2008; 358(7):667-675.
4) Gribble, SM et al. The complex nature of constitutional de novo apparently balanced translocations in patients presenting with abnormal phenotypes. *J Med Genet.* 2005; 42(1):8-16.

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