Integrated GENETICS
LabCorp Specialty Testing Group

LabCorp
Laboratory Corporation of America

Reveal
SNP Microarray

SAMPLE REPORT

City Hospital
123 City Avenue
Anywhere, ST 12345

LCLS Specimen Number:	**123-456-7891-0**	Account Number:	12345678
Patient Name:	**Doe, John**	Ordering Physician:	Ordering Doctor, MD
Date of Birth:	00/00/2011	Specimen Type:	**BLOOD**
Gender:	M	Date Collected:	02/01/2012
Patient ID:		Date Received:	02/02/2012
Lab Number:		CoPath Number:	
Indications:	Autism	Client Reference:	
Test:	**Chromosome Microarray**	Date Reported:	**02/11/2012**
Genotyping Targets:	2695000	Array Type:	SNP

MICROARRAY RESULT: 536 KB INTERSTITIAL DUPLICATION OF 7Q36.3->Q36.3

INTERPRETATION: POSSIBLE FAMILIAL VARIANT

arr 7q36.3(158,583,829-159,119,707)x3

The whole genome chromosome SNP microarray (REVEAL) analysis has identified an interstitial duplication of the chromosomal segment listed above. This interval includes one OMIM annotated gene*, *VIPR2*. At this time, no clinically established disorders have been reported with imbalance in this region, although this could change as studies progress.

In order to further evaluate clinical relevance, parental metaphase FISH analysis utilizing interval specific BAC probes may be considered to determine whether this variation has a familial derivation or is a de novo change more likely to be clinically significant. **In general, duplications are clinically tolerated better than deletions, and thus are found more frequently as familial variants.** Genetic counseling is recommended.

No other alterations were detected within the present reporting criteria.

The follow-up parental blood (green top sodium heparin) should be submitted under test code 510320. There is no charge associated with this follow-up test for up to two family members. Policy details are available for view on www.labcorp.com. The current sample will be retained for 13 months as a positive control for potential parental FISH follow-up studies. Please provide a new specimen on this patient if submitting parental samples after this date.

**For OMIM genes listed on NCBI, please bookmark the following URL: http://1.usa.gov/pkjEDG; click on the desired chromosome number, then enter start and end linear positions in the upper and lower boxes on the left menu bar, and click "Go" for the inclusive list.*

Methodology

SNP microarray analysis was performed using the Affymetrix Cytoscan HD platform which uses over 743,000 SNP probes and 1,953,000 NPCN probes with a median spacing of 0.88 kb. 250ng of total genomic DNA extracted from lymphocytes was digested with NspI and then ligated to NspI adaptors, respectively, and amplified using Titanium Taq with a GeneAmp

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Lab Number:		CoPath Number:	
Indications:	Autism	Client Reference:	

PCR System 9700. PCR products were purified using AMPure beads and quantified using NanoDrop 8000. Purified DNA was fragmented and biotin labeled and hybridized to the Affymetrix Cytoscan HD GeneChip. Data was analyzed using Chromosome Analysis Suite. The analysis is based on the GRCh37/hg19 assembly.

Positive evaluation criteria include:

* DNA copy gain/loss within a known clinically significant gene region of 50 Kb or greater.
* DNA copy number loss of >200 kb or gain >500 kb outside known clinically significant regions with at least one OMIM annotated gene or within a region of clear clinical significance.
* UPD testing is recommended for patient results demonstrating a long contiguous region of homozygosity in a single chromosome of >20 Mb interstitially or >10 Mb telomerically (15 and 8 Mb, respectively, for imprinted chromosomes).
* Contiguous homozygosity of >8 Mb within multiple chromosomes suggests common descent. These regions of potential recessive allele risk are designated.
* A high level of allele homozygosity due to numerous contiguous short runs (associated with a geographically or socially limited gene pool) is reported at the 99th percentile.

Truly balanced chromosome alterations will not be detected by this analysis. The threshold for mosaicism is variable, depending on the size of segment. Empiric studies have detected whole chromosome 22 mosaicism below 10.0%. CNVs cited in the Database of Genomic Variants are not reported.

This test was developed and its performance characteristics determined by Laboratory Corporation of America Holdings (LabCorp). It has not been cleared or approved by the Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

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Board Certified Cytogeneticist

Test Site: LabCorp
1904 Alexander Drive, RTP, NC 27709-0153 (800) 533-0567