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, Jane**	Ordering Physician:	Ordering Doctor, MD
Date of Birth:	00/00/2010	Specimen Type:	**BLOOD**
Gender:	F	Date Collected:	02/01/2012
Patient ID:		Date Received:	02/02/2012
Lab Number:		CoPath Number:	
Indications:	Congenital Heart Defect; Developmental Delay	Client Reference:	
Test:	**Chromosome Microarray**	Date Reported:	**02/11/2012**
Genotyping Targets:	2695000	Array Type:	SNP

MICROARRAY RESULT: NORMAL FEMALE DOSAGE; LONG CONTIGUOUS REGIONS OF HOMOZYGOSITY IN MULTIPLE CHROMOSOMES

INTERPRETATION: APPARENT COMMON DESCENT

arr (1-22,X)x2

The whole genome chromosome SNP microarray (REVEAL)analysis did not demonstrate significant DNA copy number changes within the clinically significant criteria for this analysis indicated below.

There are, however, extended contiguous regions of allele homozygosity (>8 Mb) observed in multiple chromosomes that is consistent with common descent (related parents). These may be added to provide a measure of identity by descent which in this case is equivalent to **first cousin** parentage. Multiple generations of consanguinity can increase the levels of allele homozygosity.

If an autosomal recessive disorder is being considered in the differential diagnosis, candidate genes may be checked for inclusion in homozygotic regions. A candidate gene found in a homozygotic region increases the correlation with that recessive disorder (long contiguous regions are listed below).

Genetic counseling is recommended.

Bp linear position (start-end):

chr1: 59,716,213 - 84,855,605
chr2: 176,798,160 - 192,017,771
chr2: 195,356,527 - 229,022,909
chr4: 23,326,312 - 42,205,264
chr4: 115,510,414 - 138,371,982
chr7: 128,797,066 - 142,338,469
chr7: 7,427,443 - 32,815,508
chr11: 78,889,853 - 101,156,501
chr12: 118,005,882 - 128,927,440
chr20: 29,448,795 - 38,242,301
chr20: 15,481,965 - 26,289,925
chr21: 22,033,862 - 30,280,016
chr21: 34,291,639 - 43,453,305
Total: 224.89 Mb (6.3% of autosomal genome)

**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.*

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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 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