Integrated GENETICS
LabCorp Specialty Testing Group

Reveal SNP Microarray

Client/Sending Facility:
LABCORP OF AMERICA
CMBP
1912 ALEXANDER DR
RTP, NC 27709
Ph: (919)361-7700
Fax: (919) 361-7296 NCB-13

LCLS Specimen Number:	**106-225-9016-0**	Account Number:	90001555
Patient Name:	**REPORT, SAMPLE**	Ordering Physician:	
Date of Birth:	01/01/2011	Specimen Type:	**BLOOD**
Gender:	M	Client Reference:	
Patient ID:		Date Collected:	04/15/2024
Lab Number:	YU24-40012 G	Date Received:	04/15/2024
Indications:	Autism	Date Reported:	**05/17/2024**
Test:	**Chromosome Microarray**		
Genotyping Targets:	2772571	Array Type:	SNP

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

INTERPRETATION: VARIANT OF UNCERTAIN SIGNIFICANCE

arr[hg19]7q36.3(158,583,829-159,119,707)x3

The whole genome SNP microarray (Reveal) analysis has detected an interstitial duplication of the chromosomal segment listed above. This interval includes 3 OMIM genes (*ESYT2, DYNC2I1, VIPR2*). At this time, no clinically established disorders have been reported with duplication of this region, although this could change as studies progress. In general, duplications are clinically tolerated better than deletions, and thus are found more frequently as familial variants.

In order to further evaluate clinical relevance, parental analysis is necessary to determine whether this alteration represents a familial variant or a *de novo* change more likely to be clinically significant.

No other DNA copy number changes or copy neutral ROH were detected within the present reporting criteria. **Genetic counseling is recommended.**

The follow-up parental blood (green top sodium heparin) should be submitted under test code **511780 (qPCR). There is no charge associated with this follow-up test for up to two family members.** Please reference the proband name, date of birth, and specimen number when submitting parental or familial samples. Billing 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 follow-up studies. Please provide a new specimen on this patient if submitting parental samples after this date. **Note to New York clients:** the current sample will only be retained for 60 days from receipt date in the lab for potential parental/familial follow-up studies.

Methodology:
SNP microarray analysis was performed using the Cytoscan ® HD Accel platform which uses 2,029,441 nonpolymorphic copy number probes and 743,130 SNP probes for LOH/AOH analysis and relationship assessment. The array has an average intragenic spacing of 0.818 kb and average intergenic spacing of 1.51 kb. Total genomic DNA was extracted from the sample type provided, digested with Xce1, and then ligated to Xce1 adaptors. PCR products were purified and quantified. Purified DNA was fragmented, biotin labeled, and hybridized to the Cytoscan ® HD Accel Gene Chip. Data were analyzed using

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Chromosome Analysis Suite. The analysis is based on the GRCh37/hg19 assembly. This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug administration.

Positive evaluation criteria include:
* DNA copy number loss of >200 kb or gain >500 kb outside known clinically significant regions with at least one OMIM gene.
* DNA copy gain/loss within or including a known clinically significant gene of 25 kb or greater.
* 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 one region that is >8 Mb but below reporting criteria for UPD will be reported as increased risk of recessive disorder.
* 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.

SNP chromosomal microarray cannot detect:
* Truly balanced chromosome alterations
* Sequence variants
* Small insertions and deletions (indels)
* Changes in regions not represented by probes on the array
* Tetraploidy
* Low level mosaicism
* Whole chromosome uniparental heterodisomy without parental specimens
* Imbalances in the mitochondrial genome

Single gene partial or intragenic copy number variants (CNVs) detected by an independent technology such as next generation sequencing (NGS) may not be detectable by microarray. The ability to detect the CNV is dependent on size and probe coverage. The threshold for mosaicism is variable, depending on the size of the segment and array quality. Empiric studies have detected mosaicism for trisomy of a whole autosome below 10.0%. CNVs that are known to be common in the population may not be reported.

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Gloria Haskell

GLORIA HASKELL, PHD, FACMG

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