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
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LCLS Specimen Number:	**146-225-5001-0**	Account Number:	90001555
Patient Name:	**SAMPLE REPORT, 511966**	Ordering Physician:	
Date of Birth:	10/11/1989	Specimen Type:	**AMNIOTIC FLUID**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	05/24/2019
Lab Number:	YU19-43601 MG	Date Received:	05/24/2019
Indications:		Date Reported:	**05/24/2019**
Test:	**Chromosome Microarray**		
Genotyping Targets:	2695000	Array Type:	SNP

MICROARRAY RESULT: 3.64 MB INTERSTITIAL DELETION OF 17P11.2->P11.2

INTERPRETATION: FEMALE WITH SMITH-MAGENIS SYNDROME

arr[hg19] 17p11.2(16,772,264-20,413,433)x1

The whole genome SNP microarray (Reveal) analysis has identified a female with an interstitial deletion of the chromosome segment listed above. The deleted region includes numerous OMIM genes [start:*TNFRSF13B* to end:*SPECC1*], including *RAI1*, the gene responsible for features of Smith-Magenis syndrome. Smith-Magenis syndrome is characterized by distinctive physical features, developmental delay, cognitive impairment, and behavioral abnormalities (see reference below).

Parental FISH follow-up analysis is recommended to confirm a *de novo* origin and rule out a balanced rearrangement with high recurrence risk.

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 **511770 (FISH). Charges will apply.** 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.

Maternal cell contamination studies will be reported under separate cover, if ordered.

Reference:

Smith ACM, Boyd KE, Elsea SH, et al. Smith-Magenis Syndrome. 2001 Oct 22 [Updated 2012 Jun 28]. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2013. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1310/

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

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DNA 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:

* Copy numbers gains >2Mb and losses >1Mb, including at least one OMIM gene are reported in this analysis.
* Gains/losses of >50 Kb within clinically significant genes or regions. On request, candidate genes can be analyzed at a much lower threshold, depending on the gene specific marker density.
* UPD testing is recommended for patient results demonstrating a long contiguous region of homozygosity (ROH) 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 short ROH (associated with a geographically or socially limited gene pool) is reported at the 99th percentile.
* Triploid DNA that normalizes to 2 copies in standard CGH array analysis, is detectable in this allele specific microarray by 2:1 allele dosage ratios generated within each chromosome.

Truly balanced chromosome alterations will not be detected by this analysis, although cryptic imbalance associated with some translocations are readily detected due to the dense whole genome probe coverage. 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 LabCorp. It has not been cleared or approved by the Food and Drug administration.

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Test: **FISH Amnio Rfx CMA or Chromo**

(Updated Report)

This report amends previous report dated 05/24/2019

Cells Counted: 50 Cells Analyzed: 50

FISH RESULT: NORMAL FEMALE BY FISH

INTERPRETATION:

nuc ish Xcen(DXZ1x2),Ycen(DYZ3x0),13q14(RB1x2),
18cen(D18Z1x2),21q22.12(RCAN1x2)

Fluorescence in situ hybridization (FISH) of uncultured amniocytes has revealed two hybridization signals for chromosomes 13, 18, 21, and X in 50 interphase cells analyzed. No cells with a Y hybridization signal were observed. This is consistent with a normal female disomic for chromosomes 13, 18, and 21.

Results should be interpreted within the context of a full cytogenetic analysis, family history and ultrasound findings.

Threshold:
Fewer than 10% of interphase cells with three signals is considered background while greater than 60% is consistent with full trisomy.

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 U.S. Food and Drug Administration. The DNA probe vendor for this study was Kreatech (Leica BioSystems).

Director, PhD

GNEAS1

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Technical component performed by Laboratory Corporation of America Holdings,
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