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:	**146-225-5000-0**	Account Number:	90001555
Patient Name:	**SAMPLE REPORT, 052104**	Ordering Physician:	
Date of Birth:	06/12/1985	Specimen Type:	**AMNIOTIC FLUID**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	05/24/2019
Lab Number:	YU19-43595 GA	Date Received:	05/24/2019
Indications:		Date Reported:	**05/24/2019**

Test: **Chromosome Amnio RFX CMA**

(Updated Report)

This report amends previous report dated 05/24/2019

Cells Counted:	15	Cells Karyotyped:	2
Colonies Counted:	15	Band Resolution:	450
Cells Analyzed:	15		

CYTOGENETIC RESULT: 46,XX

INTERPRETATION: NORMAL FEMALE KARYOTYPE

Cytogenetic analysis of cultured amniocytes has revealed a FEMALE karyotype with an apparently normal GTG banding pattern in all in-situ colonies or subcultured metaphases analyzed.

This result does not exclude the possibility of subtle rearrangements below the resolution of cytogenetics or congenital anomalies due to other etiologies.

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:	**146-225-5000-0**	Account Number:	90001555
Patient Name:	**SAMPLE REPORT, 052104**	Ordering Physician:	
Date of Birth:	06/12/1985	Specimen Type:	**AMNIOTIC FLUID**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	05/24/2019
Lab Number:	YU19-43595 GA	Date Received:	05/24/2019
Indications:		Date Reported:	**05/24/2019**
Test:	**Chromosome Microarray**		
Genotyping Targets:	2695000	Array Type:	snp

MICROARRAY RESULT: 1.40 MB INTERSTITIAL DELETION OF 17Q12->Q12

INTERPRETATION: FEMALE WITH 17Q12 MICRODELETION

arr[hg19] 17q12(34,815,072-36,215,917)x1

The whole genome SNP microarray (Reveal) analysis revealed a female with an interstitial deletion of the chromosomal segment listed above. This interval includes numerous OMIM genes [start:*ZNHIT3* to end:*HNF1B]*.

Deletion of this region that includes the *HNF1B* gene (OMIM:189907) is associated with a variable phenotype that may include one or more of the following: cystic renal disease, pancreatic atrophy, liver abnormalities, cognitive impairment and structural brain abnormalities, maturity-onset diabetes, and epilepsy. Prenatal cases have also been reported with ultrasound abnormalities such as cystic kidney, poly/oligohydramnios and diaphragmatic hernia (see references). ***Due to the variability of this disorder, precise prenatal prediction of a phenotype is not possible.***

Parental follow-up analysis is recommended to determine whether this deletion represents an inherited or *de novo* change.

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 **511810 (qPCR). There is no charge for qPCR follow-up studies to prenatal arrays.** If parental studies are negative, consistent with a *de novo* CNV, parental FISH may be considered to rule out a rare balanced rearrangement (test code 511680). Parental follow-up for *de novo* CNVs is available at a charge and may take up to 56 days for results. Please reference the prenatal specimen number when submitting parental or familial samples. Billing policy details are available for view on www.labcorp.com.

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:	**146-225-5000-0**	Account Number:	90001555
Patient Name:	**SAMPLE REPORT, 052104**	Ordering Physician:	
Date of Birth:	06/12/1985	Specimen Type:	**AMNIOTIC FLUID**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	05/24/2019
Lab Number:	YU19-43595 GA	Date Received:	05/24/2019

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

References:

Goumy et al.,(2015) Am J Med Genet A 167A(1):250-3. PMID# 25425496
George et al., (2012) Mol Syndromol 2(2):72-75.PMID#22511894
Nik-Zainal et al.,(2011) J Med Genet 48(3):197-204.PMID#21278390
Moreno-De-Luca et al.,(2010) Am J Hum Genet 87(5):618-30.PMID# 21055719
Nagamani et al.,(2010) Eur J Hum Genet 18(3):278-84. PMID# 19844256

Hendrix et al.,(2012) Fetal Diagn Ther 31(2):129-33. PMID# 22178801
Chen et al., (2013) Taiwan J Obstet Gynecol 52(4):551-7.PMID#24411042

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 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 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.
* Triploid DNA that normalizes to 2 copies in standard CGH array analysis, are 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 imbalances 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.

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:	**146-225-5000-0**	Account Number:	90001555
Patient Name:	**SAMPLE REPORT, 052104**	Ordering Physician:	
Date of Birth:	06/12/1985	Specimen Type:	**AMNIOTIC FLUID**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	05/24/2019
Lab Number:	YU19-43595 GA	Date Received:	05/24/2019

Director, PhD

GNEAS1

Arundhati Chatterjee, MD
Medical Director
Stuart Schwartz, PhD
National Director of Cytogenetics

Technical component performed by Laboratory Corporation of America Holdings,
1904 TW Alexander Drive , RTP , NC , 27709-0153 (800) 345-4363