Prenatal Chromosome Microarray

Specimen Type

Comment: AMNIOTIC FLUID

# of Genotyping Targets

Comment: 2695000

Array Type

SNP

Diagnosis

NORMAL FEMALE

Interpretation

Comment: arr(1-22,X)x2

The whole genome chromosome SNP microarray (Reveal) analysis was normal. No significant DNA copy number changes or copy neutral regions within the 2.695 million region specific SNP and structural targets were detected under the present reporting criteria indicated below.

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

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 Chromeosme Analysis Suite. The analysis is based on the GRCh37/hg19 assembly.
<table>
<thead>
<tr>
<th>TESTS</th>
<th>RESULT</th>
<th>FLAG</th>
<th>UNITS</th>
<th>REFERENCE INTERVAL</th>
<th>LAB</th>
</tr>
</thead>
</table>

Positive evaluation criteria include:
* Copy numbers gains >2Mb and losses >1Mb, including at least one OMIM annotated 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, 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.

Director Review: M. Katharine Rudd, PhD, FACMG
Preauthorization: Will Follow

For inquiries, the physician may contact Branch: 800−222−7566 Lab: 800−735−4087

Dir: Arundhati Chatterjee, MD
1904 TW Alexander Drive Suite C, RTP, NC 27709−0153
MICROARRAY RESULT: NORMAL FEMALE

INTERPRETATION:

arr(1-22,X)x2

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LCLS Specimen Number: 238-225-9009-0
Patient Name: SAMPLE REPORT, 510100
Date of Birth: 05/22/1946
Gender: F
Patient ID: 
Lab Number: YU16-67590

Account Number: 9000999
Ordering Physician: 
Specimen Type: AMNIOTIC FLUID
Client Reference: 
Date Collected: 08/25/2016
Date Received: 08/26/2016

M. Katharine Rudd, PhD, FACMG
Board Certified Cytogeneticist

Arundhati Chatterjee, MD
Medical Director
Peter Papenhausen, 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

Professional Component performed by LabCorp CLIA 34D1008914, 1904 TW Alexander Dr, Research Triangle Park, NC 27709. Medical Director, Arundhati Chatterjee, MD. Integrated Genetics is a brand used by Esoterix Genetic Laboratories, LLC, a wholly-owned subsidiary of Laboratory Corporation of America Holdings.

This document contains private and confidential health information protected by state and federal law.
The whole genome SNP microarray (Reveal) copy number analysis has identified a female with an interstitial deletion of the chromosomal segment listed above. This interval includes 14 OMIM genes [start:ZNF74 to end:HIC2], and spans LCR22 B−D.

The deletion is localized to the distal end of the 2.5 Mb DiGeorge/VCF region, and does not include the critical genes implicated in the pathogenesis of those syndromes. However, a variable phenotype associated with this "central" 22q11 microdeletion has been described. Common features include developmental delay, intellectual disability, cardiac defects, and dysmorphic features (see references). Clinically normal carriers have also been reported suggesting a reduced penetrance and/or variable expressivity, which may be influenced by genetic and/or environmental factors. Therefore, prenatal prediction is difficult.

Parental follow-up analysis is recommended to determine inheritance.
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 tube) should be submitted under the test code 511810 (qPCR). There is no charge for this parental follow up of array results. Please reference the prenatal specimen number when submitting parental or familial samples. Billing policy details are available for view on www.labcorp.com.

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**Director Review**
M. Katharine Rudd, PhD, FACMG

**Preauthorization**
Will Follow

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For inquiries, the physician may contact Branch: 800−222−7566 Lab: 800−735−4087
Test: Prenatal Chromosome Microarray

Genotyping Targets: 2695000

Array Type: SNP

MICROARRAY RESULT: 1.08 MB INTERSTITIAL DELETION OF 22Q11.2->Q11.2

INTERPRETATION: FEMALE WITH 22q11.21 CENTRAL MICRODELETION: LCR22 B-D

$$\text{arr[hg19]} \ 22q11.21\ (20,717,654-21,798,907) \times 1$$

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