

City Hospital
123 City Avenue
Anywhere, ST 12345

LCLS Specimen Number: 123-456-7891-0

Patient Name: **Doe, Jane**

Date of Birth: 00/00/1980

Gender: F

Patient ID:

Lab Number:

Indications: Cystic Hygroma

Account Number: 12345678

Ordering Physician: Ordering Doctor, MD

Specimen Type: **AMNIOTIC FLUID**

Date Collected: 02/01/2012

Date Received: 02/02/2012

CoPath Number:

Client Reference:

Test: **Prenatal Chromosome
Microarray**

Date Reported: **02/11/2012**

Genotyping Targets: 2695000

Array Type: SNP

MICROARRAY RESULT: NORMAL FEMALE

INTERPRETATION:

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.

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:

- 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 custom clinically significant gene set. On request candidate genes can be analyzed at a much lower threshold, depending on 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 >8Mb within multiple chromosomes suggests common descent. These regions of potential recessive allele risk are designated prenatally only when the total is consistent with first cousin parentage or greater.
- 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. Complete moles are accurately detected by the presence of whole genome allele homozygosity.

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%.

This test is not designed as a SNP-based disease association linkage analysis assay. 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

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