Integrated GENETICS
LabCorp Specialty Testing Group

LabCorp
Laboratory Corporation of America

Reveal
SNP Microarray

SAMPLE REPORT

City Hospital
123 City Avenue
Anywhere, ST 12345

LCLS Specimen Number:	**123-456-7891-0**	Account Number:	12345678
Patient Name:	**Doe, Jane**	Ordering Physician:	Ordering Doctor, MD
Date of Birth:	00/00/1974	Specimen Type:	**POC**
Gender:	F	Date Collected:	01/30/2012
Patient ID:		Date Received:	02/02/2012
Lab Number:		CoPath Number:	
Indications:	Molar Pregnancy	Client Reference:	
Test:	**POC/Tissue Microarray**	Date Reported:	**02/11/2012**
Genotyping Targets:	2695000	Array Type:	SNP

MICROARRAY RESULT: NORMAL FEMALE DOSAGE: COMPLETE CONTIGUOUS HOMOZYGOSITY ON ALL CHROMOSOMES OBSERVED

INTERPRETATION: COMPLETE MOLAR PREGNANCY

arr(1-22,X)x2 hmz

The whole genome SNP microarray (REVEAL) analysis demonstrated no copy number change with the present reporting criteria indicated below.

There was, however, total genomic allele homozygosity which is associated with a complete molar pregnancy. In most cases the androgenic origin of a complete mole is the result of a duplication of a haploid paternal X sperm (23,X) penetrating an "empty ovum" lacking functional maternal DNA. Thus the presence of a double haploid paternal sperm would be identified by the complete homozygosity as observed in this patient.

Patients with a complete hydatidiform mole are at an increased risk of development of persistent gestational trophoblastic disease (PGTD). Choriocarcinoma, a malignant neoplasm of the trophoblast, occurs in about 2 to 3% of patients with a complete mole (Joneborg U, et. al. J Reprod Med. 2011;56(11-12):511-4).

There is a small but significant risk of recurrence. Recurrence can be either a complete or partial mole (Al-Ghamdi AA.J Family Community Med. 2011;18(3):159-61).

Prenatal diagnosis is recommended for all future pregnancies. Genetic counseling is also recommended.

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 human genome version GRCh37/hg19.

Positive evaluation criteria include:

* Copy numbers gains >2 Mb and losses >1 Mb, including at least one OMIM annotated gene are reported in this analysis.
* Gains/losses of >100 Kb within a custom clinically significant gene set. On request, candidate genes can be analyzed at a much lower threshold, depending on the gene specific marker density.
* DNA copy gain/loss of whole chromosomes with at least 10% fetal origin of the DNA tested.
* Maternal cell contamination (MCC) is detected by comparison of abnormal dosage allele combinations as well as normal dosage mixes of fetal and maternal alleles.

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* Complete moles are accurately detected by the presence of whole genome allele homozygosity (~50% hmz in rare dispermy moles).
* Triploid tissue that normalizes to 2 copies in standard array analysis, are detectable in this allele specific microarray by 2:1 heterozygote allele ratios generated within each chromosome by the software.
Truly balanced chromosome alterations (generally not the cause of miscarriage) will not be detected by this analysis. 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 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.

Board Certified Cytogeneticist

Test Site: LabCorp
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