Integrated GENETICS
LabCorp Specialty Testing Group

Reveal SNP Microarray

Client/Sending Facility:
LABCORP OF AMERICA
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1912 ALEXANDER DR
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Ph: (919)361-7700
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LCLS Specimen Number:	**106-225-9011-0**	Account Number:	90001555
Patient Name:	**REPORT, SAMPLE**	Ordering Physician:	
Date of Birth:	03/18/1990	Specimen Type:	**POC**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	04/15/2024
Lab Number:	YU24-40002 G	Date Received:	04/15/2024
Indications:	Molar Pregnancy	Date Reported:	**05/10/2024**
Test:	**POC/Tissue Reveal(SM) IG CMA**		
Genotyping Targets:	2772571	Array Type:	SNP

MICROARRAY RESULT: NORMAL FEMALE DOSAGE WITH TOTAL GENOMIC ALLELE HOMOZYGOSITY

INTERPRETATION: COMPLETE HYDATIDIFORM MOLE

arr(X,1-22)x2 hmz

The whole genome SNP microarray (Reveal) analysis has identified a female with no copy number changes within 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 from duplication of a haploid paternal X sperm (23,X) and fertilization of an oocyte lacking functional maternal DNA. The presence of a duplicated haploid sperm is identified by the complete homozygosity, as observed in this tissue.

Patients with a complete hydatidiform mole are at 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 complete mole (see references). There is a small but significant risk of recurrence. Recurrence can be either complete or partial mole (see references).

Prenatal diagnosis for all future pregnancies and genetic counseling are recommended.

References:

Joneborg U, et al. Choriocarcinoma following ovarian hydatidiform mole: a case report. J Reprod Med 2011 Nov-Dec; 56(11-12):511-4. PMID: 22195336.

Al-Ghamdi AA. Recurrent hydatidiform mole: A case report of six consecutive molar pregnancies complicated by choriocarcinoma, and review of the literature. J Family Community Med 2011 Sep;18(3):159-61. PMID 22175045.

Methodology:

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SNP microarray analysis was performed using the Cytoscan ® HD Accel platform which uses 2,029,441 nonpolymorphic copy number probes and 743,130 SNP probes for LOH/AOH analysis and relationship assessment. The array has an average intragenic spacing of 0.818 kb and average intergenic spacing of 1.51 kb. Total genomic DNA was extracted from the sample type provided, digested with Xce1, and then ligated to Xce1 adaptors. PCR products were purified and quantified. Purified DNA was fragmented, biotin labeled, and hybridized to the Cytoscan ® HD Accel Gene Chip. Data were analyzed using Chromosome Analysis Suite. The analysis is based on the GRCh37/hg19 assembly. This test was developed and its performance characteristics determined by LabCorp. It has not been cleared or approved by the Food and Drug administration.
Positive evaluation criteria include:
* DNA copy number loss of >1 Mb or gain >2 Mb outside known clinically significant regions with at least one OMIM gene.
* DNA copy gain/loss within or including a known clinically significant gene of 25 kb or greater.
* 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.
* 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 one region that is >8 Mb but below reporting criteria for UPD will be reported as increased risk of recessive disorder.
* 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.
* Complete moles are detected by the presence of whole genome allele homozygosity (~50% hmz in rare dispermy moles).
* Triploid DNA normalizes to 2 copies in array analysis, but is detectable in this allele specific SNP microarray by the characteristic 2:1 allele ratios and pattern generated within each autosome.

SNP chromosomal microarray cannot detect:
* Truly balanced chromosome alterations
* Sequence variants
* Small insertions and deletions (indels)
* Changes in regions not represented by probes on the array
* Tetraploidy
* Low level mosaicism
* Whole chromosome uniparental heterodisomy without parental specimens
* Imbalances in the mitochondrial genome

Single gene partial or intragenic copy number variants (CNVs) detected by an independent technology such as next generation sequencing (NGS) may not be detectable by microarray. The ability to detect the CNV is dependent on size and probe coverage. The threshold for mosaicism is variable, depending on the size of the segment and array quality. Empiric studies have detected mosaicism for trisomy of a whole autosome below 10.0%. CNVs that are known to be common in the population may not be reported.

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Gloria Haskell

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