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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LCLS Specimen Number:	**106-225-9012-0**	Account Number:	90001555
Patient Name:	**REPORT, SAMPLE**	Ordering Physician:	
Date of Birth:	11/15/1987	Specimen Type:	**AMNIOTIC FLUID**
Gender:	F	Client Reference:	
Patient ID:		Date Collected:	04/15/2024
Lab Number:	YU24-40006 G	Date Received:	04/15/2024
Indications:	Increased nuchal translucency; Hypothorax	Date Reported:	**05/15/2024**
Test:	**Prenatal Chromosome Microarray**		
Genotyping Targets:	2772571	Array Type:	SNP

MICROARRAY RESULT: NORMAL DOSAGE; WHOLE CHROMOSOME 14 HOMOZYGOSITY

INTERPRETATION: FEMALE WITH UNIPARENTAL ISODISOMY 14

arr(14)x2 hmz

The whole genome SNP microarray (Reveal) analysis was normal female in respect to the copy number criteria indicated below.

There was, however, complete allele homozygosity observed in the chromosome 14 analysis which is diagnostic of uniparental isodisomy. There are confirmed imprinted genes on chromosome 14 which, in cases of UPD 14, result in either maternal or paternal UPD-associated syndromes.

Maternal UPD14 is characterized by intrauterine growth problems, developmental delay, short stature, small hands and feet, scoliosis, hypotonia, obesity, distinctive facial appearance, and precocious puberty (see references).

Paternal UPD14 is characterized by polyhydramnios, characteristic facial anomalies, severe neurologic involvement, growth problems, and skeletal anomalies (including small thorax, abnormal ribs, and short limbs; see references).

There are additional risks for recessive allele pairing in the homologues and possible residual effects of an early developmental presence of aneusomy for chromosome 14 during the usual rescue etiology to UPD. There was no evidence of low-level dosage mosaicism for chromosome 14 in this analysis.

No other DNA copy number changes or copy neutral ROH were detected within the present reporting criteria. **Genetic counseling is recommended.**

To determine parent of origin, UPD testing is available. Charges will apply and follow-up parental bloods (lavender top tube) should be submitted if that testing is desired. Please reference the proband name, date of birth, and specimen number when submitting

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parental samples. Billing policy details are available for view on www.labcorp.com.

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

References:

Ogata T, Kagami M. Molecular mechanisms leading to the phenotypic development in paternal and maternal uniparental disomy for chromosome 14. Clin Pediatr Endocrinol. 2008;17(4):103-11. PMID: 24790371.

Murphy SK, et al. Epigenetic detection of human chromosome 14 uniparental disomy. Hum Mutat. 2003 Jul;22(1):92-7. PMID: 12815599.

Kamnasaran D, Cox DW. Current status of human chromosome 14. J Med Genet. 2002 Feb;39(2):81-90. PMID: 11836355.

Methodology:

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.
* 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 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

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* 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.

Gloria Haskell

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