Report date: -
Case file ID: -
Patient: -

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Formerly Gene Security Network

Microarray Chromosome Analysis with Parental Support ™

Patient: - Date of birth: -	Attending physician: -	Clinic: - - - - -	Samples collected: - Samples received: - Sample type: Products of Conception (POC)

Sample	Results/Details	Parental Origin of Abnormality
301089.2-2-P **POC6615-DNA**	**RESULT: Normal Female** **MICROARRAY RESULT: arr(1-22, X)x2** **Clinical Interpretation:** Normal female result. Maternal cell contamination (MCC) has been ruled out.	-

If you have questions or wish to discuss the results, please contact us at 650-249-9092 and ask for the genetic counselor on call.

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REPORT TERMINOLOGY DEFINITIONS

UPD

Uniparental Disomy (UPD) is defined as having two copies of a given chromosome from one parent and none from the other. This testing can detect UPD due to heterodisomy (two homologous or 'unmatched' chromosomes from one parent). UPD of a single chromosome due to isodisomy (two identical or 'matched' chromosomes from one parent) will be detected but reported as Monosomy since it is indistinguishable from Monosomy with this testing technology. UPD of every chromosome due to isodisomy will be reported as full UPD. UPD detection is based on a statistical model validated on single cells from a UPD cell line.

Gains/Losses

Gains and losses occur when a segment of a chromosome is missing (loss) or repeated (gain). Losses are indicative of a deletion and gains are indicative of a duplication. Gains and losses can occur as isolated de novo events or through inheritance of unbalanced rearrangements. Gains (duplications) and losses (deletions) diagnosed in a fetus or livebirth are generally associated with an abnormal phenotype. The criteria for a reportable gain and/or loss include: (a) Gain or loss greater than 5 MB in size, (b) One of the known deletion/duplication syndromes listed†, (c) Terminal gain and/or loss detectable by this testing technology, (d) All gains and losses < 5Mb in size and > 1 MB in size are reviewed and will be reported based on clinical relevance. The cytogenetic band breakpoints reported are based on microarray probe location using SNP Human Genome Build hg18 and are the minimally deleted and/or duplicated regions.

Limitations

There remain certain chromosome abnormalities that this analysis will not detect. These include:

- Balanced chromosome rearrangements (balanced translocations or inversions).
- Some gains or losses of chromosome material less than 5 MB in size not including those deletion/duplication syndromes listed below.†
- Specific genes and conditions caused by single gene mutations.
- Copy number variants (CNVs).
- Tetraploidy (four copies of the complete set of chromosomes) if maternal and paternal chromosome contributions are equal.
- Low levels of chromosome mosaicism.
- Full trisomy cannot be distinguished from trisomy due to a Robertsonian translocation (involves chromosomes 13, 14, 15, 21, or 22) or isochromosome. Depending on patient history, parental chromosomes may be considered to rule out a Robertsonian translocation or isochromosome in one of the parents.

Reporting of certain chromosome abnormalities will be as follows:

- UPD due to isodisomy will be reported as monosomy (as described above).
- Two copies of the Y chromosome will always be reported as a single Y.
- In most cases, mosaicism will be reported as a mixture of disomic, monosomic and trisomic cell lines.

† Table of Chromosome Microdeletions and Microduplications routinely reported

Chromosome Number	Deletion or Duplication	Syndrome Name	Chromosome Region
1	Deletion	1p36 deletion	1p36
1	Deletion	1q21.1 deletion	1q21.1
2	Deletion	2q37 deletion	2q37
3	Deletion	3q29 deletion	3q29
4	Deletion	Wolf-Hirschhorn syndrome	4p16.3
5	Deletion	Cri du Chat syndrome	5p15.2
7	Deletion	Williams syndrome	7q11.23
8	Deletion	Langer Giedon syndrome	8q23.2-8q24.1
9	Deletion	9q34 deletion	9q34
10	Deletion	DiGeorge syndrome - region 2	10p13-p14
11	Deletion	WAGR syndrome	11p13-14
11	Deletion	Jacobsen syndrome	11q24.1
15	Deletion	Prader-Willi/Angelman syndrome	15q11-q13
17	Deletion	Smith-Magenis syndrome	17p11.2
17	Deletion	Miller Dieker syndrome	17p13.3
22	Deletion	DiGeorge syndrome/VCFS	22q11.2
22	Deletion	Phelan-McDermid syndrome	22q13
22	Duplication	22q11.2 duplication	22q11.2

Non-paternity may produce inconclusive test results and may be reported out as no results. Natera has a policy of non-disclosure for non-paternity.

Natera's POC testing can also be performed on prior losses preserved in paraffin. If there is interest in testing a previous loss, please contact Natera for details.

These results should always be interpreted by a clinician in the context of clinical and familial data.

Approved by:

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Clinical Geneticist

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