Case #: 012345 **Cell Line:** Sample Name

Results: The results listed using the International System for Human Cytogenomic Nomenclature (ISCN). Any calls from the call table that are above the aberration size criteria will be listed.

Microarray Analysis Report: 012345

Date Reported: Tuesday, August 1, 2023
Cell Line: Sample Name
Submitted Passage #: None Given
Date of Sample: 7/13/2023
Specimen: Human T Cells

Aberration Criteria: Gain or Loss: >400kb LOH: >5Mb

Indicates a mosaic call

Cell Line Sex: Male
Reason for Testing: QC testing
Barcode: 012345678900
Position: R06C01
Investigator: WiCell Stem Cell Bank, WiCell

Call Table: All calls found during analysis are listed in this table.

Microarray Results: arr[GRCh37] 14q11.2(22510249_22964246)x0,14q11.2q32.33(23029263_106864851)x2 hmz[0.15]

hmz: denotes an AOH in the results

Shows the number of copies observed

Call Table:

Chromosome Region (GRCh37/hg19)	Cytoband	Event (% mosaic)	Estimated Copy Number	Length (Base Pairs)	Gene Count
chr3:165,264,514-165,296,562	3q26.1	CN Loss	1	32,049	0
chr7:38,293,984-38,368,834	7p14.1	Homozygous Copy Loss	0	74,851	1
chr7:38,370,141-38,393,479	7p14.1	CN Loss	1	23,339	1
chr7:142,327,013-142,475,348	7q34	CN Loss	1	148,336	1
chr13:57,866,157-57,889,148	13q21.1	CN Loss	1	22,992	0
chr14:21,357,953-21,416,425	14q11.2	CN Loss	1	58,473	2
chr14:22,449,161-22,509,182	14q11.2	CN Loss	1	60,022	0
chr14:22,510,249-22,964,246	**14q11.2**	**Homozygous Copy Loss**	**0**	**453,998**	**1**
chr14:22,964,937-23,000,062	14q11.2	CN Loss	1	35,126	0
chr14:23,029,263-106,864,851	**14q11.2 - q32.33**	**AOH (15%)**	**2**	**83,835,589**	**849**

Bolded calls are above the aberration size criteria and are reportable

Interpretation:

- *There was 1 reportable copy number changes as well as 1 reportable regions of LOH identified.*
- *A 0.454Mb loss on chromosome 14 was observed. The copy number variant encompasses a homozygous deletion of this region.*
- *A 83.8Mb region of LOH on chromosome 14 was observed. The ratio shift of this copy number change indicates possible low-level mosaicism of approximately 15%.*
- *The UCSC Genome Browser (https://genome.ucsc.edu/) and DECIPHER (https://decipher.sanger.ac.uk/search) ma information on regions of interest.*

Interpretation: A more in depth explanation of the results and what the calls might mean.

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Case #: 012345 ***Cell Line: Sample Name***

Results Completed by: TECHNOLOGIST NAME

Reviewed and Interpreted by: DIRECTOR NAME

Specifications:

· Platform: Illumina Global Diversity Array with Cytogenetics-8 (GDACyto)

· Marker coverage: 1,825,277 spanning whole human genome

· Analysis software: NxClinical (Via) 6.1 Software

· Array design, genomic position, genes and chromosome banding are based on genome build GRCh37/hg19.

· Aberrant copy number genomic regions are identified by log R ratio (LRR) and B allele frequency (BAF). LRR is the log ratio of observed probe intensity to expected intensity, deviations from zero are evidence for copy number change. BAF is the proportion of hybridized sample that carries the B allele: 0.0, 0.5, and 1.0 are expected for each locus in a normal sample. Deviations from this expectation are indicative of aberrant copy number.

· Quality assurance monitors: 1) Call Rate; 2) Confidence Threshold; 3) LogRDev; 4) Illumina sample dependent/independent QC measures

· Reportable copy number changes are gains or losses greater than 400kb. Reportable regions of LOH are greater than 5Mb. See Interpretation for copy number changes and regions of LOH that meet these criteria. See Call Table for all copy number changes identified by the analysis software. If mosaicism is detected, the approximate percentage of mosaicism is listed in the ‘Variant Type (% mosaic)’ column.

· The assay is currently validated for the detection of copy number losses greater than 20-kilobases (kb) in size and copy number gains 50-kb in size (smaller changes may be detected depending on gene content and probe number but will not be included in the Call Table). From internal validation studies, abnormalities present in a mosaic state are reliably detected if the mosaicism level (percentage of abnormal cells) is 20% or higher.

· Sample intensities were compared to standard cluster file intensities comprised of over 100 samples from Caucasian(CEU), Asian(CHB+JPT), and Yoruban(YRI) HapMap populations.

Limitations: This assay will detect aneuploidy, deletions, and duplications of represented loci, and regions of loss/absence of heterozygosity (LOH), but will not detect balanced alterations (reciprocal translocations, Robertsonian translocations, inversions, and insertions), or point mutations. Based on the results of internal validation studies, abnormalities present in a mosaic state are reliably detected if the mosaicism level (percentage of abnormal cells) is 20% or higher. The failure to detect an alteration at any locus does not exclude all anomalies at that locus. Significance of the number of probes used to detect an aberration has not been determined and confirmational testing may be informative. Actual chromosomal localization of copy number change is not determined by this assay. Other mapping procedures are required for determining chromosomal localization.

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Date:_____ Sent By:_____ Sent To:_____ QC Review By:_____

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