

Case #: 012345

**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.

Cell Line: Sample Name



# Microarray Analysis Report: 012345

**Date Reported:** Tuesday, August 1, 2023

**Cell Line Sex:** Male

**Cell Line:** Sample Name

**Reason for Testing:** QC testing

**Submitted Passage #:** None Given

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

**Barcode:** 012345678900

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

**Date of Sample:** 7/13/2023

**Position:** R06C01

**Specimen:** Human T Cells

**Investigator:** WiCell Stem Cell Bank, WiCell

**Microarray Results:** arr(X,Y)x1,(1-22)x2

Shows the number of copies observed

**Call Table:**

Chromosome Region (GRCh37/hg19)	Cytoband	Event (% mosaic)	Estimated Copy Number	Length (Base pairs)	Gene Count
chr7:38,311,831-38,338,251	7p14.1	Homozygous Copy Loss	0	26,421	1
chr7:142,023,396-142,046,091	7q34	CN Loss	1	22,696	0
chr12:7,991,000-8,113,376	12p13.31	CN Gain	3	122,377	2
chr14:22,790,594-22,961,867	14q11.2	Homozygous Copy Loss	0	171,274	1
chr14:106,029,590-106,066,706	14q32.33	CN Loss	1	37,117	0
chr14:106,067,118-106,127,700	14q32.33	Homozygous Copy Loss	0	60,583	2
chr14:106,133,067-106,194,562	14q32.33	CN Loss	1	61,496	1
chr17:44,165,803-44,399,988	17q21.31	CN Gain	3	234,186	4
chr19:6,890,202-7,105,830	19p13.3p13.2	CN Gain	3	215,629	9

### Interpretation:

- There were 0 reportable copy number changes as well as 0 reportable regions of LOH identified.
- The UCSC Genome Browser (<https://genome.ucsc.edu/>) and DECIPHER (<https://decipher.sanger.ac.uk/search>) may provide additional information on regions of interest.

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

**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

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· 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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