The Jackson Laboratory for Genomic Medicine
10 Discovery Drive
Farmington, CT 06032

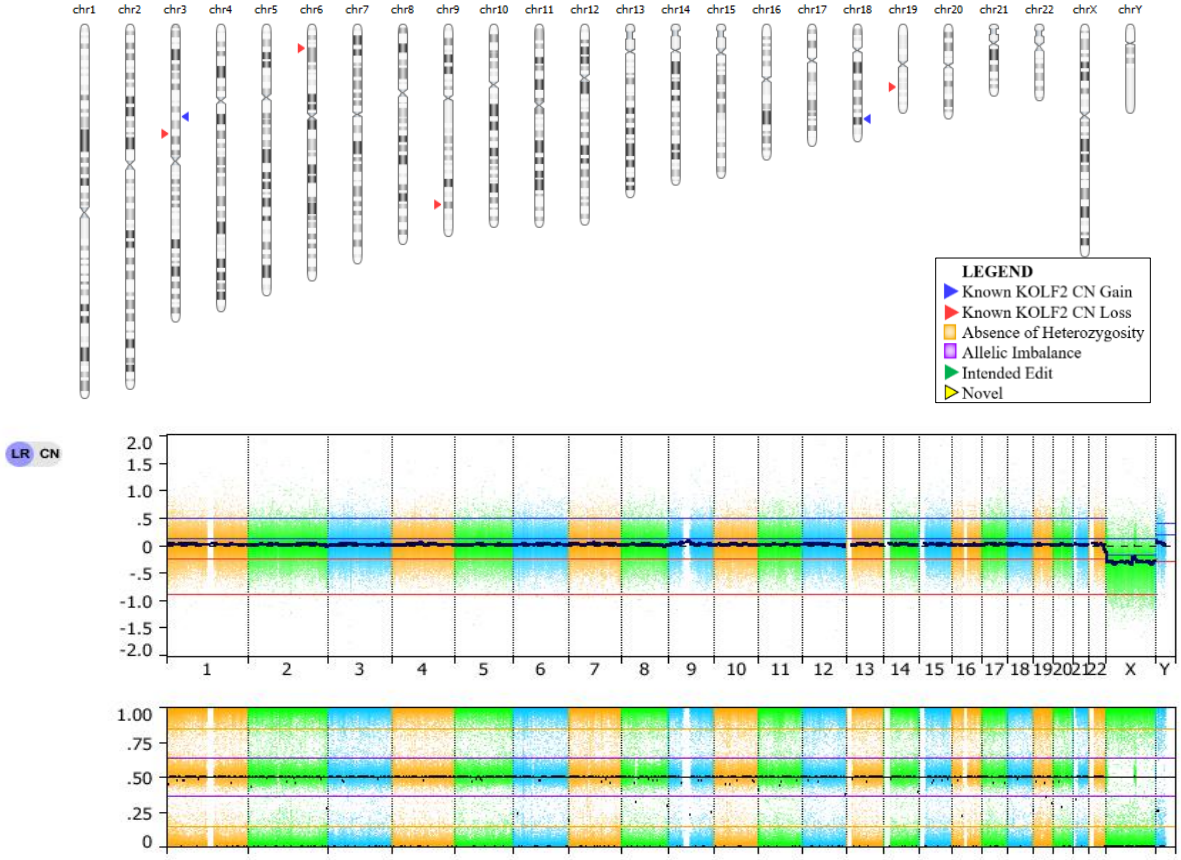
Sample ID: 11_KIF5A_C03_KOLF2.1J **Sample Barcode**: 208138750029 R02C01 020824

Cell Line: KOLF2.1J, Male

Array Result: PASS

Target Edit: KIF5A N256S **at hg19:** chr12:57,962,798

Whole Genome View



Novel Sample-Specific Regions: None detected

Known KOLF2-C1 CNVs

Below are additional CNVs that have been identified. These regions are known aberrations in the KOLF2-C1 cell line, which is the original iPSC line from which KOLF2.1J was generated. These CNVs are present in all genetically engineered lines derived from KOLF2-C1 and KOLF2.1J.

Event	Chr Region - PacBio (hg19) *	Size (bp)	Cytoband	Approx CN	Genes	Detected**
CN Gain[1]	chr3:61135038-61911274	776,236	3p14.2	3.0	FHIT, PTPRG	Automatic
CN Loss[1]	chr3:72289391-72432282	142,891	3p13	1.0	RYBP	Automatic
CN Loss[1]	chr6:15490359-15723361	233,002	6p22.3	1.0	JARID2, DTNBP1	Automatic
CN Loss[1]	chr9:119241091-119372704	131,613	9q33.1	1.0		Automatic
CN Gain[1]	chr18:62113242-62270679	157,437	18q22.1	3.0		Automatic
CN Loss[2]	chr19:41355704-41387475	31,771	19q13.2	1.0	CYP2A7	Automatic

*CNVs were confirmed with PacBio Long Read sequencing data. CN Losses were identified with **pav** (doi:10.1126/science.abf7117, PMID: 33632895). CN Gains were identified with **pbsv** (https://github.com/PacificBiosciences/pbsv).

**Known KOLF2-C1 CN events are automatically detected by VIA™ in >90% of samples *(Automatic)*. When a CN event is not automatically detected, the region is manually reviewed *(By Review)* to confirm presence of the known CN event.

1: CN events originally identified by Aziku et al. doi.org/10.1101/2023.06.26.546614 (bioRxiv preprint)

2: Unpublished CN event identified by C. Beck.

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METHODS:

The Illumina Infinium Global Diversity Array with Cytogenetics-8 (GDA Cyto) contains ~1.8 million probes throughout the human genome. It provides extensive targeted coverage of more than 4800 key genes across the genome and high exonic coverage in regions of disease relevance, providing highly accurate copy number variation calls and an average resolution of 1.5 Mb. Supplemental probes were selected based on the input from a cytogenetics research consortium and strategically localized near exons, in spatial consideration of existing probe placement, to deliver optimal performance in copy number variation analysis. The current build used is the human genome reference build hg19. Analysis was performed using VIA software v7.0 (Bionano Genomics).

The current microarray platform detects chromosomal imbalances (i.e., gains and losses), but is not designed to specifically detect balanced chromosomal rearrangements, inversions, methylation abnormalities, or aberrations involving genomic regions not targeted by this platform.

Processing Settings	
Data Type:	GxA-Cyto GTC
Processing Type:	GDA-Cyto_v8
Systematic Correction Type:	Quadratic Correction
Systematic Correction File:	Catlg_ILM_GDACyto_20047166_A1_20210504.txt
Allow manual centering:	TRUE
Recenter Probes Type:	Median
Auto Gender Detection Enable:	TRUE
Type:	SNP FASST2 Segmentation
Max Contiguous Probe Spacing (Kbp):	1000
Min number of probes per segment:	50
Significance Threshold:	1.00E-13
Segment Boundaries:	Inner probes
Amplification (4+:2):	0.5
Gain (3:2):	0.13
Loss (1:2):	-0.24
Homozygous Loss (0:2):	-0.9
M vs M, X/Y Loss (0:1):	-0.3
M vs F, X Loss (0:2):	-0.9
M vs M, X/Y Gain (2:1):	0.2
M vs F, X Gain (2:2):	-0.18
M vs M, X/Y Amplification, F vs M, X Gain (3:1):	0.41
F vs M, X Amplification (4+:1):	1.7
Homozygous Frequency Threshold:	0.96
Homozygous Value Threshold:	0.85
Heterozygous Imbalance Threshold:	0.36
Minimum LOH Length (KB):	3000
Minimum SNP Probe Density (Probes/MB):	500
Robust Variance Sample QC Calculation Percent outliers to remove:	0.5
Mosaic Labeling Type:	Aberrant Cell Fraction
Mosaic Labeling:	TRUE
Mosaic Labeling Label anything lower than (%):	65
Pre-Classification Decision Tree:	KOLF_v1
Segmental Duplications Track Version	20210331
VIA Report Version	KOLF_V1