

CUSTOM PANEL OVERVIEW	
Number of Targets:	1567 (1518 SNPs, 49 InDels)
Number of Probes:	2816
Probe Footprint (bps):	253,731 bps
Partial Targets <u>NOT</u> Covered:	3 InDels
Whole Targets <u>NOT</u> Covered:	184 (175 SNPs, 9 InDels)

PROJECT RATIONAL AND GOALS

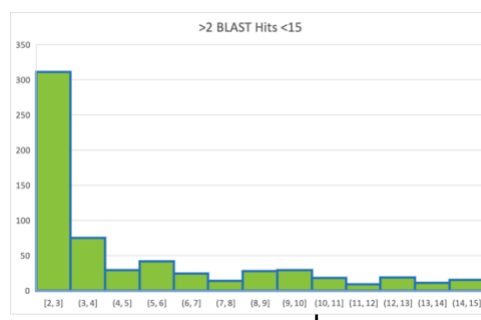
NIST seeks to validate putative low-frequency variants identified in NIST's GIAB HG002 reference material, and to further discern de novo mutations that occur overtime during cell line propagation. CHOP will sequence the capture libraries.

- 1) Design a hybrid capture panel comprised of SNPs and InDels targets ☒
- 2) Send Kit to NIST for DNA sample processing and subsequent sequencing at CHOP
- 3) Provide access to DNANexus to generate error-corrected BAM and VCF/MUT files to carry out basic mutagenesis analysis and assess CHIP clonal populations

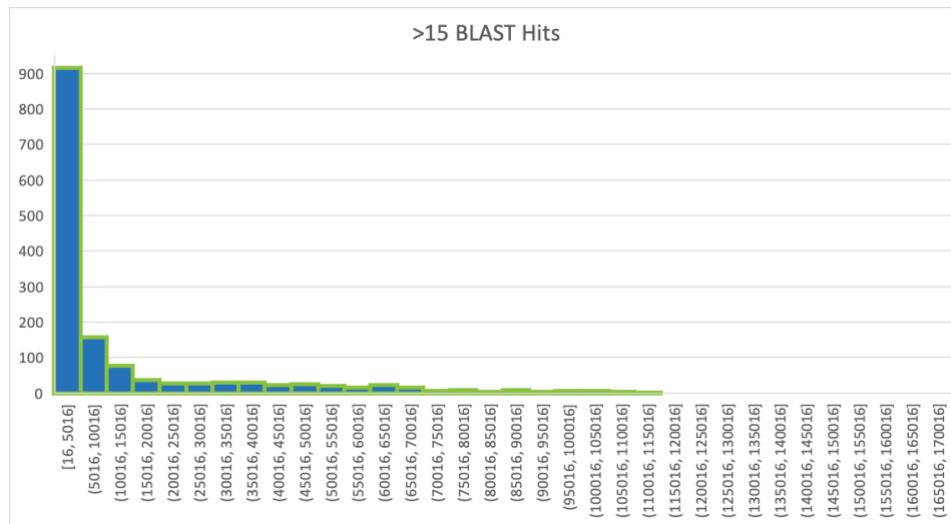
CUSTOM PANEL DESIGN DESCRIPTION

Custom Panel was designed for NIST/CHOP targeting 1,567 individual SNPs and InDels defined by NIST. Target files were provided by NIST in the human hg38 genome build which will facilitate downstream analysis using DNANexus. A total of 2,816 probes were designed to NIST's target regions after a rigorous filtration strategy. Targets were filtered as follows:

1. Probes were designed to the original 2,522 SNPs provided by NIST resulting in 4,828 probes. Probes were then BLAST-ed against the human genome (hg38).
 - a. 2,698 probes had a single BLAST hit
 - b. 624 had ≥ 2 BLAST hits ≤ 15



- c. 1,478 probes with >15 hits in the genome were filtered



- d. 1,518 SNPs remained for further analysis
e. Further filtration of SNPs and InDels for off-target effects and predicted secondary structured resulted in limited removal of a few targets, as noted in table and depicted below.

A total of 2816 probes will cover a footprint of ~254Kb of the human genome

Figure 1. No Coverage. SNP 126.

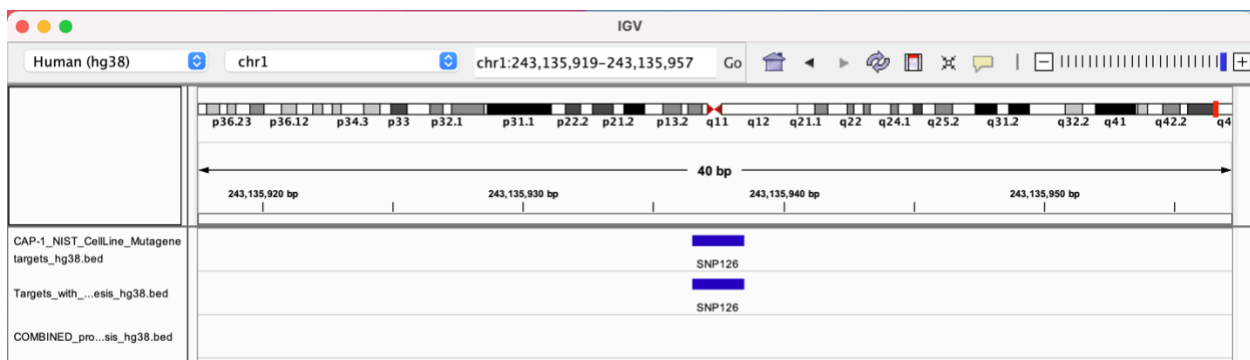


Figure 2A. Partial Coverage. InDel 9.

