CNV+ Breast Cancer NGS Panel Sample Results

NuProbe's Quantitative Amplicon Sequencing (QASeq) technology allows rapid (4-8 week) development of custom NGS panels to detect mutations at 0.2% Variant Allele Frequency (VAF) and deletions at 1.97 ploidy or amplifications at 2.04 ploidy from FFPE tissue, fresh/frozen tissue or blood cfDNA.

Here, we show sample results of applying the CNV+ Breast Cancer NGS Panel using QASeq technology to analyzing an individual cfDNA sample of a breast cancer patient purchased from Discovery Life Science. The results include copy number variations (CNVs), pathogenic mutations and figures demonstrating the detailed data.

CNV Status

Deletions and amplifications of genes, also known as copy number variations (CNVs), are present in a significant percentage of tumors, between 3% and 98% depending on the cancer type, and are clinically relevant as prognostic markers and as therapeutic targets. The table on the right reports the CNV status of this sample.

Gene or Chromosome	Ploidy	Notes
ERBB2	1.69	Non-focal
Chr17p	1.76	
Chr17	1.77	

Pathogenic Mutations

Mutation in genome (GRCh38.p12)	COSMIC ID	Reference SNP (rs) number	AA and CDS change	VAF%
Chr10:87894045G>A	COSV64307061	-	PTEN_ENST00000371953.7(c.100G>A;p.A34T)	0.3
Chr17:7675088C>T	COSV52661038	rs28934578	TP53_ENST00000269305.8(c.524G>A;p.R175H)	0.3

Figures

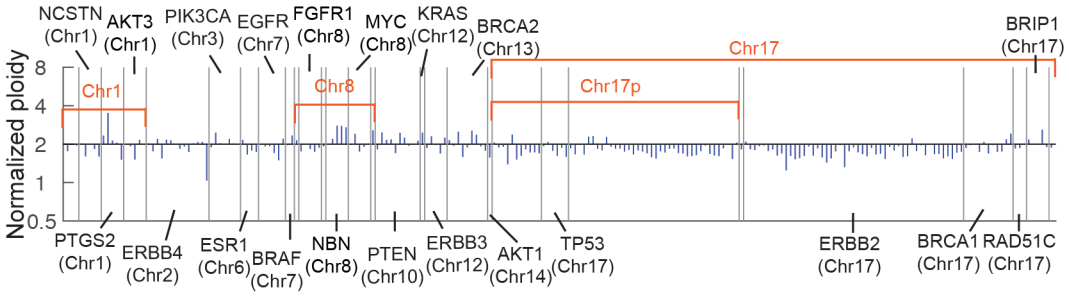

Figure 1. Ploidy summary for each amplicon. The sequencing data were processed with UMI family clustering, conversion yield normalization to obtain molecule count, and molecule count for each amplicon was then normalized with reference group to calculate the ploidy number. Reference is determined by sequential Mann-Whitney U tests on each gene of interest.

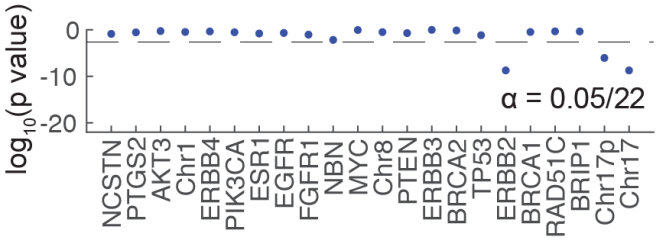

Figure 2. Calculated p-values of CNV for each gene, based on the Mann-Whitney U test for the results in Figure 1. The significance value α was set at α = 0.05/22, based on Bonferroni correction. CNVs were called for ERBB2 gene, Chr17p and Chr17 for this sample.

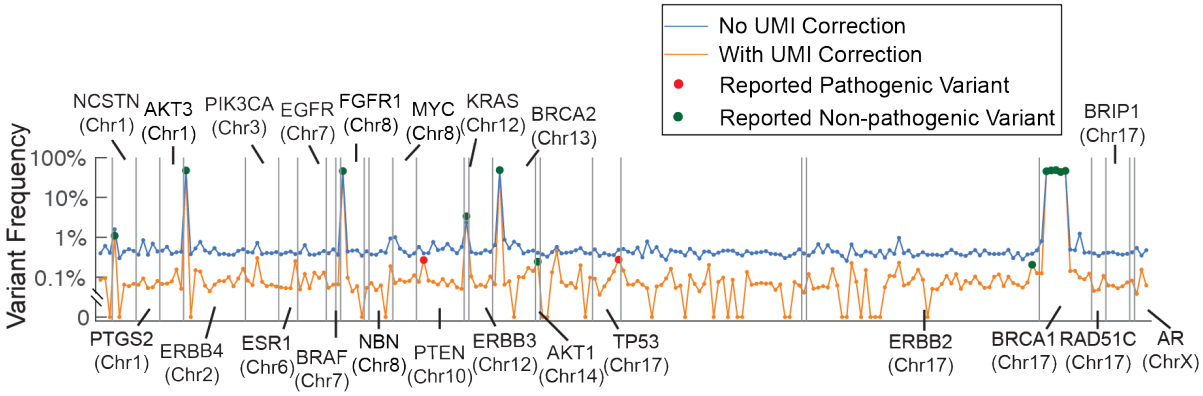

Figure 3. Variant frequency summary for each amplicon with and without UMI. Variant calls above calling threshold were highlighted in green for pathogenic variants and in red for non-pathogenic variants.