# Supplementary Material

Supplement to: Li Bao, Zhaoyang Qian, Maria Lyng et al. Coexisting genomic aberrations associated with lymph node metastasis in breast cancer.

**Content:**

cancer samples from the Chinese sample set.

Supplemental Figure 10. Comparison of mutation frequencies in ER+ primary breast cancers from patients (n=17; red bars) with and without (n=15; blue bars) lymph node metastasis from the Chinese sample set.

Supplemental Figure 11. Amplification of MYC (copy number ≥5) in 6 primary breast cancer samples from the Chinese sample set.

Supplemental Figure 12. Comparison of mutant frequency of frequently mutated genes between samples with different lymph node stages in three datasets (METABRIC, TCGA, Nik-Zainal et al.).

Supplemental Figure13. Comparison of mutation frequency for the most frequently mutated genes in the TCGA sample set between the lymph mode status N0 group and N2-3 group of breast cancer patients with ER+ (A) or ER- (B) primary tumors.

Supplemental Figure 14. Significance of correlations between CNV of genes and lymph node stages before and after normalization for age, tumor size, PR and *TP53* status.

Supplemental Figure 15. Significance of correlations between CNV and lymph node stages in selected genome regions.

Supplemental Figure 16. Significance of correlations between logR and lymph node stages across the whole genome in TCGA dataset by logistic regression analysis before and after normalization for other prognostic factors.

Supplemental Figure 17. GC normalization and profiles of copy number variations (CNVs).

Supplemental Table 1. Details of breast cancer single cells.

Supplemental Table 2. Mutations in single cells.

Supplemental Table 3. 78 SNVs specific to cancer cell-pool/single-cell.

Supplemental Table 4. 49 SNVs specific to normal mammary cell-pool/single-cell.

Supplemental Table 5. 22 SNVs presumably derived from hematopoietic cells.

Supplemental Table 6. CNV segmentation of all 7 cell pools.

Supplemental Table 7. Clinical information for the expanded sample set of 54 Chinese breast cancer patients.

Supplemental Table 8. Target genes for capture sequencing in the 54 Chinese sample set.

Supplemental Table 9. Somatic non-synonymous mutations detected in the sample set of 54 Chinese breast cancers.

Supplemental Table 10. CNV segmentation of all 54 Chinese breast cancers.

Supplemental Table 11. Genes with higher frequency of amplification in advanced lymph node stage samples among all three sample sets.

Supplemental Table 12. Genes with higher frequency of deletion in advanced lymph node stage samples among all three sample sets.

Supplemental Table 13. Correlation of other prognostic factors with lymph node metastasis in three datasets.

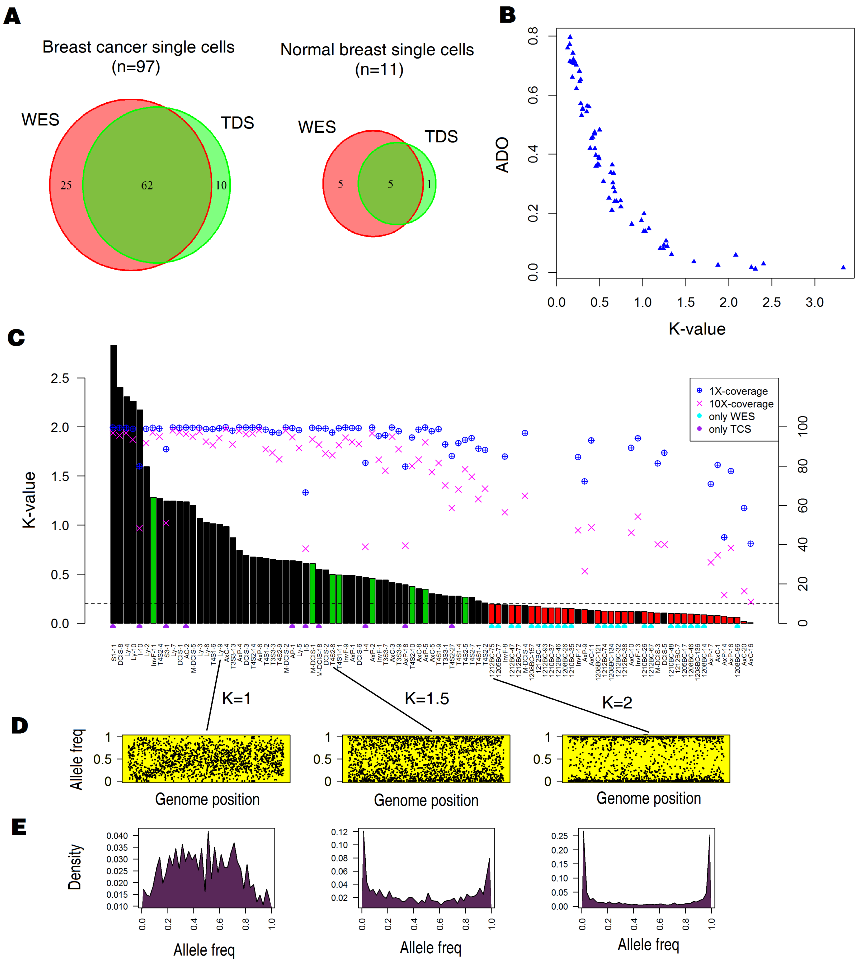
Supplemental Table 14. Genomic areas significantly associated with breast cancer lymph node metastasis identified by TCGA samples.

Supplemental Table 15. qPCR results of 170 ER+ Danish breast cancers using

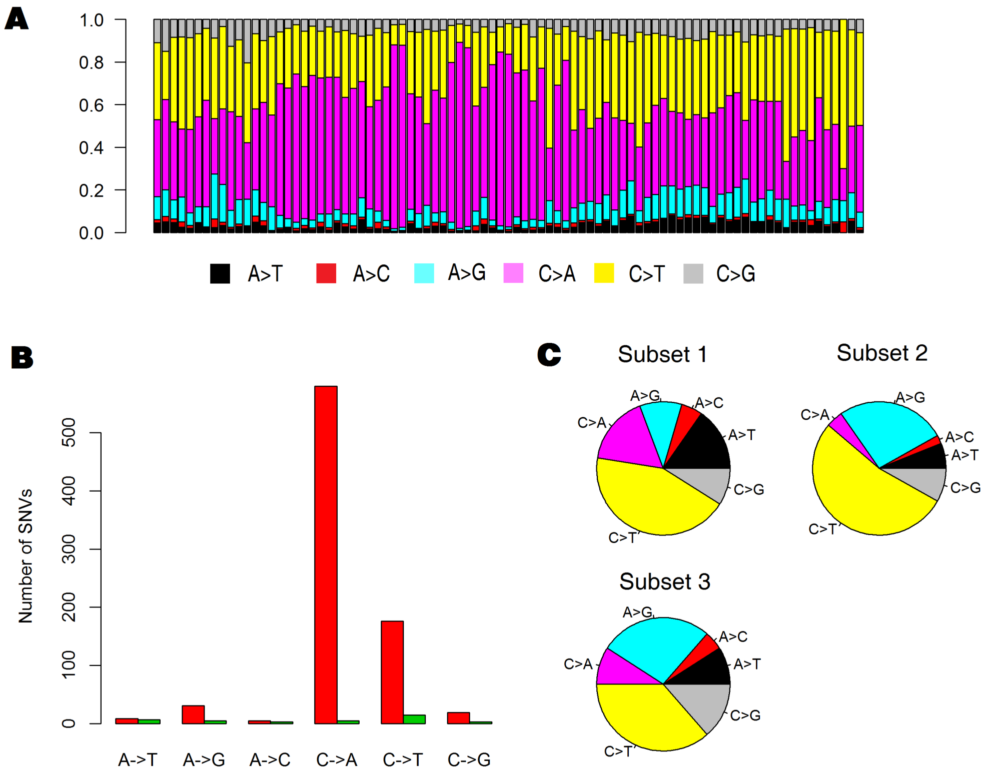
RNaseP as reference gene.

Supplemental Table 16. The qPCR data for the CNV analysis of the Danish cohort.

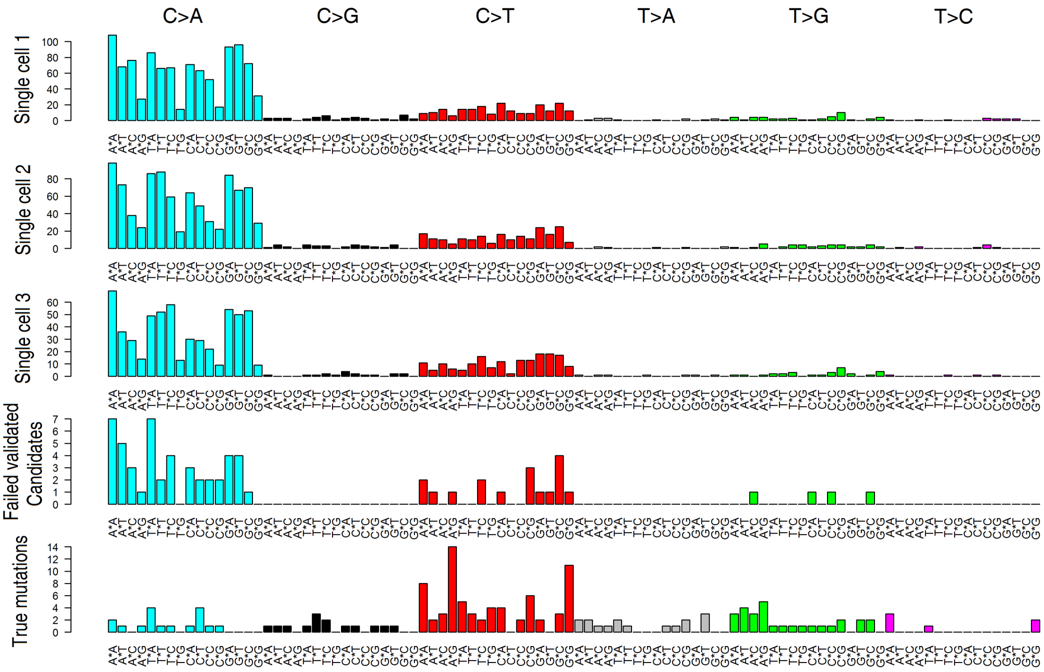
**Supplemental Figure 1. Workflow of sequencing and data analysis.**



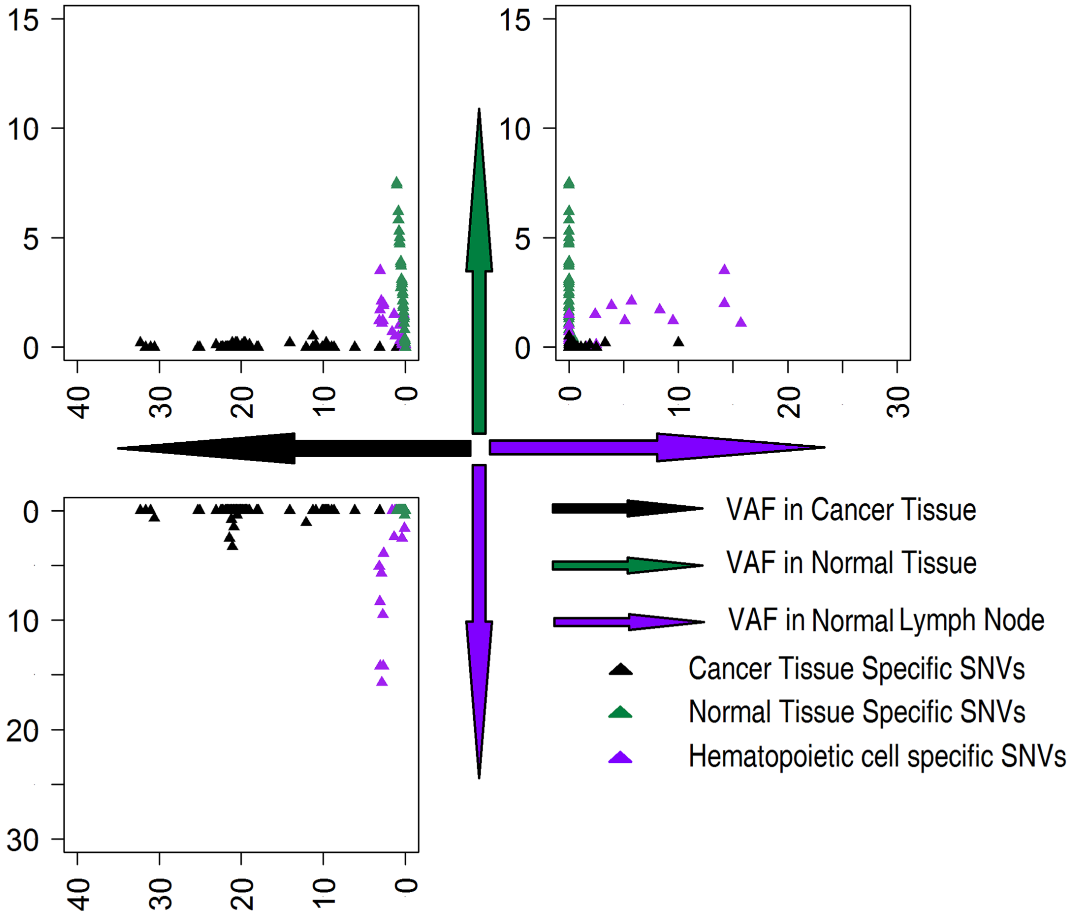
**Supplemental Figure 2**. **Single-cell sequencing profiles and quality control of single cells**. (**A**) Overview of sequencing method (WES: whole exome sequencing; TDS: target deep sequencing) used in single cells. (**B**) Correlation between K-value and ADO. (**C**, **D**, **E**) Quality of single-cell sequencing: (**C**) Barplot of K-value of 97 single cancer cells. Black bars indicate K-value of samples selected for evolutionary analysis, green bars indicate K-value of samples presumably contaminated by nuclei of normal cells, and red bars indicate low quality samples excluded from further analysis. 1X and 10X coverage rates for each cell subjected to target capture sequencing (TCS) are also shown. (**D**) Allele frequency of each SNP with a cover depth >20 is shown for 3 samples (Ly-9: K=1, T4S2-8: K=0.5 and 1212BC-75: K=0.2). The horizontal axis indicates the coordinates of each SNP in the human genome, and the vertical axis indicates the allele frequency of each SNP. (**E**) Allele frequency distribution of SNPs with a cover depth >20 is shown for the same 3 samples in **D**. The horizontal axis indicates the allele frequency of SNPs, and the vertical axis the proportions of SNPs in each allele frequency interval (allele frequency of 0-1 was separated by a total of 40 equal intervals).



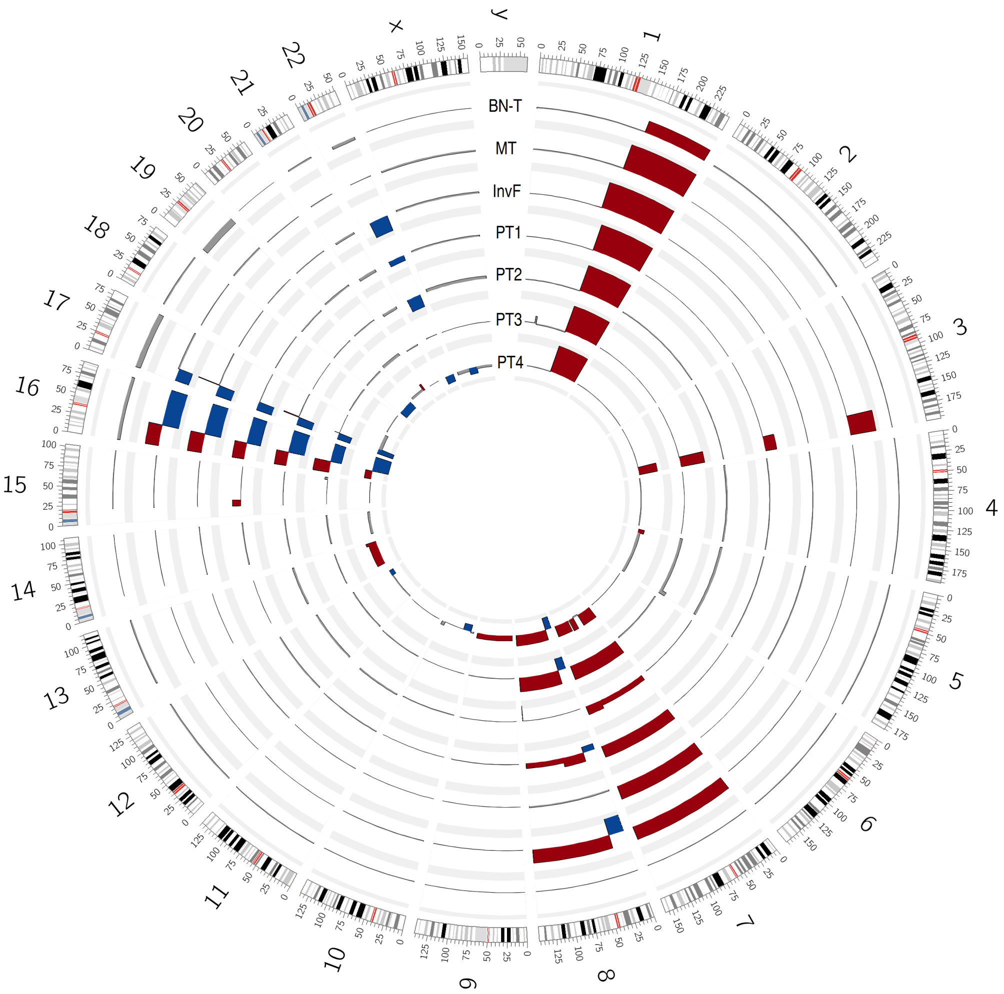
**Supplemental Figure 3. Duplex sequencing reduced false-positive variant calling.** (**A**) The preliminary detected substitutions in WES of single cells, primarily C>A and C>T substitutions. (**B**) The number of preliminary detected substitutions (red bars) and duplex verified substitutions (green bars) in a single cancer cell (InvF-9). (**C**) Distribution of substitution types for 3 subsets of verified SNVs dominated by C>T and A>G substitutions.



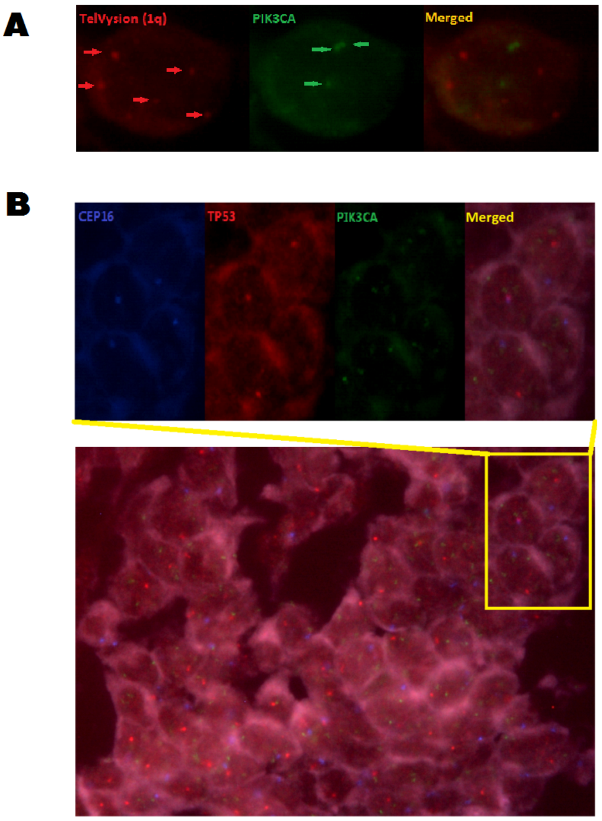
**Supplemental Figure 4. The trinucleotide mutation signature of putative amplified errors of candidate SNVs.** The trinucleotide context of putative amplified errors in 3 single cells (SC1, SC2 and SC3; total 3,341 in three cells), candidate mutations that failed validation (Failed validated candidates; 69 SNVs) and true mutations (True positives). The vertical axis displays mutation numbers of each trinucleotide signature.



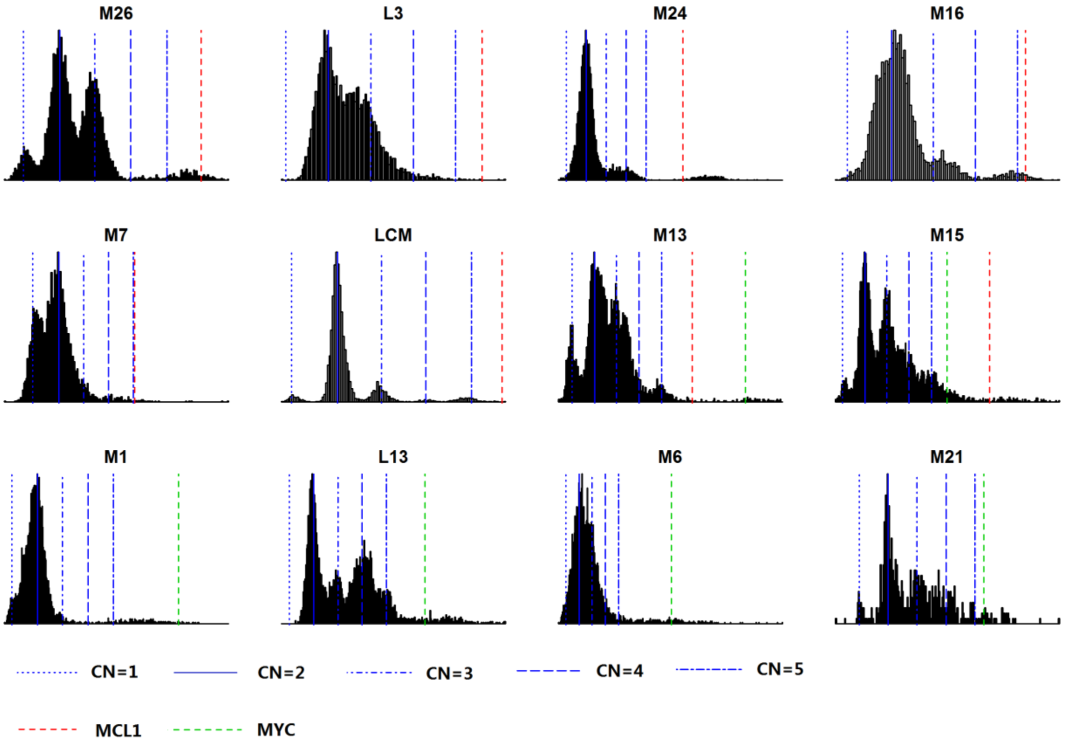
**Supplemental Figure 5. Allele frequencies of 3 SNV subsets in breast cancer cell pools, normal breast cell pools and normal lymph node tissue.** Each point represents one SNV, and black, green and purple points represent breast cancer-specific SNVs (subset 1), normal breast tissue-specific SNVs (subset 2) and SNVs likely belonging to hematopoietic cells (subset 3), respectively.



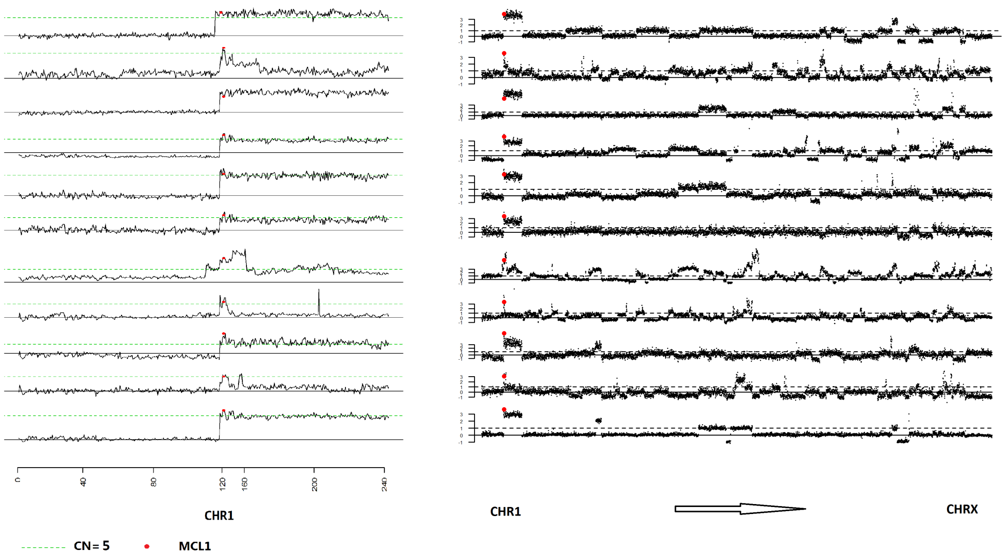
**Supplemental Figure 6. Circos map of CNVs of the 7 cell pools.** The outermost circle depicts the karyotype of the human reference genome (Hg19), while the inner 7 circles present the copy ratio of the 7 cell pools BN-T, MT, InvF, PT1, PT2, PT3 and PT4, respectively. Amplified and deleted regions are highlighted by red and blue, respectively.



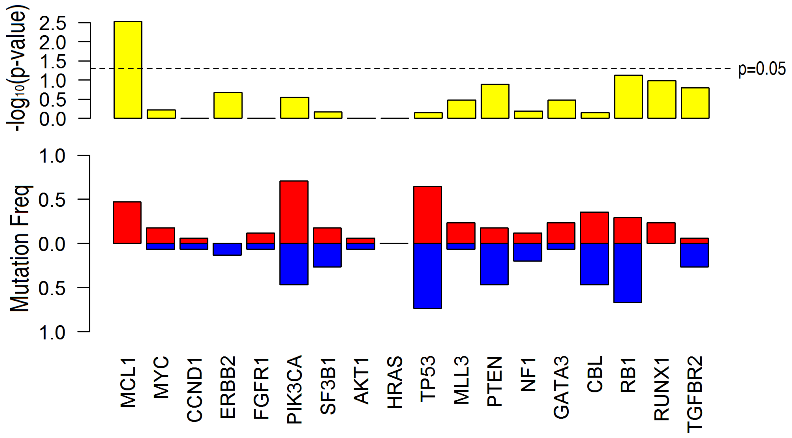
**Supplemental Figure 7. Fluorescence in situ hybridization (FISH) validation of cancer cell CNV alterations identified by sequencing.** (**A**) FISH images showed 5 copies of chr1q (red) and 3 copies of PIK3CA (green) in some cancer cells of the primary breast tumor. (**B**) FISH images showed only one copy of CEP16 (blue), one copy of TP53 (red), and 4 copies of PIK3CA (green) in some cancer cells of the lymph node metastasis.



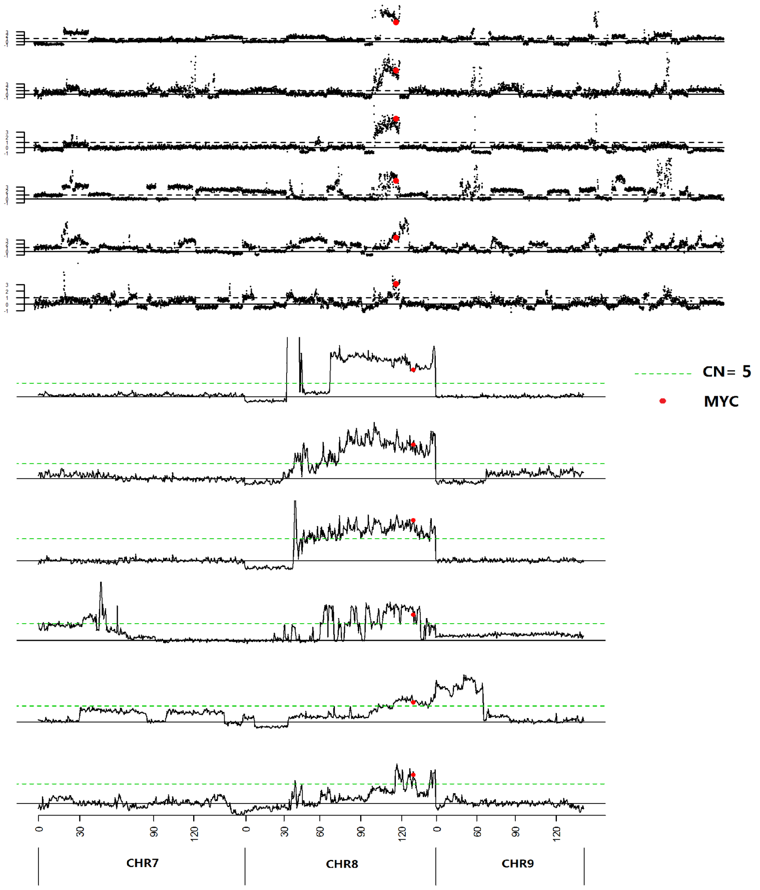
**Supplemental Figure 8. Depth distribution of 12 samples from the Chinese sample set exhibiting clear peaks and high-level (copy number ≥5) amplifications of MCL1 or MYC.** Horizontal axis: Copy ratio of bins. Vertical axis: Numbers of bins. Corresponding copy ratios of copy numbers (copy number =1-5) are shown by blue lines, while copy ratios of MCL1 and MYC are shown by red and green lines, respectively.



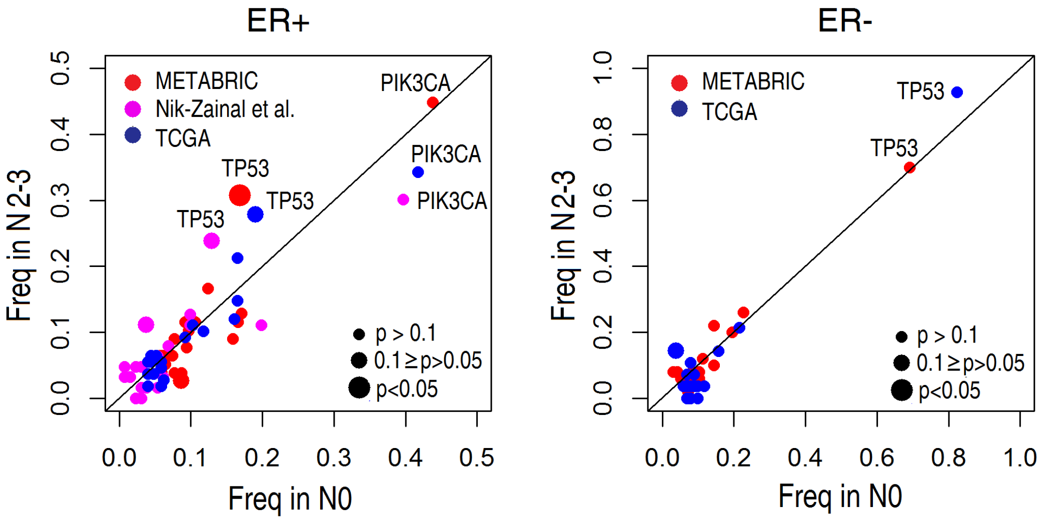
**Supplemental Figure 9**. **Amplification of MCL1 (copy number ≥5) in 11 primary breast cancer samples from the Chinese sample set.** The red points depict MCL1. The horizontal axis coordinate depicts chromosome positions of the human genome, while the vertical axis depicts copy number change.



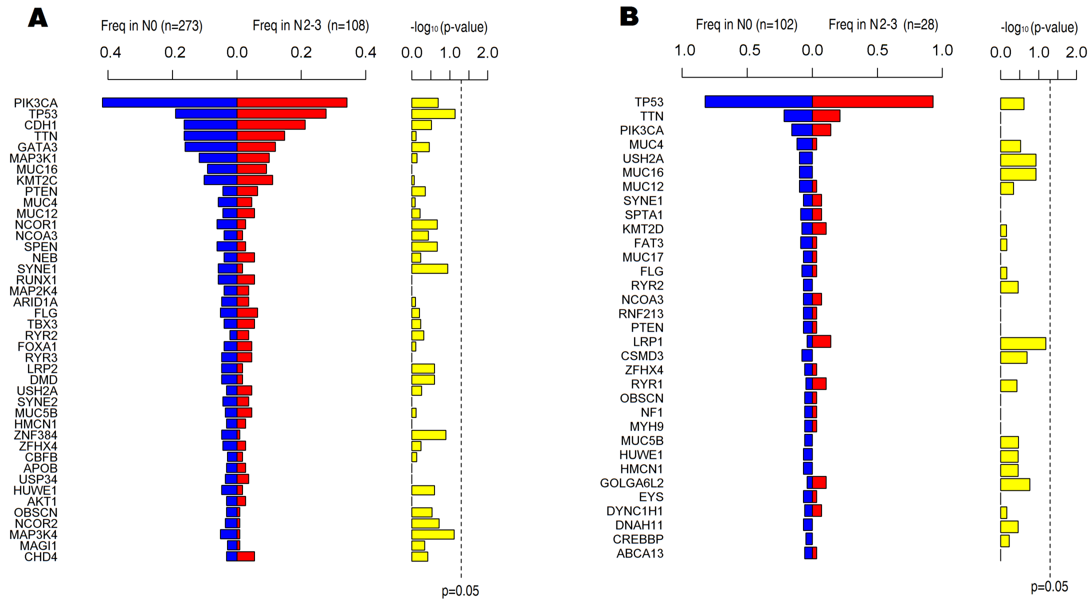
**Supplemental Figure 10. Comparison of mutation frequencies in ER+ primary breast cancers from patients with (*n* = 17; red bars) and without (*n* = 15; blue bars) lymph node metastasis from the Chinese sample set.** The yellow bar plot shows p-values (Fisher's exact test) for aberrant samples in the two subgroups for each gene. Only MCL1 was significant (*P* = 0.0029).



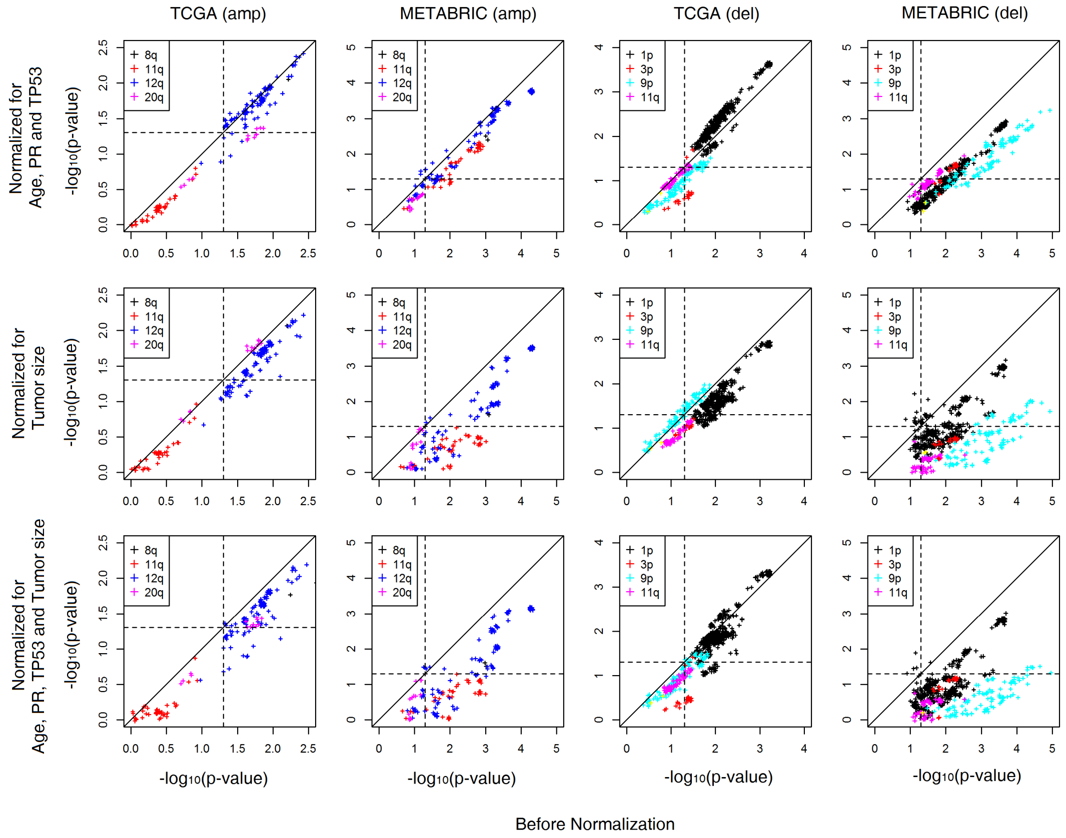
**Supplemental Figure 11. Amplification of MYC (copy number ≥5) in 6 primary breast cancer samples from the Chinese sample set.** The red points depict the MYC gene. The horizontal axis coordinate depicts chromosome positions in the human genome, while the vertical axis depicts copy number change.



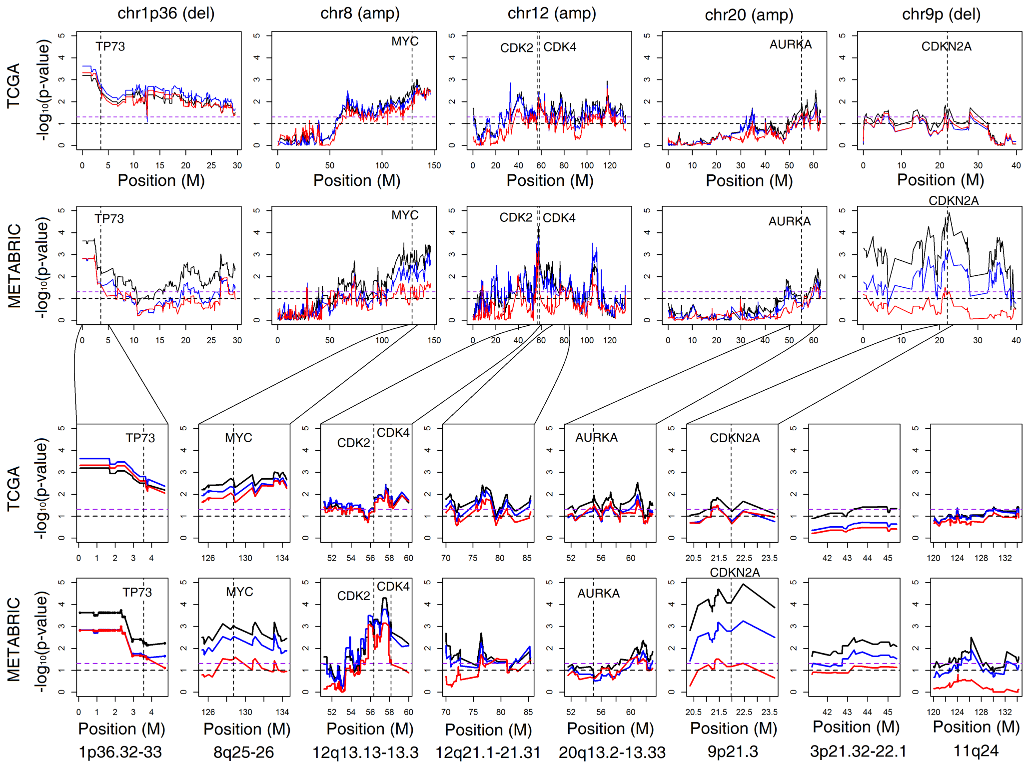
**Supplemental Figure 12. Comparison of mutant frequency of frequently mutated genes between samples with different lymph node stages in three datasets (METABRIC, TCGA, Nik-Zainal et al.).** Comparison of ER+ and ER- samples are shown in the left and right panel, respectively. For each dataset, the 20 most frequently mutated genes are shown by points, the size of which denotes different levels of *P* value (Fisher's exact test).



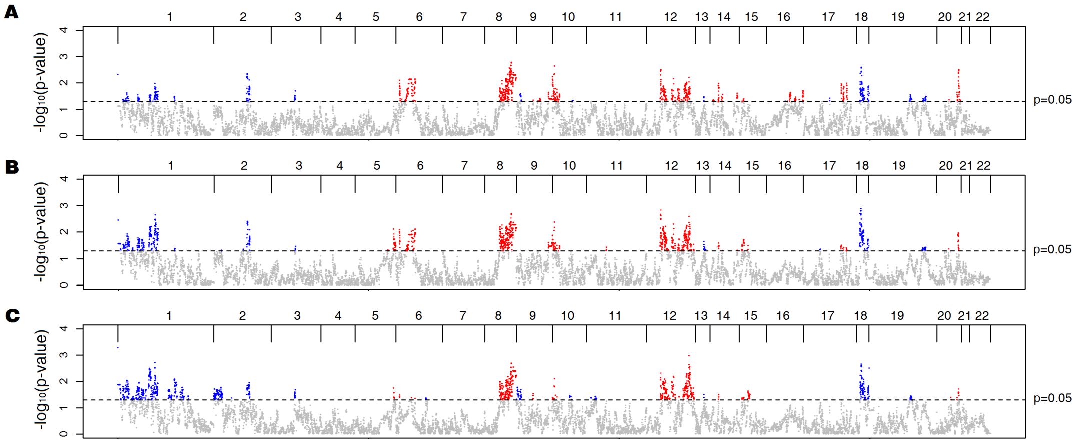
**Supplemental Figure 13. Comparison of mutation frequency for the most frequently mutated genes in the TCGA sample set between the lymph mode status N0 and N2-3 groups of breast cancer patients with ER+ (A) or ER- (B) primary tumors.** Blue bars denote mutated sample proportions in the N0 group, while red bars denote mutated sample proportions in the N2-3 group. Yellow bars denote *p*-values (Fisher's exact test) of the different proportions between the N0 and N2-3 groups.



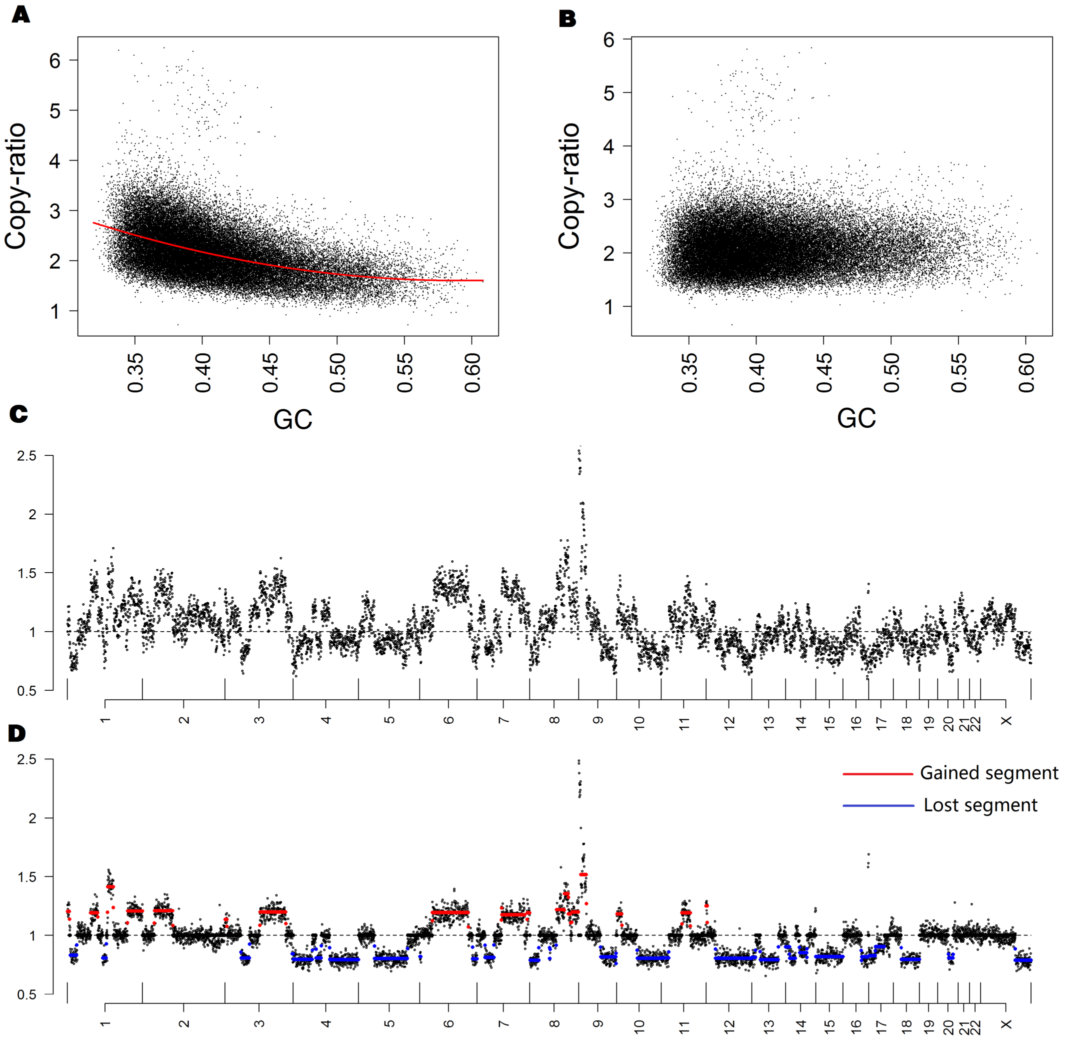
**Supplemental Figure 14. Significance of correlations between CNV of genes and lymph node stages before and after normalization for age, tumor size, PR and *TP53* status.** The significance of correlations between CNV of genes (reported in Supplemental table 11 and 12) and lymph node stages for the METABRIC and TCGA datasets were normalized for prognostic factors using a logistic regression model (see Methods for further details). The x axis displays *P* values (log mode) of genes before normalization, while the y axis displays *P* values after normalization for age, PR and TP53 status (upper panels), or tumor size (middle panels), or all four prognostic factors (lower panels). Each point represents a gene and genes in different genomic regions are highlighted by different colors. The dotted lines represent *P* value of 0.05, so genes in the upper right quartile are significant (p<0.05) both before and after normalization. After normalization for age, PR and TP53 status, the significance of genes did not appreciatively decrease, suggesting that the correlation between CNVs and lymph node metastasis is independent of these prognostic factors. After normalization for tumor size or for all four prognostic factors, an appreciatively decrease of significance was found in genes of 3p, 9p, 11q and 20q, particularly in the METABRIC dataset, while genes in 1p, 8q and 12q were less affected.



**Supplemental Figure 15. Significance of correlations between CNV and lymph node stages in selected genome regions.** Black, blue and red lines represent *P* value of genome regions before normalization, after normalization for age, PR and TP53 status, and after normalization for all four prognostic factors (two-tailed *t* test for coefficients in the logistic regression model). The dotted horizontal lines represent p-value of 0.1 (black) and 0.05 (purple). Genome regions of chr1p36.32-33 (the most significant region in chr1p), MYC at chr8q, 12q13.3 (including CDK2 and CDK4), parts of 12q21.1-21.31 and 20q13.3 remained significant ($P < 0.05$) in both the TCGA and METABRIC datasets. In contrast, chr3p21.32-22.1, 11q24 and CDKN2A at chr9p were no longer significant. The dotted vertical lines represent the position of the indicated gene(s) in the genomic region.



**Supplemental Figure 16. Significance of correlations between logR and lymph node stages across whole genome in TCGA dataset by logistic regression analysis before and after normalization for other prognostic factors.** (**A**), *P* value for genes before normalization; (**B**) *P* value for genes after normalization for age, PR and *TP53* status; (**C**) *P* value for genes after normalization for tumor size, age, PR and TP53 status. *P* value (log mode) of genes was presented by points and the dotted line denote p = 0.05. Genes with higher ($P < 0.05$) logR in N2-3 (n=301) than N0 (n=129) groups and genes with lower ($P < 0.05$) logR in N2-3 than N0 groups were highlighted by red and blue, respectively. Two-tailed *t* test of coefficients in the logistic regression model was used for testing the significance. Both before and after normalization for all four prognostic factors, genes located in genome regions of chr1p, 8q, 12q and 18q significantly correlating with lymph node stages ($P < 0.05$), indicating that these genome regions were mostly unaffected by the normalization.



**Supplemental Figure 17. GC normalization and profiles of copy number variations (CNVs).** (**A**) Correlation of copy ratio and GC content of bins before GC normalization. The red line indicates the generalized least-squares fit. (**B**) Correlation of copy ratio and GC of bins after GC normalization. (**C**) Copy ratio of bins before GC normalization. (**D**) Copy ratio of bins after GC normalization. Results of segmentation are depicted by red (gained segment) and blue lines (lost segment).

## Supplemental Table 1: Details of breast cancer single cells.

| Cell_id | Tumor | Cellular description | WES | Target_sequencing | Presumed_multi_nucle | K-value |
|---|---|---|---|---|---|---|
| DCIS-1 | PT3 | center | Y | Y | N | -0.75985082 |
| DCIS-2 | PT3 | center | Y | Y | N | -1.457784174 |
| DCIS-3 | PT3 | perifery | Y | Y | N | -1.306730608 |
| DCIS-5 | PT3 | perifery | Y | Y | N | -1.895551142 |
| DCIS-6 | PT3 | perifery | Y | Y | N | -1.523113297 |
| DCIS-7 | PT3 | perifery | Y | N |  | -1.905435455 |
| DCIS-8 | PT3 | center | Y | Y | N | 0.402315253 |
| DCIS-9 | PT3 | perifery | Y | N |  | -1.825202872 |
| InvF-1 | PT5 | inv.front | Y | Y | N | -1.559631488 |
| InvF-10 | PT5 | inv.front | N | Y | N | 0.172653115 |
| InvF-11 | PT5 | inv.front | Y | Y | Y | -0.716027869 |
| InvF-12 | PT5 | invasive front, area D | Y | Y | N | -1.859858382 |
| InvF-13 | PT5 | invasive front, area C | Y | Y | N | -1.880611243 |
| InvF-14 | PT5 | invasive front, area A | Y | N |  | -1.809023231 |
| InvF-15 | PT5 | inv.front | Y | N |  | -1.803970789 |
| InvF-20 | PT5 | inv.front | Y | N |  | -1.937882696 |
| InvF-3 | PT5 | inv.front | Y | N |  | -1.818042076 |
| InvF-4 | PT5 | inv.front | N | Y | N | -1.533986612 |
| InvF-5 | PT5 | inv.front | N | Y | N | -1.388840933 |
| InvF-6 | PT5 | inv.front | Y | N |  | -1.87592757 |
| InvF-7 | PT5 | inv.front | Y | N |  | -1.88781918 |
| InvF-8 | PT5 | inv.front | Y | Y | N | -1.813077387 |
| InvF-9 | PT5 | inv.front | Y | Y | N | -1.511498801 |
| Ly-10 | axillary metastasis | dispersed | Y | Y | N | 0.261978877 |
| Ly-3 | axillary metastasis | dispersed | Y | Y | N | -0.928705941 |
| Ly-5 | axillary metastasis | dispersed | Y | Y | N | -1.371828089 |
| Ly-6 | axillary metastasis | dispersed | Y | N |  | -1.842887258 |
| M-DCIS-1 | PT3 | malign. By DCIS | Y | Y | Y | -1.393645524 |
| M-DCIS-2 | PT3 | malign. By DCIS | Y | Y | N | -1.359413235 |
| M-DCIS-3 | PT3 | malign. By DCIS | Y | Y | N | -1.895106687 |
| M-DCIS-4 | PT3 | malign. By DCIS | Y | Y | N | -1.818969364 |
| M-DCIS-5 | PT3 | malign. By DCIS | Y | Y | N | -0.799090516 |
| M-DCIS-8 | PT3 | malign. By DCIS | N | Y | N | -1.45043714 |
| MeC-1 | axillary metastasis | center | Y | Y | N | -1.9225922 |
| MeC-10 | axillary metastasis | central area | Y | Y |  | -1.880374616 |
| MeC-11 | axillary metastasis | central area | Y | Y | N | -1.871518509 |

| | | | | | | |
|---|---|---|---|---|---|---|
| MeC-12 | axillary metastasis | center | Y | N | | -1.910135752 |
| MeC-16 | axillary metastasis | central area | Y | Y | N | -1.994832982 |
| MeC-2 | axillary metastasis | central area | N | Y | N | -0.762310387 |
| MeC-20 | axillary metastasis | central area | Y | Y | N | -1.981055634 |
| MeC-3 | axillary metastasis | center | Y | Y | N | -1.583372848 |
| MeC-4 | axillary metastasis | center | Y | Y | N | -1.016965215 |
| MeC-5 | axillary metastasis | center | Y | Y | N | -1.699949551 |
| MeC-6 | axillary metastasis | center | Y | N | | -1.871562275 |
| MeC-8 | axillary metastasis | central area | Y | Y | N | -1.647011696 |
| MeC-9 | axillary metastasis | central area | Y | N | | -1.903097335 |
| MeD-2 | axillary metastasis | dispersed | Y | Y | N | -0.406415815 |
| MeD-4 | axillary metastasis | dispersed | Y | Y | N | 0.308622424 |
| MeD-7 | axillary metastasis | dispersed | Y | Y | N | -0.755379128 |
| MeD-8 | axillary metastasis | dispersed | Y | Y | N | -0.972740402 |
| MeD-9 | axillary metastasis | dispersed | Y | Y | N | -0.990868958 |
| MeP-1 | axillary metastasis | border | Y | Y | N | -1.511511571 |
| MeP-12 | axillary metastasis | border | Y | Y | Y | -1.543698896 |
| MeP-14 | axillary metastasis | periphery, area C/cap area 3 | Y | Y | N | -1.926853886 |
| MeP-16 | axillary metastasis | periphery, area B/cap area 2 | Y | Y | N | -1.937172206 |
| MeP-17 | axillary metastasis | periphery, area A/cap area 1 | Y | Y | N | -1.918949613 |
| MeP-18 | axillary metastasis | border | N | Y | N | -1.607409286 |
| MeP-2 | axillary metastasis | border | N | Y | N | -1.36366298 |
| MeP-3 | axillary metastasis | border | Y | N | | -1.877781402 |
| MeP-4 | axillary metastasis | border | Y | Y | N | -1.258140359 |
| MeP-5 | axillary metastasis | periphery, area D/cap area 2 | Y | Y | Y | -1.652106102 |
| MeP-6 | axillary metastasis | periphery, area D/cap area 1 | Y | Y | N | -1.326512532 |
| MeP-9 | axillary metastasis | periphery, area C/cap area 2 | Y | Y | N | -1.860033978 |
| S1-1 | PT1 | solid | Y | Y | N | -1.772690566 |
| S1-10 | PT1 | solid | Y | N | | -1.849421463 |
| S1-11 | PT1 | solid | N | Y | N | 0.831562102 |
| S1-12 | PT1 | solid | Y | Y | Y | -1.508450871 |
| S1-2 | PT1 | solid | Y | Y | N | -1.339583357 |
| S1-3 | PT1 | solid | Y | N | | -1.82651608 |
| S1-4 | PT1 | solid | Y | Y | N | -1.722824892 |

| | | | | | | |
|---|---|---|---|---|---|---|
| S1-5 | PT1 | solid | Y | N | | -1.879060894 |
| S1-6 | PT1 | solid | Y | Y | N | -0.986129976 |
| S1-9 | PT1 | solid | Y | Y | N | -1.703030522 |
| S2-1 | PT2 | solid | Y | N | | -1.901747083 |
| S2-10 | PT2 | solid | Y | Y | Y | -1.625231285 |
| S2-13 | PT2 | solid | N | Y | N | -1.722179408 |
| S2-14 | PT2 | solid | Y | Y | N | -1.325602871 |
| S2-15 | PT2 | solid | Y | N | | -1.913269379 |
| S2-2 | PT2 | solid | Y | Y | N | -1.792231421 |
| S2-3 | PT2 | solid | Y | N | | -1.879618904 |
| S2-4 | PT2 | solid | Y | Y | N | -0.729499775 |
| S2-5 | PT2 | solid | Y | Y | Y | -1.732883428 |
| S2-7 | PT2 | solid | Y | Y | N | -1.737388068 |
| S2-8 | PT2 | solid | Y | Y | Y | -1.503322184 |
| S2-9 | PT2 | solid | Y | Y | N | -1.356069517 |
| S3-1 | PT3 | solid | Y | Y | N | -1.721577202 |
| S3-10 | PT3 | solid | Y | N | | -1.844822501 |
| S3-11 | PT3 | solid | Y | N | | -1.849788812 |
| S3-12 | PT3 | solid | Y | N | | -1.8816722 |
| S3-13 | PT3 | solid | Y | Y | N | -1.128722513 |
| S3-2 | PT3 | solid | N | Y | N | -0.754061828 |
| S3-3 | PT3 | solid | Y | Y | N | -1.348910399 |
| S3-4 | PT3 | solid | Y | N | | -1.813426499 |
| S3-5 | PT3 | solid | Y | N | | -1.845256606 |
| S3-7 | PT3 | solid | Y | Y | N | -1.559659601 |
| S3-8 | PT3 | solid | Y | N | | -1.899519124 |
| S3-9 | PT3 | solid | Y | Y | N | -1.597948792 |

# Supplemental Table 3: 78 SNVs specific to cancer cell-pools/single-cell.

| Gene | Function | pHGVS | chromosome | coordinate | ref | alter | All_reads_Cancer_DCS | Mut_reads_Cancer_DCS | All_reads_BN_DCS | Mut_reads_BN_DCS | Vaf_in_MT_cell_pool(%) | Vaf_in_PT4_cell_pool(%) | Vaf_in_InvF_cell_pool(%) | Vaf_in_PT1_cell_pool(%) | Vaf_in_PT2_cell_pool(%) | Vaf_in_PT3_cell_pool(%) | Vaf_in_BN_cell_pool(%) | Vaf_in_Skin-T(%) | Vaf_in_Ly-T(%) |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| RFWD2 | intron | . | chr1 | 176012469 | A | A/T | 1653 | 1016 | 257 | 0 | 60.6 | 41.5 | 69.4 | 59.2 | 66.8 | 29.9 | 0.2 | 0 | 0 |
| RYR2 | splice-5 | . | chr1 | 237580424 | G | G/A | 2197 | 1180 | 447 | 0 | 55.1 | 41 | 74 | 57.6 | 68.5 | 28.8 | 0.1 | 0 | 1.9 |
| PIK3CA | nonsense | p.Glu545* | chr3 | 178936091 | G | G/A | 1786 | 693 | 343 | 2 | 46.6 | 35 | 41.8 | 39.6 | 40.5 | 22.4 | 0.2 | 0 | 0 |
| EIF3B | missense | p.Asp656Val | chr7 | 2415101 | A | A/G | 1904 | 674 | 320 | 0 | 39.1 | 22.9 | 52.1 | 38.5 | 43.9 | 17.7 | 0 | 0 | 0.7 |
| HTRA2 | coding-synon | . | chr2 | 74757492 | T | T/C | 1117 | 274 | 109 | 0 | 23.1 | 14.8 | 38.3 | 25.2 | 37.6 | 11.8 | 0 | 0 | 0 |
| DNAH2 | missense | p.Arg1270Leu | chr17 | 7671351 | G | G/A | 1037 | 328 | 221 | 0 | 32.4 | 19.2 | 62.7 | 39.5 | 58.3 | 9.6 | 0 | 0 | 0 |
| RP1L1 | intron | . | chr8 | 10479301 | C | C/T | 1444 | 420 | 225 | 0 | 34.1 | 20.1 | 40.8 | 31.4 | 41.8 | 7.4 | 0 | 2.7 | 0 |
| SCUBE3 | intron | . | chr6 | 35212297 | A | A/C | 1710 | 430 | 422 | 0 | 26.3 | 17.6 | 37.7 | 24.3 | 38.4 | 8.3 | 0 | 0 | 0 |
| MYH11 | intron | . | chr16 | 15869796 | A | A/G | 1632 | 539 | 317 | 0 | 37.3 | 23.5 | 53.3 | 38.3 | 48.1 | 17 | 0 | 0 | 0 |
| TAF4B | intron | . | chr18 | 23847672 | T | T/A | 1317 | 297 | 400 | 0 | 29.7 | 16.9 | 31.7 | 20.9 | 31.6 | 8.3 | 0.1 | 0 | 0 |
| INPP5B | missense | p.Gly177Glu | chr1 | 38397587 | C | C/T | 1082 | 253 | 206 | 0 | 25.8 | 14.9 | 40 | 24.4 | 35.6 | 7.5 | 0 | 0 | 0 |
| KIT | coding-synon | . | chr4 | 55564710 | T | T/A | 1367 | 336 | 248 | 0 | 25 | 14.9 | 37.8 | 20.3 | 36 | 9.8 | 0.1 | 0 | 0 |
| GLT1D1 | intron | . | chr12 | 129360318 | C | C/A | 281 | 60 | 74 | 0 | 24.9 | 16.4 | 31.4 | 28.8 | 34.1 | 7.1 | 0 | 0 | 0 |
| SSFA2 | intron | . | chr2 | 182761450 | G | G/A | 1427 | 334 | 323 | 0 | 26.5 | 13.9 | 38.2 | 20.9 | 35.7 | 9.6 | 0 | 0 | 0 |
| AFAP1L1 | missense | p.Gly505Trp | chr5 | 148699178 | G | G/A | 1162 | 266 | 167 | 0 | 26.9 | 13.9 | 37.2 | 24.3 | 36.2 | 7.9 | 0 | 0 | 1.5 |
| L3HYPDH | promoter | . | chr14 | 59954739 | G | G/A | 1451 | 350 | 404 | 0 | 26.4 | 13.7 | 36.5 | 27 | 38.6 | 9.7 | 0 | 0 | 0 |
| SPATA31D1 | missense | p.Lys686Ile | chr9 | 84607442 | A | A/T | 1294 | 304 | 244 | 0 | 23.5 | 16.3 | 36.7 | 19.6 | 31.4 | 8.4 | 0 | 0 | 0 |
| SON | intron | . | chr21 | 34945784 | G | G/T | 1483 | 320 | 323 | 0 | 24.4 | 12.7 | 35.4 | 20.3 | 36.6 | 8.3 | 0.1 | 0 | 0 |
| ARID4B | coding-synon | . | chr1 | 235345135 | G | G/A | 2274 | 569 | 393 | 0 | 27.8 | 20.5 | 33.7 | 26 | 34 | 15.2 | 0 | 0 | 0 |
| GSPT2 | utr-3 | . | chrX | 51488774 | G | G/T | 1556 | 344 | 457 | 0 | 23.4 | 14.6 | 40.3 | 26.4 | 37 | 12.1 | 0 | 0 | 0 |
| SEC24A | coding-synon | . | chr5 | 133997269 | A | A/C | 1200 | 253 | 371 | 0 | 22.9 | 16.7 | 35.4 | 22.6 | 34.3 | 12.3 | 0 | 0 | 0 |
| RAP1GDS1 | missense | p.Asp139Tyr | chr4 | 99300218 | G | G/T | 1256 | 273 | 299 | 0 | 23.7 | 16.5 | 31.2 | 23.2 | 35.5 | 5.6 | 0 | 0 | 0 |
| RAD54L2 | intron | . | chr3 | 51675937 | T | T/G | 1652 | 383 | 445 | 0 | 23.2 | 17.4 | 33.8 | 18.5 | 39.1 | 8.9 | 0 | 0 | 0 |
| TMPRSS7 | coding-synon | . | chr3 | 111769579 | A | A/G | 1299 | 267 | 373 | 0 | 25.6 | 15.6 | 35.9 | 20.4 | 30.5 | 9.4 | 0.2 | 0 | 0 |
| RUNX1 | nonsense | p.Ser322* | chr21 | 36171600 | G | G/C | 1512 | 333 | 310 | 0 | 26.5 | 18.9 | 34.8 | 22.5 | 34.2 | 9.2 | 0 | 0 | 0 |
| G6PC | intron | . | chr17 | 41059362 | C | C/T | 907 | 210 | 178 | 0 | 26.9 | 13.5 | 34.6 | 24.5 | 36.8 | 7.9 | 0.1 | 0 | 0 |
| NELL2 | intron | . | chr12 | 45210076 | T | T/A | 1169 | 265 | 322 | 0 | 27.1 | 14 | 33.1 | 20.9 | 32.4 | 10.2 | 0 | 0 | 0 |
| CD248 | nonsense | p.Tyr329* | chr11 | 66083512 | G | G/T | 1372 | 339 | 247 | 0 | 24.1 | 14.5 | 37.3 | 28 | 31.6 | 8 | 0.2 | 0 | 0.4 |
| SMC3 | intron | . | chr10 | 112337693 | T | T/A | 1280 | 298 | 258 | 0 | 23.6 | 13 | 34.2 | 21.1 | 32.5 | 8.5 | 0.1 | 0 | 0 |
| WDR78 | intron | . | chr1 | 67288225 | A | A/C | 836 | 176 | 216 | 0 | 24.8 | 15.2 | 38.5 | 17.8 | 36.9 | 10.5 | 0 | 0 | 0 |
| . | . | . | chr9 | 40332891 | G | G/C | 1305 | 300 | 322 | 0 | 26.1 | 14.2 | 34.4 | 26.4 | 38.1 | 7.6 | 0 | 0 | 0 |
| S100B | utr-3 | . | chr21 | 48019239 | G | G/A | 1504 | 362 | 367 | 0 | 23.3 | 17.2 | 36.7 | 26.2 | 34.1 | 10 | 0.2 | 0 | 3.3 |
| MIR523 | promoter | . | chr19 | 54201564 | T | T/A | 1032 | 242 | 196 | 0 | 24.7 | 15.5 | 39.1 | 26.8 | 37.4 | 12.7 | 0 | 0 | 0 |
| PIEZO2 | intron | . | chr18 | 10691421 | T | T/C | 1263 | 301 | 325 | 0 | 25.8 | 16.5 | 39.9 | 26.7 | 39 | 8 | 0 | 0 | 0 |
| PAPLN | intron | . | chr14 | 73725856 | G | G/A | 1161 | 250 | 135 | 0 | 22.3 | 13.1 | 39.8 | 23.8 | 33.7 | 7.5 | 0 | 0 | 0 |
| PDE7B | intron | . | chr6 | 136500313 | G | G/A | 954 | 202 | 234 | 0 | 23.8 | 17.2 | 33.6 | 19.7 | 33.4 | 8.9 | 0.2 | 0 | 0 |
| CEP135 | intron | . | chr4 | 56886813 | A | A/G | 1308 | 252 | 360 | 0 | 23.9 | 14.3 | 33.5 | 21.4 | 34 | 6.3 | 0 | 0 | 0 |
| ABCG1 | utr-3 | . | chr21 | 43716529 | G | G/A | 1308 | 294 | 211 | 0 | 25.5 | 15.4 | 35.6 | 23.8 | 32 | 7.2 | 0 | 0 | 0 |
| SFMBT2 | coding-synon | . | chr10 | 7247868 | C | C/G | 1460 | 343 | 321 | 0 | 25.4 | 16.3 | 39.1 | 26.1 | 33.1 | 10.2 | 0 | 0 | 2.5 |
| CDX1 | missense | p.Arg183Leu | chr5 | 149562433 | G | G/A | 1591 | 385 | 199 | 0 | 22.2 | 15.1 | 37.5 | 26.1 | 35.3 | 12.2 | 0 | 0 | 0.8 |
| KIAA1731 | missense | p.Pro209Leu | chr11 | 93412579 | C | C/G | 1339 | 273 | 377 | 0 | 22.6 | 13.1 | 37.5 | 19.8 | 34.2 | 9.4 | 0 | 0 | 0 |
| MTERFD1 | nonsense | p.Trp67* | chr8 | 97270718 | C | C/G | 1854 | 352 | 446 | 0 | 20.9 | 12.8 | 31.3 | 21.3 | 31.1 | 8.2 | 0 | 0 | 0 |
| SMARCA5 | missense | p.Thr99Ile | chr4 | 144442625 | C | C/T | 1220 | 276 | 399 | 0 | 23.6 | 15.6 | 36.8 | 26.6 | 36.3 | 10.5 | 0 | 0 | 0 |
| AGXT2L1 | intron | . | chr4 | 109683896 | G | G/T | 1033 | 188 | 218 | 0 | 22.6 | 14 | 34.2 | 21.6 | 34.5 | 8.1 | 0 | 0 | 0 |
| KIF3B | coding-synon | . | chr20 | 30897952 | C | C/T | 1283 | 290 | 268 | 0 | 26.3 | 16.5 | 35.6 | 26.8 | 32.4 | 9 | 0 | 1.5 | 0 |
| ZNF566 | missense | p.Gln136Lys | chr19 | 36940733 | G | G/T | 1357 | 307 | 419 | 0 | 25 | 13.8 | 35.9 | 21.5 | 39.1 | 10.7 | 0 | 0 | 0 |
| FLT3 | missense | p.Arg436Lys | chr13 | 28611324 | C | C/G | 1517 | 349 | 376 | 0 | 22.4 | 13.2 | 34.3 | 23.6 | 38.7 | 8.5 | 0 | 0 | 0 |
| EHMT1 | intron | . | chr9 | 140657011 | G | G/A | 1230 | 301 | 241 | 0 | 23 | 17.5 | 38.6 | 22.6 | 38 | 6.5 | 0.1 | 0 | 0 |
| LCORL | intron | . | chr4 | 17910679 | C | C/T | 946 | 181 | 306 | 0 | 24.2 | 14.4 | 38.6 | 22.3 | 33.4 | 8.7 | 0 | 0 | 0 |
| SEC14L3 | missense | p.Glu388Lys | chr22 | 30856049 | C | C/T | 1433 | 371 | 272 | 0 | 23.7 | 15 | 56.2 | 26.6 | 48.2 | 7.3 | 0 | 0 | 0 |
| CD99L2 | intron | . | chrX | 149984601 | A | A/T | 1361 | 355 | 346 | 0 | 22.7 | 15.6 | 40.2 | 27.4 | 42.6 | 12.9 | 0.1 | 0 | 0 |
| CREB5 | intron | . | chr7 | 28725930 | C | C/T | 1494 | 305 | 462 | 0 | 18.6 | 14.2 | 31.1 | 18.4 | 33.4 | 10.9 | 0 | 0 | 0 |
| FAM217B | missense | p.Asn88Ile | chr20 | 58519261 | A | A/T | 1561 | 345 | 306 | 0 | 24.3 | 13.8 | 37.2 | 24.3 | 37.1 | 11.9 | 0 | 0 | 0 |
| DNAJC9 | intron | . | chr10 | 75006655 | A | A/T | 686 | 126 | 115 | 0 | 24.9 | 14.4 | 36.2 | 27.1 | 28.7 | 7.6 | 0 | 0 | 0 |
| PCYT1B | coding-synon | . | chrX | 24597532 | C | C/T | 1277 | 303 | 347 | 0 | 22.2 | 16.9 | 39.3 | 24.1 | 40.7 | 8.5 | 0 | 0 | 0 |
| PKNOX2 | intron | . | chr11 | 125299770 | G | G/A | 1122 | 272 | 193 | 0 | 22.3 | 14.4 | 35 | 23.7 | 39.4 | 9.9 | 0.2 | 0 | 0 |
| . | . | . | chr1 | 164953186 | C | C/T | 1646 | 212 | 212 | 0 | 14.4 | 10 | 16 | 13.8 | 18.8 | 6.1 | 0 | 0 | 0 |
| ZNF667 | utr-3 | . | chr19 | 56952444 | T | T/A | 1211 | 267 | 292 | 0 | 24.9 | 15.1 | 32.1 | 23.8 | 32.5 | 8.5 | 0 | 0 | 0 |
| EDARADD | utr-3 | . | chr1 | 236646875 | G | G/C | 2082 | 294 | 248 | 0 | 14.3 | 10.4 | 19 | 16.3 | 18.1 | 6.7 | 0 | 0 | 1.1 |
| DNM3 | missense | p.Arg107Leu | chr1 | 171956880 | G | G/A | 2469 | 308 | 547 | 0 | 13.5 | 10.1 | 17.1 | 11.2 | 15.9 | 8.5 | 0.1 | 0.8 | 0 |
| ANK1 | intron | . | chr8 | 41529867 | C | C/T | 1566 | 163 | 204 | 0 | 17.7 | 12.6 | 6.3 | 12.3 | 3 | 8.9 | 0 | 0 | 0 |
| CAMTA1 | intron | . | chr1 | 7151574 | C | C/A | 1658 | 216 | 349 | 0 | 24.9 | 12.2 | 6.4 | 15.8 | 4.4 | 7.5 | 0 | 0 | 0 |
| AKR1CL1 | intron | . | chr10 | 5200766 | G | G/A | 1375 | 190 | 447 | 0 | 23.6 | 13.8 | 6.5 | 14.4 | 2.3 | 9.1 | 0 | 0 | 0 |
| F11 | nonsense | p.Arg497* | chr4 | 187207577 | C | C/T | 1459 | 197 | 418 | 0 | 23 | 14.9 | 9.3 | 17.3 | 3.7 | 8.4 | 0 | 0.5 | 0 |
| ARHGAP6 | missense | p.Arg450His | chrX | 11197553 | C | C/T | 1487 | 198 | 310 | 0 | 25.3 | 12.8 | 3.7 | 12.1 | 3.5 | 8 | 0 | 0 | 0 |
| RFX3 | intron | . | chr9 | 3270833 | C | C/A | 1353 | 174 | 435 | 0 | 26.2 | 9.8 | 7.9 | 11.7 | 3.5 | 8.2 | 0.2 | 0 | 0 |
| TRIM32 | utr-5 | . | chr9 | 119459942 | A | A/G | 1592 | 193 | 340 | 0 | 23 | 13.5 | 5.3 | 13.6 | 4.5 | 8.4 | 0 | 0 | 0 |
| CD5 | intron | . | chr11 | 60885566 | C | C/T | 817 | 98 | 144 | 0 | 26.8 | 13.3 | 7.8 | 13 | 4.3 | 13.3 | 0.5 | 0 | 0 |
| ZMYND8 | coding-synon | . | chr20 | 45905326 | G | G/A | 1249 | 170 | 294 | 0 | 22.7 | 11.2 | 7.5 | 11.6 | 5.9 | 7.9 | 0 | 0 | 0 |
| MYH6 | missense | p.Arg696His | chr14 | 23866253 | C | C/T | 640 | 13 | 45 | 0 | 7.3 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| GATAD2A | nonsense | p.Glu110* | chr19 | 19603173 | G | G/A | 796 | 24 | 55 | 0 | 19.3 | 0 | 4.9 | 0 | 0.1 | 0 | 0 | 2.7 | 0 |
| MAGEC3 | missense | p.Pro33Leu | chrX | 140926199 | C | C/A | 1249 | 9 | 247 | 0 | 2.4 | 0 | 0.2 | 0 | 0 | 0 | 0.1 | 0 | 0 |
| OR10K2 | nonsense | p.Ser133* | chr1 | 158390259 | G | G/A | 1946 | 152 | 240 | 0 | 0 | 0 | 26.5 | 10.7 | 25.8 | 0 | 0 | 0 | 0 |
| CHIC1 | intron | . | chrX | 72797180 | A | A/T | 1267 | 92 | 411 | 0 | 0.1 | 0 | 28 | 7.7 | 7 | 0.5 | 0 | 0 | 0 |
| PDE1C | intron | . | chr7 | 31847965 | A | A/G | 1702 | 176 | 408 | 0 | 0 | 0 | 45 | 16.2 | 37.3 | 0 | 0.2 | 0 | 0 |
| ARHGEF28 | intron | . | chr5 | 73072247 | G | G/T | 1369 | 20 | 380 | 0 | 0 | 3.4 | 0 | 0 | 0 | 1.3 | 0 | 0 | 0 |
| UBR5 | intron | . | chr8 | 103309664 | G | G/T | 1203 | 1 | 219 | 0 | 0 | 0 | 0 | 0 | 0.3 | 0 | 0.2 | 0 | 0 |
| KIF3C | intron | . | chr2 | 26178354 | G | G/T | 857 | 1 | 98 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0.1 | 0 | 0 |

# Supplemental Table 4: 49 SNVs specific to normal mammary cell-pool/single-cell.

| Gene | Function | pHGVS | chrom | coordinate | ref | alter | All_reads_Cancer_DCS | Mut_reads_Cancer_DCS | All_reads_BN_DCS | Mut_reads_BN_DCS | Vaf_in_MT_cell_pool(%) | Vaf_in_PT4_cell_pool(%) | Vaf_in_InvF_cell_pool(%) | Vaf_in_PT1_cell_pool(%) | Vaf_in_PT2_cell_pool(%) | Vaf_in_PT3_cell_pool(%) | Vaf_in_BN_cell_pool(%) | Vaf_in_Skin-T(%) | Vaf_in_Ly-T(%) |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| . | . | . | chrX | 151821894 | G | G/A | 1748 | 0 | 424 | 26 | 0.1 | 0 | 0 | 0 | 0 | 0 | 5.3 | 0 | 0 |
| HECTD4 | coding-synon | . | chr12 | 112646352 | C | C/T | 1362 | 0 | 357 | 19 | 0.7 | 0.2 | 0.2 | 0.2 | 0.2 | 0 | 2.7 | 0.9 | 0 |
| FAM127A | utr-3 | . | chrX | 134167162 | C | C/G | 1318 | 0 | 315 | 11 | 0.2 | 0 | 0 | 0 | 0 | 0 | 2.4 | 0 | 0 |
| ADCY8 | nonsense | p.Tyr1190* | chr8 | 131792822 | G | G/A | 1399 | 0 | 200 | 4 | 0 | 0 | 0 | 0 | 0 | 0 | 2.7 | 0 | 0 |
| KIAA1549 | coding-synon | . | chr7 | 138603490 | G | G/A | 1066 | 0 | 120 | 6 | 0.2 | 0.2 | 0 | 0.1 | 0 | 0 | 2.1 | 0 | 0 |
| XPO5 | intron | . | chr6 | 43535248 | A | A/G | 1642 | 0 | 438 | 6 | 0 | 0 | 0 | 0 | 0 | 0 | 3.7 | 0 | 0 |
| . | . | . | chr6 | 26427877 | C | C/G | 1432 | 0 | 256 | 4 | 0 | 0 | 0 | 0 | 0 | 0 | 1.9 | 0 | 0 |
| FAM120B | intron | . | chr6 | 170626416 | T | T/C | 1753 | 0 | 368 | 8 | 0 | 0 | 0 | 0 | 0 | 0 | 1.6 | 0 | 0 |
| ARHGEF28 | intron | . | chr5 | 73141983 | A | A/G | 976 | 0 | 222 | 6 | 0.1 | 0 | 0 | 0.3 | 0 | 0 | 4.8 | 0 | 0 |
| DDX60 | missense | p.Val1479Ile | chr4 | 169157501 | C | C/T | 1264 | 0 | 370 | 4 | 0 | 0 | 0 | 0 | 0 | 0 | 1.3 | 0 | 0 |
| PIK3CA | missense | p.His1047Leu | chr3 | 178952085 | A | A/G | 2188 | 1 | 482 | 12 | 0 | 0.1 | 0.1 | 0 | 0 | 0 | 3.8 | 0 | 0 |
| RFPL1 | missense | p.Arg203Leu | chr22 | 29837765 | G | G/A | 1110 | 0 | 253 | 11 | 0 | 0 | 0 | 0.2 | 0 | 0 | 3.9 | 1.1 | 0 |
| RBM12 | coding-synon | . | chr20 | 34242158 | T | T/A | 1582 | 0 | 410 | 18 | 0 | 0 | 0 | 0 | 0 | 0 | 5 | 0 | 0 |
| TYW5 | coding-synon | . | chr2 | 200801136 | T | T/C | 1189 | 0 | 391 | 17 | 0.1 | 0 | 0 | 0 | 0 | 0 | 4.7 | 0 | 0 |
| NLRP8 | coding-synon | . | chr19 | 56466570 | C | C/T | 1261 | 0 | 310 | 14 | 0 | 0 | 0 | 0 | 0 | 0 | 6.2 | 0 | 0 |
| ACOX1 | missense | p.Val534Met | chr17 | 73945426 | C | C/T | 1262 | 0 | 281 | 15 | 0.1 | 0 | 0 | 0 | 0 | 0 | 7.5 | 0 | 0 |
| ARSG | intron | . | chr17 | 66343089 | A | A/G | 1246 | 0 | 261 | 2 | 0 | 0 | 0 | 0 | 0.2 | 0 | 2.9 | 0 | 0 |
| USP7 | intron | . | chr16 | 8994508 | T | T/C | 1988 | 0 | 414 | 30 | 0 | 0 | 0 | 0 | 0 | 0 | 5.8 | 0 | 0 |
| ADNP2 | missense | p.Gln862Leu | chr18 | 77895881 | A | A/T | 1009 | 0 | 166 | 5 | 0 | 0 | 0 | 0 | 0 | 0 | 2.5 | 2.9 | 0 |
| OR7C1 | coding-synon | . | chr19 | 14910151 | T | T/C | 1058 | 0 | 205 | 2 | 0 | 0 | 0 | 0 | 0 | 0 | 0.8 | 0 | 0 |
| KIAA1468 | missense | p.Arg590Leu | chr18 | 59919932 | G | G/A | 1497 | 0 | 434 | 16 | 0 | 0.3 | 0 | 0 | 0 | 0 | 5.8 | 0.8 | 0 |
| BCMO1 | intron | . | chr16 | 81303746 | A | A/G | 697 | 0 | 122 | 6 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 0 | 0 |
| SLFN5 | missense | p.Ile58Phe | chr17 | 33585881 | A | A/G | 1510 | 0 | 353 | 30 | 0 | 0 | 0 | 0 | 0 | 0 | 7.4 | 0 | 0 |
| NMRAL1 | intron | . | chr16 | 4519161 | G | G/A | 680 | 0 | 97 | 3 | 0 | 0 | 0 | 0.3 | 0 | 0 | 1.8 | 0 | 0 |
| C2CD3 | missense | p.Pro766Thr | chr11 | 73814460 | G | G/A | 1455 | 0 | 338 | 2 | 0 | 0 | 0 | 0 | 0 | 0 | 1.6 | 0 | 0 |
| BTG4 | splice-3 | . | chr11 | 111338407 | C | C/A | 1472 | 0 | 262 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 2.1 | 0 | 0 |
| RORA | coding-synon | . | chr15 | 60803463 | T | T/C | 1454 | 0 | 434 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 1.1 | 0 | 0 |
| SLC11A2 | intron | . | chr12 | 51386791 | C | C/T | 1059 | 0 | 291 | 11 | 0.1 | 0.2 | 0 | 0 | 0 | 0 | 3.1 | 0 | 0 |
| AGL | coding-synon | . | chr1 | 100356847 | T | T/A | 1330 | 0 | 434 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| KIAA0907 | missense | p.Gly286Asp | chr1 | 155895459 | C | C/T | 2324 | 0 | 342 | 0 | 0 | 0 | 0 | 0.1 | 0 | 0 | 0.2 | 0 | 0 |
| CLK1 | intron | . | chr2 | 201719959 | G | G/A | 59 | 0 | 5 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1.4 | 0 | 0 |
| MECOM | intron | . | chr3 | 169099360 | C | C/T | 1866 | 0 | 418 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| . | . | . | chr4 | 101051130 | G | G/A | 861 | 0 | 184 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| BBS7 | intron | . | chr4 | 122784286 | T | T/C | 1398 | 0 | 440 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| ARAP3 | missense | p.Val444Ile | chr5 | 141052174 | C | C/T | 1060 | 0 | 129 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1.6 | 0 | 0 |
| AKR1B15 | missense | p.Glu78Val | chr7 | 134252992 | A | A/C | 1824 | 0 | 378 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| ADAM28 | intron | . | chr8 | 24167395 | G | G/A | 930 | 0 | 292 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| PKHD1L1 | missense | p.Arg1654Leu | chr8 | 110457059 | G | G/A | 1562 | 0 | 415 | 0 | 0.1 | 0 | 0 | 0 | 0 | 0 | 0 | 0.4 | 0 |
| IFNA16 | missense | p.Ser139Tyr | chr9 | 21216889 | G | G/C | 1418 | 0 | 366 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| PSAT1 | missense | p.Arg213Leu | chr9 | 80923397 | G | G/A | 1247 | 0 | 190 | 0 | 0.1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| GSN | coding-synon | . | chr9 | 124064411 | C | C/T | 1216 | 0 | 124 | 0 | 0.1 | 0.1 | 0 | 0.1 | 0 | 0 | 0.3 | 1 | 0 |
| . | . | . | chr11 | 55839004 | G | G/C | 1294 | 0 | 333 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| TUT1 | intron | . | chr11 | 62356789 | C | C/T | 1511 | 0 | 411 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| HEPHL1 | nonsense | p.Arg801* | chr11 | 93826773 | C | C/T | 1421 | 0 | 359 | 0 | 0.1 | 0 | 0 | 0 | 0 | 0 | 0.3 | 0 | 0 |
| GABRB3 | intron | . | chr15 | 26825646 | G | G/A | 1510 | 0 | 411 | 0 | 0.1 | 0 | 0 | 0 | 0 | 0 | 0 | 1.2 | 0 |
| FOXN1 | coding-synon | . | chr17 | 26856144 | C | C/A | 1500 | 0 | 236 | 0 | 0 | 0 | 0 | 0 | 0.1 | 0.2 | 0.4 | 0 | 0.4 |
| ZNF254 | missense | p.Lys245Asn | chr19 | 24309537 | A | A/G | 1133 | 0 | 350 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| ERCC1 | missense | p.Thr252Asn | chr19 | 45917240 | G | G/A | 1433 | 0 | 345 | 0 | 0 | 0.1 | 0 | 0 | 0 | 0 | 0 | 1.5 | 0 |
| PREX1 | coding-synon | . | chr20 | 47309328 | C | C/T | 1278 | 0 | 220 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |

## Supplementary Table 5: 22 SNVs presumably derived from hematopoietic cells.

| Gene | Function | pHGVS | chrom | coordinate | ref | alter | All_reads_Cancer_DCS | Mut_reads_Cancer_DCS | All_reads_BN_DCS | Mut_reads_BN_DCS | Vaf_in_MT_cell_pool(%) | Vaf_in_PT4_cell_pool(%) | Vaf_in_InvF_cell_pool(%) | Vaf_in_PT1_cell_pool(%) | Vaf_in_PT2_cell_pool(%) | Vaf_in_PT3_cell_pool(%) | Vaf_in_BN_cell_pool(%) | Vaf_in_Skin-T(%) | Vaf_in_Ly-T(%) |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| TET2 | missense | p.Ser792Leu | chr4 | 106157474 | C | C/A | 1488 | 3 | 461 | 0 | 0.2 | 0.2 | 0 | 0.5 | 0.3 | 0 | 0.1 | 1.6 | 0 |
| KDM5C | missense | p.Phe511Tyr | chrX | 53239909 | A | A/C | 1523 | 53 | 224 | 0 | 3 | 4 | 2.5 | 3.7 | 2 | 3.9 | 1.1 | 4.8 | 15.7 |
| CNKSR2 | missense | p.Arg59Cys | chrX | 21444725 | C | C/T | 1284 | 42 | 276 | 2 | 3.8 | 3.3 | 1.8 | 4.3 | 1.3 | 2.4 | 1.9 | 3.1 | 3.9 |
| CIR1 | missense | p.Asn303Lys | chr2 | 175213669 | G | G/A | 1643 | 50 | 390 | 8 | 2.6 | 2.8 | 2.5 | 3.9 | 2.2 | 2.9 | 2 | 2.4 | 14.2 |
| ST6GALNAC2 | missense | p.Ile368Asn | chr17 | 74562208 | A | A/G | 1730 | 53 | 307 | 3 | 2.7 | 4.3 | 2.1 | 3.8 | 2.5 | 2.8 | 1.2 | 2.1 | 9.5 |
| NUP214 | intron | . | chr9 | 134090452 | T | T/C | 1615 | 50 | 425 | 16 | 2.2 | 4.7 | 1.7 | 2.8 | 3.1 | 3.7 | 3.5 | 12.5 | 14.2 |
| STXBP5L | missense | p.His149Leu | chr3 | 120764358 | A | A/T | 1349 | 41 | 485 | 17 | 2.2 | 3.9 | 3.1 | 2.2 | 2.8 | 4.2 | 2.1 | 0.7 | 5.7 |
| INPP5J | coding-synon | . | chr22 | 31522431 | G | G/A | 1036 | 38 | 160 | 3 | 2.6 | 3.9 | 3.7 | 4.8 | 3.5 | 2.7 | 1.2 | 0 | 5.1 |
| OTUD5 | intron | . | chrX | 48792335 | G | G/C | 1633 | 47 | 303 | 3 | 3.4 | 4 | 1.8 | 3.1 | 2.1 | 5.8 | 1.7 | 0 | 8.3 |
| ARHGAP21 | missense | p.His899Gln | chr10 | 24893265 | G | G/A | 1129 | 18 | 366 | 0 | 0.5 | 2.1 | 0.8 | 3.1 | 0.7 | 3.5 | 0.7 | 1.8 | 0 |
| LAMB4 | intron | . | chr7 | 107720040 | G | G/T | 1797 | 15 | 430 | 8 | 0.9 | 0.6 | 1.1 | 0.3 | 0.7 | 0.5 | 1 | 2.1 | 0 |
| AMHR2 | missense | p.Cys551Phe | chr12 | 53825187 | G | G/C | 1506 | 17 | 391 | 2 | 1.2 | 1.7 | 0.8 | 1.1 | 1 | 2.3 | 1.5 | 0 | 2.4 |
| MAGEC3 | intron | . | chrX | 140969602 | A | A/G | 1445 | 24 | 305 | 0 | 1.9 | 1.1 | 0.9 | 0.9 | 1.3 | 2.2 | 0.5 | 0 | 0 |
| WDPCP | missense | p.Val306Ile | chr2 | 63631702 | C | C/T | 1370 | 8 | 352 | 4 | 0.7 | 0.4 | 0.2 | 0.9 | 0 | 0.2 | 0.4 | 0 | 0 |
| C7orf26 | coding-synon | . | chr7 | 6639734 | C | C/G | 1492 | 5 | 181 | 0 | 0.6 | 0.1 | 0.8 | 0.5 | 0.1 | 1 | 0.2 | 0 | 0 |
| LTBP1 | intron | . | chr2 | 33614215 | A | A/G | 1086 | 1 | 260 | 0 | 0 | 0.2 | 0 | 0.3 | 0 | 0.2 | 0 | 2.5 | 0.4 |
| ZNF333 | missense | p.Asn460Ile | chr19 | 14829518 | A | A/G | 1125 | 7 | 216 | 2 | 0.3 | 0.5 | 0.7 | 0.6 | 1.6 | 1.7 | 0.5 | 0 | 0 |
| ZNF878 | missense | p.His383Gln | chr19 | 12155067 | G | G/A | 1310 | 4 | 332 | 0 | 0.3 | 0.4 | 0.3 | 0.1 | 0.8 | 0.8 | 0.2 | 0 | 0 |
| CYP19A1 | coding-synon | . | chr15 | 51529096 | G | G/A | 1576 | 4 | 412 | 0 | 0.8 | 0.7 | 0.3 | 0.1 | 0 | 0.4 | 0.2 | 0 | 0 |
| MGA | intron | . | chr15 | 42049973 | T | T/A | 1435 | 10 | 322 | 0 | 0.7 | 1 | 1 | 1.1 | 0.4 | 0 | 0.3 | 0 | 0 |
| ANXA9 | missense | p.Gly101Val | chr1 | 150956791 | G | G/A | 2373 | 1 | 282 | 0 | 0.1 | 0 | 0.1 | 0 | 0.2 | 0 | 0.3 | 0 | 0 |
| DDR2 | intron | . | chr1 | 162725140 | A | A/G | 1988 | 0 | 344 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 1.5 | 0 | 0 |

## Supplemental Table 6: CNV segmentation of all 7 cell pools.

| Sample_id | Chr | Start | End | Bins_number | Log2_Copy_ratio | Copy_ratio | Copy_Number |
|---|---|---|---|---|---|---|---|
| BN-T | 1 | 700000 | 156800000 | 1296 | 0.007649246 | 1.005316134 | 2 |
| BN-T | 1 | 156900000 | 248900000 | 917 | 0.112535076 | 1.081126303 | 3 |
| BN-T | 2 | 100000 | 242900000 | 2360 | 0.010972316 | 1.007634425 | 2 |
| BN-T | 3 | 0 | 197600000 | 1942 | 0.004360513 | 1.00302705 | 2 |
| BN-T | 4 | 100000 | 190800000 | 1863 | 0.006392782 | 1.004440971 | 2 |
| BN-T | 5 | 200000 | 180700000 | 1756 | 0.009692354 | 1.006740846 | 2 |
| BN-T | 6 | 700000 | 170900000 | 1621 | 0.008104712 | 1.005633568 | 2 |
| BN-T | 7 | 500000 | 158700000 | 1525 | -0.004900102 | 0.99660927 | 2 |
| BN-T | 8 | 100000 | 146200000 | 1413 | 0.00278909 | 1.00193512 | 2 |
| BN-T | 9 | 200000 | 140800000 | 1100 | 0.000995554 | 1.000690303 | 2 |
| BN-T | 10 | 300000 | 135100000 | 1278 | -0.003897902 | 0.997301827 | 2 |
| BN-T | 11 | 300000 | 134800000 | 1304 | 0.007441052 | 1.005171068 | 2 |
| BN-T | 12 | 300000 | 133300000 | 1300 | -0.011505096 | 0.992056989 | 2 |
| BN-T | 13 | 19100000 | 114800000 | 954 | 0.01276197 | 1.008885165 | 2 |
| BN-T | 14 | 19100000 | 106800000 | 867 | 0.000777701 | 1.000539206 | 2 |
| BN-T | 15 | 20400000 | 102300000 | 781 | -0.004593337 | 0.996821204 | 2 |
| BN-T | 16 | 400000 | 90200000 | 764 | -0.028194449 | 0.980646822 | 2 |
| BN-T | 17 | 100000 | 80800000 | 759 | -0.048512125 | 0.966933029 | 2 |
| BN-T | 18 | 100000 | 77900000 | 745 | 0.01275792 | 1.008882332 | 2 |
| BN-T | 19 | 500000 | 59100000 | 550 | -0.061502599 | 0.958265545 | 2 |
| BN-T | 20 | 400000 | 62900000 | 594 | -0.000135006 | 0.999906425 | 2 |
| BN-T | 21 | 10900000 | 47700000 | 334 | 0.012949307 | 1.009016179 | 2 |
| BN-T | 22 | 16800000 | 50900000 | 332 | -0.026870731 | 0.981547009 | 2 |
| BN-T | X | 2800000 | 154400000 | 1466 | 0.001583446 | 1.001098164 | 2 |
| PT4 | 1 | 700000 | 142600000 | 1191 | -0.013529735 | 0.99066574 | 2 |
| PT4 | 1 | 142600000 | 248900000 | 1023 | 0.775893004 | 1.712249577 | 7 |
| PT4 | 2 | 100000 | 243000000 | 2361 | 0.008110792 | 1.005637805 | 2 |
| PT4 | 3 | 200000 | 167400000 | 1638 | 0.003778318 | 1.002622363 | 2 |
| PT4 | 3 | 167600000 | 197800000 | 302 | 0.451230868 | 1.367206223 | 5 |
| PT4 | 4 | 300000 | 190900000 | 1862 | -0.02066057 | 0.985781239 | 2 |
| PT4 | 5 | 300000 | 14500000 | 142 | 0.148307566 | 1.108268593 | 3 |
| PT4 | 5 | 14900000 | 180300000 | 1605 | 0.0632739 | 1.044834109 | 2 |
| PT4 | 6 | 200000 | 170700000 | 1624 | -0.008702112 | 0.99398631 | 2 |
| PT4 | 7 | 100000 | 55700000 | 556 | 0.289526852 | 1.222239365 | 4 |
| PT4 | 7 | 55900000 | 77500000 | 166 | -0.028143427 | 0.980681504 | 2 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| PT4 | 7 | 77600000 | 97200000 | 196 | 0.288788948 | 1.221614378 | 4 |
| PT4 | 7 | 97300000 | 103000000 | 55 | 0.030100722 | 1.02108341 | 2 |
| PT4 | 7 | 103300000 | 159100000 | 553 | 0.269968359 | 1.205781383 | 4 |
| PT4 | 8 | 500000 | 24200000 | 223 | -0.295310258 | 0.814897066 | 0 |
| PT4 | 8 | 24500000 | 146200000 | 1183 | 0.255604561 | 1.193835915 | 4 |
| PT4 | 9 | 200000 | 140800000 | 1100 | 0.125366975 | 1.090785158 | 3 |
| PT4 | 10 | 200000 | 11400000 | 112 | 0.02951557 | 1.020669346 | 2 |
| PT4 | 10 | 11800000 | 43100000 | 274 | -0.160173672 | 0.894917334 | 1 |
| PT4 | 10 | 43500000 | 122200000 | 757 | -0.004065335 | 0.997186091 | 2 |
| PT4 | 10 | 122300000 | 135100000 | 127 | 0.095189614 | 1.068205793 | 3 |
| PT4 | 11 | 100000 | 134800000 | 1306 | 0.005975341 | 1.00415038 | 2 |
| PT4 | 12 | 300000 | 110400000 | 1072 | -0.027973775 | 0.980796833 | 2 |
| PT4 | 12 | 110400000 | 133400000 | 229 | -0.105832561 | 0.929268519 | 1 |
| PT4 | 13 | 19100000 | 104200000 | 850 | 0.195372058 | 1.145019413 | 3 |
| PT4 | 13 | 104200000 | 114900000 | 105 | 0.256545274 | 1.194614612 | 4 |
| PT4 | 14 | 19000000 | 107100000 | 871 | 0.05351059 | 1.037787159 | 2 |
| PT4 | 15 | 20400000 | 102500000 | 783 | -0.012371348 | 0.991461497 | 2 |
| PT4 | 16 | 500000 | 32100000 | 300 | 0.16353379 | 1.120027216 | 3 |
| PT4 | 16 | 32800000 | 90100000 | 457 | -0.419807296 | 0.747524466 | 0 |
| PT4 | 17 | 400000 | 19300000 | 188 | -0.472740047 | 0.720594704 | 0 |
| PT4 | 17 | 20000000 | 81000000 | 564 | -0.077958199 | 0.94739752 | 2 |
| PT4 | 18 | 300000 | 77900000 | 743 | 0.012799313 | 1.008911279 | 2 |
| PT4 | 19 | 300000 | 59100000 | 552 | -0.193167927 | 0.874682947 | 1 |
| PT4 | 20 | 100000 | 55900000 | 527 | 0.054582342 | 1.038558398 | 2 |
| PT4 | 20 | 55900000 | 62800000 | 69 | 0.176790736 | 1.130366595 | 3 |
| PT4 | 21 | 9400000 | 48100000 | 346 | 0.005230863 | 1.003632339 | 2 |
| PT4 | 22 | 16900000 | 51200000 | 334 | -0.184090095 | 0.880204044 | 1 |
| PT4 | X | 3000000 | 53400000 | 496 | -0.07210345 | 0.951250063 | 2 |
| PT4 | X | 53400000 | 85400000 | 286 | -0.159095051 | 0.895586663 | 1 |
| PT4 | X | 85500000 | 154800000 | 685 | -0.055291158 | 0.962400196 | 2 |
| PT1 | 1 | 725000 | 142625000 | 4121 | -0.009030874 | 0.993759826 | 2 |
| PT1 | 1 | 142700000 | 249175000 | 3058 | 0.717221574 | 1.644012849 | 6 |
| PT1 | 2 | 125000 | 243000000 | 8825 | 0.003499393 | 1.002428538 | 2 |
| PT1 | 3 | 275000 | 167300000 | 6149 | 0.000353795 | 1.000245262 | 2 |
| PT1 | 3 | 167475000 | 197850000 | 1142 | 0.27600073 | 1.210833697 | 4 |
| PT1 | 4 | 100000 | 190800000 | 7051 | -0.017081222 | 0.988230013 | 2 |
| PT1 | 5 | 100000 | 180550000 | 6594 | -0.017853151 | 0.987701393 | 2 |
| PT1 | 6 | 225000 | 170825000 | 6143 | 0.007927592 | 1.005510113 | 2 |
| PT1 | 7 | 375000 | 159075000 | 5473 | 0.288795634 | 1.22162004 | 4 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| PT1 | 8 | 225000 | 24500000 | 861 | -0.143685025 | 0.905204069 | 2 |
| PT1 | 8 | 24700000 | 68275000 | 1531 | 0.219947984 | 1.164691593 | 4 |
| PT1 | 8 | 68400000 | 146225000 | 2897 | 0.121305844 | 1.087718957 | 2 |
| PT1 | 9 | 50000 | 140900000 | 3993 | 0.009844737 | 1.006847188 | 2 |
| PT1 | 10 | 175000 | 135350000 | 4745 | 0.000525372 | 1.000364226 | 2 |
| PT1 | 11 | 175000 | 134900000 | 4592 | 0.020623408 | 1.01439772 | 2 |
| PT1 | 12 | 150000 | 133450000 | 4761 | -0.029754683 | 0.979586853 | 2 |
| PT1 | 13 | 19025000 | 114950000 | 3610 | 3.17E-05 | 1.000021973 | 2 |
| PT1 | 14 | 19775000 | 107275000 | 3186 | -0.022981569 | 0.984196596 | 2 |
| PT1 | 15 | 20125000 | 33025000 | 365 | 0.208091927 | 1.155159388 | 4 |
| PT1 | 15 | 33025000 | 102225000 | 2541 | -0.000921626 | 0.999361381 | 2 |
| PT1 | 16 | 75000 | 33825000 | 937 | 0.295266332 | 1.22711149 | 4 |
| PT1 | 16 | 34375000 | 90100000 | 1613 | -0.339104971 | 0.790531595 | 1 |
| PT1 | 17 | 225000 | 17950000 | 654 | -0.403337587 | 0.756107051 | 0 |
| PT1 | 17 | 18025000 | 81125000 | 1916 | -0.010444264 | 0.992786729 | 2 |
| PT1 | 18 | 250000 | 77875000 | 2841 | 0.014413782 | 1.010040947 | 2 |
| PT1 | 19 | 300000 | 58975000 | 1432 | -0.026986006 | 0.981468584 | 2 |
| PT1 | 20 | 150000 | 62825000 | 2065 | 0.040128837 | 1.028205644 | 2 |
| PT1 | 21 | 9525000 | 48100000 | 1212 | -0.067161728 | 0.954514006 | 2 |
| PT1 | 22 | 17100000 | 51100000 | 1102 | -0.130003904 | 0.913828977 | 2 |
| PT1 | X | 2800000 | 148350000 | 5238 | 0.032162687 | 1.022543832 | 2 |
| MT | 1 | 700000 | 144900000 | 1199 | 0.020568238 | 1.014358929 | 2 |
| MT | 1 | 145700000 | 249200000 | 1010 | 0.806968745 | 1.749531627 | 7 |
| MT | 2 | 200000 | 242900000 | 2359 | 0.000798379 | 1.000553548 | 2 |
| MT | 3 | 200000 | 167300000 | 1637 | -0.001117933 | 0.999225408 | 2 |
| MT | 3 | 167700000 | 197900000 | 302 | 0.604306445 | 1.52024774 | 6 |
| MT | 4 | 0 | 191100000 | 1866 | -0.002070712 | 0.998565721 | 2 |
| MT | 5 | 400000 | 180300000 | 1750 | -0.000650744 | 0.99954904 | 2 |
| MT | 6 | 200000 | 170900000 | 1626 | 0.009862219 | 1.006859388 | 2 |
| MT | 7 | 400000 | 158900000 | 1528 | 0.313510413 | 1.242727871 | 4 |
| MT | 8 | 200000 | 23900000 | 223 | -0.44212214 | 0.736051113 | 0 |
| MT | 8 | 24300000 | 146300000 | 1186 | 0.310627914 | 1.240247383 | 4 |
| MT | 9 | 500000 | 140900000 | 1098 | -0.005369412 | 0.996285125 | 2 |
| MT | 10 | 300000 | 135400000 | 1281 | -0.010483891 | 0.992759461 | 2 |
| MT | 11 | 400000 | 134700000 | 1302 | -0.001916428 | 0.998672515 | 2 |
| MT | 12 | 200000 | 133800000 | 1304 | 0.006580312 | 1.004571543 | 2 |
| MT | 13 | 19300000 | 114700000 | 951 | 0.002456089 | 1.001703881 | 2 |
| MT | 14 | 19000000 | 107200000 | 872 | 0.007094207 | 1.00492944 | 2 |
| MT | 15 | 20500000 | 102500000 | 782 | 0.011574396 | 1.008055028 | 2 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| MT | 16 | 400000 | 33000000 | 308 | 0.314503547 | 1.243583644 | 4 |
| MT | 16 | 33600000 | 90200000 | 452 | -0.413823534 | 0.750631358 | 0 |
| MT | 17 | 500000 | 19700000 | 191 | -0.331095391 | 0.794932688 | 0 |
| MT | 17 | 20100000 | 81000000 | 563 | 0.031350253 | 1.021968163 | 2 |
| MT | 18 | 300000 | 77600000 | 740 | -0.016935394 | 0.988329909 | 2 |
| MT | 19 | 200000 | 58700000 | 549 | 0.033373463 | 1.023402358 | 2 |
| MT | 20 | 0 | 62800000 | 597 | -0.017518912 | 0.987930246 | 2 |
| MT | 21 | 9600000 | 48000000 | 343 | 0.015239931 | 1.010619506 | 2 |
| MT | 22 | 16900000 | 51200000 | 334 | -0.010707312 | 0.99260573 | 2 |
| MT | X | 2800000 | 154700000 | 1469 | -0.038401625 | 0.973733158 | 2 |
| PT2 | 1 | 725000 | 144700000 | 4193 | -0.020302496 | 0.986025938 | 2 |
| PT2 | 1 | 145125000 | 248475000 | 2820 | 0.897565081 | 1.862919172 | 6 |
| PT2 | 2 | 250000 | 242950000 | 8790 | -0.028514738 | 0.980429135 | 2 |
| PT2 | 3 | 125000 | 197875000 | 7363 | -0.014617165 | 0.989919308 | 2 |
| PT2 | 4 | 350000 | 190775000 | 6993 | -0.045007597 | 0.969284713 | 2 |
| PT2 | 5 | 50000 | 180600000 | 6573 | -0.068345676 | 0.953731005 | 2 |
| PT2 | 6 | 475000 | 170925000 | 6163 | -0.013866239 | 0.990434697 | 2 |
| PT2 | 7 | 675000 | 120775000 | 4226 | 0.160433987 | 1.117623288 | 2 |
| PT2 | 7 | 120775000 | 159100000 | 1332 | 0.259282572 | 1.196883367 | 4 |
| PT2 | 8 | 300000 | 145900000 | 5310 | 0.004239949 | 1.002943231 | 2 |
| PT2 | 8 | 145925000 | 146300000 | 13 | -0.465053516 | 0.724444204 | 2 |
| PT2 | 9 | 200000 | 141100000 | 4015 | 0.000932387 | 1.00064649 | 2 |
| PT2 | 10 | 200000 | 135475000 | 4749 | -0.020741134 | 0.985726192 | 2 |
| PT2 | 11 | 350000 | 112300000 | 3839 | 0.041965525 | 1.029515482 | 2 |
| PT2 | 12 | 175000 | 133375000 | 4755 | -0.047274604 | 0.967762804 | 2 |
| PT2 | 13 | 19050000 | 114925000 | 3643 | 0.054726985 | 1.038662529 | 2 |
| PT2 | 14 | 20225000 | 107225000 | 3191 | -0.025801915 | 0.982274454 | 2 |
| PT2 | 15 | 20125000 | 102400000 | 2936 | 0.012517373 | 1.008714131 | 2 |
| PT2 | 16 | 375000 | 33725000 | 909 | 0.364779529 | 1.287684832 | 4 |
| PT2 | 16 | 34400000 | 90075000 | 1518 | -0.541998967 | 0.686818608 | 0 |
| PT2 | 17 | 25000 | 18850000 | 690 | -0.599424561 | 0.66001716 | 0 |
| PT2 | 17 | 18850000 | 20075000 | 20 | 0.50316179 | 1.417316332 | 2 |
| PT2 | 17 | 20075000 | 80900000 | 1886 | 0.023035305 | 1.016095008 | 2 |
| PT2 | 18 | 125000 | 77875000 | 2854 | 0.004868854 | 1.003380534 | 2 |
| PT2 | 19 | 325000 | 58950000 | 1377 | 0.021530775 | 1.015035915 | 2 |
| PT2 | 20 | 175000 | 62650000 | 2076 | 0.032101342 | 1.022500354 | 2 |
| PT2 | 21 | 9550000 | 48000000 | 1192 | -0.077725812 | 0.947550138 | 2 |
| PT2 | 22 | 16200000 | 51125000 | 1208 | -0.382206247 | 0.767263353 | 1 |
| PT2 | X | 2975000 | 154675000 | 5425 | 0.082786741 | 1.059061773 | 2 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| PT3 | 1 | 725000 | 47425000 | 1737 | 0.005916971 | 1.004109754 | 2 |
| PT3 | 1 | 47425000 | 51725000 | 169 | 0.069368944 | 1.049257623 | 4 |
| PT3 | 1 | 51725000 | 144700000 | 2759 | -0.00958383 | 0.993379011 | 2 |
| PT3 | 1 | 144925000 | 249175000 | 3916 | 0.293646043 | 1.225734097 | 6 |
| PT3 | 2 | 100000 | 243025000 | 9268 | 0.001505186 | 1.00104386 | 2 |
| PT3 | 3 | 150000 | 167325000 | 6480 | -0.001663556 | 0.998847575 | 2 |
| PT3 | 3 | 167350000 | 197900000 | 1210 | 0.222918306 | 1.167092013 | 5 |
| PT3 | 4 | 100000 | 190900000 | 7380 | 8.56E-05 | 1.000059318 | 2 |
| PT3 | 5 | 125000 | 169675000 | 6540 | 0.024152093 | 1.01688187 | 2 |
| PT3 | 5 | 169675000 | 180725000 | 414 | 0.064064027 | 1.045406494 | 4 |
| PT3 | 6 | 275000 | 170925000 | 6421 | -0.011248744 | 0.992233283 | 2 |
| PT3 | 7 | 50000 | 159050000 | 5988 | 0.132346976 | 1.096075347 | 4 |
| PT3 | 8 | 225000 | 24500000 | 898 | -0.111790569 | 0.925438761 | 2 |
| PT3 | 8 | 24575000 | 146225000 | 4642 | 0.129885678 | 1.094206991 | 4 |
| PT3 | 9 | 150000 | 141100000 | 4309 | 0.004320952 | 1.002999545 | 2 |
| PT3 | 10 | 150000 | 135475000 | 5049 | 0.000643507 | 1.000446144 | 2 |
| PT3 | 11 | 200000 | 134825000 | 5112 | 0.007873794 | 1.005472619 | 2 |
| PT3 | 12 | 175000 | 133775000 | 5135 | -0.010547943 | 0.992715385 | 2 |
| PT3 | 13 | 19075000 | 115025000 | 3788 | 0.007432434 | 1.005165064 | 2 |
| PT3 | 14 | 19075000 | 107275000 | 3439 | 0.012400824 | 1.008632645 | 2 |
| PT3 | 15 | 20075000 | 95100000 | 2785 | 0.000885892 | 1.000614242 | 2 |
| PT3 | 15 | 95175000 | 102375000 | 287 | 0.029454071 | 1.020625838 | 4 |
| PT3 | 16 | 125000 | 33775000 | 1184 | 0.150906368 | 1.110266774 | 4 |
| PT3 | 16 | 33775000 | 90175000 | 1742 | -0.102223541 | 0.93159607 | 2 |
| PT3 | 17 | 450000 | 19625000 | 728 | -0.124867145 | 0.917088492 | 0 |
| PT3 | 17 | 19625000 | 20050000 | 17 | 0.048634197 | 1.0342853 | 2 |
| PT3 | 17 | 20050000 | 80975000 | 2166 | 0.001012326 | 1.000701937 | 2 |
| PT3 | 18 | 75000 | 77950000 | 2950 | 0.008914404 | 1.006198124 | 2 |
| PT3 | 19 | 300000 | 59050000 | 1931 | 0.010703585 | 1.00744675 | 2 |
| PT3 | 20 | 100000 | 62875000 | 2323 | 0.02528037 | 1.017677446 | 2 |
| PT3 | 21 | 9525000 | 48050000 | 1342 | -0.009918391 | 0.993148673 | 2 |
| PT3 | 22 | 16850000 | 51150000 | 1254 | 0.005572544 | 1.003870063 | 2 |
| PT3 | X | 2725000 | 154875000 | 5707 | 0.001628049 | 1.001129115 | 2 |
| InvF | 1 | 725000 | 121225000 | 4415 | -0.00916308 | 0.993668764 | 2 |
| InvF | 1 | 142800000 | 248600000 | 3475 | 0.980987894 | 1.973816529 | 6 |
| InvF | 2 | 275000 | 243000000 | 8833 | -0.007721638 | 0.994662066 | 2 |
| InvF | 3 | 100000 | 197900000 | 7368 | 0.005914116 | 1.004107767 | 2 |
| InvF | 4 | 125000 | 190925000 | 6782 | -0.019308137 | 0.986705778 | 2 |
| InvF | 5 | 375000 | 180650000 | 6489 | -0.029293353 | 0.979900145 | 2 |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| InvF | 6 | 350000 | 170825000 | 6109 | -0.001048078 | 0.999273792 | 2 |
| InvF | 7 | 25000 | 158900000 | 5769 | 0.414496826 | 1.332833739 | 4 |
| InvF | 8 | 200000 | 146225000 | 5340 | 0.040256559 | 1.028296676 | 2 |
| InvF | 9 | 400000 | 140875000 | 4120 | -0.003438104 | 0.997619725 | 2 |
| InvF | 10 | 275000 | 135325000 | 4863 | -0.001398004 | 0.999031447 | 2 |
| InvF | 11 | 275000 | 134525000 | 4841 | 0.007227545 | 1.005022322 | 2 |
| InvF | 12 | 200000 | 133725000 | 4873 | -0.020117821 | 0.986152165 | 2 |
| InvF | 13 | 19025000 | 115050000 | 3449 | 0.001265336 | 1.000877449 | 2 |
| InvF | 14 | 20125000 | 106950000 | 3242 | -0.015888879 | 0.989047093 | 2 |
| InvF | 15 | 20050000 | 102400000 | 3030 | 0.007485198 | 1.005201827 | 2 |
| InvF | 16 | 100000 | 33825000 | 1094 | 0.431834852 | 1.348948109 | 4 |
| InvF | 16 | 34325000 | 90050000 | 1441 | -0.586972811 | 0.665738353 | 0 |
| InvF | 17 | 25000 | 18850000 | 673 | -0.624439466 | 0.648671758 | 0 |
| InvF | 17 | 18850000 | 19875000 | 22 | 0.693392169 | 1.61708125 | 2 |
| InvF | 17 | 19875000 | 81000000 | 2082 | 0.023862387 | 1.016677692 | 2 |
| InvF | 18 | 350000 | 77825000 | 2764 | -0.017114793 | 0.988207018 | 2 |
| InvF | 19 | 250000 | 59050000 | 1816 | 0.021862144 | 1.015269082 | 2 |
| InvF | 20 | 325000 | 62850000 | 2266 | 0.018662353 | 1.013019786 | 2 |
| InvF | 21 | 9850000 | 48025000 | 1208 | -0.057428433 | 0.960975508 | 2 |
| InvF | 22 | 16875000 | 51075000 | 1243 | -0.478083866 | 0.71793052 | 0 |
| InvF | X | 2700000 | 154575000 | 5413 | 0.047438078 | 1.033428143 | 2 |

## Supplemental Table 7: Clinical information for the expanded sample set of 54 Chinese breast cancer patients.

| Sample_id | American Joint Committee on Cancer Tumor Stage Code | Lymph Node Stage American Joint Committee on Cancer Code | American Joint Committee on Cancer Metastasis Stage Code | Neoplasm Disease Stage American Joint Committee on Cancer Code | ER ihc | HER2 ihc score | HER2 fish | Positive Finding Lymph Node/Lymph Node(s) Examined |
|---|---|---|---|---|---|---|---|---|
| M1 | 3 | 3 | 2 | IV | 3 | 1+ | Negative | — |
| M2 | 2 | 3a | 1 | IV | 0 | 0 | Negative | — |
| M3 | 3 | 3 | 1 | IV | 0 | 1 | Negative | — |
| M4 | 2b | 3b | 1 | IV | 0 | 2 | Negative | — |
| M5 | 2 | 3 | 1 | IV | 3+ | 2+ | Negative | — |
| M6 | 2 | 3a | 1 | IV | 0 | 1+ | Negative | — |
| M7 | 2a | 2 | 1 | IV | 0 | 0 | Negative | — |
| M8 | 2 | 3a | 0 | IIIC | 0 | 0 | Negative | 35/36 |
| M9 | 1c | 3a | 0 | IIIC | 3 | 2 | Negative | 14/14 |
| M10 | 2 | 3 | 0 | IIIC | 3+ | 0 | Negative | 10/14 |
| M11 | 2 | 3a | 0 | IIIC | 0 | 3+ | Positive | 13/19 |
| M12 | 1c | 3a | 0 | IIIC | 2+ | 2+ | Negative | 13/22 |
| M13 | 2 | 3 | 0 | IIIC | 3 | 2 | Negative | 10/22 |
| M14 | 1c | 2 | 0 | IIIA | 0 | 0 | Negative | 6/19 |
| M15 | 3 | 2a | 0 | IIIA | 0 | 1 | Negative | 7/19 |
| M16 | 1c | 2a | 0 | IIIA | 3 | 2 | Negative | 5/19 |
| M17 | 1c | 2 | 0 | IIIA | 3+ | 2+ | Negative | 4/18 |
| M18 | 2 | 1 | 0 | IIB | 0 | 3+ | Positive | 3/15 |
| M19 | 2 | 1a | 0 | IIB | 0 | 3 | Positive | 3/15 |
| M20 | 1c | 1 | 0 | IIA | 3+ | 2+ | Negative | 2/20 |
| M21 | 2 | 1a | 0 | IIB | 1 | 2 | Positive | 2/25 |
| M22 | 2 | 2a | 0 | IIIA | 3 | 1 | Negative | 2/21 |
| M23 | 2 | 1 | 0 | IIB | 0 | 0 | Negative | 2/17 |
| M24 | 2 | 1 | 0 | IIB | 3+ | 1 | Negative | 1/4 |
| M25 | 1c | 1 | 1 | IV | 3+ | 0 | Negative | 1/21 |
| M26 | 1c | 1 | 0 | IIA | 2+ | 2+ | Negative | 1/20 |
| M27 | 1b | 1 | 1 | IV | 3+ | 2+ | Negative | 1/3 |
| M28 | 1c | 1a | 0 | IIA | 3 | 2 | Positive | 1/17 |
| L1 | 1b | 0 | 0 | IA | 0 | 2+ | Positive | 0/11 |
| L2 | is | 0 | 0 | 0 | 3+ | 1 | Negative | 0/17 |

|  |  |  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|---|---|
| L3 | 1c | 0 | 0 | IA | 0 | 1+ | Negative | 0/14 |
| L4 | 1c | 0 | 0 | IA | 0 | 1+ | Negative | 0/10 |
| L5 | 1 | 0 | 0 | IA | 3+ | 1 | Negative | 0/11 |
| L6 | 1c | 0 | 0 | IA | 3+ | 2+ | Negative | 0/18 |
| L7 | 1c | 0 | 0 | IA | 3+ | 2+ | Negative | 0/3 |
| L8 | 1c | 0 | 0 | IA | 2+ | 0 | Negative | 0/18 |
| L9 | 2 | 0 | 0 | IIA | 1+ | 1 | Negative | 0/7 |
| L10 | 1c | 0 | 0 | IA | 3+ | 0 | Negative | 0/16 |
| L11 | 2 | 0 | 0 | IIA | — | — | — | 0/22 |
| L12 | 0 | 0 | 0 | 0 | 1 | 3+ | Positive | 0/12 |
| L13 | 2 | 0 | 0 | IIA | 3+ | 0 | Negative | 0/5 |
| L14 | 1c | 0 | 0 | IA | 1+ | 2+ | Negative | 0/22 |
| L15 | 1c | 0 | 0 | IA | 2+ | 2+ | Negative | 0/5 |
| L16 | is | 0 | 0 | 0 | 3 | 1 | Negative | 0/12 |
| L17 | is | 0 | 0 | 0 | 0 | 2+ | Positive | 0/18 |
| L18 | 1c | 0 | 0 | IA | 0 | 3+ | Positive | 0/17 |
| L19 | 2 | 0 | 0 | IIA | 0 | 3+ | Positive | 0/16 |
| L20 | 1c | 0 | 0 | IA | 0 | 3+ | Positive | 0/7 |
| L21 | 1c | 0 | 0 | IA | 0 | 2+ | Negative | 0/29 |
| L22 | 2 | 0 | 0 | IIA | 0 | 1 | Negative | 0/17 |
| L23 | 2 | 0 | 0 | IIA | 0 | 0 | Negative | 0/12 |
| L24 | 1b | 0 | 0 | IA | 3 | 3 | Positive | 0/2 |
| L25 | 1c | 1 | 1 | IV | 3 | 0 | Negative | 0/16 |
| L26 | 2 | 0 | 0 | IIA | 3+ | 1+ | Negative | 0/15 |

## Supplemental Table 8: Target genes for capture sequencing in the 54 Chinese sample set.

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| ABL1 | BRIP1 | CTLA4 | FANCG | H3F3A | KRAS | NCOA1 | PMS2 | RUNX1 | TBX3 |
| ABL2 | BTG1 | CTNNA1 | FANCI | H3F3C | LCK | NCOA2 | PNRC1 | RUNX1T1 | TEK |
| ACVR1B | BTK | CTNNB1 | FANCL | HCK | LIMK1 | NCOR1 | POLQ | RXRA | TERT |
| ACVR2A | C11orf30 | CUL4A | FANCM | HDAC1 | LRRK2 | NEK11 | PPP2R1A | RXRB | TET2 |
| AJUBA | C1QA | CUL4B | FAT3 | HDAC2 | LYN | NF1 | PRDM1 | RXRG | TFG |
| AKT1 | C1R | CYLD | FBXW7 | HDAC3 | MALAT1 | NF2 | PRKAA1 | SDHAF2 | TGFBR2 |
| AKT2 | C1S | CYP17A1 | FCGR1A | HDAC4 | MAP2K1 | NFE2L2 | PRKAR1A | SDHB | TIPARP |
| AKT3 | CARD11 | DAXX | FCGR2A | HDAC6 | MAP2K2 | NFE2L3 | PRKCA | SDHC | TLR4 |
| ALK | CASP8 | DDR1 | FCGR2B | HDAC8 | MAP2K4 | NFKBIA | PRKCB | SDHD | TMEM127 |
| ALOX12B | CBFB | DDR2 | FCGR2C | HGF | MAP3K1 | NKX2-1 | PRKCG | SEMA3A | TNFAIP3 |
| ANGPT1 | CBL | DIS3 | FCGR3A | HIF1A | MAP3K13 | NKX3-1 | PRKDC | SEMA3E | TNFRSF14 |
| ANGPT2 | CBLB | DNMT1 | FCGR3B | HIST1H1C | MAPK1 | NOTCH1 | PRSS8 | SETBP1 | TNFRSF8 |
| APC | CBR1 | DNMT3A | FGF10 | HIST1H2BD | MAPK3 | NOTCH2 | PSMB1 | SETD2 | TNFSF11 |
| APCDD1 | CCND1 | DOT1L | FGF12 | HIST1H3B | MAPK8 | NOTCH3 | PSMB2 | SF1 | TNFSF13B |
| AR | CCND2 | DUSP6 | FGF14 | HNF1A | MAPK8IP1 | NOTCH4 | PSMB5 | SF3B1 | TOP1 |
| ARAF | CCND3 | EDNRA | FGF19 | HRAS | MAX | NPM1 | PTCH1 | SH2B3 | TOP2A |
| ARFRP1 | CCNE1 | EGFR | FGF23 | HRH2 | MC1R | NR3C1 | PTCH2 | SIN3A | TOP2B |
| ARHGAP35 | CD79A | EGR3 | FGF3 | HSD17B3 | MCL1 | NRAS | PTEN | SLAMF7 | TP53 |
| ARID1A | CD79B | EIF4A2 | FGF4 | HSD3B2 | MDM2 | NSD1 | PTP4A3 | SLC4A1 | TRAF7 |
| ARID1B | CDC25C | ELAC2 | FGF6 | HSP90AA1 | MDM4 | NTRK1 | PTPN11 | SLIT2 | TSC1 |
| ARID2 | CDC42 | ELF3 | FGF7 | HSPA4 | MECOM | NTRK2 | PTPRD | SMAD2 | TSC2 |
| ARID5B | CDC73 | EML4 | FGFR1 | IDH1 | MED12 | NTRK3 | RAC1 | SMAD3 | TSHR |
| ASXL1 | CDH1 | EP300 | FGFR2 | IDH2 | MEF2B | NUP93 | RAC2 | SMAD4 | TSHZ2 |
| ATM | CDK12 | EPCAM | FGFR3 | IFNAR1 | MEN1 | PAK3 | RAD21 | SMARCA1 | TSHZ3 |
| ATR | CDK2 | EPHA2 | FGFR4 | IFNAR2 | MET | PAK7 | RAD50 | SMARCA4 | TUBA1A |
| ATRX | CDK4 | EPHA3 | FH | IGF1 | MIR142 | PALB2 | RAD51 | SMARCB1 | TUBB |
| AURKA | CDK6 | EPHA5 | FLCN | IGF1R | MITF | PARP1 | RAD51B | SMARCD1 | TUBD1 |
| AURKB | CDK8 | EPHB1 | FLT1 | IGF2 | MLH1 | PARP2 | RAD51C | SMC1A | TUBE1 |
| AXIN1 | CDKN1A | EPHB2 | FLT3 | IKBKB | MLH3 | PARP3 | RAD51D | SMC3 | TUBG1 |
| AXIN2 | CDKN1B | EPHB6 | FLT4 | IKBKE | MLL | PARP4 | RAD52 | SMO | TYR |
| AXL | CDKN2A | EPPK1 | FNTA | IKZF1 | MLL2 | PAX5 | RAD54L | SOCS1 | U2AF1 |
| B2M | CDKN2B | ERBB2 | FOXA1 | IL7R | MLL3 | PBRM1 | RAF1 | SOX10 | USP9X |
| B4GALT3 | CDKN2C | ERBB3 | FOXA2 | INHBA | MLL4 | PCBP1 | RARA | SOX17 | VEGFA |
| BACH1 | CDX2 | ERBB4 | FOXL2 | IRF4 | MPL | PCM1 | RARB | SOX2 | VEGFB |
| BAK1 | CEBPA | ERCC2 | FPGS | IRS2 | MRE11A | PDGFRA | RARG | SOX9 | VEZF1 |
| BAP1 | CFLAR | ERCC3 | FUBP1 | ITGB2 | MS4A1 | PDGFRB | RB1 | SPEN | VHL |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| BARD1 | CHD1 | ERG | FYN | JAK1 | MSH2 | PDK1 | REL | SPOP | WHSC1L1 |
| BCL2 | CHD2 | ESR1 | GAB2 | JAK2 | MSH3 | PHF6 | RET | SPRY4 | WISP3 |
| BCL2A1 | CHD4 | ETV1 | GATA1 | JAK3 | MSH4 | PIGF | RHEB | SRC | WT1 |
| BCL2L1 | CHEK1 | ETV6 | GATA2 | JUN | MSH5 | PIK3C2A | RICTOR | SRD5A2 | WWP1 |
| BCL2L11 | CHEK2 | EWSR1 | GATA3 | KAT6A | MSH6 | PIK3C2B | RNASEL | SRSF2 | XIAP |
| BCL2L2 | CHUK | EXT1 | GID4 | KDM5A | MSR1 | PIK3C2G | RNF43 | SSTR2 | XPA |
| BCL6 | CIC | EXT2 | GNA11 | KDM5C | MTOR | PIK3C3 | ROBO1 | STAG2 | XPC |
| BCOR | CRBN | EZH2 | GNA13 | KDM6A | MUC1 | PIK3CA | ROBO2 | STAT4 | XPO1 |
| BCORL1 | CREBBP | FAM123B | GNAQ | KDR | MUTYH | PIK3CB | ROS1 | STAT5B | XRCC3 |
| BCR | CRIPAK | FAM46C | GNAS | KEAP1 | MYC | PIK3CG | RPA1 | STK11 | YES1 |
| BLM | CRKL | FANCA | GNRHR | KIF1B | MYCL1 | PIK3R1 | RPL22 | SUFU | ZNF217 |
| BMPR1A | CRLF2 | FANCC | GPR124 | KIF5B | MYCN | PIK3R2 | RPL5 | SUZ12 | ZNF703 |
| BRAF | CROT | FANCD2 | GRIN2A | KIT | MYD88 | PLK1 | RPS14 | SYK | ZRSR2 |
| BRCA1 | CSF1R | FANCE | GRM3 | KLF4 | NAV3 | PML | RPS6KB1 | TAF1 | |
| BRCA2 | CTCF | FANCF | GSK3B | KLHL6 | NBN | PMS1 | RPTOR | TBL1XR1 | |

## Supplemental Table 9: Somatic non-synonymous mutations detected in the sample set of 54 Chinese breast cancers.

| Sample_id | chr | coordinate | ref | alter | normal_reads1 | Gene | ExIn_ID | Function | pHGVS |
|---|---|---|---|---|---|---|---|---|---|
| L1 | chr17 | 7577570 | C | T | 286 | TP53 | EX7 | missense | p.M237I |
| L1 | chr3 | 178952085 | A | G | 417 | PIK3CA | EX21E | missense | p.H1047R |
| L1 | chr3 | 52610695 | G | A | 161 | PBRM1 | EX23 | nonsense | p.R1160* |
| L10 | chr10 | 63661474 | G | T | 267 | ARID5B | EX1 | missense | p.E2D |
| L10 | chr15 | 99478132 | G | A | 279 | IGF1R | EX16 | missense | p.M1012I |
| L10 | chr3 | 178952077 | T | A | 385 | PIK3CA | EX21E | missense | p.N1044K |
| L10 | chr7 | 86986843 | A | G | 112 | CROT | EX4 | missense | p.I46V |
| L11 | chr1 | 118166353 | T | +G | 562 | FAM46C | EX2E | frameshift | p.E289Gfs*10 |
| L11 | chr17 | 7579356 | G | -ACGGAA | 217 | TP53 | EX4 | cds-del | p.F109_R110del |
| L11 | chr3 | 178952085 | A | G | 274 | PIK3CA | EX21E | missense | p.H1047R |
| L11 | chr3 | 30713294 | C | T | 402 | TGFBR2 | EX5 | missense | p.R232W |
| L12 | chr12 | 4385368 | C | G | 242 | CCND2 | EX2 | missense | p.I131M |
| L12 | chr19 | 15276812 | G | C | 421 | NOTCH3 | EX30 | missense | p.A1818G |
| L12 | chr5 | 67591107 | A | -ACCAGACCTTATCCAGCTGAG | 624 | PIK3R1 | EX13 | cds-del | p.P568_R574del |
| L12 | chr8 | 144942032 | C | T | 531 | EPPK1 | EX1E | nonsense | p.W1797* |
| L12 | chrX | 129148815 | C | G | 513 | BCORL1 | EX3 | missense | p.I689M |
| L13 | chr1 | 156846246 | C | A | 735 | NTRK1 | EX14 | missense | p.L563M |
| L13 | chr11 | 108159727 | C | T | 461 | ATM | EX28 | missense | p.P1378L |
| L13 | chr17 | 7574018 | G | A | 444 | TP53 | EX10 | missense | p.R337C |
| L13 | chr3 | 178936082 | G | A | 123 | PIK3CA | EX10 | missense | p.E542K |
| L13 | chr5 | 79950736 | C | G | 49 | MSH3 | EX1 | missense | p.P64A |
| L14 | chr10 | 89711893 | C | T | 729 | PTEN | EX6 | nonsense | p.Q171* |
| L14 | chr17 | 70117588 | C | T | 209 | SOX9 | EX1 | missense | p.S19F |
| L14 | chr17 | 7579337 | C | +CAGAATG | 154 | TP53 | EX4 | frameshift | p.G117Afs*34 |
| L14 | chr8 | 55372120 | G | T | 47 | SOX17 | EX2E | missense | p.M270I |
| L15 | chr1 | 10434945 | C | A | 293 | KIF1B | EX45 | missense | p.F1664L |
| L15 | chr1 | 11303223 | G | C | 291 | MTOR | EX9 | missense | p.R454G |
| L15 | chr1 | 193218898 | G | A | 309 | CDC73 | EX16 | missense | p.D486N |
| L15 | chr1 | 206649604 | G | A | 295 | IKBKE | EX6 | missense | p.E147K |
| L15 | chr1 | 32745484 | C | G | 244 | LCK | EX11 | missense | p.H362D |
| L15 | chr10 | 61665989 | G | A | 119 | CCDC6 | EX1 | missense | p.S65L |
| L15 | chr11 | 92577781 | G | A | 314 | FAT3 | EX18 | missense | p.E3750K |
| L15 | chr12 | 121416713 | G | C | 237 | HNF1A | EX1 | missense | p.E48Q |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| L15 | chr12 | 40722167 | G | A | 196 | LRRK2 | EX39 | missense | p.G1888S |
| L15 | chr13 | 32890618 | G | C | 222 | BRCA2 | EX2 | missense | p.E7D |
| L15 | chr13 | 32893253 | C | T | 196 | BRCA2 | EX3 | missense | p.S36F |
| L15 | chr14 | 75513863 | G | C | 350 | MLH3 | EX2 | missense | p.F832L |
| L15 | chr15 | 75685087 | C | G | 263 | SIN3A | EX15 | missense | p.D783H |
| L15 | chr15 | 91295158 | C | G | 192 | BLM | EX4 | missense | p.S314C |
| L15 | chr16 | 10031954 | G | A | 326 | GRIN2A | EX4 | missense | p.A290V |
| L15 | chr16 | 23700564 | C | G | 183 | PLK1 | EX8 | missense | p.Q426E |
| L15 | chr16 | 24196431 | G | C | 214 | PRKCB | IVS13 | splice-3 | . |
| L15 | chr16 | 50827525 | G | A | 280 | CYLD | EX18 | missense | p.D807N |
| L15 | chr17 | 29483067 | C | G | 256 | NF1 | EX2 | missense | p.L43V |
| L15 | chr17 | 38546414 | G | A | 193 | TOP2A | EX34 | nonsense | p.Q1424* |
| L15 | chr17 | 47679339 | C | T | 266 | SPOP | EX11 | missense | p.E290K |
| L15 | chr17 | 47696733 | T | A | 219 | SPOP | EX6 | missense | p.N72I |
| L15 | chr17 | 56440659 | C | G | 260 | RNF43 | EX5 | missense | p.E187Q |
| L15 | chr17 | 7574034 | C | T | 196 | TP53 | IVS9 | splice-3 | . |
| L15 | chr18 | 45374893 | G | T | 316 | SMAD2 | EX7 | missense | p.S287Y |
| L15 | chr19 | 2180755 | G | A | 126 | DOT1L | EX2 | missense | p.R42Q |
| L15 | chr19 | 42795012 | G | A | 234 | CIC | EX10 | missense | p.E698K |
| L15 | chr2 | 202025343 | C | G | 331 | CFLAR | EX9 | missense | p.H328D |
| L15 | chr2 | 204736158 | C | T | 261 | CTLA4 | EX3 | missense | p.S172L |
| L15 | chr2 | 29436902 | G | A | 179 | ALK | EX24 | missense | p.R1231W |
| L15 | chr20 | 39742762 | C | G | 270 | TOP1 | EX15 | missense | p.I535M |
| L15 | chr20 | 54945597 | C | G | 198 | AURKA | EX10 | missense | p.G325R |
| L15 | chr22 | 41536260 | G | C | 242 | EP300 | EX9 | missense | p.R626P |
| L15 | chr3 | 119263418 | C | A | 270 | CD80 | EX3 | nonsense | p.E133* |
| L15 | chr3 | 178952085 | A | G | 284 | PIK3CA | EX21E | missense | p.H1047R |
| L15 | chr3 | 185161345 | G | C | 284 | MAP3K13 | EX4 | missense | p.D258H |
| L15 | chr3 | 47161916 | G | C | 250 | SETD2 | EX3 | missense | p.L1404V |
| L15 | chr3 | 52442080 | G | A | 214 | BAP1 | EX5 | missense | p.S90F |
| L15 | chr4 | 20555546 | G | C | 264 | SLIT2 | EX26 | missense | p.D894H |
| L15 | chr5 | 112179254 | G | C | 347 | APC | EX17E | missense | p.E2655Q |
| L15 | chr5 | 98212189 | G | C | 272 | CHD1 | EX23 | nonsense | p.S1104* |
| L15 | chr6 | 106552889 | G | C | 303 | PRDM1 | EX5 | missense | p.R285T |
| L15 | chr6 | 18130928 | C | G | 137 | TPMT | EX9E | missense | p.E237Q |
| L15 | chr6 | 31726328 | G | C | 193 | MSH5 | EX14 | missense | p.D383H |
| L15 | chr6 | 33288238 | C | G | 246 | DAXX | EX4 | missense | p.E402D |
| L15 | chr7 | 87183103 | C | G | 263 | ABCB1 | EX10 | missense | p.E325Q |
| L15 | chr7 | 98478824 | G | C | 203 | TRRAP | EX2 | missense | p.L17F |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| L15 | chr8 | 119123251 | G | C | 242 | EXT1 | EX1 | nonsense | p.S12* |
| L15 | chr8 | 55372155 | C | G | 92 | SOX17 | EX2E | missense | p.S282W |
| L15 | chrX | 129189975 | C | A | 262 | BCORL1 | EX12E | missense | p.T1667N |
| L16 | chr14 | 105246551 | C | T | 494 | AKT1 | EX3 | missense | p.E17K |
| L16 | chr2 | 198266834 | T | C | 616 | SF3B1 | EX15 | missense | p.K700E |
| L17 | chr11 | 534286 | C | G | 224 | HRAS | EX2 | missense | p.G13R |
| L17 | chr6 | 31726370 | C | T | 255 | MSH5 | EX14 | missense | p.P397S |
| L18 | chr6 | 112405450 | C | G | 152 | TUBE1 | IVS3 | splice-3 | . |
| L19 | chr17 | 37627863 | C | T | 108 | CDK12 | EX2 | missense | p.S593F |
| L19 | chr17 | 7577588 | G | -GTGGTACAGTCAGAGCCAAC | 99 | TP53 | EX7 | frameshift | p.V225Hfs*8 |
| L19 | chr3 | 178952085 | A | G | 117 | PIK3CA | EX21E | missense | p.H1047R |
| L2 | chr2 | 198266834 | T | C | 360 | SF3B1 | EX15 | missense | p.K700E |
| L2 | chr5 | 98224896 | C | T | 261 | CHD1 | EX15 | missense | p.G743S |
| L20 | chr10 | 8115865 | G | -CCACATCT | 834 | GATA3 | EX6E | frameshift | p.H406Afs*99 |
| L21 | chr1 | 161143840 | C | T | 82 | B4GALT3 | IVS4 | splice-3 | . |
| L21 | chr17 | 7579453 | T | +C | 118 | TP53 | EX4 | frameshift | p.A79Sfs*70 |
| L22 | chr17 | 7578212 | G | A | 200 | TP53 | EX6 | nonsense | p.R213* |
| L22 | chr3 | 168810805 | A | C | 180 | MECOM | EX14 | missense | p.I1035M |
| L22 | chr8 | 117864799 | C | T | 153 | RAD21 | EX10 | missense | p.R437H |
| L23 | chr13 | 110437323 | G | A | 13 | IRS2 | EX1 | missense | p.R360W |
| L23 | chr16 | 2137891 | G | A | 189 | TSC2 | EX39 | missense | p.V1673I |
| L23 | chr17 | 7577548 | C | T | 241 | TP53 | EX7 | missense | p.G245S |
| L23 | chr7 | 87178749 | C | T | 199 | ABCB1 | EX15 | missense | p.R547H |
| L23 | chr8 | 48746814 | C | G | 307 | PRKDC | EX59 | missense | p.D2699H |
| L23 | chr9 | 36846874 | G | C | 328 | PAX5 | EX9 | missense | p.N355K |
| L24 | chr12 | 52345593 | C | +G | 31 | ACVR1B | EX1 | frameshift | p.S24Vfs*24 |
| L24 | chr14 | 45644457 | G | A | 468 | FANCM | EX14 | missense | p.E834K |
| L24 | chr15 | 75682063 | C | G | 527 | SIN3A | EX16 | missense | p.R984T |
| L24 | chr17 | 15965539 | G | A | 468 | NCOR1 | EX36 | missense | p.S1756F |
| L24 | chr3 | 178952085 | A | G | 409 | PIK3CA | EX21E | missense | p.H1047R |
| L25 | chr12 | 12871250 | T | C | 491 | CDKN1B | IVS1 | splice-5 | . |
| L25 | chr3 | 178936091 | G | A | 491 | PIK3CA | EX10 | missense | p.E545K |
| L25 | chr5 | 1255456 | C | T | 635 | TERT | EX14 | missense | p.V1035I |
| L3 | chr17 | 7577514 | G | -T | 128 | TP53 | EX7 | frameshift | p.T256Hfs*89 |
| L3 | chr2 | 29917773 | A | T | 167 | ALK | EX3 | missense | p.S299T |
| L3 | chr7 | 151848649 | C | A | 156 | MLL3 | EX50 | missense | p.D4182Y |
| L4 | chr1 | 161143489 | C | G | 81 | B4GALT3 | EX6 | missense | p.V237L |
| L4 | chr17 | 16097825 | T | G | 50 | NCOR1 | EX2 | missense | p.Y20S |
| L4 | chr17 | 41276061 | A | C | 38 | BRCA1 | EX2 | missense | p.M18R |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| L4 | chr17 | 7578212 | G | A | 67 | TP53 | EX6 | nonsense | p.R213* |
| L4 | chr9 | 35078206 | C | G | 83 | FANCG | EX4 | missense | p.A148P |
| L4 | chrX | 76939599 | G | C | 115 | ATRX | EX9 | missense | p.I383M |
| L5 | chr10 | 89717697 | T | C | 271 | PTEN | EX7 | missense | p.F241S |
| L5 | chr2 | 128038108 | T | C | 234 | ERCC3 | EX9 | missense | p.N481S |
| L5 | chr2 | 198266834 | T | C | 247 | SF3B1 | EX15 | missense | p.K700E |
| L5 | chr7 | 151891142 | A | C | 322 | MLL3 | EX31 | missense | p.L1538V |
| L6 | chr10 | 8115907 | C | +GTGGT | 223 | GATA3 | EX6E | frameshift | p.P420Wfs*58 |
| L6 | chr12 | 92537902 | C | G | 286 | BTG1 | EX2E | missense | p.R157T |
| L6 | chr3 | 178936091 | G | A | 284 | PIK3CA | EX10 | missense | p.E545K |
| L6 | chr9 | 139438555 | C | G | 162 | NOTCH1 | IVS1 | splice-3 | . |
| L7 | chr1 | 27023489 | C | T | 69 | ARID1A | EX1 | nonsense | p.Q199* |
| L7 | chr2 | 198266834 | T | C | 340 | SF3B1 | EX15 | missense | p.K700E |
| L7 | chr6 | 26032031 | C | A | 227 | HIST1H3B | EX1E | missense | p.Q86H |
| L8 | chr10 | 89653846 | C | G | 79 | PTEN | EX2 | missense | p.N48K |
| L8 | chr17 | 29654534 | C | -CAAGTAACTT | 443 | NF1 | EX38 | frameshift | p.V1764Qfs*7 |
| L8 | chr17 | 7578416 | C | A | 453 | TP53 | EX5 | missense | p.V172F |
| L8 | chr3 | 178936094 | C | G | 87 | PIK3CA | EX10 | missense | p.Q546E |
| L9 | chr1 | 22987373 | C | T | 127 | C1QB | EX3E | missense | p.P86S |
| L9 | chr10 | 6060023 | G | A | 85 | IL2RA | EX7 | missense | p.R263W |
| L9 | chr12 | 50027868 | C | G | 112 | PRPF40B | EX10 | missense | p.P269A |
| L9 | chr13 | 25016762 | G | A | 52 | PARP4 | EX29 | missense | p.T1170I |
| L9 | chr4 | 148457110 | C | G | 159 | EDNRA | EX5 | missense | p.L277V |
| M1 | chr11 | 119149280 | G | A | 321 | CBL | EX9 | missense | p.V430M |
| M1 | chr17 | 29585447 | C | T | 267 | NF1 | EX32 | missense | p.S1420L |
| M1 | chr3 | 178916937 | T | -GAA | 424 | PIK3CA | EX2 | cds-del | p.E109[2>1] |
| M10 | chr17 | 7577100 | T | C | 141 | TP53 | EX8 | missense | p.R280G |
| M10 | chr22 | 24175842 | C | T | 135 | SMARCB1 | EX8 | missense | p.T357I |
| M12 | chr10 | 8115718 | C | +T | 229 | GATA3 | EX6E | frameshift | p.M357Yfs*15 |
| M12 | chr17 | 12028681 | T | A | 277 | MAP2K4 | EX8 | missense | p.I295N |
| M12 | chr20 | 30310018 | A | G | 225 | BCL2L1 | EX2 | missense | p.S2P |
| M12 | chr3 | 25622139 | C | T | 235 | RARB | EX5 | missense | p.R238C |
| M13 | chr3 | 178952085 | A | T | 576 | PIK3CA | EX21E | missense | p.H1047L |
| M13 | chr7 | 151864230 | C | A | 387 | MLL3 | IVS42 | splice-5 | . |
| M13 | chrX | 70678212 | C | T | 485 | TAF1 | EX35 | missense | p.T1707I |
| M14 | chr1 | 16261368 | C | T | 527 | SPEN | EX11 | missense | p.A2878V |
| M14 | chr1 | 204402496 | C | T | 449 | PIK3C2B | EX27 | missense | p.D1301N |
| M14 | chr10 | 104592775 | A | G | 448 | CYP17A1 | EX5 | missense | p.L315P |
| M14 | chr11 | 119167693 | T | C | 496 | CBL | EX13 | missense | p.M701T |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| M14 | chr11 | 64534458 | G | C | 166 | SF1 | EX12 | missense | p.P624R |
| M14 | chr12 | 6692073 | G | A | 473 | CHD4 | EX28 | missense | p.R1393W |
| M14 | chr17 | 7578382 | G | C | 419 | TP53 | EX5 | nonsense | p.S183* |
| M14 | chr9 | 32635534 | A | -CGGTAGCTGCTGCCCTCAGCAGCAAAT | 222 | TAF1L | EX1E | cds-del | p.D6_T14del |
| M15 | chr10 | 43622046 | C | -ACTCCATCTG | 395 | RET | EX19 | frameshift | p.S1024* |
| M15 | chr11 | 118352779 | G | C | 515 | MLL | EX7 | missense | p.K1328N |
| M15 | chr11 | 92531574 | C | T | 463 | FAT3 | EX9 | nonsense | p.R1799* |
| M15 | chr15 | 93528887 | G | A | 439 | CHD2 | EX26 | missense | p.D1133N |
| M15 | chr17 | 16097870 | C | A | 133 | NCOR1 | EX2 | missense | p.G5V |
| M15 | chr17 | 7578370 | C | A | 327 | TP53 | IVS5 | splice-5 | . |
| M15 | chr3 | 181430489 | G | C | 329 | SOX2 | EX1E | missense | p.R114P |
| M16 | chr12 | 115118880 | T | G | 466 | TBX3 | EX2 | missense | p.D154A |
| M16 | chr14 | 105246551 | C | T | 234 | AKT1 | EX3 | missense | p.E17K |
| M16 | chr18 | 45368211 | G | A | 263 | SMAD2 | EX10E | missense | p.S434L |
| M16 | chr2 | 198266834 | T | C | 422 | SF3B1 | EX15 | missense | p.K700E |
| M16 | chr21 | 36206878 | C | -ATCT | 235 | RUNX1 | EX7 | frameshift | p.D211Ifs*25 |
| M16 | chr7 | 152009005 | A | -C | 208 | MLL3 | EX5 | frameshift | p.V206Yfs*4 |
| M17 | chr1 | 156844747 | C | T | 594 | NTRK1 | EX11 | missense | p.T434M |
| M17 | chr12 | 7177230 | A | C | 429 | C1S | EX12E | missense | p.N448H |
| M17 | chr17 | 41243479 | C | G | 416 | BRCA1 | EX10 | missense | p.E1357Q |
| M17 | chr17 | 41243515 | C | T | 515 | BRCA1 | EX10 | missense | p.E1345K |
| M17 | chr17 | 41244049 | C | T | 528 | BRCA1 | EX10 | missense | p.E1167K |
| M17 | chr17 | 41244843 | T | -C | 618 | BRCA1 | EX10 | frameshift | p.E902Nfs*98 |
| M17 | chr19 | 18272827 | G | T | 187 | PIK3R2 | EX7 | missense | p.Q289H |
| M17 | chr3 | 178936091 | G | A | 84 | PIK3CA | EX10 | missense | p.E545K |
| M17 | chr3 | 78649381 | G | C | 541 | ROBO1 | EX30 | nonsense | p.S1608* |
| M17 | chrX | 41077649 | T | -CGTCACAGCAAGAATGTA | 501 | USP9X | EX37 | cds-del | p.H2080_R2085del |
| M18 | chr17 | 7577142 | C | G | 345 | TP53 | EX8 | missense | p.G266R |
| M2 | chr14 | 23548798 | C | A | 852 | ACIN1 | EX6 | missense | p.R640S |
| M2 | chr17 | 7577570 | C | T | 304 | TP53 | EX7 | missense | p.M237I |
| M2 | chr2 | 142238072 | A | C | 603 | LRP1B | EX3 | missense | p.L79W |
| M20 | chr2 | 198267361 | T | C | 369 | SF3B1 | EX14 | missense | p.K666E |
| M20 | chr3 | 178952085 | A | G | 411 | PIK3CA | EX21E | missense | p.H1047R |
| M21 | chr1 | 156452282 | A | -CTTGAGCAG | 53 | MEF2D | EX3 | cds-del | p.L66_K68del |
| M21 | chr1 | 16259377 | G | C | 487 | SPEN | EX11 | missense | p.E2214D |
| M21 | chr1 | 65332747 | A | T | 611 | JAK1 | EX7 | missense | p.H264Q |
| M21 | chr10 | 112350871 | C | T | 471 | SMC3 | EX17 | missense | p.T598I |
| M21 | chr12 | 56481621 | T | G | 515 | ERBB3 | EX6 | missense | p.F219C |
| M21 | chr13 | 28913428 | C | G | 522 | FLT1 | EX17 | missense | p.E789Q |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| M21 | chr14 | 45657033 | G | C | 377 | FANCM | EX19 | missense | p.M1574I |
| M21 | chr15 | 90631963 | C | A | 424 | IDH2 | EX4 | missense | p.K130N |
| M21 | chr16 | 50783634 | G | T | 520 | CYLD | EX4 | nonsense | p.E9* |
| M21 | chr17 | 7577538 | C | T | 378 | TP53 | EX7 | missense | p.R248Q |
| M21 | chr19 | 15291635 | A | T | 347 | NOTCH3 | EX19 | missense | p.L1000Q |
| M21 | chr2 | 212812242 | C | G | 482 | ERBB4 | EX3 | missense | p.E112Q |
| M21 | chr2 | 47630335 | C | A | 170 | MSH2 | EX1 | missense | p.A2E |
| M21 | chr2 | 48026114 | C | G | 667 | MSH6 | EX4 | nonsense | p.S331* |
| M21 | chr2 | 61147244 | G | A | 385 | REL | EX8 | missense | p.V308I |
| M21 | chr21 | 44837433 | C | G | 185 | SIK1 | EX13 | missense | p.E656Q |
| M21 | chr3 | 47058721 | C | A | 482 | SETD2 | EX21E | missense | p.K2519N |
| M21 | chr3 | 89391064 | G | C | 580 | EPHA3 | EX5 | missense | p.S377T |
| M21 | chr7 | 86415644 | G | C | 462 | GRM3 | EX3 | missense | p.S179T |
| M21 | chr9 | 35076457 | G | C | 503 | FANCG | EX8 | missense | p.L350V |
| M21 | chrX | 76855036 | C | A | 383 | ATRX | EX25 | missense | p.G1934W |
| M22 | chr3 | 10114944 | A | C | 89 | FANCD2 | EX28 | missense | p.K871N |
| M22 | chr3 | 178921548 | G | A | 147 | PIK3CA | EX5 | missense | p.V344M |
| M22 | chr3 | 178936094 | C | A | 342 | PIK3CA | EX10 | missense | p.Q546K |
| M23 | chr17 | 7577120 | C | T | 469 | TP53 | EX8 | missense | p.R273H |
| M24 | chr11 | 119077276 | C | T | 84 | CBL | EX1 | missense | p.T50M |
| M24 | chr11 | 77991847 | C | T | 610 | GAB2 | EX2 | missense | p.R59Q |
| M24 | chr17 | 29685515 | C | +T | 436 | NF1 | EX55 | frameshift | p.K2664* |
| M24 | chr17 | 7578394 | T | C | 500 | TP53 | EX5 | missense | p.H179R |
| M24 | chr2 | 48936128 | C | A | 452 | LHCGR | EX8 | missense | p.M213I |
| M24 | chr3 | 178936091 | G | A | 98 | PIK3CA | EX10 | missense | p.E545K |
| M24 | chrX | 63413091 | C | A | 523 | FAM123B | EX2E | nonsense | p.E26* |
| M27 | chr19 | 42792017 | G | A | 198 | CIC | EX6 | missense | p.R274Q |
| M27 | chr3 | 178921548 | G | A | 218 | PIK3CA | EX5 | missense | p.V344M |
| M27 | chr3 | 178951964 | G | C | 247 | PIK3CA | EX21E | missense | p.G1007R |
| M27 | chr3 | 178952085 | A | G | 272 | PIK3CA | EX21E | missense | p.H1047R |
| M27 | chr5 | 149514369 | G | -T | 350 | PDGFRB | EX4 | frameshift | p.T192Pfs*27 |
| M27 | chr5 | 56170910 | A | T | 251 | MAP3K1 | EX10 | nonsense | p.R580* |
| M27 | chr5 | 56176630 | G | A | 149 | MAP3K1 | IVS12 | splice-5 | |
| M28 | chr3 | 178921553 | T | A | 430 | PIK3CA | EX5 | missense | p.N345K |
| M28 | chr3 | 178921559 | T | A | 411 | PIK3CA | EX5 | missense | p.N347K |
| M28 | chr3 | 178952139 | A | T | 349 | PIK3CA | EX21E | missense | p.H1065L |
| M3 | chr17 | 7577121 | G | A | 243 | TP53 | EX8 | missense | p.R273C |
| M3 | chr21 | 36206761 | C | T | 172 | RUNX1 | EX7 | missense | p.A251T |
| M4 | chr1 | 32741208 | G | C | 73 | LCK | EX6 | missense | p.R139P |

| | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| M4 | chr13 | 28893613 | G | C | 214 | FLT1 | EX24 | missense | p.T1078S |
| M4 | chr17 | 7577058 | C | A | 198 | TP53 | EX8 | nonsense | p.E294* |
| M4 | chr7 | 98501118 | C | G | 213 | TRRAP | EX12 | missense | p.I338M |
| M4 | chr8 | 87460692 | G | C | 252 | WWP1 | EX20 | missense | p.L741F |
| M5 | chr10 | 8115842 | C | +T | 317 | GATA3 | EX6E | frameshift | p.S398Ffs*110 |
| M5 | chr2 | 198266834 | T | C | 329 | SF3B1 | EX15 | missense | p.K700E |
| M5 | chr3 | 178936091 | G | A | 284 | PIK3CA | EX10 | missense | p.E545K |
| M6 | chr17 | 7578556 | T | C | 149 | TP53 | IVS4 | splice-3 | . |
| M7 | chr1 | 11182145 | G | C | 566 | MTOR | EX48 | missense | p.S2234W |
| M7 | chr10 | 88679009 | C | T | 413 | BMPR1A | EX10 | missense | p.L317F |
| M7 | chr10 | 89624261 | A | T | 224 | PTEN | EX1 | missense | p.N12I |
| M7 | chr13 | 49030469 | A | -CT | 373 | RB1 | EX19 | frameshift | p.L649Vfs*3 |
| M7 | chr17 | 7577099 | C | A | 483 | TP53 | EX8 | missense | p.R280I |
| M7 | chr3 | 178952085 | A | G | 467 | PIK3CA | EX21E | missense | p.H1047R |
| M7 | chr7 | 151960129 | G | C | 161 | MLL3 | EX9 | nonsense | p.S424* |
| M7 | chr8 | 118825139 | T | C | 378 | EXT1 | EX8 | missense | p.D565G |
| M7 | chr8 | 41792213 | C | A | 518 | KAT6A | EX18E | missense | p.L1175F |
| M8 | chr11 | 119167638 | G | A | 442 | CBL | EX13 | missense | p.V683M |
| M8 | chr13 | 49030486 | G | T | 410 | RB1 | IVS19 | splice-5 | . |
| M8 | chr14 | 36987064 | G | A | 202 | NKX2-1 | EX3E | missense | p.R209C |
| M8 | chr16 | 9892253 | T | C | 515 | GRIN2A | EX12 | missense | p.K746R |
| M8 | chr17 | 63530100 | T | A | 558 | AXIN2 | EX10 | missense | p.R779W |
| M8 | chr17 | 7577610 | T | C | 342 | TP53 | IVS6 | splice-3 | . |
| M9 | chr12 | 6697486 | C | A | 360 | CHD4 | EX23 | missense | p.W1148L |
| M9 | chr16 | 67063716 | C | -G | 74 | CBFB | IVS2 | splice-5 | . |
| M9 | chr16 | 67070551 | G | C | 489 | CBFB | EX3 | missense | p.A59P |
| M9 | chr17 | 56435939 | C | T | 390 | RNF43 | EX9 | missense | p.G400R |
| M9 | chr3 | 178936082 | G | A | 428 | PIK3CA | EX10 | missense | p.E542K |
| M9 | chr9 | 80537112 | T | A | 241 | GNAQ | EX2 | missense | p.T96S |
| M9 | chrX | 123185068 | A | T | 347 | STAG2 | EX12 | missense | p.K372M |

## Supplemental Table 11. Genes with higher frequency of amplification in advanced lymph node stage samples among all three sample sets.

| Chr | Position | gene | -log10(p-value)(METABRIC) | -log10(p-value)(TCGA) | -log10(p-value)(Nik-Zainal et al.) |
|---|---|---|---|---|---|
| 8 | 116420723 | TRPS1 | 2.206321372 | 1.091837529 | 1.051548353 |
| 8 | 128748314 | MYC | 2.410596629 | 1.205449152 | 1.06438342 |
| 11 | 67033904 | ADRBK1 | 2.784005203 | 1.350516004 | 1.170034847 |
| 11 | 67056761 | ANKRD13D | 2.745365676 | 1.104191344 | 1.170034847 |
| 11 | 67070918 | SSH3 | 2.745365676 | 1.104191344 | 1.170034847 |
| 11 | 67118235 | POLD4 | 2.745365676 | 1.104191344 | 1.170034847 |
| 11 | 67131638 | CLCF1 | 2.745365676 | 1.350516004 | 1.170034847 |
| 11 | 67159422 | RAD9A | 2.745365676 | 1.350516004 | 1.170034847 |
| 11 | 67165651 | PPP1CA | 2.697708808 | 1.350516004 | 1.170034847 |
| 11 | 67171383 | TBC1D10C | 2.745365676 | 1.350516004 | 1.170034847 |
| 11 | 67195934 | RPS6KB2 | 2.745365676 | 1.372845546 | 1.170034847 |
| 11 | 67202980 | PTPRCAP | 2.745365676 | 1.372845546 | 1.170034847 |
| 11 | 67205517 | CORO1B | 2.745365676 | 1.372845546 | 1.170034847 |
| 11 | 67218771 | GPR152 | 2.745365676 | 1.372845546 | 1.170034847 |
| 11 | 67222817 | CABP4 | 2.745365676 | 1.372845546 | 1.170034847 |
| 11 | 67231818 | TMEM134 | 2.745365676 | 1.372845546 | 1.170034847 |
| 11 | 67250504 | AIP | 2.281832365 | 1.669884209 | 1.170034847 |
| 11 | 67259238 | PITPNM1 | 2.697708808 | 1.669884209 | 1.170034847 |
| 11 | 67273967 | CDK2AP2 | 2.745365676 | 1.824007484 | 1.170034847 |
| 11 | 67286417 | CABP2 | 2.745365676 | 1.824007484 | 1.170034847 |
| 11 | 67351065 | GSTP1 | 2.213954758 | 1.39287069 | 1.170034847 |
| 11 | 67374322 | NDUFV1 | 2.251724187 | 1.39287069 | 1.170034847 |
| 11 | 67395409 | NUDT8 | 2.251724187 | 1.39287069 | 1.170034847 |
| 11 | 67398773 | TBX10 | 2.452994648 | 1.39287069 | 1.170034847 |
| 11 | 67410025 | ACY3 | 2.452994648 | 1.39287069 | 1.170034847 |
| 11 | 67429632 | ALDH3B2 | 2.477225955 | 1.39287069 | 1.170034847 |
| 11 | 67758574 | UNC93B1 | 2.281832365 | 1.133095539 | 1.358428917 |
| 11 | 67776047 | ALDH3B1 | 2.213954758 | 1.133095539 | 1.358428917 |
| 11 | 67798083 | NDUFS8 | 1.838269456 | 1.133095539 | 1.358428917 |
| 11 | 67806461 | TCIRG1 | 1.661875794 | 1.120547945 | 1.358428917 |
| 11 | 67820325 | CHKA | 1.838269456 | 1.133095539 | 1.358428917 |
| 11 | 69455872 | CCND1 | 1.354509525 | 1.136137182 | 1.434983831 |
| 11 | 69480330 | ORAOV1 | 1.354509525 | 1.031649549 | 1.434983831 |
| 11 | 69587796 | FGF4 | 1.49815608 | 1.044407547 | 1.236155735 |

| | | | | | |
|---|---|---|---|---|---|
| 11 | 69624735 | FGF3 | 1.229012095 | 1.266459497 | 1.236155735 |
| 11 | 69924407 | ANO1 | 1.661262499 | 1.051538438 | 1.236155735 |
| 11 | 70049268 | FADD | 1.687793432 | 1.051538438 | 1.871317444 |
| 11 | 70116805 | PPFIA1 | 1.854048165 | 1.162178819 | 1.871317444 |
| 11 | 70244611 | CTTN | 1.581381205 | 1.293017027 | 1.871317444 |
| 11 | 71145456 | DHCR7 | 2.426787984 | 1.372845546 | 1.146103576 |
| 11 | 71164216 | NADSYN1 | 2.74800954 | 1.372845546 | 1.146103576 |
| 11 | 71238312 | KRTAP5-7 | 2.74800954 | 1.234388804 | 1.146103576 |
| 11 | 71249070 | KRTAP5-8 | 2.699859752 | 1.234388804 | 1.146103576 |
| 11 | 71259465 | KRTAP5-9 | 2.699859752 | 1.234388804 | 1.146103576 |
| 11 | 76927602 | GDPD4 | 1.170845753 | 1.272325023 | 1.146103576 |
| 12 | 43748011 | ADAMTS20 | 1.758139258 | 1.47352975 | 1.033860521 |
| 12 | 51157788 | ATF1 | 1.812498404 | 1.570418844 | 1.033860521 |
| 12 | 51236700 | TMPRSS12 | 1.812498404 | 1.570418844 | 1.033860521 |
| 12 | 51318533 | METTL7A | 1.812498404 | 1.256690622 | 1.033860521 |
| 12 | 51347781 | HIGD1C | 1.812498404 | 1.543403964 | 1.033860521 |
| 12 | 51379774 | SLC11A2 | 1.812498404 | 1.543403964 | 1.033860521 |
| 12 | 51442081 | LETMD1 | 1.812498404 | 1.543403964 | 1.033860521 |
| 12 | 51454987 | CSRNP2 | 1.812498404 | 1.543403964 | 1.033860521 |
| 12 | 51487538 | TFCP2 | 1.401468333 | 1.543403964 | 1.033860521 |
| 12 | 51580718 | POU6F1 | 1.401468333 | 1.570418844 | 1.338457228 |
| 12 | 51632507 | DAZAP2 | 1.401468333 | 1.773911204 | 1.338457228 |
| 12 | 51639132 | SMAGP | 1.401468333 | 1.773911204 | 1.338457228 |
| 12 | 51674821 | BIN2 | 1.401468333 | 1.744080455 | 1.338457228 |
| 12 | 51722226 | CELA1 | 1.401468333 | 1.570418844 | 1.338457228 |
| 12 | 51745832 | GALNT6 | 1.21747724 | 1.570418844 | 1.338457228 |
| 12 | 51818593 | SLC4A8 | 1.688800058 | 1.744080455 | 1.338457228 |
| 12 | 51985019 | SCN8A | 1.27098903 | 1.41473928 | 1.338457228 |
| 12 | 54574141 | SMUG1 | 1.21747724 | 1.118568862 | 1.104040312 |
| 12 | 54730999 | MIR148B | 1.691953511 | 1.118568862 | 1.104040312 |
| 12 | 54891494 | NCKAP1L | 1.21747724 | 1.238273588 | 1.104040312 |
| 12 | 56473808 | ERBB3 | 2.82486947 | 1.238273588 | 1.104040312 |
| 12 | 56498102 | PA2G4 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56510373 | RPL41 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56512029 | ZC3H10 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56521985 | ESYT1 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56546203 | MYL6B | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56552044 | MYL6 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56555635 | SMARCC2 | 2.976175806 | 1.238273588 | 1.104040312 |

| | | | | | |
|---|---|---|---|---|---|
| 12 | 56596287 | RNF41 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56623819 | SLC39A5 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56631590 | ANKRD52 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56660641 | COQ10A | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56665482 | CS | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56704211 | CNPY2 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56710006 | PAN2 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56732662 | IL23A | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56735380 | STAT2 | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56754354 | APOF | 2.976175806 | 1.238273588 | 1.104040312 |
| 12 | 56810156 | TIMELESS | 2.82486947 | 1.238273588 | 1.104040312 |
| 12 | 56843285 | MIP | 2.82486947 | 1.238273588 | 1.104040312 |
| 12 | 56862300 | SPRYD4 | 2.82486947 | 1.238273588 | 1.104040312 |
| 12 | 56864735 | GLS2 | 2.82486947 | 1.238273588 | 1.104040312 |
| 12 | 56915608 | RBMS2 | 2.82486947 | 1.238273588 | 1.104040312 |
| 12 | 56989379 | BAZ2A | 2.682088752 | 1.238273588 | 1.104040312 |
| 12 | 57031958 | ATP5B | 2.82486947 | 1.238273588 | 1.336677299 |
| 12 | 57038810 | SNORD59A | 2.82486947 | 1.238273588 | 1.336677299 |
| 12 | 57057124 | PTGES3 | 2.82486947 | 1.238273588 | 1.336677299 |
| 12 | 57106210 | NACA | 2.82486947 | 1.238273588 | 1.336677299 |
| 12 | 57125363 | PRIM1 | 2.82486947 | 1.238273588 | 1.336677299 |
| 12 | 57157107 | HSD17B6 | 3.279255202 | 1.238273588 | 1.336677299 |
| 12 | 57316937 | SDR9C7 | 3.756563571 | 1.238273588 | 1.336677299 |
| 12 | 57345214 | RDH16 | 3.756563571 | 1.238273588 | 1.336677299 |
| 12 | 57388354 | GPR182 | 3.756563571 | 1.238273588 | 1.336677299 |
| 12 | 57392616 | ZBTB39 | 3.756563571 | 1.238273588 | 1.336677299 |
| 12 | 57403780 | TAC3 | 3.756563571 | 1.238273588 | 1.336677299 |
| 12 | 57422300 | MYO1A | 3.756563571 | 1.238273588 | 1.336677299 |
| 12 | 57482676 | NAB2 | 3.756563571 | 1.256690622 | 1.336677299 |
| 12 | 57489186 | STAT6 | 3.756563571 | 1.256690622 | 1.336677299 |
| 12 | 57522281 | LRP1 | 3.756563571 | 1.256690622 | 1.458756171 |
| 12 | 57610577 | NXPH4 | 3.756563571 | 1.543403964 | 1.458756171 |
| 12 | 57623355 | SHMT2 | 3.756563571 | 1.543403964 | 1.458756171 |
| 12 | 57628685 | NDUFA4L2 | 3.756563571 | 1.543403964 | 1.458756171 |
| 12 | 57637241 | STAC3 | 3.756563571 | 1.543403964 | 1.458756171 |
| 12 | 57647547 | R3HDM2 | 3.756563571 | 1.543403964 | 1.458756171 |
| 12 | 57828542 | INHBC | 3.279255202 | 1.570418844 | 1.458756171 |
| 12 | 57849095 | INHBE | 3.279255202 | 1.570418844 | 1.458756171 |
| 12 | 57853933 | GLI1 | 3.279255202 | 1.41473928 | 1.458756171 |

| | | | | | |
|---|---|---|---|---|---|
| 12 | 57866037 | ARHGAP9 | 3.279255202 | 1.41473928 | 1.458756171 |
| 12 | 57881735 | MARS | 3.279255202 | 1.41473928 | 1.458756171 |
| 12 | 57910370 | DDIT3 | 3.279255202 | 1.41473928 | 1.458756171 |
| 12 | 57916658 | MBD6 | 3.122879723 | 1.570418844 | 1.458756171 |
| 12 | 57923832 | DCTN2 | 3.122879723 | 1.384403 | 1.458756171 |
| 12 | 57943846 | KIF5A | 2.974780697 | 1.214289363 | 1.458756171 |
| 12 | 57984941 | PIP4K2C | 2.974780697 | 1.192034599 | 1.458756171 |
| 12 | 57998603 | DTX3 | 2.974780697 | 1.192034599 | 1.458756171 |
| 12 | 58013692 | SLC26A10 | 2.974780697 | 1.192034599 | 1.210703553 |
| 12 | 58019550 | B4GALNT1 | 2.834346085 | 1.192034599 | 1.210703553 |
| 12 | 58087737 | OS9 | 3.576187353 | 1.192034599 | 1.210703553 |
| 12 | 58118075 | AGAP2 | 3.576187353 | 1.192034599 | 1.210703553 |
| 12 | 58138783 | TSPAN31 | 3.576187353 | 1.192034599 | 1.210703553 |
| 12 | 58141509 | CDK4 | 3.576187353 | 1.192034599 | 1.210703553 |
| 12 | 58156116 | CYP27B1 | 3.425958667 | 1.192034599 | 1.210703553 |
| 12 | 58162350 | METTL1 | 3.425958667 | 1.192034599 | 1.210703553 |
| 12 | 58176527 | TSFM | 2.988797382 | 1.359871652 | 1.210703553 |
| 12 | 58191158 | AVIL | 2.988797382 | 1.359871652 | 1.210703553 |
| 12 | 58213709 | CTDSP2 | 2.988797382 | 1.359871652 | 1.210703553 |
| 12 | 58218391 | MIR26A2 | 1.294694244 | 1.359871652 | 1.210703553 |
| 12 | 58335444 | XRCC6BP1 | 3.127353439 | 1.077429467 | 1.210703553 |
| 12 | 62860596 | MON2 | 2.086150325 | 1.192034599 | 1.210703553 |
| 12 | 62997465 | MIRLET7I | 1.185455265 | 1.192034599 | 1.210703553 |
| 12 | 67663060 | CAND1 | 2.492727754 | 1.024678587 | 1.080402767 |
| 12 | 70637493 | CNOT2 | 2.326610837 | 1.39287069 | 1.044005732 |
| 12 | 70760061 | KCNMB4 | 2.326610837 | 1.39287069 | 1.044005732 |
| 12 | 70910629 | PTPRB | 1.84690148 | 1.410938927 | 1.044005732 |
| 12 | 71031852 | PTPRR | 2.15634325 | 1.410816037 | 1.044005732 |
| 12 | 71518876 | TSPAN8 | 1.81940477 | 1.154171265 | 1.245241601 |
| 12 | 71833812 | LGR5 | 1.81940477 | 1.009333071 | 1.245241601 |
| 12 | 72332625 | TPH2 | 1.854513224 | 1.173900948 | 1.693247121 |
| 12 | 72666528 | TRHDE | 1.725159742 | 1.188940352 | 1.338457228 |
| 12 | 75433857 | KCNC2 | 1.512882119 | 1.256690622 | 1.392371258 |
| 12 | 75669758 | CAPS2 | 1.512882119 | 1.26057383 | 1.166825126 |
| 12 | 75728462 | GLIPR1L1 | 1.512882119 | 1.26057383 | 1.166825126 |
| 12 | 75784849 | GLIPR1L2 | 1.512882119 | 1.26057383 | 1.166825126 |
| 12 | 75874512 | GLIPR1 | 1.985915641 | 1.26057383 | 1.166825126 |
| 12 | 75891418 | KRR1 | 1.985915641 | 1.26057383 | 1.166825126 |
| 12 | 76419226 | PHLDA1 | 2.086150325 | 1.118568862 | 1.166825126 |

| | | | | | |
|---|---|---|---|---|---|
| 12 | 76438671 | NAP1L1 | 2.351136795 | 1.118568862 | 1.166825126 |
| 12 | 79257772 | SYT1 | 1.759839119 | 1.039484789 | 1.338457228 |
| 12 | 79985744 | PAWR | 1.817341085 | 1.317012747 | 1.166825126 |
| 12 | 85253266 | SLC6A15 | 2.351136795 | 1.192174057 | 1.166825126 |
| 12 | 85408093 | TSPAN19 | 1.866749533 | 1.192174057 | 1.166825126 |
| 12 | 85430098 | LRRIQ1 | 1.866749533 | 1.344707846 | 1.166825126 |
| 20 | 44098393 | WFDC2 | 1.23793312 | 1.109460516 | 1.534858912 |
| 20 | 44141100 | SPINT3 | 1.23793312 | 1.001120566 | 1.534858912 |
| 20 | 44162835 | WFDC6 | 1.23793312 | 1.001120566 | 1.534858912 |
| 20 | 45313003 | TP53RK | 1.391083595 | 1.092163631 | 1.322880006 |
| 20 | 54572412 | CBLN4 | 1.702749761 | 1.318303169 | 1.088064467 |
| 20 | 54823787 | MC3R | 1.682582604 | 1.197616782 | 1.088064467 |
| 20 | 54944444 | AURKA | 1.517574847 | 1.195788738 | 1.088064467 |
| 20 | 54967573 | CSTF1 | 1.502173461 | 1.195788738 | 1.088064467 |
| 20 | 54987167 | CASS4 | 1.502173461 | 1.188602821 | 1.088064467 |
| 20 | 56725982 | C20orf85 | 1.834468 | 1.195788738 | 1.088064467 |
| 20 | 56807832 | PPP4R1L | 1.854048165 | 1.082357273 | 1.088064467 |
| 20 | 56884770 | RAB22A | 1.854048165 | 1.188602821 | 1.088064467 |
| 20 | 56964174 | VAPB | 1.868570292 | 1.188602821 | 1.088064467 |

**Supplemental Table 12. Genes with higher frequency of deletion in advanced lymph node stage samples among all three sample sets.**

| Chr | Position | gene | -log10(p-value)(METABRIC) | -log10(p-value)(TCGA) | -log10(p-value)(Nik-zainal et al.) |
|---|---|---|---|---|---|
| 1 | 69090 | OR4F5 | 3.349045488 | 2.665908013 | 1.012496149 |
| 1 | 803450 | FAM41C | 3.297569987 | 2.665908013 | 1.012496149 |
| 1 | 861120 | SAMD11 | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 879582 | NOC2L | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 895966 | KLHL17 | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 901876 | PLEKHN1 | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 934343 | HES4 | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 948846 | ISG15 | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 955502 | AGRN | 3.236411833 | 2.665908013 | 1.012496149 |
| 1 | 1017197 | C1orf159 | 3.236411833 | 2.665908013 | 1.210120027 |
| 1 | 1109285 | TTLL10 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1138887 | TNFRSF18 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1146705 | TNFRSF4 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1152287 | SDF4 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1167628 | B3GALT6 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1177825 | FAM132A | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1189291 | UBE2J2 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1215815 | SCNN1D | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1227763 | ACAP3 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1243993 | PUSL1 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1246964 | CPSF3L | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1266725 | TAS1R3 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1270657 | DVL1 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1288070 | MXRA8 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1309109 | AURKAIP1 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1321090 | CCNL2 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1337275 | MRPL20 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1370902 | VWA1 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1385068 | ATAD3C | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1407163 | ATAD3B | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1447522 | ATAD3A | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1477052 | SSU72 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1550794 | MIB2 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1567559 | MMP23B | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1631377 | MMP23A | 3.297569987 | 2.665908013 | 1.210120027 |

| | | | | | |
|---|---|---|---|---|---|
| 1 | 1658823 | SLC35E2 | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1682670 | NADK | 3.297569987 | 2.665908013 | 1.210120027 |
| 1 | 1716724 | GNB1 | 3.297569987 | 2.348614417 | 1.210120027 |
| 1 | 1846265 | CALML6 | 3.297569987 | 2.348614417 | 1.210120027 |
| 1 | 1849028 | TMEM52 | 3.297569987 | 2.348614417 | 1.210120027 |
| 1 | 1950767 | GABRD | 3.297569987 | 2.348614417 | 1.210120027 |
| 1 | 1981908 | PRKCZ | 3.297569987 | 2.348614417 | 1.210120027 |
| 1 | 2160133 | SKI | 3.297569987 | 2.516272588 | 1.012496149 |
| 1 | 2252695 | MORN1 | 3.297569987 | 2.516272588 | 1.012496149 |
| 1 | 2323213 | RER1 | 3.349045488 | 2.516272588 | 1.012496149 |
| 1 | 2336240 | PEX10 | 3.05534957 | 2.516272588 | 1.012496149 |
| 1 | 2407753 | PLCH2 | 3.112279965 | 2.516272588 | 1.012496149 |
| 1 | 2439974 | PANK4 | 3.112279965 | 2.516272588 | 1.012496149 |
| 1 | 2460183 | HES5 | 3.112279965 | 2.516272588 | 1.012496149 |
| 1 | 2487804 | TNFRSF14 | 3.112279965 | 2.516272588 | 1.012496149 |
| 1 | 2522080 | MMEL1 | 3.112279965 | 2.516272588 | 1.171180394 |
| 1 | 2938045 | ACTRT2 | 2.145775286 | 2.321917381 | 1.171180394 |
| 1 | 2985741 | PRDM16 | 2.189138675 | 2.164398296 | 1.171180394 |
| 1 | 3371146 | ARHGEF16 | 2.189138675 | 2.001673014 | 1.595383498 |
| 1 | 3404505 | MEGF6 | 2.189138675 | 2.001673014 | 1.595383498 |
| 1 | 3541555 | TPRG1L | 2.145775286 | 2.001673014 | 1.595383498 |
| 1 | 3569128 | TP73 | 1.949043178 | 2.001673014 | 1.595383498 |
| 1 | 3668964 | CCDC27 | 1.949043178 | 2.001673014 | 1.595383498 |
| 1 | 3696783 | LRRC47 | 1.949043178 | 1.853861459 | 1.595383498 |
| 1 | 3773844 | DFFB | 1.925359729 | 1.69438039 | 1.595383498 |
| 1 | 3805696 | C1orf174 | 1.925359729 | 1.837078151 | 1.595383498 |
| 1 | 4715104 | AJAP1 | 1.949043178 | 1.69438039 | 1.382610127 |
| 1 | 5922869 | NPHP4 | 2.213954758 | 1.432131397 | 1.337135821 |
| 1 | 6105980 | KCNAB2 | 1.838269456 | 1.432131397 | 1.270851447 |
| 1 | 6161846 | CHD5 | 1.838269456 | 1.432131397 | 1.270851447 |
| 1 | 6245079 | RPL22 | 1.838269456 | 1.432131397 | 1.270851447 |
| 1 | 6266188 | RNF207 | 1.838269456 | 1.432131397 | 1.270851447 |
| 1 | 6281252 | ICMT | 1.838269456 | 1.432131397 | 1.270851447 |
| 1 | 6304251 | HES3 | 1.855422009 | 1.432131397 | 1.270851447 |
| 1 | 6307405 | GPR153 | 1.855422009 | 1.432131397 | 1.270851447 |
| 1 | 6324331 | ACOT7 | 1.855422009 | 1.432131397 | 1.270851447 |
| 1 | 6475291 | HES2 | 1.320294256 | 1.432131397 | 1.270851447 |
| 1 | 6484847 | ESPN | 1.320294256 | 1.432131397 | 1.270851447 |
| 1 | 6521210 | TNFRSF25 | 1.320294256 | 1.432131397 | 1.278519418 |

|  |  |  |  |  |  |
|---|---|---|---|---|---|
| 1 | 6526151 | PLEKHG5 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6581406 | NOL9 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6615337 | TAS1R1 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6640062 | ZBTB48 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6650783 | KLHL21 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6673755 | PHF13 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6684924 | THAP3 | 1.320294256 | 1.432131397 | 1.278519418 |
| 1 | 6694227 | DNAJC11 | 1.307762306 | 1.432131397 | 1.278519418 |
| 1 | 6845383 | CAMTA1 | 1.868238343 | 1.432131397 | 1.278519418 |
| 1 | 10509775 | CORT | 1.158122081 | 1.551478257 | 1.031776382 |
| 1 | 10520587 | DFFA | 1.158122081 | 1.551478257 | 1.031776382 |
| 1 | 10535002 | PEX14 | 1.007447959 | 1.567580778 | 1.031776382 |
| 1 | 13910251 | PDPN | 1.055534274 | 1.422767263 | 1.051060636 |
| 1 | 14031349 | PRDM2 | 1.078944559 | 1.673521016 | 1.256079676 |
| 1 | 15479027 | TMEM51 | 1.240709291 | 1.543530965 | 1.230450538 |
| 1 | 15573767 | FHAD1 | 1.069061787 | 1.824126863 | 1.031776382 |
| 1 | 15736390 | EFHD2 | 1.355568315 | 1.824126863 | 1.031776382 |
| 1 | 15764937 | CTRC | 1.355568315 | 1.824126863 | 1.031776382 |
| 1 | 15783222 | CELA2A | 1.355568315 | 1.824126863 | 1.031776382 |
| 1 | 15802595 | CELA2B | 1.355568315 | 1.673521016 | 1.031776382 |
| 1 | 15818768 | CASP9 | 1.355568315 | 1.673521016 | 1.031776382 |
| 1 | 15853351 | DNAJC16 | 1.196946765 | 1.543530965 | 1.031776382 |
| 1 | 15898193 | AGMAT | 1.196946765 | 1.543530965 | 1.031776382 |
| 1 | 15943952 | DDI2 | 1.208325724 | 1.543530965 | 1.031776382 |
| 1 | 15986363 | RSC1A1 | 1.355568315 | 1.543530965 | 1.031776382 |
| 1 | 16010826 | PLEKHM2 | 1.355568315 | 1.543530965 | 1.209084091 |
| 1 | 16062808 | SLC25A34 | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16068916 | TMEM82 | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16090993 | FBLIM1 | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16174358 | SPEN | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16268363 | ZBTB17 | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16330730 | C1orf64 | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16340522 | HSPB7 | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16348485 | CLCNKA | 1.403235435 | 1.681162488 | 1.209084091 |
| 1 | 16370230 | CLCNKB | 1.392222813 | 1.681162488 | 1.209084091 |
| 1 | 16384263 | FAM131C | 1.392222813 | 1.681162488 | 1.209084091 |
| 1 | 16450831 | EPHA2 | 1.392222813 | 1.681162488 | 1.209084091 |
| 1 | 16524598 | ARHGEF19 | 1.392222813 | 1.681162488 | 1.401482476 |
| 1 | 16576558 | FBXO42 | 1.392222813 | 1.824126863 | 1.401482476 |

| | | | | | |
|---|---|---|---|---|---|
| 1 | 16725137 | SPATA21 | 1.392222813 | 1.824126863 | 1.401482476 |
| 1 | 16767166 | NECAP2 | 1.392222813 | 1.824126863 | 1.401482476 |
| 1 | 16890411 | NBPF1 | 1.208325724 | 1.543530965 | 1.401482476 |
| 1 | 17017712 | ESPNP | 1.392222813 | 1.543530965 | 1.977909106 |
| 1 | 17248444 | CROCC | 1.392222813 | 1.543530965 | 1.977909106 |
| 1 | 17300996 | MFAP2 | 1.392222813 | 1.543530965 | 1.977909106 |
| 1 | 17312452 | ATP13A2 | 1.403235435 | 1.543530965 | 1.977909106 |
| 1 | 17345216 | SDHB | 1.403235435 | 1.681162488 | 1.977909106 |
| 1 | 17393255 | PADI2 | 1.593911675 | 1.681162488 | 1.977909106 |
| 1 | 17531620 | PADI1 | 1.428994378 | 1.681162488 | 2.026703827 |
| 1 | 17575592 | PADI3 | 1.428994378 | 1.681162488 | 2.026703827 |
| 1 | 17634689 | PADI4 | 1.428994378 | 1.956607407 | 2.026703827 |
| 1 | 17698740 | PADI6 | 1.428994378 | 1.956607407 | 2.026703827 |
| 1 | 17733250 | RCC2 | 1.428994378 | 1.956607407 | 2.026703827 |
| 1 | 17866329 | ARHGEF10L | 1.448826729 | 1.974659366 | 2.026703827 |
| 1 | 18081807 | ACTL8 | 1.448826729 | 1.974659366 | 2.567461983 |
| 1 | 18434239 | IGSF21 | 1.121813606 | 1.956607407 | 2.567461983 |
| 1 | 18807423 | KLHDC7A | 1.448826729 | 1.956607407 | 2.026703827 |
| 1 | 18957499 | PAX7 | 1.783666762 | 1.681162488 | 2.026703827 |
| 1 | 19166092 | TAS1R2 | 1.996771178 | 2.128169022 | 2.458068902 |
| 1 | 19197923 | ALDH4A1 | 1.996771178 | 1.837078151 | 2.458068902 |
| 1 | 19230773 | IFFO2 | 1.996771178 | 1.837078151 | 2.458068902 |
| 1 | 19400999 | UBR4 | 1.761750664 | 1.837078151 | 2.458068902 |
| 1 | 19578074 | MRTO4 | 1.761750664 | 1.837078151 | 2.528672544 |
| 1 | 19592475 | AKR7L | 1.761750664 | 1.824126863 | 2.528672544 |
| 1 | 19609056 | AKR7A3 | 1.761750664 | 1.824126863 | 2.528672544 |
| 1 | 19629201 | AKR7A2 | 2.128765977 | 1.824126863 | 2.528672544 |
| 1 | 19638739 | PQLC2 | 2.128765977 | 1.824126863 | 2.528672544 |
| 1 | 19665266 | CAPZB | 2.324321272 | 1.824126863 | 2.528672544 |
| 1 | 19969722 | NBL1 | 2.291244356 | 1.673521016 | 2.528672544 |
| 1 | 19991779 | HTR6 | 2.291244356 | 1.673521016 | 2.528672544 |
| 1 | 20008705 | TMCO4 | 2.092269511 | 1.543530965 | 2.458068902 |
| 1 | 20140521 | RNF186 | 2.324321272 | 1.543530965 | 2.458068902 |
| 1 | 20208887 | OTUD3 | 2.324321272 | 1.543530965 | 2.458068902 |
| 1 | 20246799 | PLA2G2E | 2.324321272 | 1.543530965 | 2.458068902 |
| 1 | 20301923 | PLA2G2A | 2.349490985 | 1.543530965 | 2.458068902 |
| 1 | 20396700 | PLA2G5 | 2.128765977 | 1.543530965 | 2.458068902 |
| 1 | 20438431 | PLA2G2D | 2.128765977 | 1.543530965 | 2.458068902 |
| 1 | 20465822 | PLA2G2F | 2.092246607 | 1.543530965 | 2.458068902 |

| | | | | | |
|---|---|---|---|---|---|
| 1 | 20490483 | PLA2G2C | 2.092246607 | 1.681162488 | 2.458068902 |
| 1 | 20512577 | UBXN10 | 2.092246607 | 1.543530965 | 2.528672544 |
| 1 | 20617411 | VWA5B1 | 2.092246607 | 1.543530965 | 2.528672544 |
| 1 | 20808883 | CAMK2N1 | 2.092246607 | 1.543530965 | 2.528672544 |
| 1 | 20825940 | MUL1 | 2.092246607 | 1.543530965 | 2.528672544 |
| 1 | 20878931 | FAM43B | 1.916956246 | 1.543530965 | 2.528672544 |

**Supplemental Table 13: Correlation of other prognostic factors with lymph node metastasis in three datasets.**

| p-value | Age | Tumor size (T stage 1-3) | PR | HER2 | TP53 mutation |
|---|---|---|---|---|---|
| Nik-Zainal et al. (N0 vs N2-3; n=131 vs 63) | 1.0 | 1.3e-06 | 0.24 | 1.0 | 0.065 |
| TCGA (N0 vs N2-3; n=301 vs 129) | 0.0046 (negatively) | <2.0e-11 | 0.31 | 0.66 | 0.084 |
| METABRIC (Stage 1 vs Stage 3-4; n=403 vs 78) | 7.5e-6 (positively) | <1.0e-15 | 0.00129 | 0.32 | 0.0070 |

Tumor size (T stages) significantly correlated with lymph node status in all datasets. Age negatively correlated with lymph node metastasis in the TCGA dataset, but positively in METABRIC dataset. HER2 status did not correlate with lymph node metastasis in any of the three datasets and was removed from the subsequent regression analysis. PR and TP53 mutation status only correlated with lymph node metastasis in the METABRIC dataset. However, we found a trend toward increased TP53 mutation frequency in N2-3 groups compared with the N0 group in Nik-Zainal et al.'s dataset (23.8% vs 13.0%) and TCGA dataset (26.4% vs 17.8%).

## Supplemental Table 14: Genomic areas significantly associated with breast cancer lymph node metastasis identified by TCGA samples.

| chr | start | end | cytoband | Gain/loss | genes |
|---|---|---|---|---|---|
| chr1 | 0 | 2522080 | 1p36.32-36.33 | loss | FAM138A,OR4F5,LOC100132287,OR4F29,OR4F16,LOC100133331,LOC643837,FAM41C,SAMD11,NOC2L,KLHL17,PLEKHN1,C1orf170,HES4,ISG15,AGRN,C1orf159,MIR200B,MIR200A,MIR429,TTLL10,TNFRSF18,TNFRSF4,SDF4,B3GALT6,FAM132A,UBE2J2,SCNN1D,ACAP3,PUSL1,CPSF3L,GLTPD1,TAS1R3,DVL1,MXRA8,AURKAIP1,CCNL2,LOC148413,MRPL20,TMEM88B,VWA1,ATAD3C,ATAD3B,ATAD3A,SSU72,MIB2,MMP23B,MMP23A,SLC35E2,NADK,GNB1,CALML6,TMEM52,KIAA1751,GABRD,PRKCZ,C1orf86,SKI,MORN1,LOC100129534,RER1,PEX10,PLCH2,PANK4,HES5,LOC115110,TNFRSF14,MMEL1 |
| chr1 | 8921058 | 9712667 | 1p36.22-36.23 | loss | ENO1,CA6,SLC2A7,SLC2A5,GPR157,MIR34A,H6PD,SPSB1,SLC25A33,TMEM201,PIK3CD,C1orf200 |
| chr1 | 17698740 | 18807423 | 1p36.13 | loss | PADI6,RCC2,ARHGEF10L,ACTL8,IGSF21,KLHDC7A |
| chr1 | 31882411 | 41492870 | 1p34.2-35.2 | loss | SERINC2,LOC284551,TINAGL1,HCRTR1,PEF1,COL16A1,BAI2,SPOCD1,PTP4A2,KHDRBS1,TMEM39B,KPNA6,TXLNA,CCDC28B,IQCC,DCDC2B,EIF3I,FAM167B,LCK,HDAC1,MARCKSL1,LOC100128071,TSSK3,BSDC1,ZBTB8B,ZBTB8A,ZBTB8OS,RBBP4,SYNC,KIAA1522,YARS,S100PBP,FNDC5,HPCA,TMEM54,RNF19B,AK2,ADC,TRIM62,ZNF362,PHC2,ZSCAN20,CSMD2,HMGB4,C1orf94,GJB5,GJB4,GJB3,GJA4,C1orf212,DLGAP3,ZMYM6,ZMYM1,SFPQ,ZMYM4,KIAA0319L,NCDN,TFAP2E,PSMB2,C1orf216,CLSPN,EIF2C4,EIF2C1,EIF2C3,TEKT2,ADPRHL2,COL8A2,TRAPPC3,MAP7D1,THRAP3,STK40,LSM10,OSCP1,MRPS15,CSF3R,GRIK3,ZC3H12A,MEAF6,SNIP1,DNALI1,GNL2,RSPO1,C1orf109,CDCA8,EPHA10,MANEAL,YRDC,C1orf122,MTF1,INPP5B,SF3A3,FHL3,UTP11L,POU3F1,RRAGC,MYCBP,GJA9,RHBDL2,AKIRIN1,NDUFS5,MACF1,KIAA0754,BMP8A,PABPC4,SNORA55,HEYL,NT5C1A,HPCAL4,PPIE,BMP8B,OXCT2,TRIT1,MYCL1,MFSD2A,CAP1,PPT1,RLF,TMCO2,ZMPSTE24,COL9A2,SMAP2,ZNF643,ZNF642,ZNF684,RIMS3,LOC100130557,NFYC,MIR30E,MIR30C1,KCNQ4,CITED4,SLFNL1 |
| chr1 | 48998526 | 56960418 | 1p32.3 | loss | AGBL4,BEND5,ELAVL4,DMRTA2,FAF1,CDKN2C,C1orf185,RNF11,TTC39A,EPS15,OSBPL9,NRD1,RAB3B,TXNDC12,KTI12,BTF3L4,ZFYVE9,CC2D1B,PRPF38A,ZCCHC11,GPX7,FAM159A,ZYG11B,ZYG11A,ECHDC2,SCP2,PODN,SLC1A7,CPT2,C1orf123,MAGOH,LRP8,DMRTB1,GLIS1,TMEM48,YIPF1,DIO1,HSPB11,LRRC42,LDLRAD1,TMEM59,CDCP2,CYB5RL,MRPL37,SSBP3,ACOT11,FAM151A,TTC4,PARS2,TTC22,C1orf177,DHCR24,TMEM61,BSND,PCSK9,USP24,PPAP2B |
| chr1 | 70225857 | 94335013 | 1p22.1-31.1 | loss | LRRC7,LRRC40,ANKRD13C,HHLA3,CTH,PTGER3,ZRANB2,MIR186,NEGR1,LRRIQ3,FPGT,TNNI3K,C1orf173,CRYZ,TYW3,LHX8,SLC44A5,ACADM,RABGGTB,SNORD45C,SNORD45A,SNORD45B,MSH4,ASB17,ST6GALNAC3,ST6GALNAC5,PIGK,AK5,ZZZ3,USP33,FAM73A,NEXN,FUBP1,DNAJB4,GIPC2,MGC27382,PTGFR,IFI44L,IFI44,ELTD1,LPHN2,TTLL7,PRKACB,SAMD13,UOX,DNASE2B,RPF1,GNG5,SPATA1,CTBS,C1orf180,SSX2IP,LPAR3,MCOLN2,MCOLN3,WDR63,SYDE2,C1orf52,BCL10,DDAH1,CYR61,ZNHIT6,COL24A1,ODF2L,CLCA2,CLCA1,CLCA4,CLCA3P,SH3GLB1,SEP15,HS2ST1,LOC339524,LMO4,PKN2,GTF2B,CCBL2,RBMXL1,GBP3,GBP1,GBP2,GBP7,GBP4,GBP5,GBP6,LRRC8B,LRRC8C,LRRC8D,ZNF326,BARHL2,ZNF644,HFM1,CDC7,HSP90B3P,TGFBR3,BRDT,EPHX4,BTBD8,KIAA1107,C1orf146,GLMN,RPAP2,GFI1,EVI5,RPL5,SNORD21,SNORA66,FAM69A,MTF2,TMED5,CCDC18,DR1,FNBP1L,BCAR3,MIR760,DNTTIP2 |
| chr1 | 97187174 | 109102970 | 1p13.3-21.3 | loss | PTBP2,DPYD,MIR137,SNX7,LPPR5,LPPR4,PALMD,FRRS1,AGL,SLC35A3,HIAT1,SASS6,LRRC39,DBT,MIR553,CDC14A,GPR88,VCAM1,EXTL2,SLC30A7,DPH5,S1PR1,OLFM3,COL11A1,RNPC3,AMY2B,AMY2A,AMY1A,AMY1B,AMY1C,PRMT6,NTNG1,VAV3,SLC25A24,NBPF4,NBPF6,FAM102B |
| chr1 | 111682248 | 113615830 | 1p13.2 | loss | CEPT1,DENND2D,CHI3L2,CHIA,OVGP1,WDR77,ATP5F1,C1orf162,ADORA3,RAP1A,DDX20,KCND3,CTTNBP2NL,WNT2B,ST7L,CAPZA1,MOV10,RHOC,PPM1J,FAM19A3,SLC16A1,LRIG2 |
| chr2 | 148602569 | 157291964 | 2q23.1-24.1 | loss | ACVR2A,MBD5,EPC2,KIF5C,LYPD6B,LYPD6,MMADHC,RND3,RBM43,NMI,TNFAIP6,RIF1,NEB,ARL5A,CACNB4,STAM2,FMNL2,PRPF40A,ARL6IP6,RPRM,GALNT13,KCNJ3,NR4A2,GPD2 |
| chr2 | 162164785 | 168810529 | 2q24.2-24.3 | loss | PSMD14,TBR1,SLC4A10,DPP4,GCG,FAP,IFIH1,GCA,KCNH7,FIGN,GRB14,COBLL1,SLC38A11,SCN3A,SCN2A,CSRNP3,GALNT3,TTC21B,SCN1A,SCN9A,SCN7A,XIRP2,B3GALT1,STK39 |
| chr3 | 193310932 | 197476623 | 3q29 | gain | OPA1,LOC100128023,HES1,LOC100131551,CPN2,LRRC15,GP5,ATP13A3,TMEM44,LSG1,FAM43A,ACAP2,PPP1R2,APOD,MIR570,MUC20,MUC4,TNK2,TFRC,ZDHHC19,PCYT1A,TCTEX1D2,TM4SF19,UBXN7,RNF168,C3orf43,WDR53,FBXO45,LRRC33,PIGX,PAK2,SENP5,NCBP2,LOC152217,PIGZ,MFI2,DLG1,BDH1,LOC220729,KIAA0226,MIR922,FYTTD1 |
| chr6 | 26365397 | 26597170 | 6p22.2 | gain | BTN3A2,BTN2A2,BTN3A1,BTN3A3,BTN2A1,BTN1A1,HCG11,HMGN4,ABT1 |
| chr6 | 30227338 | 33762448 | 6p21.32-21.33 | gain | HLA-L,HCG18,TRIM39,RPP21,HLA-E,GNL1,PRR3,ABCF1,MIR877,PPP1R10,MRPS18B,C6orf136,DHX16,NRM,MDC1,TUBB,FLOT1,IER3,DDR1,GTF2H4,VARS2,SFTA2,DPCR1,MUC21,HCG22,C6orf15,PSORS1C1,CDSN,PSORS1C2,CCHCR1,TCF19,POU5F1,PSORS1C3,HCG27,HLA -C,HLA-B,MICA,HCP5,HCG26,MICB,MCCD1,SNORD117,SNORD84,ATP6V1G2,NFKBIL1,LTA,TNF,LTB,LST1,NCR3,AIF1,SNORA38,APOM,C6orf47,CSNK2B,LY6G5B,LY6G5C,LY6G6F,LY6G6E,LY6G6D,LY6G6C,C6orf25,DDAH2,CLIC1,MSH5,VARS,LSM2,HSPA1L,HSPA1A,HSPA1B,C6orf48,SNORD48,SNORD52,NEU1,SLC44A4,EHMT2,ZBTB12,C2,CFB,RDBP,SKIV2L,DOM3Z,STK19,C4A,TNXA,C4B,CYP21A2,TNXB,ATF6B,FKBPL,PRRT1,PPT2,EGFL8,AGPAT1,RNF5,AGER,PBX2,GPSM3,NOTCH4,C6orf10,BTNL2,HLA -DRA,HLA-DRB5,HLA-DRB6,HLA-DRB1,HLA-DQA1,HLA-DQB1,HLA-DQA2,HLA-DQB2,HLA-DOB,TAP2,PSMB8,TAP1,PSMB9,HLA-DMB,HLA-DMA,BRD2,HLA-DOA,HLA-DPA1,HLA-DPB1,HLA-DPB2,COL11A2,RXRB,SLC39A7,HSD17B8,MIR219-1,RING1,VPS52,RPS18,B3GALT4,WDR46,PFDN6,RGL2,TAPBP,ZBTB22,DAXX,KIFC1,PHF1,CUTA,SYNGAP1,ZBTB9,BAK1,GGNBP1,ITPR3,IP6K3,LEMD2,MLN |
| chr8 | 123793900 | 146277823 | 8q24.13-24.3 | gain | ZHX2,DERL1,WDR67,FAM83A,LOC100131726,C8orf76,ZHX1,ATAD2,MIR548D1,WDYHV1,FBXO32,KLHL38,ANXA13,FAM91A1,FER1L6,TMEM65,TRMT12,RNF139,TATDN1,NDUFB9,MTSS1,LOC157381,ZNF572,SQLE,KIAA0196,NSMCE2,TRIB1,FAM84B,POU5F1B,LOC727677,MYC,PVT1,GSDMC,FAM49B,ASAP1,ADCY8,EFR3A,OC90,HHLA1,KCNQ3,HPYR1,LRRC6,TMEM71,PHF20L1,TG,SLA,WISP1,NDRG1,ST3GAL1,ZFAT,MIR30B,MIR30D,LOC286094,KHDRBS3,FAM135B,COL22A1,KCNK9,TRAPPC9,CHRAC1,EIF2C2,PTK2,DENND3,SLC45A4,LOC731779,GPR20,PTP4A3,FLJ43860,TSNARE1,BAI1,ARC,JRK,PSCA,LY6K,SLURP1,LYPD2,LYNX1,LY6D,GML,CYP11B1,CYP11B2,LOC100133669,LY6E,C8orf31,LY6H,GPIHBP1,ZFP41,GLI4,ZNF696,TOP1MT,RHPN1,MAFA,ZC3H3,GSDMD,C8orf73,NAPRT1,EEF1D,TIGD5,PYCRL,TSTA3,ZNF623,ZNF707,BREA2,MAPK15,FAM83H,SCRIB,MIR937,PUF60,NRBP2,EPPK1,MIR661,PARP10,GRINA,SPATC1,OPLAH,EXOSC4,GPAA1,CYC1,SHARPIN,MAF1,KIAA1875,HEATR7A,SCXB,BOP1,SCXA,HSF1,DGAT1,SCRT1,FBXL6,ADCK5,CPSF1,MIR939,SLC39A4,VPS28,CYHR1,KIFC2,FOXH1,PPP1R16A,GPT,MFSD3,RECQL4,LRRC14,LRRC24,ZNF251,ZNF34,RPL8,ZNF517,ZNF7,COMMD5,ZNF250,ZNF16,C8orf33 |
| chr9 | 0 | 16409500 | 9p22.3-24.3 | loss | WASH1,FAM138C,FOXD4,CBWD1,C9orf66,DOCK8,KANK1,DMRT1,DMRT3,DMRT2,SMARCA2,FLJ35024,VLDLR,KCNV2,KIAA0020,RFX3,GLIS3,SLC1A1,PPAPDC2,CDC37L1,AK3,RCL1,MIR101 -2,JAK2,INSL6,INSL4,RLN2,RLN1,CD274,PDCD1LG2,KIAA1432,ERMP1,MLANA,KIAA2026,RANBP6,IL33,TPD52L3,UHRF2,GLDC,KDM4C,C9orf123,PTPRD,TYRP1,MPDZ,NFIB,ZDHHC21,CER1,FREM1,LOC389705,TTC39B,SNAPC3,PSIP1,BNC2 |
| chr9 | 21006364 | 32629451 | 9p21 | loss | PTPLAD2,IFNB1,IFNW1,IFNA21,IFNA4,IFNA7,IFNA10,IFNA16,IFNA17,IFNA14,IFNA5,KLHL9,IFNA6,IFNA13,IFNA2,IFNA8,IFNA1,IFNE,MIR31,MTAP,C9orf53,CDKN2A,CDKN2B,DMRTA1,ELAVL2,TUSC1,PLAA,IFT74,LRRC19,TEK,IFNK,C9orf72,LINGO2,MIR876,MIR873,ACO1,DDX58,TOPORS,NDUFB6,TAF1L |
| chr10 | 7200585 | 13685705 | 10p13-14 | gain | SFMBT2,ITIH5,ITIH2,KIN,ATP5C1,TAF3,FLJ45983,GATA3,SFTA1P,LOC254312,USP6NL,ECHDC3,C10orf47,UPF2,DHTKD1,SEC61A2,NUDT5,CDC123,CAMK1D,LOC283070,CCDC3,OPTN,MCM10,UCMA,PHYH,SEPHS1,BEND7,PRPF18,FRMD4A |
| chr10 | 112052797 | 112631552 | 10q25.2 | loss | SMNDC1,DUSP5,SMC3,RBM20,LOC282997,PDCD4 |
| chr11 | 5757677 | 7020548 | 11p15.4 | loss | OR56B1,OR52N4,OR52N5,OR52N1,OR52N2,OR52E6,OR52E8,OR52E4,OR56A3,OR56A5,OR52L1,OR56A4,OR56A1,OR56B4,OR52B2,OR52W1,C11orf42,FAM160A2,CNGA4,CCKBR,PRKCDBP,SMPD1,APBB1,HPX,TRIM3,ARFIP2,FXC1,DNHD1,RRP8,ILK,TAF10,TPP1,DCHS1,MRPL17,OR2AG2,OR2AG1,OR6A2,OR10A5,OR10A2,OR10A4,OR2D2,OR2D3,ZNF215,ZNF214 |
| chr12 | 69004618 | 69753531 | 12q15 | gain | RAP1B,NUP107,SLC35E3,MDM2,CPM,CPSF6,LYZ,YEATS4 |
| chr16 | 28834368 | 31885078 | 16p11.2 | loss | ATXN2L,TUFM,SH2B1,ATP2A1,RABEP2,CD19,NFATC2IP,SPNS1,LAT,LOC606724,BOLA2,SULT1A4,LOC388242,LOC440354,SLC7A5P1,SPN,QPRT,C16orf54,MAZ,PRRT2,MVP,CDIPT,LOC440356,SEZ6L2,ASPHD1,KCTD13,TMEM219,TAOK2,HIRIP3,INO80E,DOC2A,C16orf92,FAM57B,ALDOA,PPP4C,TBX6,YPEL3,GDPD3,MAPK3,CORO1A,BOLA2B,SULT1A3,LOC613038,LOC613037,LOC595101,CD2BP2,TBC1D10B,MYLPF,SEPT1,ZNF48,ZNF771,DCTPP1,SEPHS2,ITGAL,ZNF768,ZNF747,ZNF764,ZNF688,ZNF785,ZNF689,PRR14,FBRS,SRCAP,SNORA30,PHKG2,C16orf93,RNF40,ZNF629,BCL7C,CTF1,FBXL19,ORAI3,SETD1A,HSD3B7,STX1B,STX4,ZNF668,ZNF646,VKORC1,BCKDK,PRSS8,PRSS36,FUS,PYCARD,TRIM72,PYDC1,ITGAM,ITGAX,ITGAD,COX6A2,ZNF843,ARMC5,TGFB1I1,SLC5A2,C16orf58,AHSP,CSDAP1,ZNF720,ZNF267 |
| chr17 | 27030533 | 27071022 | 17q11.2 | loss | PROCA1,RAB34,RPL23A,SNORD42B,SNORD4A,SNORD42A,SNORD4B,TLCD1,NEK8,TRAF4 |
| chr17 | 41476352 | 42018171 | 17q21.31 | loss | ARL4D,DHX8,ETV4,MEOX1,SOST,DUSP3,C17orf105,MPP3,CD300LG,MPP2,PPY |
| chr18 | 25530929 | 31158540 | 18q12.1 | loss | CDH2,DSC3,DSC2,DSC1,DSG1,DSG4,DSG3,DSG2,TTR,B4GALT6,RNF125,RNF138,MEP1B,FAM59A,WBP11P1,KLHL14,ASXL3 |

## Supplemental Table 15: qPCR results of 170 ER+ Danish breast cancers using RNaseP as reference gene.

| Sample_id | Numbers of lymph node metastases | Recurrence | TANC1 | MCL1 | MYC | BCL2L1 |
|---|---|---|---|---|---|---|
| 24 | 0 | R | 2.51 | 3.92 | 4.82 | 2.83 |
| 49 | 31 | R | 1.74 | 11.38 | 4.69 | 3.77 |
| 60 | 2 | R | 2.1 | 4.26 | 2.86 | 2.39 |
| 102 | 1 | R | 1.41 | 4.97 | 2.7 | 2.17 |
| 166 | 1 | R | 1.88 | 5.95 | 2.57 | 2.21 |
| 170 | 5 | R | 0.93 | 8.39 | 1.8 | 2.88 |
| 171 | 1 | R | 1.84 | 3.44 | 2.22 | 1.93 |
| 183 | 7 | R | 2.71 | 3.54 | 3.69 | 2.16 |
| 185 | 3 | R | 1.47 | 6.78 | 2.27 | 1.31 |
| 188 | 4 | R | 1.9 | 3.54 | 1.99 | 1.81 |
| 200 | 2 | R | 2.32 | 5.3 | 3.65 | 1.95 |
| 204 | 1 | R | 2.62 | 4.91 | 2.38 | 2.54 |
| 211 | 2 | R | 0.68 | 4.38 | 1.26 | 1.35 |
| 227 | 1 | R | 1.98 | 3.72 | 2.43 | 2.17 |
| 250 | 3 | R | 1.81 | 4.94 | 3.69 | 1.72 |
| 260 | 13 | R | 1.75 | 2.97 | 1.43 | 1.86 |
| 272-1 | 1 | R | 5.33 | 10.17 | 4.1 | 4.17 |
| 272-2 | 1 | R | 5 | NA | 3.6 | 2.99 |
| 288 | 0 | R | 1.41 | 5.33 | 1.52 | 2.3 |
| 300 | 19 | R | 1.63 | 8.95 | 2.67 | 2.52 |
| 305 | 2 | R | 2.22 | 5.1 | 6.06 | 3.85 |
| 395 | 2 | R | 1.78 | 8.1 | 8.85 | 2.16 |
| 424 | 0 | R | 1.23 | 2.71 | 3.4 | 1.72 |
| 452 | 0 | R | 1.16 | 3.91 | 2.52 | 2.67 |
| 458 | 0 | R | 2.49 | 4.74 | 5.12 | 4.56 |
| 501 | 0 | R | 1.74 | 4.09 | 5.04 | 2.13 |
| 516 | 15 | R | 1.5 | 6.73 | 3.17 | 2.11 |
| 525 | 10 | R | 1.93 | 4.76 | 3.19 | 2.66 |
| 543 | 0 | R | 1.05 | 2.35 | 3.34 | 1.55 |
| 564 | 1 | R | 2.26 | 4.16 | 5.72 | 2.75 |
| 570 | 3 | R | 1.26 | 3.1 | 3.5 | 2.32 |
| 594 | 0 | R | 2.45 | 2.98 | 2.16 | 2.54 |
| 595 | 0 | R | 1.84 | 4.35 | 3.35 | 3.43 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 644 | 0 | R | 1.15 | 3.82 | 5.61 | 3.08 |
| 646 | 2 | R | 1.24 | 4.68 | 2.24 | 2.1 |
| 686 | 5 | R | 2.35 | 3.26 | 4.47 | 3.18 |
| 693 | 24 | R | 2.23 | 3.63 | 2.68 | 2.12 |
| 702 | 10 | R | 2.59 | 4.9 | 5.13 | 2.18 |
| 703 | 1 | R | 0.94 | 5.16 | 2.68 | 1.61 |
| 704 | 1 | R | 2.21 | 4.78 | 2.45 | 2.19 |
| 706 | 1 | R | 1.91 | 7.22 | 3.41 | 2.4 |
| 707 | 1 | R | 1.58 | 2.99 | 2.05 | 1.92 |
| 709 | 1 | R | 0.95 | 3.62 | 2.45 | 1.61 |
| 712 | 1 | R | 2.04 | 4.67 | 2.25 | 1.98 |
| 714 | 2 | R | 2.4 | 5.69 | 2.73 | 3.12 |
| 729 | 2 | R | 2.3 | 5.26 | 2.52 | 2.17 |
| 741 | 7 | R | 1.84 | 6.21 | 2.27 | 2.16 |
| 744 | 2 | R | 1.67 | 8.64 | 4.39 | 3.22 |
| 747 | 14 | R | 1.77 | 5.18 | 2.22 | 1.88 |
| 748 | 2 | R | 2.12 | 3.12 | 3.49 | 1.61 |
| 750 | 1 | R | 2.49 | 3.89 | 3.14 | 2.92 |
| 751 | 5 | R | 1.92 | 5.74 | 4.22 | 1.66 |
| 753 | 0 | R | 2.29 | 5.77 | 2.15 | 1.79 |
| 757 | 2 | R | 1.21 | 6.46 | 1.57 | 1.4 |
| 766 | 0 | R | 3.11 | 6.89 | 5.6 | 3.57 |
| 768 | 26 | R | 1.97 | 5.25 | 2.2 | 1.95 |
| 785 | 3 | R | 1.26 | 4.69 | 1.7 | 1.99 |
| 794 | 0 | R | 2.15 | 5.78 | 2.55 | 3.31 |
| 809 | 0 | R | 3.34 | 7.75 | 5.42 | 2.89 |
| 815 | 4 | R | 1.98 | 10.86 | 2.26 | 2.72 |
| 841 | 2 | R | 1.5 | 6.31 | 2.75 | 2.75 |
| 847 | 3 | R | 1.15 | 9.1 | 2.29 | 3.51 |
| 891 | 1 | R | 2.38 | 3.83 | 3.68 | 2.09 |
| 962 | 18 | R | 2.03 | 0.99 | 4.53 | 1.28 |
| 964 | 3 | R | 2.28 | 1.34 | 5.1 | 1.86 |
| 971 | 0 | R | 3.44 | 1.91 | 2.92 | 2.08 |
| 979 | 0 | R | 3.09 | 3.94 | 3.15 | 2.16 |
| 993 | 13 | R | 2.3 | 2.78 | 2.3 | 1.8 |
| 995 | 3 | R | 4.76 | 4.68 | 3.67 | 2.9 |
| 996 | 0 | R | 3.22 | 2.93 | 2.97 | 2.46 |
| 1008 | 4 | R | 2.39 | 7.2 | 82.71 | 3.15 |
| 1015 | 0 | R | 2.26 | 6.4 | 2.47 | 2.19 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 1016 | 23 | R | 1.38 | 4.09 | 2.92 | 2.3 |
| 1035 | 2 | R | 2.38 | 2.94 | 2.33 | 2.47 |
| 1042 | 13 | R | 2.72 | 3.79 | 2.76 | 3.46 |
| 1047 | 6 | R | 1.97 | 3.74 | 4.07 | 2.81 |
| 1050 | 2 | R | 4.72 | 6.49 | 8.31 | 5.14 |
| 1063 | 22 | R | 1.23 | 3.57 | 2.28 | 2.06 |
| 1067 | 1 | R | 1.25 | 4.62 | 2.18 | 2.73 |
| 1113 | 0 | R | 1.78 | 3.19 | 2.06 | 1.84 |
| 1115 | 1 | R | 1.9 | 8.1 | 2.46 | 2.95 |
| 1116 | 0 | R | 2.15 | 3.1 | 2.48 | 1.91 |
| 1132 | 5 | R | 1.35 | 4.04 | 2.96 | 3.19 |
| 1137 | 1 | R | 1.31 | 6.36 | 2.65 | 1.36 |
| 1144 | 14 | R | 3.67 | 4.74 | 4.28 | 4.12 |
| 1150 | 40 | R | 1.06 | 5.15 | 2.8 | 1.36 |
| 1188 | 12 | R | 3.87 | 9.34 | 4.2 | 2.69 |
| 1196 | 3 | R | 1.24 | 12.82 | NA | 2.66 |
| 1208 | 0 | R | 2.39 | 3.89 | 3.35 | 2.36 |
| 1226 | 4 | R | 1.71 | 3.59 | 2.9 | 2.78 |
| 1249 | 4 | R | 1.82 | 5.07 | 2.22 | 2.13 |
| 1268 | 0 | R | 2.11 | 5.76 | NA | 2.28 |
| 1269 | 16 | R | 1.3 | 2.65 | 4.42 | 1.45 |
| 1270 | 8 | R | 1.35 | 5.07 | 6.46 | 4.5 |
| 1280 | 3 | R | 1.43 | 3.5 | 4.31 | 1.64 |
| 1281 | 0 | R | 1.24 | 7.16 | 3.44 | 2.4 |
| 1293 | 6 | R | 2.63 | 1.99 | 2.59 | 2.02 |
| 1310 | 0 | R | 1.42 | 4.98 | 6.68 | 2.9 |
| 1311 | 5 | R | 1.62 | 7.89 | 3.47 | 2.57 |
| 1319 | 18 | R | 1.46 | 5.42 | 2.16 | 2.46 |
| 1336 | 8 | R | 1.42 | 5.88 | 4.33 | 1.83 |
| 1373 | 3 | R | 1.41 | 6.57 | 3.69 | 3.92 |
| 1374 | 0 | R | 2.28 | 5.16 | 1.84 | 2.79 |
| 199 | 4 | N | 2.2 | 4.54 | 2.39 | 1.99 |
| 333 | 1 | N | 1.73 | 7.39 | 3.59 | 2.71 |
| 340 | 0 | N | 1.75 | 4.77 | 2.25 | 2.21 |
| 419 | 1 | N | 2.05 | 4.48 | 6.05 | 2.73 |
| 494 | 10 | N | 3.24 | 3.3 | 2.89 | 2.56 |
| 496 | 10 | N | 2.22 | 3.2 | 3.18 | 2.86 |
| 652 | 5 | N | 1.69 | 5.33 | 3.51 | 2.14 |
| 715 | 0 | N | 2.22 | 3.99 | 3.14 | 2.43 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 716 | 1 | N | 1.74 | 3.98 | 2.49 | 2.41 |
| 717 | 2 | N | 1.18 | 9.01 | 1.7 | 1.91 |
| 718 | 3 | N | 2.31 | 5.15 | 2.36 | 2.53 |
| 721 | 3 | N | 2.05 | 4.49 | 2.4 | 2.14 |
| 722 | 0 | N | 1.5 | 3.2 | 4.16 | 1.43 |
| 724 | 3 | N | 2.47 | 5.92 | 2.7 | 2.51 |
| 730 | 1 | N | 1.33 | 2.85 | 1.42 | 1.53 |
| 731 | 3 | N | 2.28 | 6.36 | 3.23 | 2.49 |
| 732 | 0 | N | 1.22 | 4.95 | 2.38 | 2.01 |
| 733 | 0 | N | 2.67 | 5.72 | 2.81 | 3.24 |
| 734 | 1 | N | 2.15 | 5.96 | 3.37 | 2.62 |
| 735 | 1 | N | 1.97 | 6.56 | 3.74 | 2.02 |
| 739 | 5 | N | 2.53 | 4.62 | 4.34 | 2.32 |
| 742 | 0 | N | 2.04 | 6.16 | 2.51 | 3.3 |
| 743 | 3 | N | 1.87 | 3.57 | 4.44 | 3.21 |
| 745 | 2 | N | 1.91 | 6.93 | 1.93 | 1.73 |
| 752 | 1 | N | 1.96 | 3.65 | 3.31 | 1.99 |
| 754 | 0 | N | 2.39 | 2.65 | 1.85 | 1.76 |
| 755 | 1 | N | 1.9 | 5.78 | 2.3 | 2.07 |
| 756 | 0 | N | 1.48 | 4.24 | 1.91 | 1.53 |
| 760 | 1 | N | 2.13 | 7.65 | 5.2 | 2.71 |
| 762 | 0 | N | 3.17 | 5.11 | 2.77 | 2.26 |
| 763 | 2 | N | 2.63 | 7.01 | 2.88 | 1.82 |
| 764 | 0 | N | 2.15 | 3.54 | 2.74 | 1.8 |
| 765 | 0 | N | 2.06 | 6.11 | 2.7 | 2.07 |
| 769 | 0 | N | 2 | 4.89 | 1.92 | 2.04 |
| 770 | 3 | N | 2.2 | 6.72 | 7.26 | 3.05 |
| 771 | 4 | N | 2.23 | 3.83 | 2.49 | 2.19 |
| 773 | 0 | N | 2.19 | 4.55 | 2.8 | 1.95 |
| 774 | 0 | N | 2 | 3.03 | 2.14 | 1.61 |
| 775 | 0 | N | 2.08 | 8.68 | 2.29 | 1.75 |
| 778 | 0 | N | 2.48 | 8.96 | 2.12 | 1.89 |
| 779 | 0 | N | 1.73 | 6.46 | 2.69 | 1.68 |
| 781 | 1 | N | 2.16 | 4.44 | 2.46 | 2.03 |
| 782 | 1 | N | 2.84 | 5.3 | 2.62 | 2.27 |
| 783 | 2 | N | 2.33 | 7.82 | 3.45 | 1.91 |
| 784 | 0 | N | 1.25 | 17.65 | 8.51 | 1.39 |
| 787 | 2 | N | 1.91 | 5.8 | 3.91 | 2.56 |
| 789 | 0 | N | 2.62 | 7.5 | 3.05 | 2.09 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 790 | 0 | N | 2.77 | 3.44 | 4.86 | 2.27 |
| 792 | 0 | N | 1.48 | 3.86 | 2.79 | 3.14 |
| 796 | 4 | N | 2.5 | 8.05 | 5.29 | 2.36 |
| 798 | 10 | N | 2.21 | 3.91 | 1.29 | 2.14 |
| 799 | 1 | N | 2.38 | 4.83 | 2.31 | 2.03 |
| 803 | 1 | N | 1.69 | 6.33 | 2.14 | 1.61 |
| 805 | 0 | N | 2.23 | 4.3 | 2.38 | 2.23 |
| 806 | 1 | N | 1.82 | 3.46 | 2.54 | 2.32 |
| 808 | 0 | N | 2.35 | 5.27 | 2.44 | 2.17 |
| 814 | 0 | N | 2.99 | 7.86 | 2.8 | 2.82 |
| 953 | 1 | N | 1.61 | 1.58 | 2.4 | 1.34 |
| 1105 | 1 | N | 1.31 | 15.25 | 3.16 | 1.71 |
| 1171 | 3 | N | 1.81 | 10.72 | 2.32 | 2.69 |
| 1176 | 5 | N | 1.85 | 6.87 | 2.1 | 2.66 |
| 1181 | 0 | N | 1.79 | 10.9 | 8.02 | 2.11 |
| 1182 | 2 | N | 1.85 | 4.46 | 3.97 | 2.96 |
| 1229 | 2 | N | 2.12 | 4.03 | 2.38 | 1.83 |
| 1296 | 0 | N | 2.03 | 5.92 | 2.24 | 2.38 |
| 1300 | 0 | N | 1.76 | 5.45 | 2.16 | 2.2 |
| 1349 | 0 | N | 1.78 | 3.47 | 3.7 | 2.4 |