

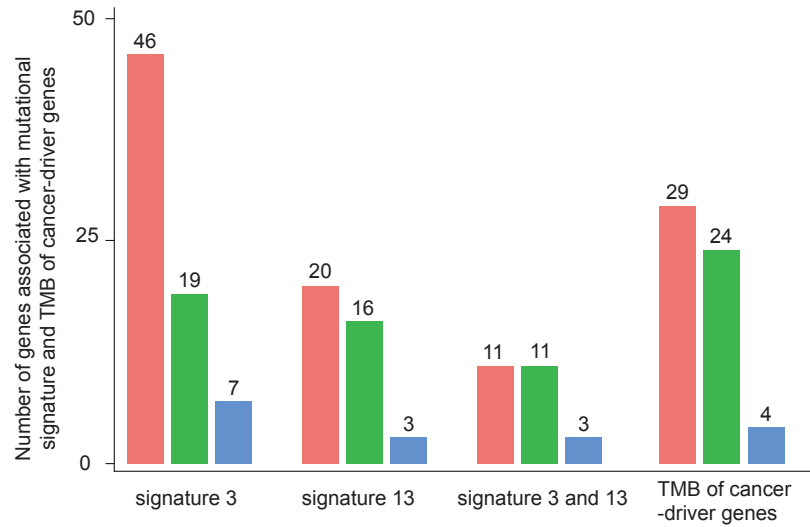
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**Supplemental Data**

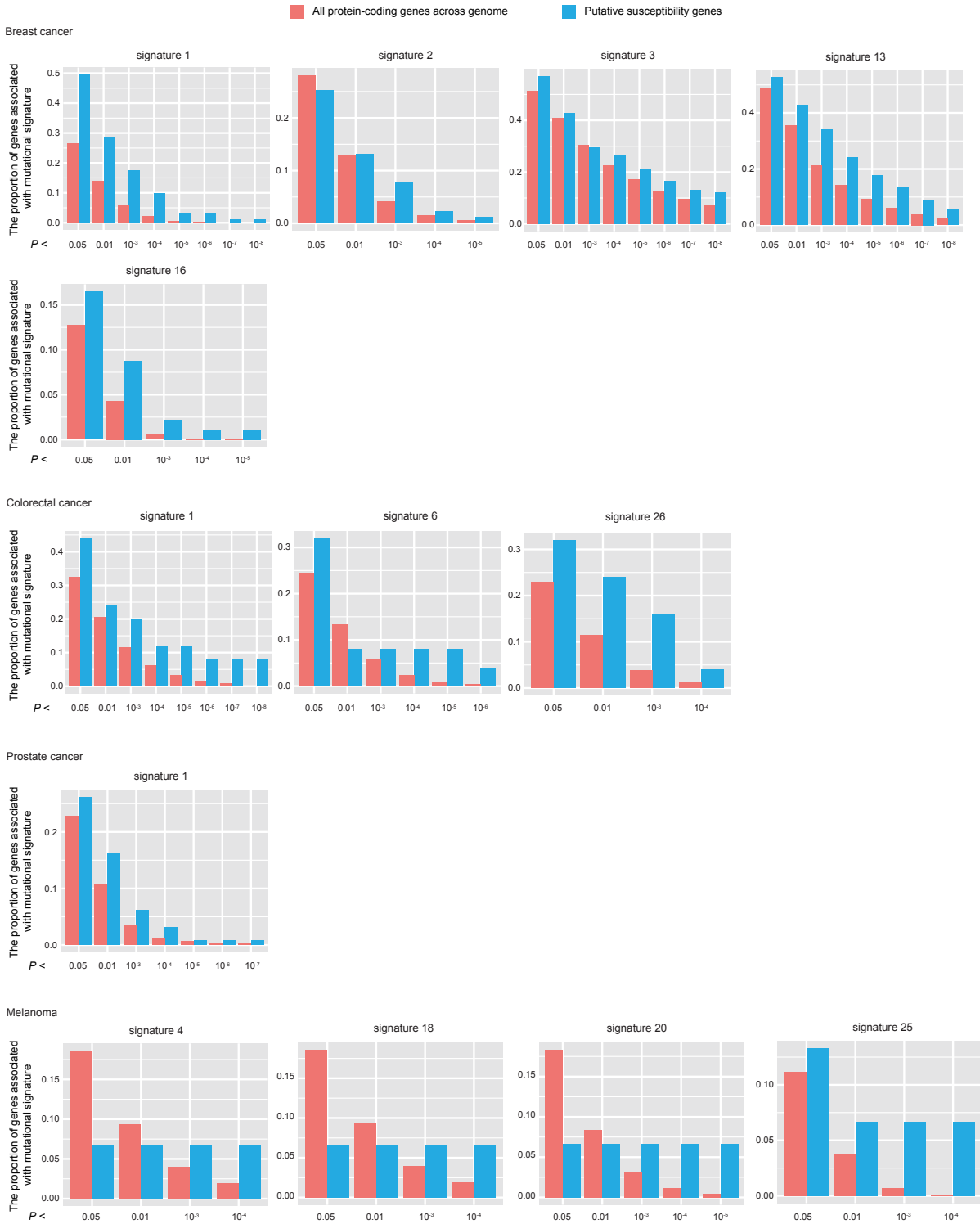
**Identifying Putative Susceptibility Genes and  
Evaluating Their Associations with  
Somatic Mutations in Human Cancers**

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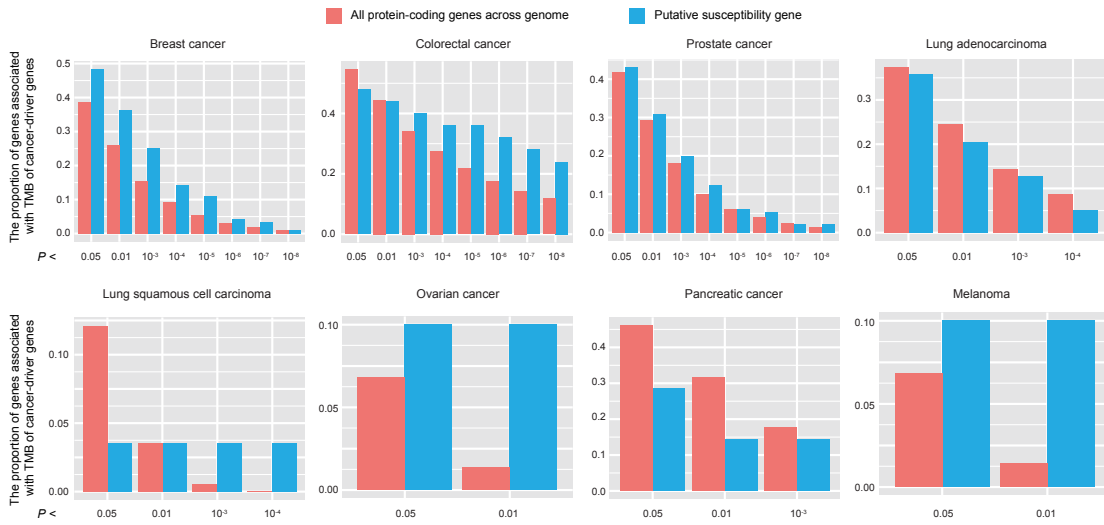
## Supplemental Data



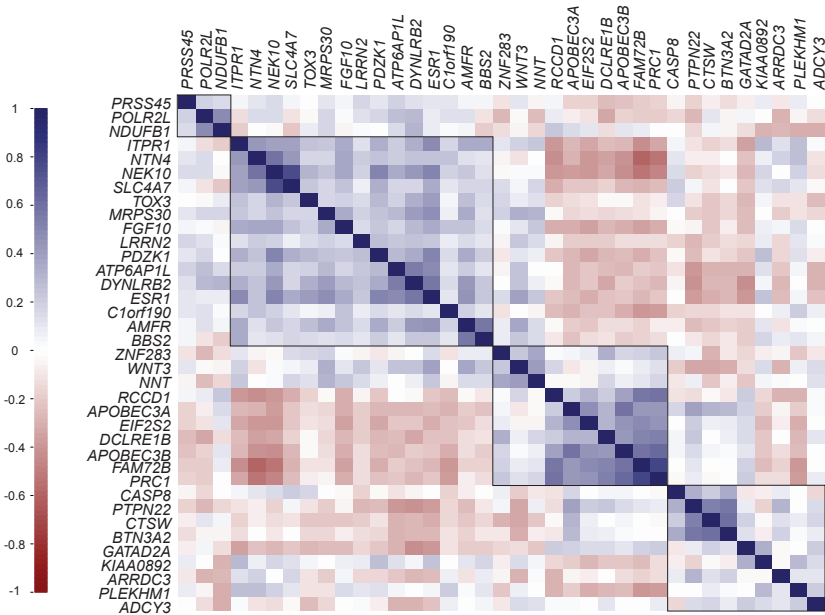
**Figure S1. Numbers of putative susceptibility genes associated with mutational signatures and TMB of cancer-driver genes in breast cancer.** The red and green colors refer to numbers of significant associations for the genes identified in our previous <sup>1</sup> and Fachal et al's <sup>2</sup> studies, respectively (Bonferroni-corrected  $P < 0.05$ ). The blue colors refer to numbers of significant associations that were commonly observed in the two gene sets.



**Figure S2. The proportion of genes associated with mutational signatures at different significance thresholds for each cancer type.** The red and blue colors refer to the proportion of significant associations for all protein-coding genes across genome and significant associations for putative susceptibility genes, respectively.



**Figure S3. The proportion of genes associated with TMB of cancer-driver genes at different significance thresholds for each cancer type.** The red and blue colors refer to the proportion of significant associations for all protein-coding genes across genome and significant associations for putative susceptibility genes, respectively.



**Figure S4.** The expression correlations of putative susceptibility genes that were associated with mutation signatures and TMB of cancer-driver genes in breast cancer.

**Table S1:** A list of 294 GWAS-identified risk SNPs for each of six cancer types.

**Table S2:** A list of eQTL target genes for each of six cancer types.

**Table S3:** The identified eQTL target genes for index SNPs supported by evidence of cis-regulation by putative functional variants via proximal promoter or distal enhancer-promoter interactions.

**Table S4:** Associations between putative susceptibility gene expression levels and mutational signatures across seven cancer types (nominal  $P < 0.05$ ).

**Table S5:** Associations between putative susceptibility gene expression levels and TMB of cancer-driver genes across seven cancer types.

**Table S6:** Spearman correlation between mutational signatures and TMB of cancer-driver genes for each cancer type.

**Table S7:** A list of genes with evidence of regulation by putative functional variants, associated with specific mutational signatures and TMB of cancer-driver genes.

**Table S8:** Associations of the genes identified in the Fachal et al's study <sup>2</sup> with mutational signatures and TMB of cancer-driver genes in breast cancer (Bonferroni-corrected  $P < 0.05$ ).

### **Supplemental References**

1. Guo, X., Lin, W., Bao, J., Cai, Q., Pan, X., Bai, M., Yuan, Y., Shi, J., Sun, Y., Han, M.R., et al. (2018). A Comprehensive cis-eQTL Analysis Revealed Target Genes in Breast Cancer Susceptibility Loci Identified in Genome-wide Association Studies. *Am J Hum Genet* 102, 890-903.
2. Fachal, L., Aschard, H., Beesley, J., Barnes, D.R., Allen, J., Kar, S., Pooley, K.A., Dennis, J., Michailidou, K., Turman, C., et al. (2019). Fine-mapping of 150 breast cancer risk regions identifies 178 high confidence target genes. *bioRxiv*, 521054.