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Supplemental Data

A Comprehensive *cis*-eQTL Analysis Revealed Target Genes in Breast Cancer Susceptibility Loci Identified in Genome-wide Association Studies

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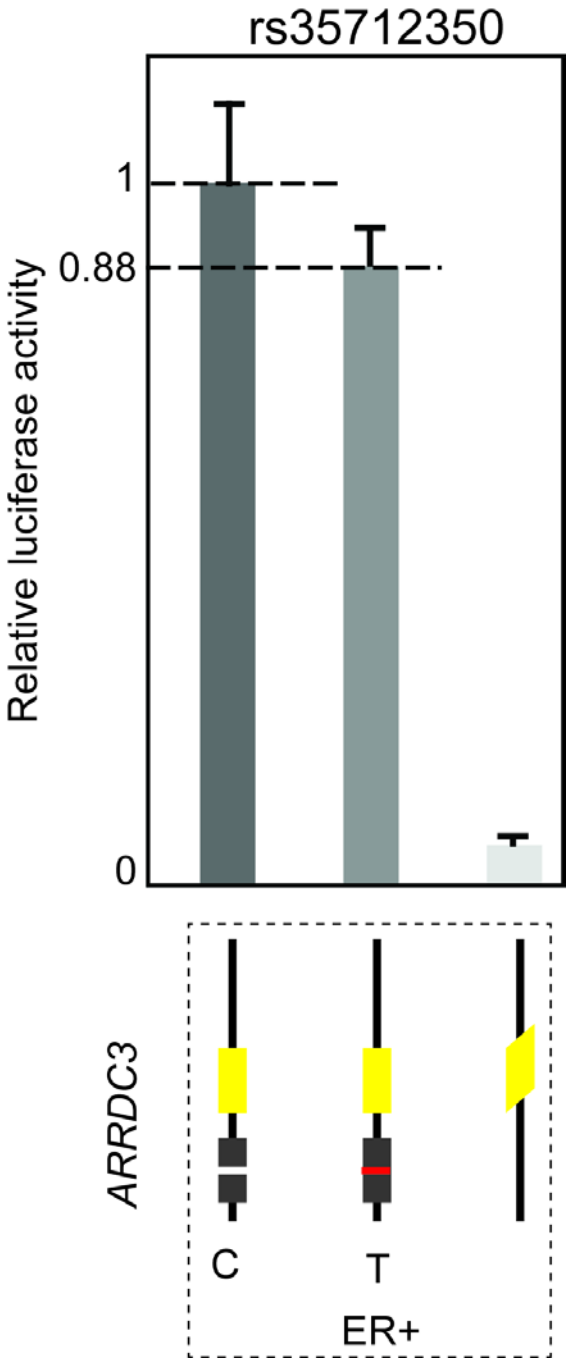

Figure S1: The result of luciferase reporter assays for rs35712350 (surrogate for lead SNP rs10474352; *ARRDC3*). The error bars represent the standard deviation of promoter activities of target genes. At the bottom of each panel: the alternative allele of functional SNPs' changing promoter activities using luciferase reporter assays in the ER+ MCF-7 and ER- SK-BR3 breast cancer cell lines. The fragment containing the reference allele of each SNP was cloned downstream for luciferase construct and an alternative allele was engineered into it.

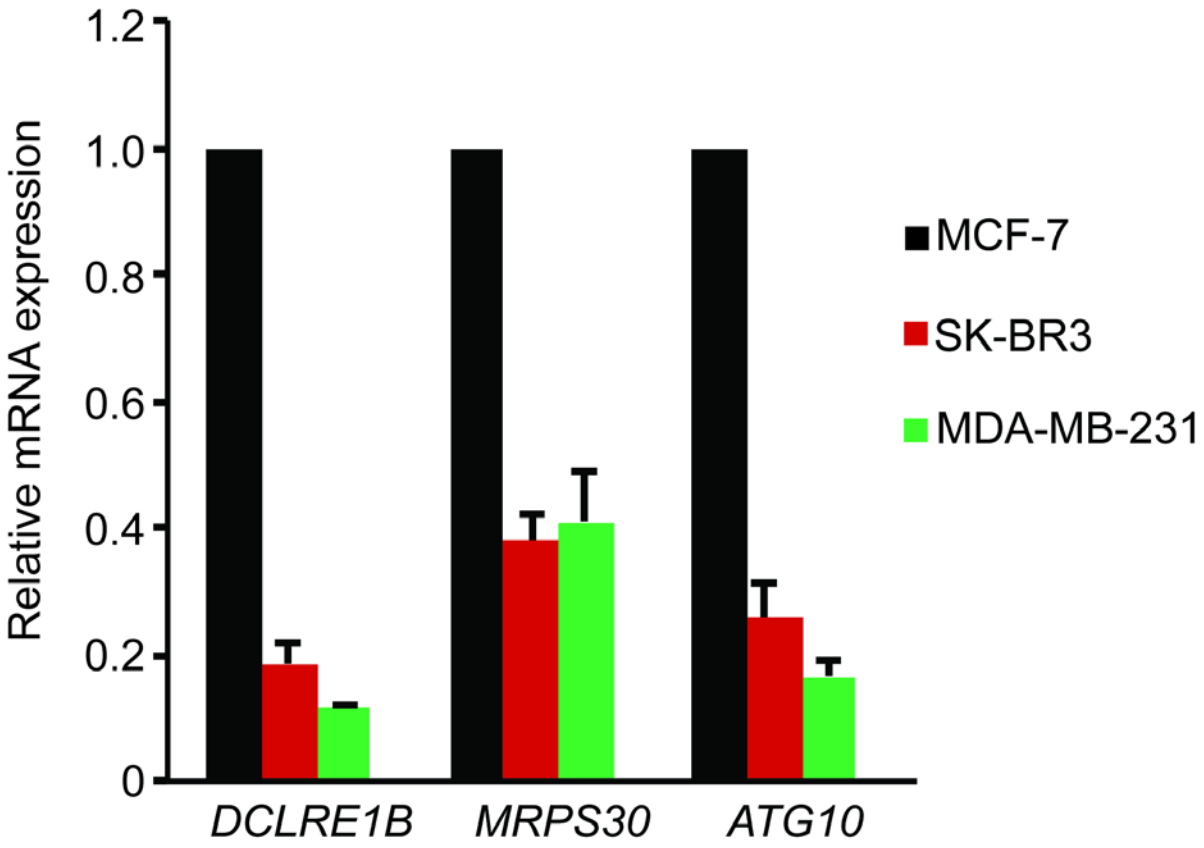

Figure S2: The relative mRNA levels of *DCLRE1B*, *MPRS30*, and *ATG10* in breast cancer cell lines relative to GAPDH were determined by quantitative RT-PCR. The mRNA levels in both knockdown and control cells were measured in technical triplicates. The error bars represent the standard deviation of the mRNA expression.

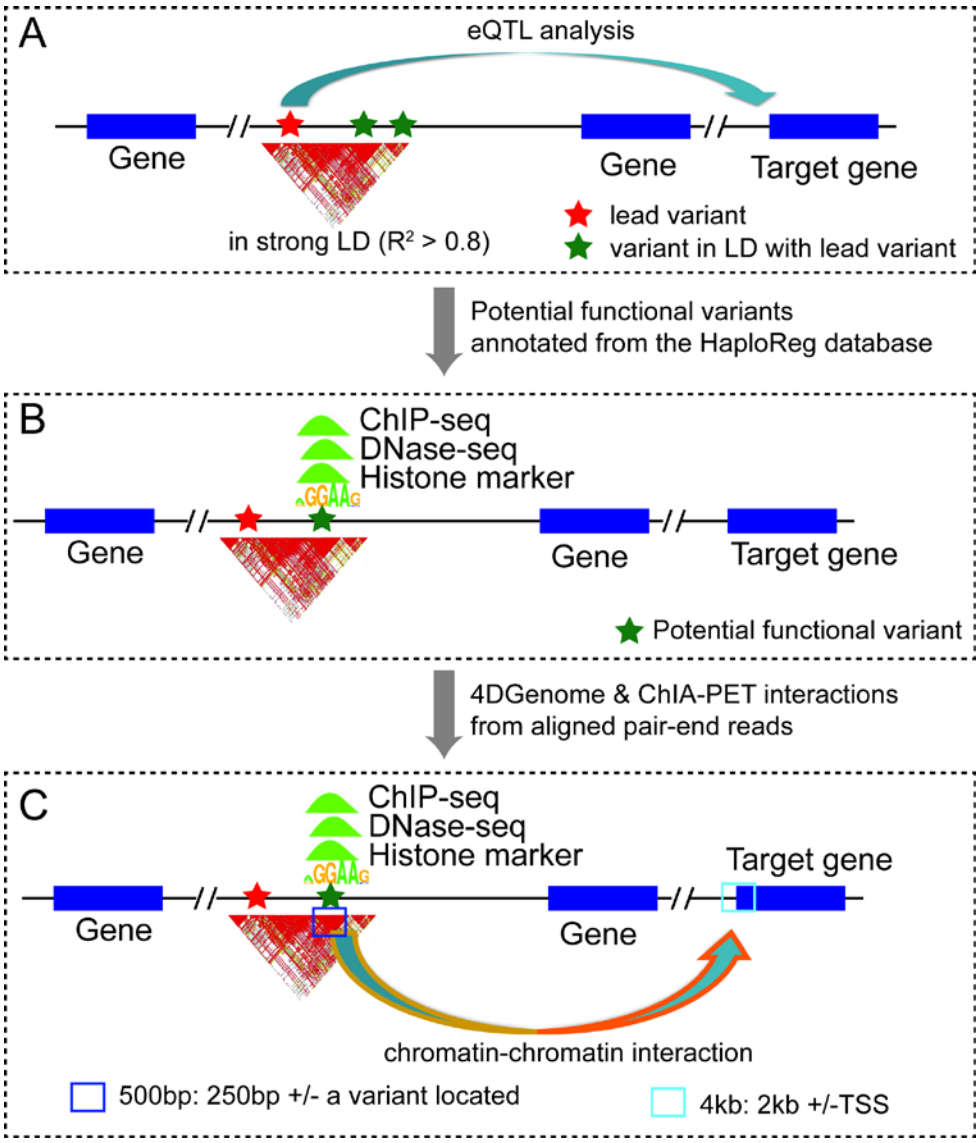

Figure S3. A flow chart to illustrate chromatin-interaction data analysis to search for evidence of the target gene and lead SNP. Panels from top to bottom: A) eQTL analysis to identify target genes for lead SNP; B) the functional annotation for SNPs in strong LD with lead SNP using Haploreg database; C) the illustration of chromatin-chromatin interactions between potential function variants and the target gene. To analyze chromatin interactions between the regions for potential functional variants and promoter regions of the identified candidate target genes, we examined ±250 bp nearby regions of functional variants and ±2kb nearby regions of the gene transcription start site (TSS).

Table S1: A collection of 159 lead SNPs for breast cancer risk.

Primers	Sequence
***ATG10*_NheI_P_S**	CTAGCTAGCGGTGAACCTCAGCGATCAGA
***ATG10*_NcoI_P_A**	CATGCCATGG ATACTCACCCCTCTCCGCTC
***ATG10*_BamHI_E_S**	CGGGATCCGATGGTGGGGGAGCTTTCTA
***ATG10*_SalI_E_A**	ACGCGTCGACAGTGATGAACTATGCAATAACATGA
***ATG10*_M_S**	AATGTAAACTGAATATTGATAGAGAAGGGA
***ATG10*_M_A**	CTATCAATATTCAGTTTACATTGAAACCCT
***PAX9*_ KpnI_P_S**	GGGGTACCTATAGGTGGCGCTGTGACAAG
***PAX9*_ NcoI_P_A**	CATGCCATGGGGGGGATGGCGGCTAAAAGG
***PAX9*_BamHI_E_S**	CGGGATCCGGACAGCCCCAGTAGTTAGT
***PAX9*_SalI_E_A**	ACGCGTCGACCATTTGAACTTCCTGCCTGAGC
***PAX9*_M_S**	GTGCAGCGTCTACCCCCGCACTCTCGCGGA
***PAX9*_M_A**	GAGAGTGCGGGGGTAGACGCTGCACATCCA
***DCLRE1B*_KpnI_P_S**	GGGGTACCGGTCTCCTACTGGAACCAACTG
***DCLRE1B*_HindIII_P_A**	CCC AAGCTTGGGAAACATCGGCCTAGCTTACTT
***DCLRE1B*_M_S**	TCTTGCATCGTCACCTACAGGTATGGGGCT
***DCLRE1B*_M_A**	CCCATACCTGTAGGTGACGATGCAAGAGGT
***DCLRE1B*_ATG_M_S**	AACCCTACCACCTTGAATTCCTGATCCCC
***DCLRE1B*_ATG_M_A**	GGGATCAGGAATTCAAGGTGGTAGGGTTG
***MRPS30*_ XhoI_P_S**	CCGCTCGAGACAGCCTCCTTCCTTGGTTCA
***MRPS30*_HindIII_P_A**	CCCAAGCTTCCCCAGATAGGAACGAAAGGACTA
***MRPS30*_ M_S**	CAGAAACGACCTCCCAAGACGTCGCGGCGA
***MRPS30*_ M_A**	GACGTCTTGGGAGGTCGTTTCTGTAGCCGT
***MRPS30*_ ATG_M_S**	GAATCGCGGGCAAAGTTGGCGGCGGCCAG
***MRPS30*_ ATG_M_A**	GCCGCCGCCAACTTTGCCCGCGATTCCGGA
***SSBP4*_BglII_P_S**	GAAGATCTAGGCTGGAGCGCAATCTTGG
***SSBP4*_HindIII_P_A**	CCCAAGCTTCGCTCCACACAGCAAAAGTG
***SSBP4*_ M_S**	TCCGAAGTGCTGGGaCTACAGGCACACGCT
***SSBP4*_ M_A**	AGCGTGTGCCTGTAGtCCCAGCACTTCGGA

Table S2. Primer pairs used for the construction and mutation of LUC expression vectors. “S” represents sense primers; “A” represents anti-sense primers; “P” represents the associated primers for the promoter sequence cloning; “E” represents the associated primers for the enhancer sequence cloning; “M” represents the associated primers for site-directed mutagenesis; “ATG” represents the associated premiers for initial codon site-directed mutagenesis. The coordinates (hg38) of cloning sequences: ***ATG10,*** chr5:81970347-81972314 (P), 82074325-82075574 (E); ***PAX9,*** chr14:36656034-36658001 (P), 36662767-36664064 (E); ***DCLRE1B,*** chr1:113905616-113907995 (P), 113903813-113906037 (E); ***MRPS30,*** chr5: 44807527-44809701 (P); ***SSBP4,*** chr19:18416066-18419590 (P).

Primers	Sequence
GAPDH_Q_F1	CTCCAAAATCAAGTGGGGCG
GAPDH_Q_R1	ATGGTTCACACCCATGACGA
DCLRE1B_Q_F1	TTGGAACCAGACCCACCCTA
DCLRE1B_Q_R1	GCACGAAGCTCGGAGTAAGA
MRPS30_Q_F	ACGGCTACAGAAACGACCTG
MRPS30_Q_R	GAAGGTCTGCGGGTAAACCA

Table S3. **Primer pairs used for silencing gene expressions of *DCLRE1B* and *MRPS30,* using siRNAs in breast cancer cell lines.**

Table S4: A list of target genes identified to be associated with lead SNPs based on eQTL analysis, using data from METABRIC, TCGA and GTEx.

Table S5: A functional annotation of SNPs in strong LDs ($R^2 > 0.8$ in European population based on the 1000 Genomes project) with 51 lead SNPs for target genes.

Table S6: The list of eQTL target genes found to be the nearest genes of functional SNPs, or supported by chromatin interactions between their promoters and the regions of functional SNPs.