

Supplementary Information

Identification of two novel breast cancer loci through large-scale genome-wide association study in the Japanese population

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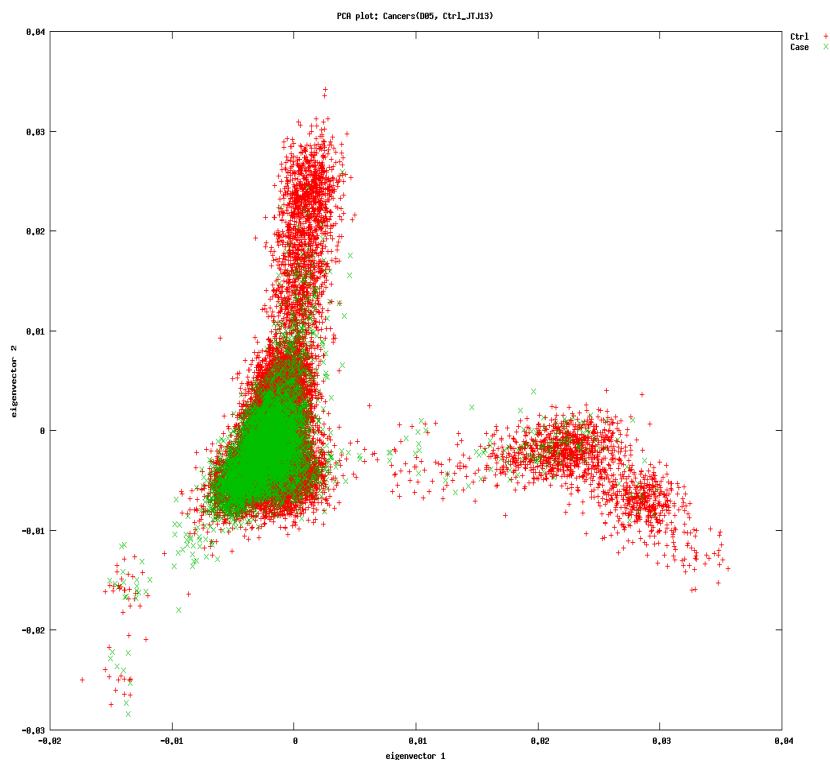
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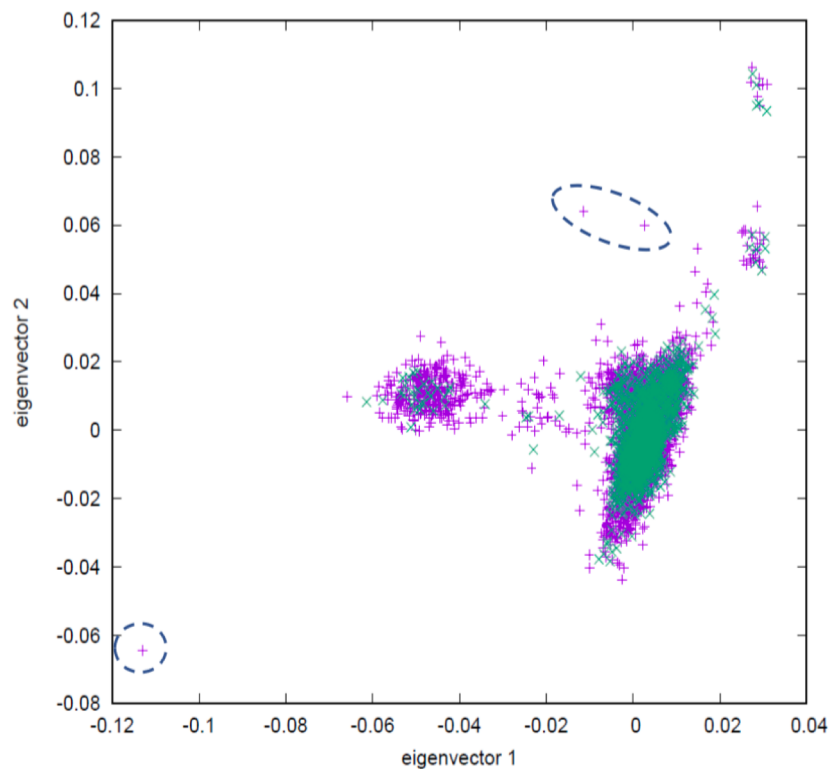
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GWAS of Phase I+II

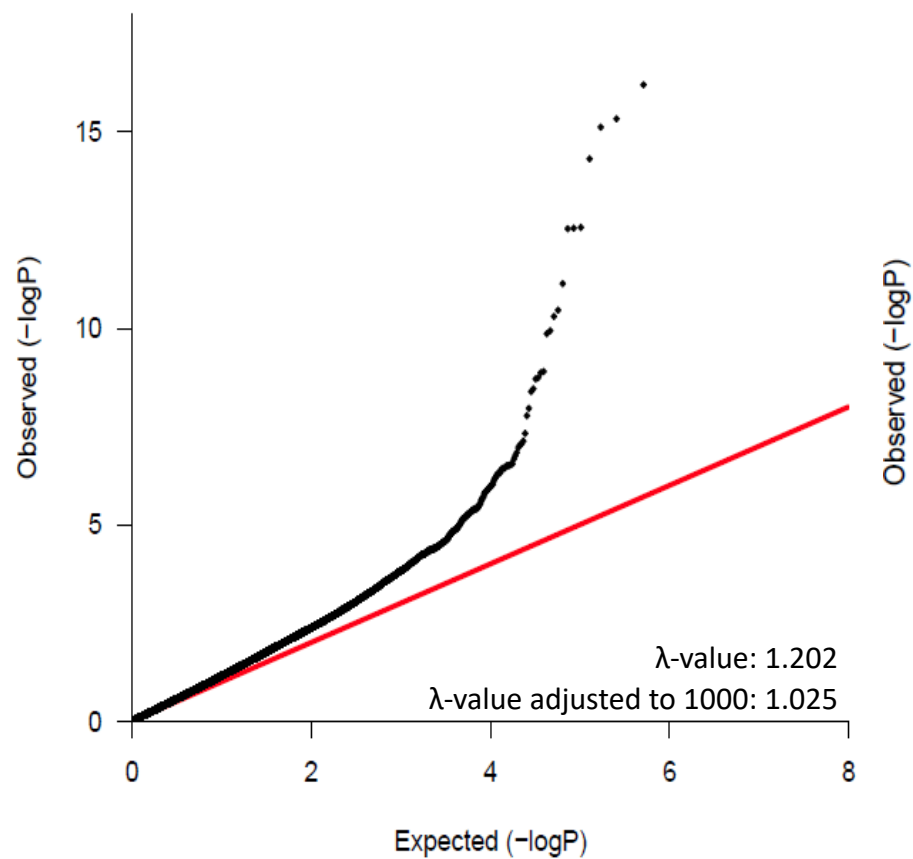


GWAS of Phase III

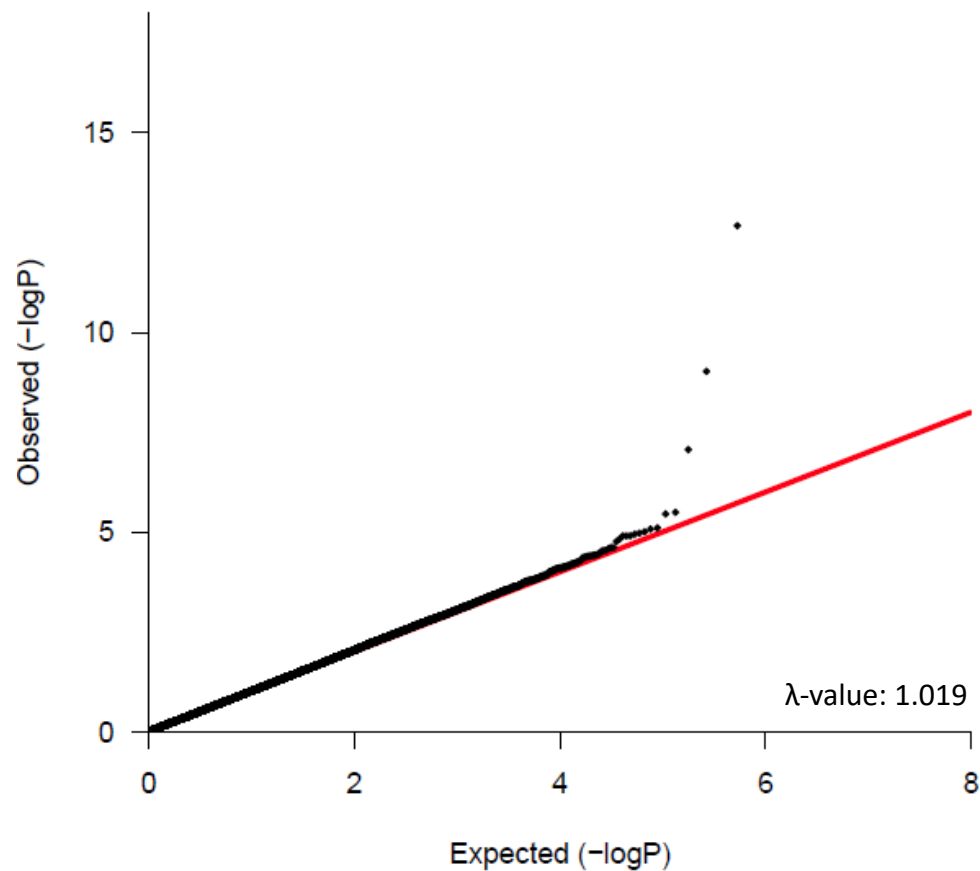


Supplementary Figure 1: Principal component analysis to evaluate population structure for GWAS of breast cancer in Japanese population. From the results of PCA analysis for GWAS Phase III, three control outliers were excluded from the study.

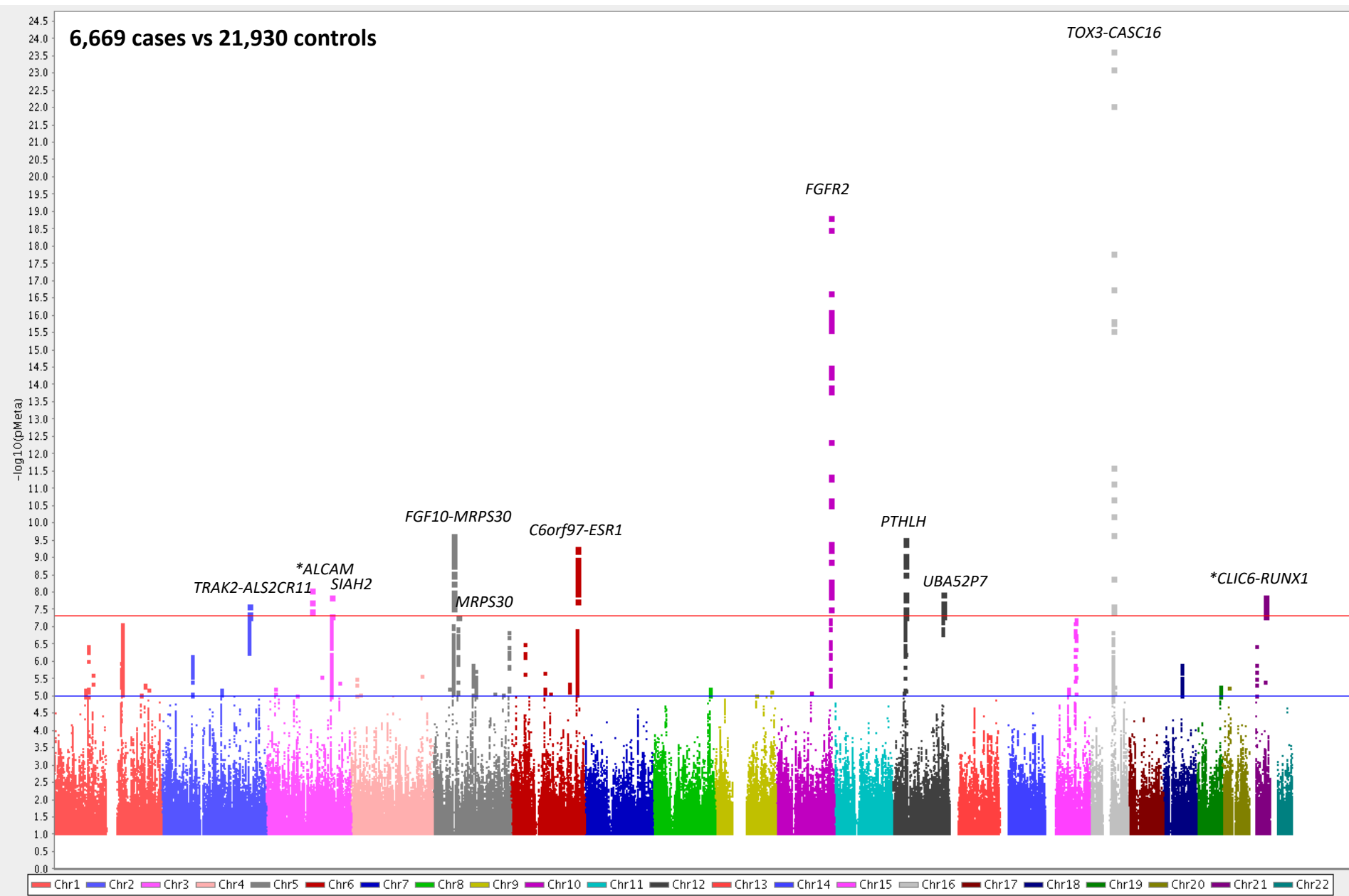
GWAS of Phase I+II



GWAS of Phase III

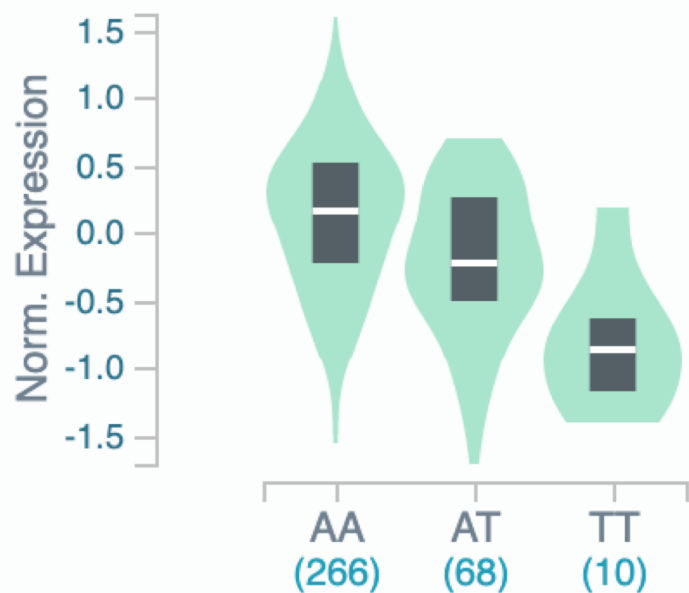


Supplementary Figure 2: Quantile-quantile plots for the GWAS of breast cancer.

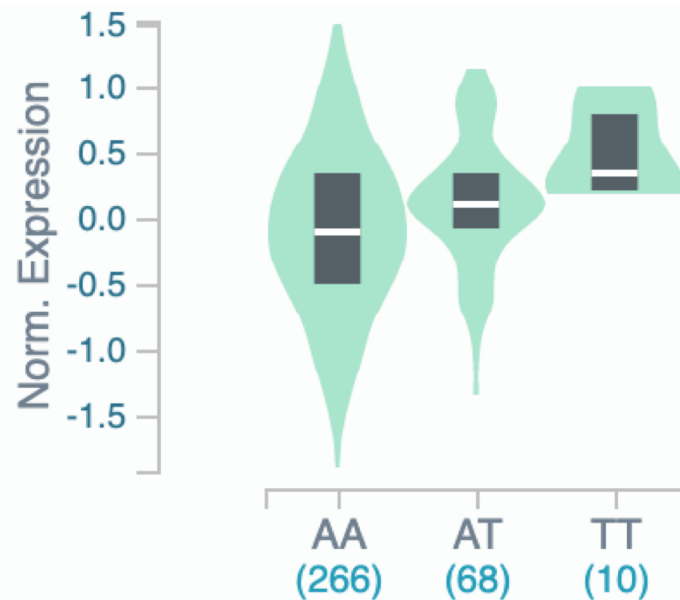


Supplementary Figure 3: Manhattan plot for genome-wide meta-analyses and 1000genome imputation analysis for the GWAS of breast cancer in Japanese population. *indicate novel loci identified from this study.

CASP8
rs2714486
Breast – Mammary Tissue



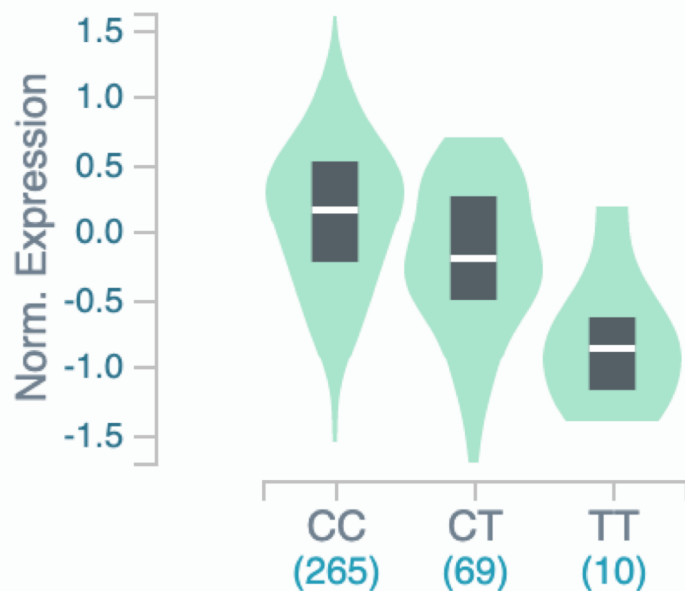
ALS2CR12
rs2714486
Breast – Mammary Tissue



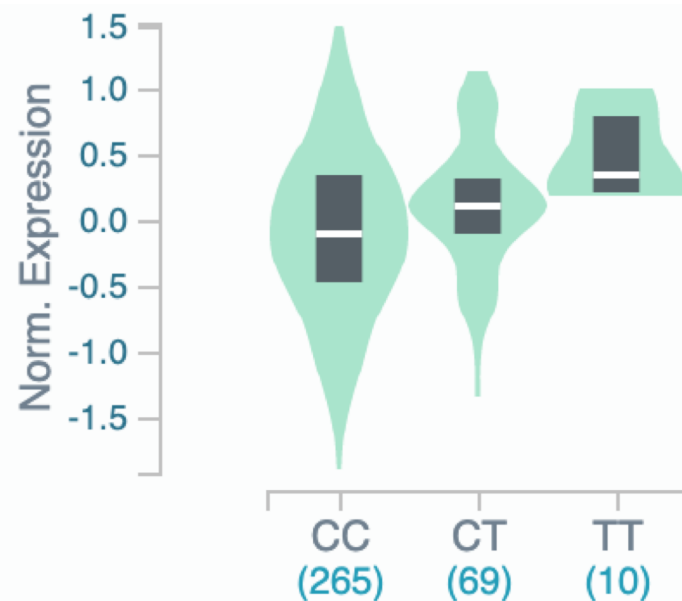
Gencode Id	Gene Symbol	Variant Id	SNP Id	P-Value	NES	Tissue
ENSG00000064012.21	<i>CASP8</i>	chr2_201384985_A_T_b38	rs2714486	4.20E-12	-0.44	Breast - Mammary Tissue
ENSG00000155749.12	<i>ALS2CR12</i>	chr2_201384985_A_T_b38	rs2714486	5.20E-07	0.35	Breast - Mammary Tissue

Supplementary Figure 4a: GTEx eQTL results of rs2540431-linked SNP rs2714486 in breast mammary tissue. rs2714486-A is the risk allele. Number of samples per genotype is stated in brackets.

CASP8
rs2540334
Breast – Mammary Tissue

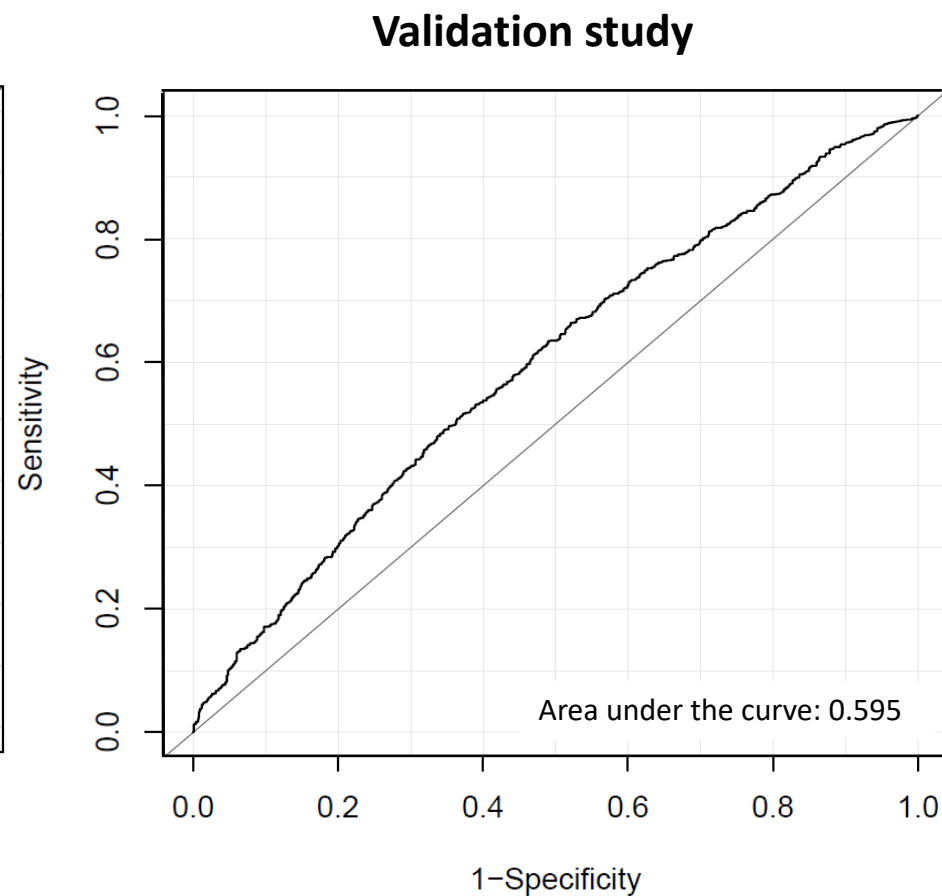
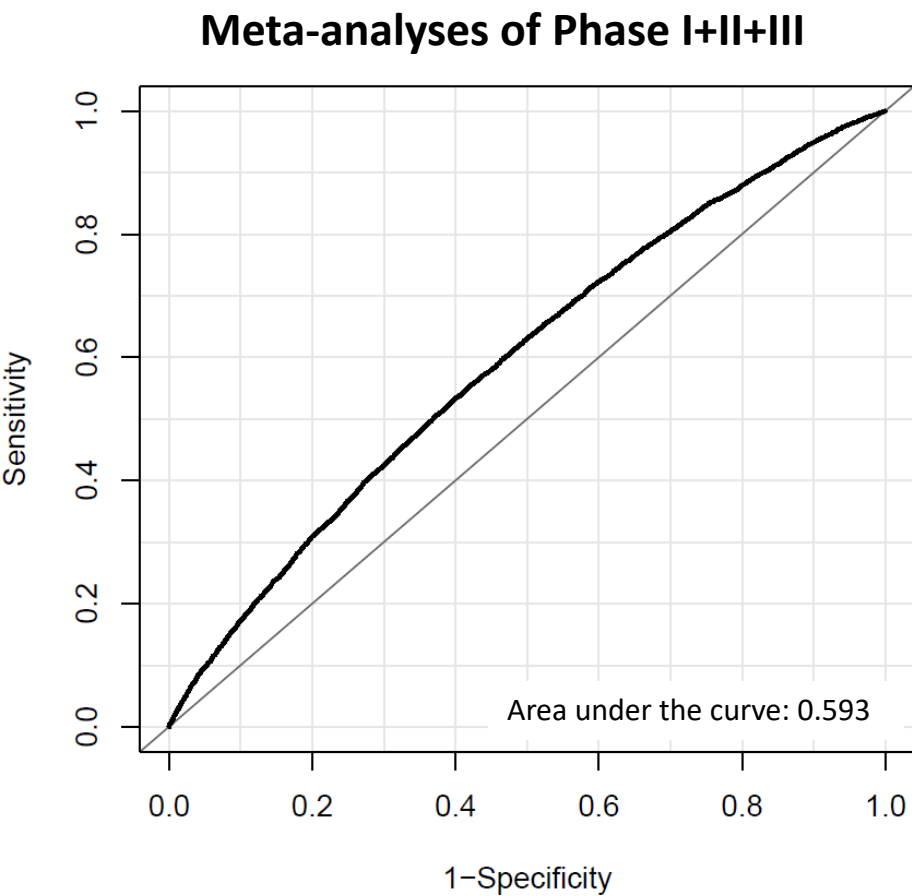


ALS2CR12
rs2540334
Breast – Mammary Tissue



Gencode Id	Gene Symbol	Variant Id	SNP Id	P-Value	NES	Tissue
ENSG00000064012.21	CASP8	chr2_201406624_C_T_b38	rs2540334	6.90E-12	-0.43	Breast - Mammary Tissue
ENSG00000155749.12	ALS2CR12	chr2_201406624_C_T_b38	rs2540334	7.80E-07	0.34	Breast - Mammary Tissue

Supplementary Figure 4b: GTEx eQTL results of rs2540431-linked SNP rs2540334 in breast mammary tissue. rs2540334-T is the risk allele. Number of samples per genotype is stated in brackets.



Supplementary Figure 5: ROC curve of wGRS prediction model to evaluate the cumulative effects of 12 significantly associated SNPs.