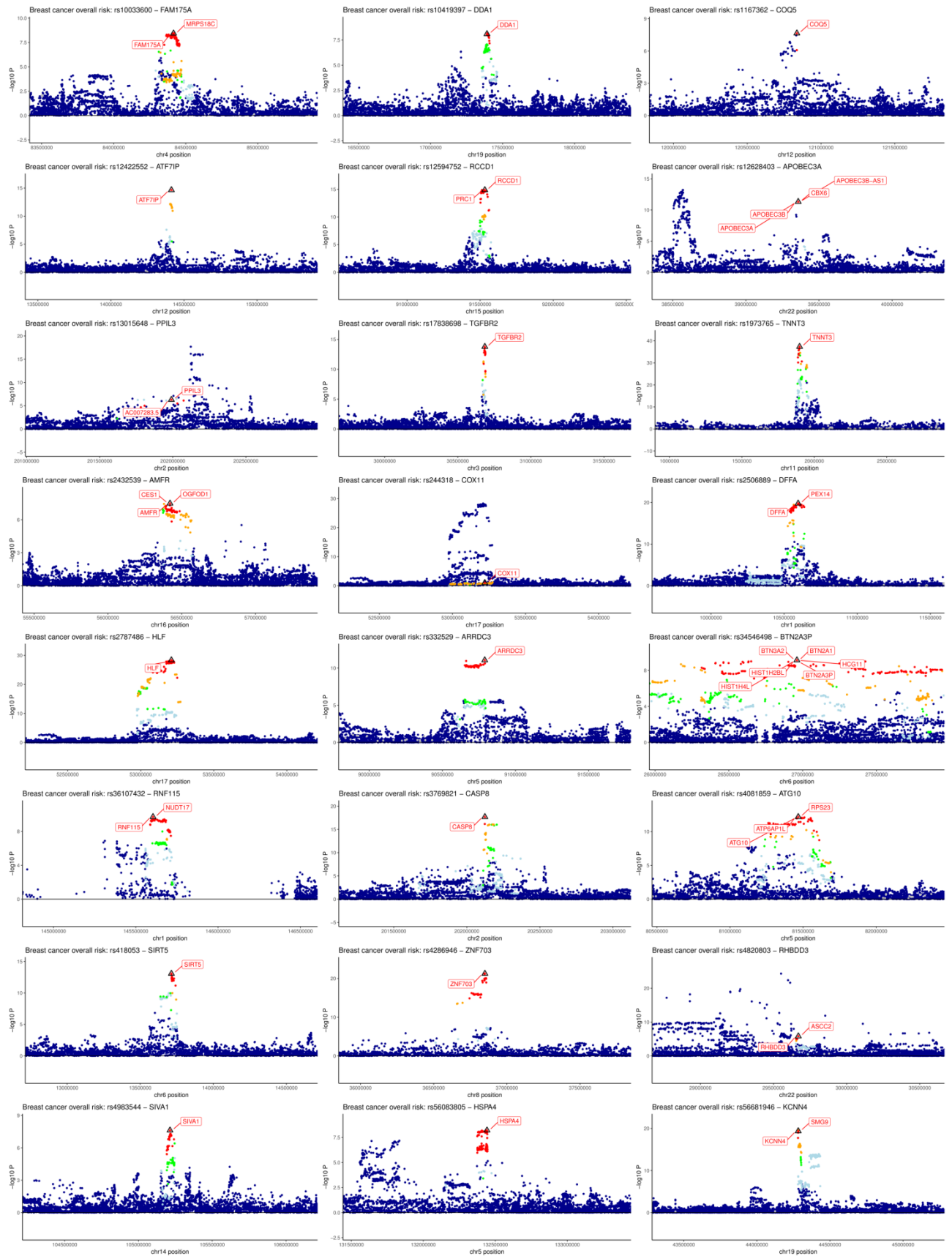
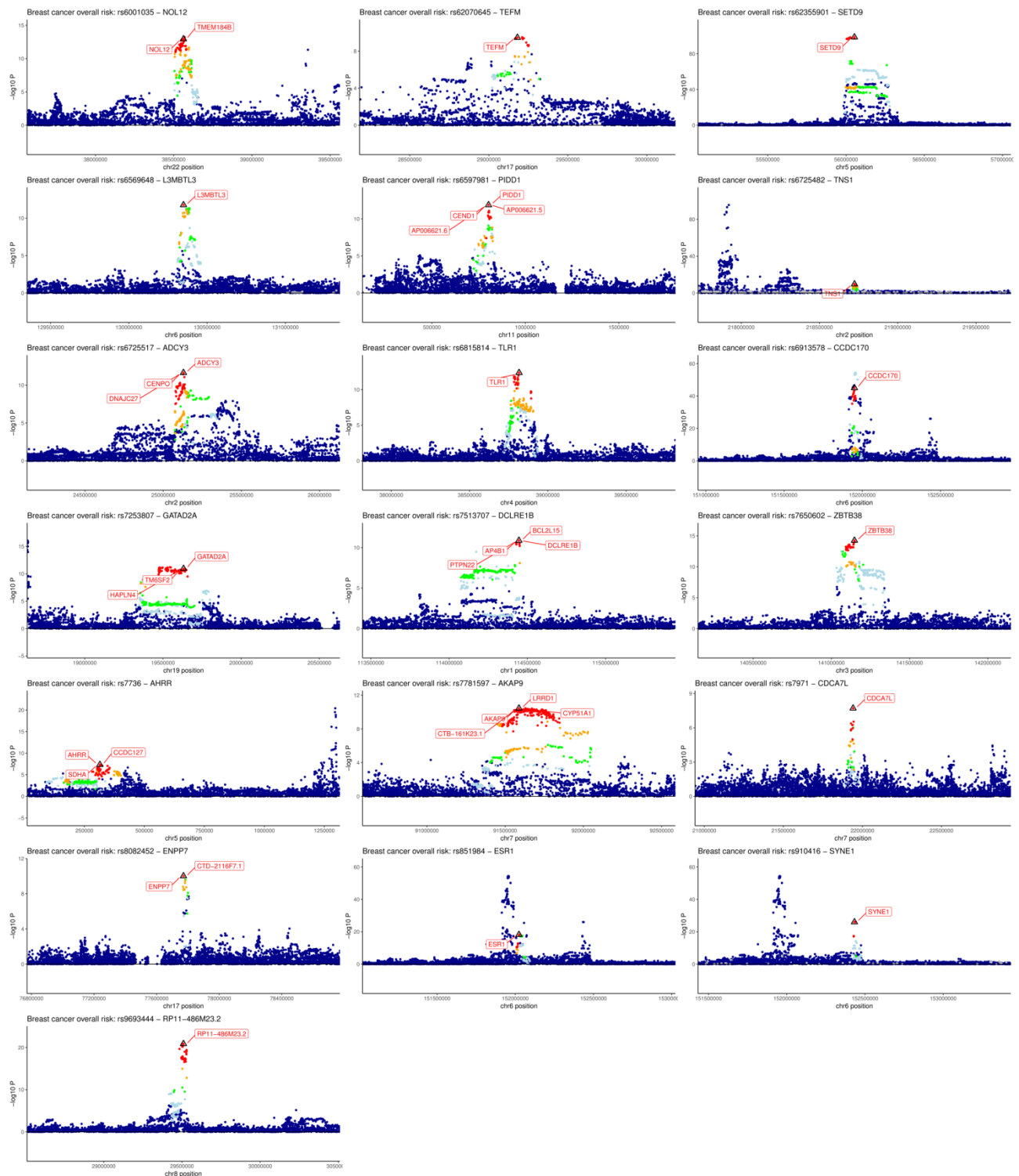


Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer

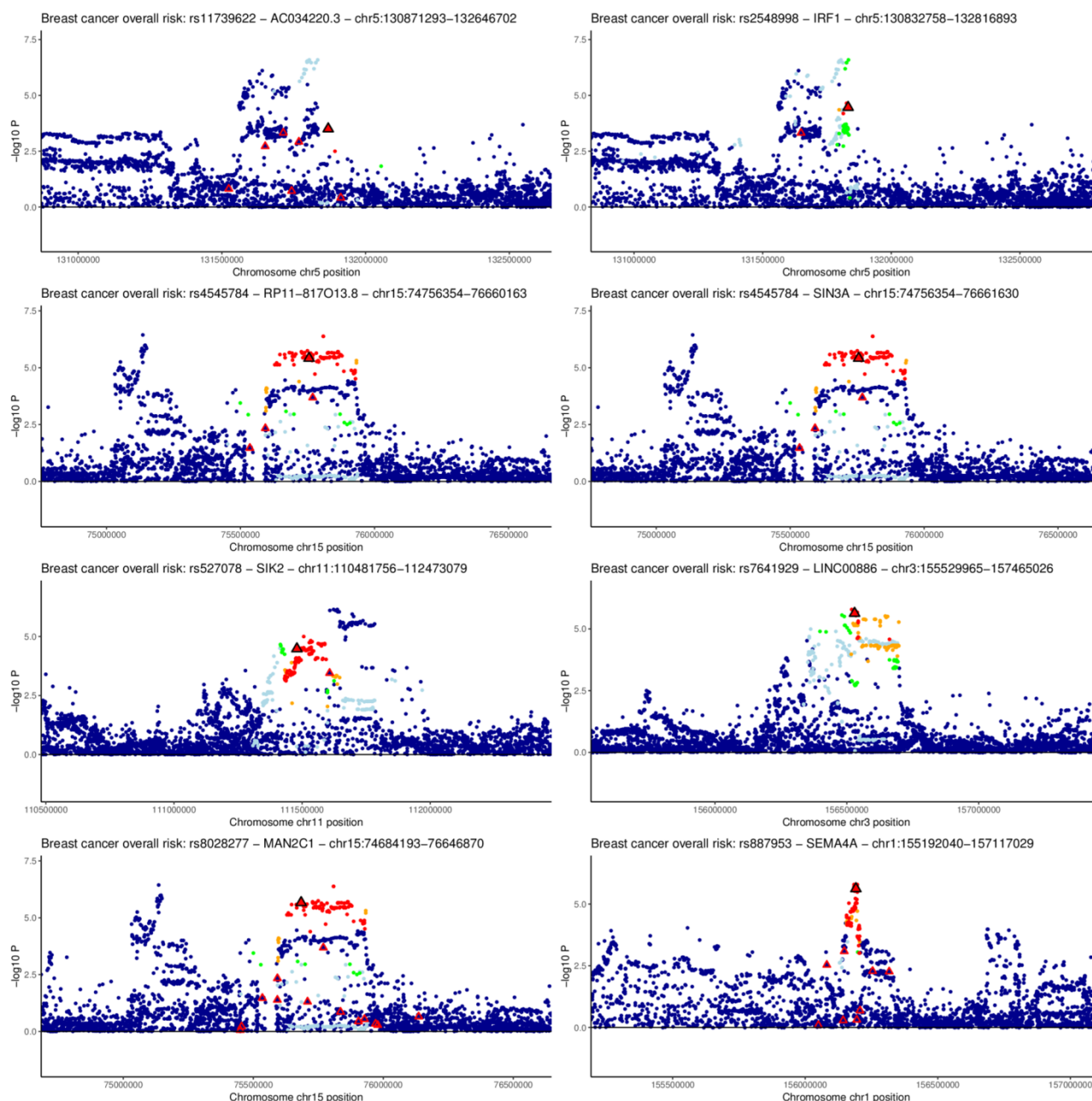
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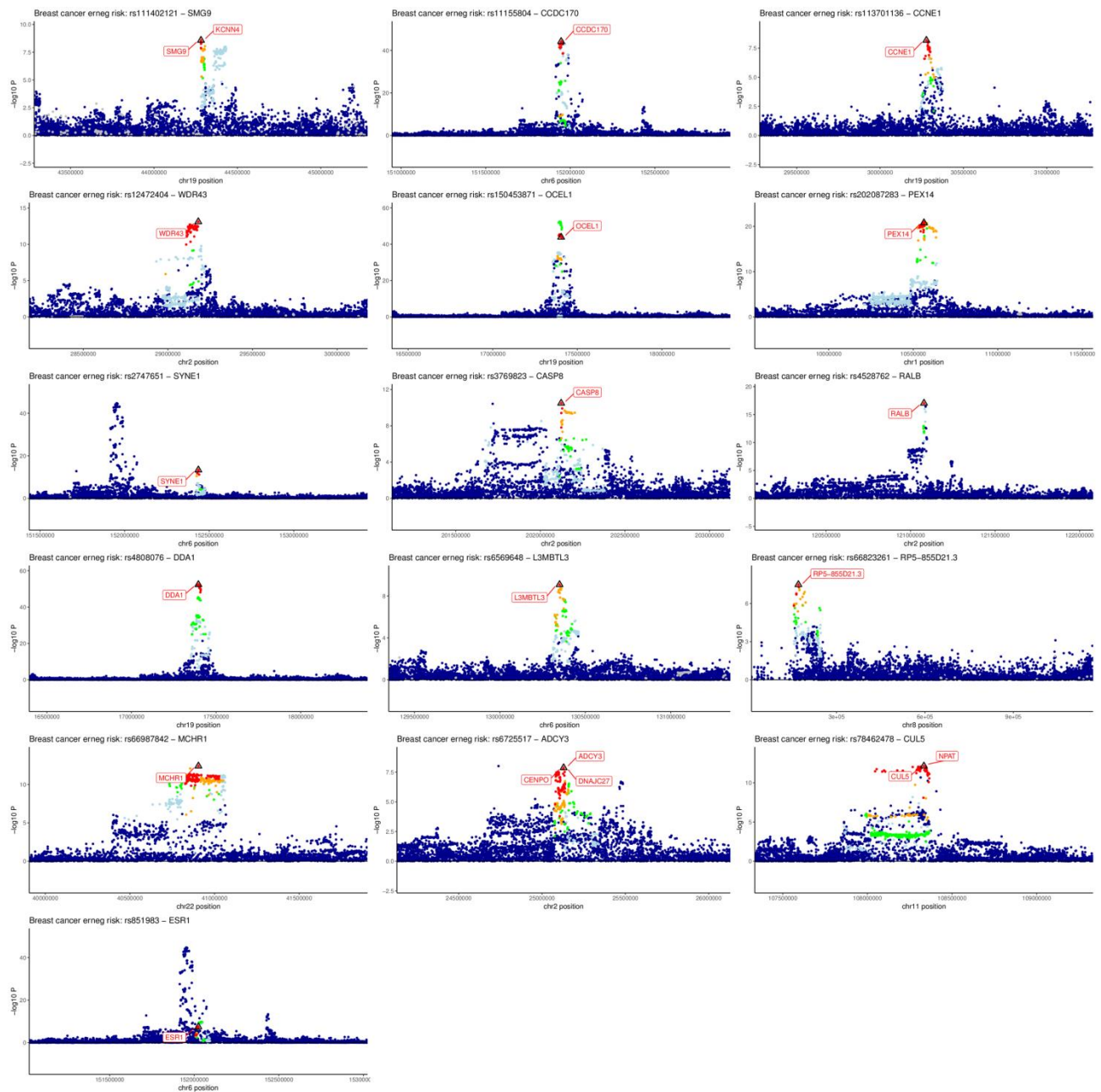
Supplementary Figure 1. Previously unreported target gene predictions at known risk loci for overall breast cancer.

Variants are represented by points coloured according to the LD with the sentinel risk variant (red: ≥ 0.8 , orange: 0.6-0.8, green: 0.4-0.6, light blue: 0.2-0.4, and dark blue: < 0.2). Sentinel risk variants (triangles) were identified based on joint association analysis¹. Figure shows on the y-axis the evidence for breast cancer association ($-\log_{10}$ of the P-value in the original published GWAS results²), and on the x-axis chromosomal position.



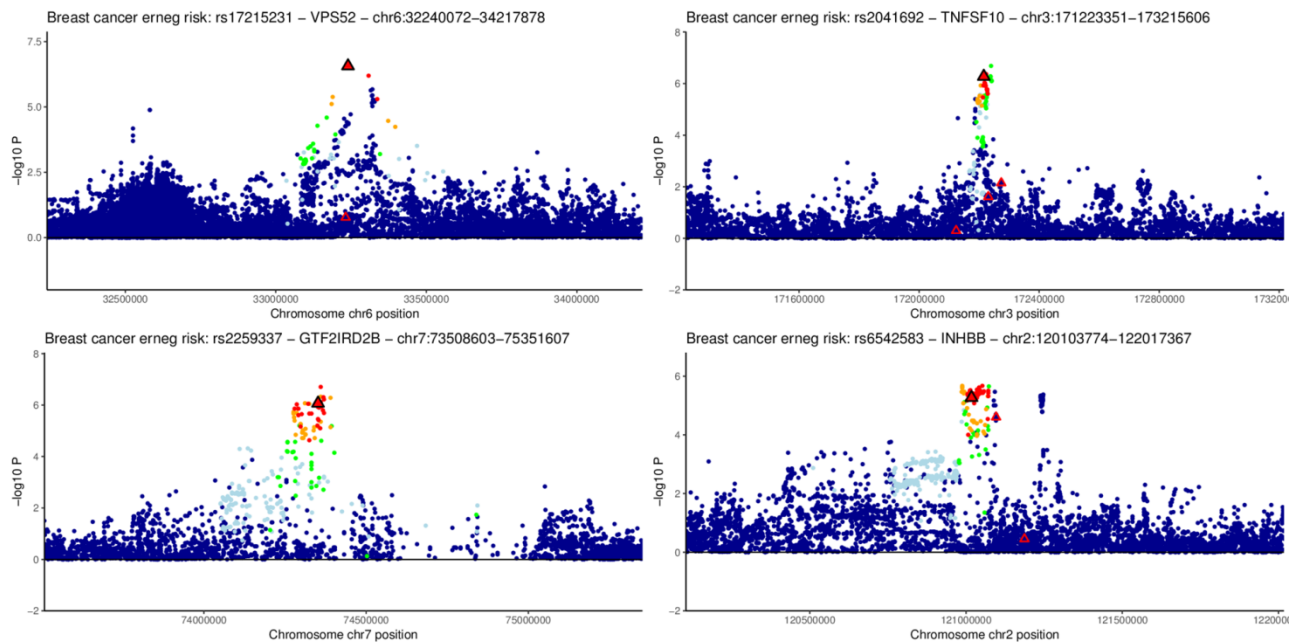
Supplementary Figure 2. Significant gene-based associations at previously unreported risk loci for overall breast cancer.

Variants are represented by points coloured according to the LD with the sentinel risk variant (red: ≥ 0.8 , orange: 0.6-0.8, green: 0.4-0.6, light blue: 0.2-0.4, and dark blue: < 0.2). Sentinel eQTL included in the EUGENE analysis (triangles) were identified from published eQTL studies of five different tissue types. Figure shows on the y-axis the evidence for breast cancer association ($-\log_{10}$ of the P-value in the published GWAS after adjusting for the association with the sentinel risk variants), and on the x-axis chromosomal position. The sentinel eQTL most associated with breast cancer risk is depicted by a black triangle; other sentinel eQTL are depicted by red triangles.



Supplementary Figure 3. Previously unreported target gene predictions at known risk loci for ER-negative breast cancer.

Variants are represented by points coloured according to the LD with the sentinel risk variant (red: ≥ 0.8 , orange: 0.6-0.8, green: 0.4-0.6, light blue: 0.2-0.4, and dark blue: < 0.2). Sentinel risk variants (triangles) were identified based on joint association analysis¹. Figure shows on the y-axis the evidence for breast cancer association ($-\log_{10}$ of the P-value in the original published GWAS results³), and on the x-axis chromosomal position.



Supplementary Figure 4. Significant gene-based associations at previously unreported risk loci for ER-negative breast cancer.

Variants are represented by points coloured according to the LD with the sentinel risk variant (red: ≥ 0.8 , orange: 0.6-0.8, green: 0.4-0.6, light blue: 0.2-0.4, and dark blue: < 0.2). Sentinel risk variants (triangles) were identified based on joint association analysis¹. Figure shows on the y-axis the evidence for ER-negative breast cancer association ($-\log_{10}$ of the P-value in the original published GWAS results³), and on the x-axis chromosomal position.

Supplementary References

1. Yang, J. *et al.* Conditional and joint multiple-SNP analysis of GWAS summary statistics identifies additional variants influencing complex traits. *Nat Genet* **44**, 369-75, S1-3 (2012).
2. Michailidou, K. *et al.* Association analysis identifies 65 new breast cancer risk loci. *Nature* **551**, 92-94 (2017).
3. Milne, R.L. *et al.* Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genet* **49**, 1767-1778 (2017).