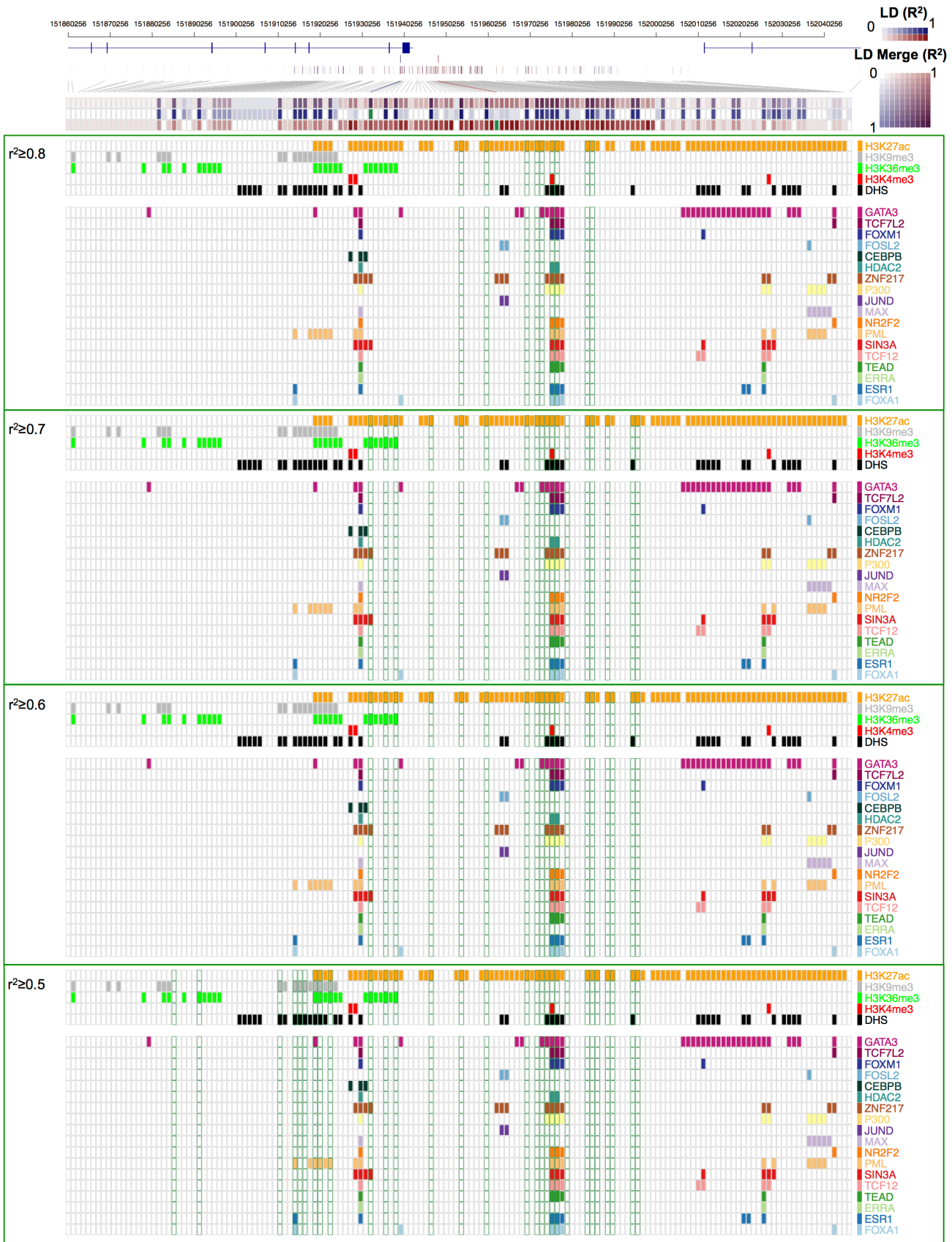
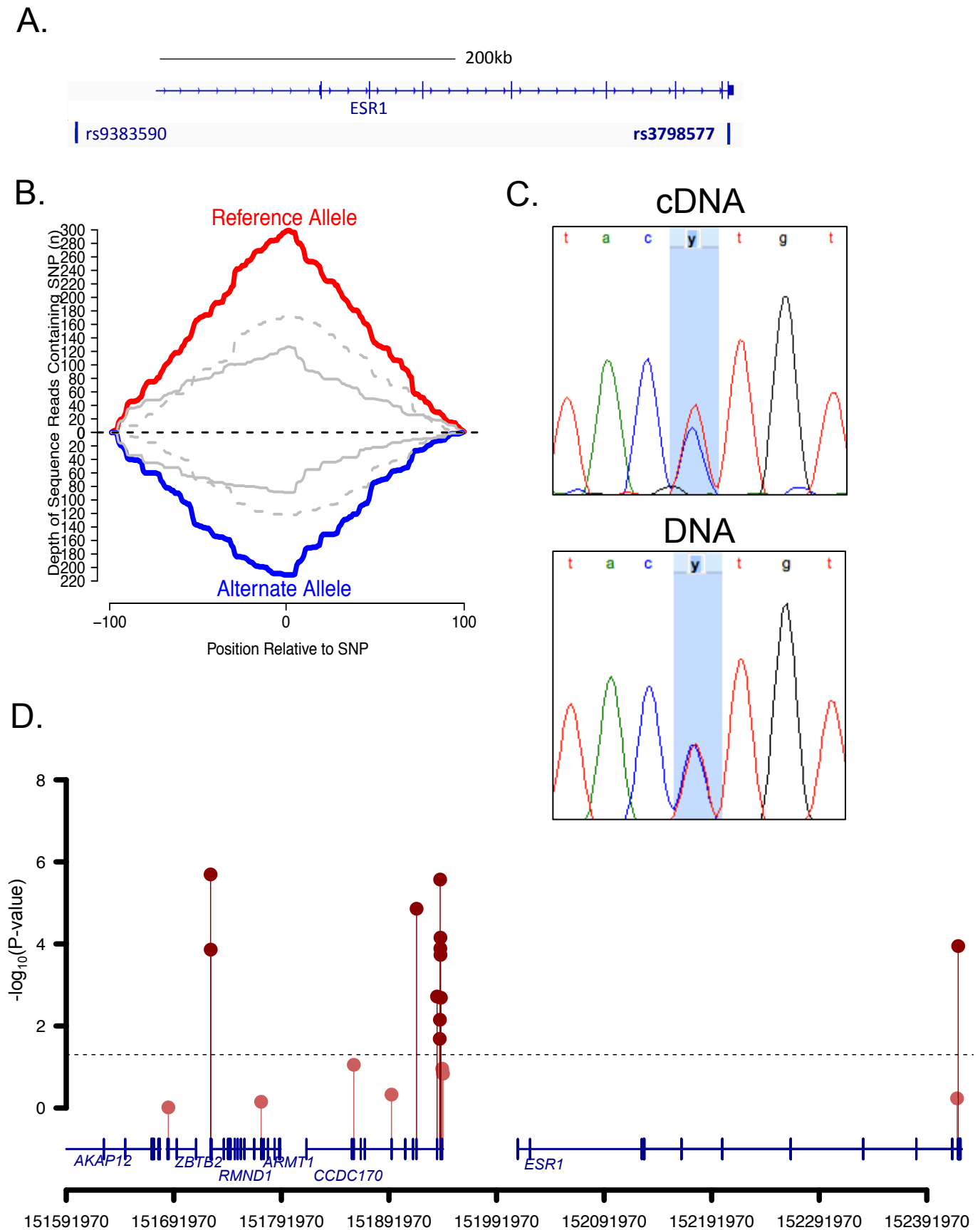


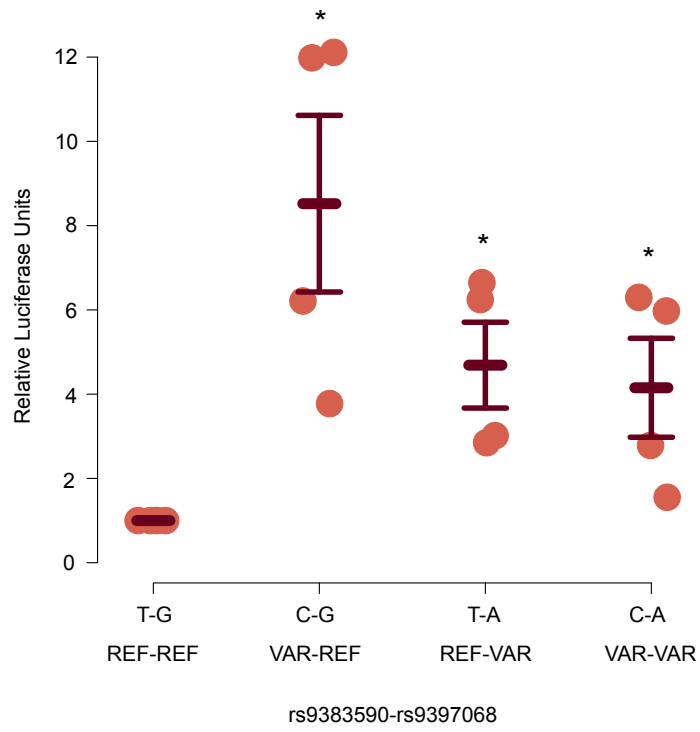
Supplementary Figure 1: Schematic representation of the population-specific LD merge and overlapping chromatin annotation data presented in Figure 1 and Supplementary Figure 2. The LD between the population-specific lead SNVs is calculated in the appropriate reference sample (blue = Europeans and red = East Asians) and merged (purple). The population-specific lead SNVs are filled in green in their respective plots. SNVs in strong LD with the lead SNVs in each separate population are outlined in green. The chromatin features overlapping each SNV in the region (top) are presented as boxes (bottom) below the LD merge.



Supplementary Figure 2: The shared linkage disequilibrium (LD) between the European and East Asian lead SNVs. The combined strength of the LD (LD Merge) between nearby SNVs and the European and East Asian lead SNVs is shown (purple). The European LD (LD EUR) pattern for the rs3734805 SNVs (blue) and the East Asian LD (LD EAS) pattern for rs2046210 (red) are shown. The squares corresponding to the population-specific lead SNVs (rs3734805 and rs2046210) are filled in green. Each panel represents different LD thresholds ($r^2 \geq 0.8, 0.7, 0.6$ and 0.5 , respectively). The SNVs passing the threshold in both the European and the East Asian populations are outlined in green boxes. The overlap of the SNVs with functional annotations (DHS, histones modifications and transcription factor binding) observed in breast cancer cells (MCF-7 or T-47D) profiled by the ENCODE project a represented as boxes and coloured according to the legend (right).



Supplementary Figure 3: Verification of the allele-biased expression of *ESR1* in HCC1419 cells. A) Location of the rs9383590 SNP and the marker rs3798577 SNP. B) Observed distribution of RNA-Seq reads containing each allele of the rs3798577 SNP. The pass both the strand distribution and the strand position tests. C) Sanger sequencing verification of allelic imbalance in *ESR1* expression in HCC1419 cells. D) Allelic imbalance of genes at the *ESR1* locus in the heterozygous HCC1419 cells. Heterozygous SNVs displaying a significant allelic imbalance (dark red) are shown together with their position relative to the genes (dark blue). Significance was determined using a binomial probability test. The dashed line indicates nominal significance ($p < 0.05$).



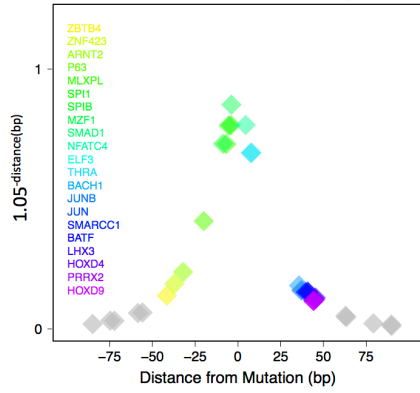
Supplementary Figure 4: Luciferase assays for the different rs9383590 and rs9397068 haplotypes. * indicates $p < 0.05$.



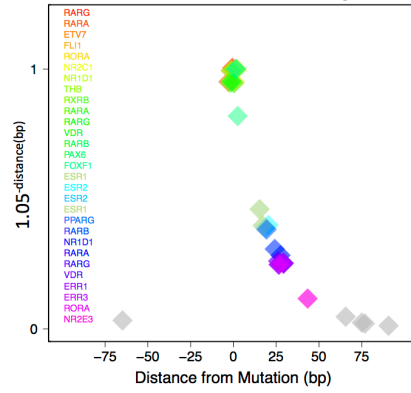
Supplementary Figure 5: Validation of the C3D predicted interactions at the *ESR1* locus. The C3D called interactions are shown in red. The ChIA-PET interactions are shown in purple. Only those interactions overlapping at least one end of the predicted interactions are shown. The 3C verified interactions are shown in pink.

A.

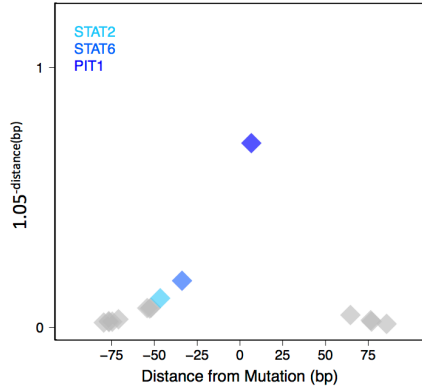
chr6:151923842:G>T (TNBC)



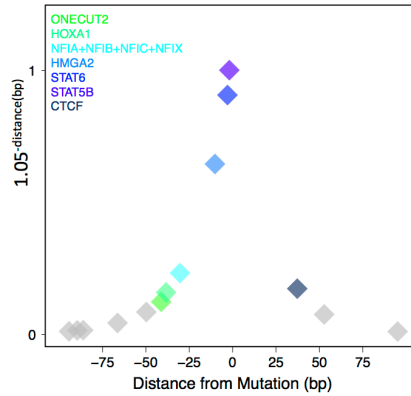
chr6:151937492:G>A (ESR1)



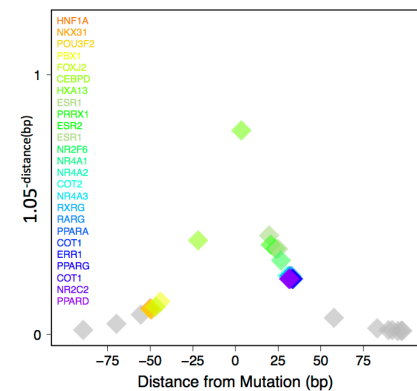
chr6:151937744:G>C (ESR1)



chr6:151954506:C>A (TNBC)

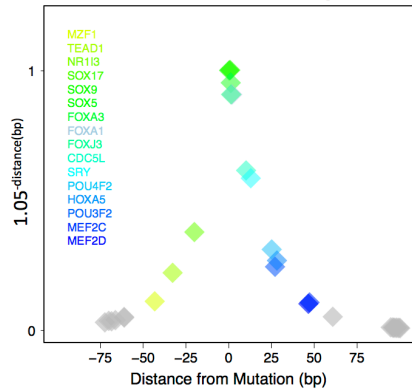


chr6:151979376:T>G (TNBC)

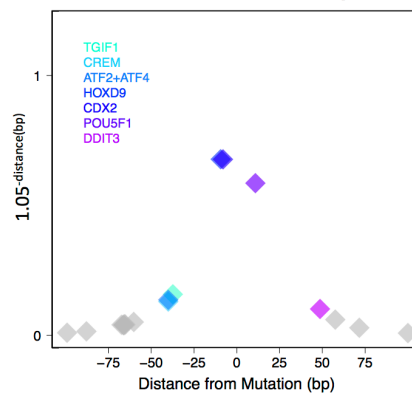


B.

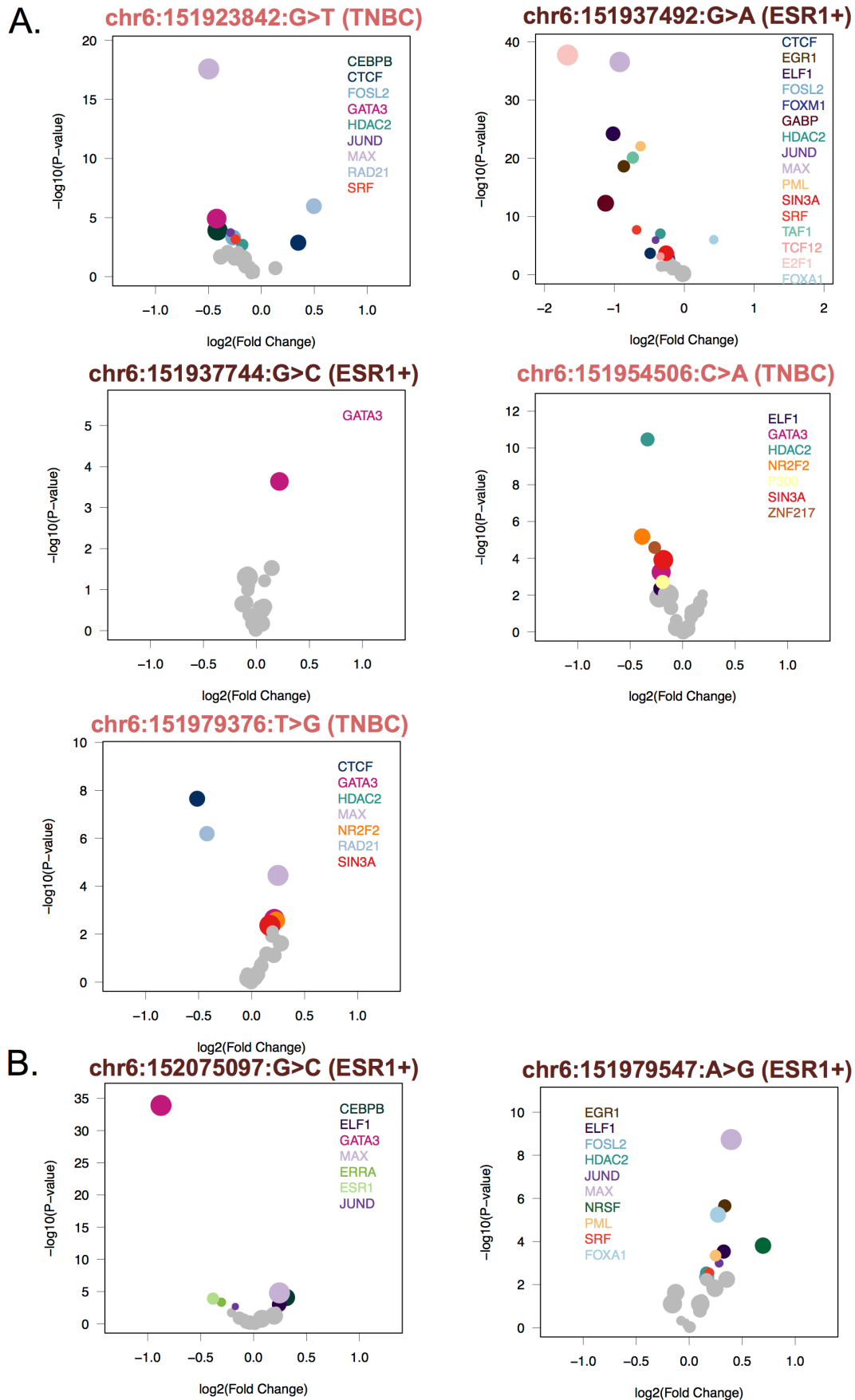
chr6:151979547:A>G (ESR1+)



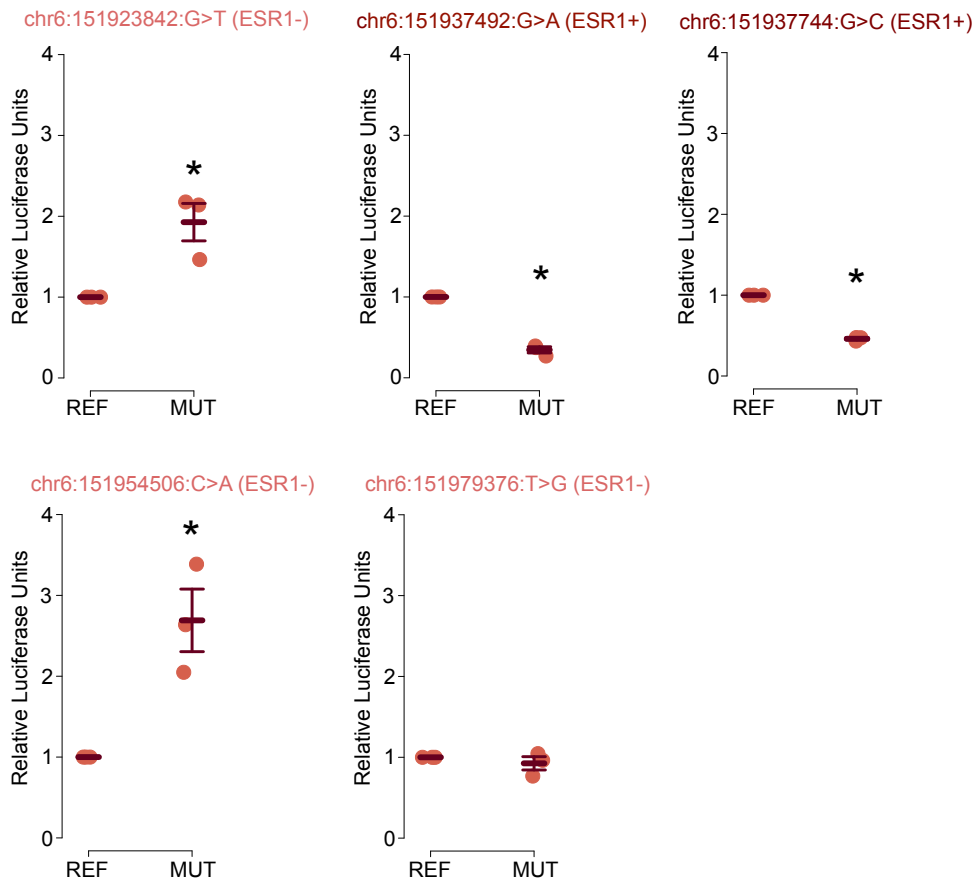
chr6:152075097:G>C (ESR1+)



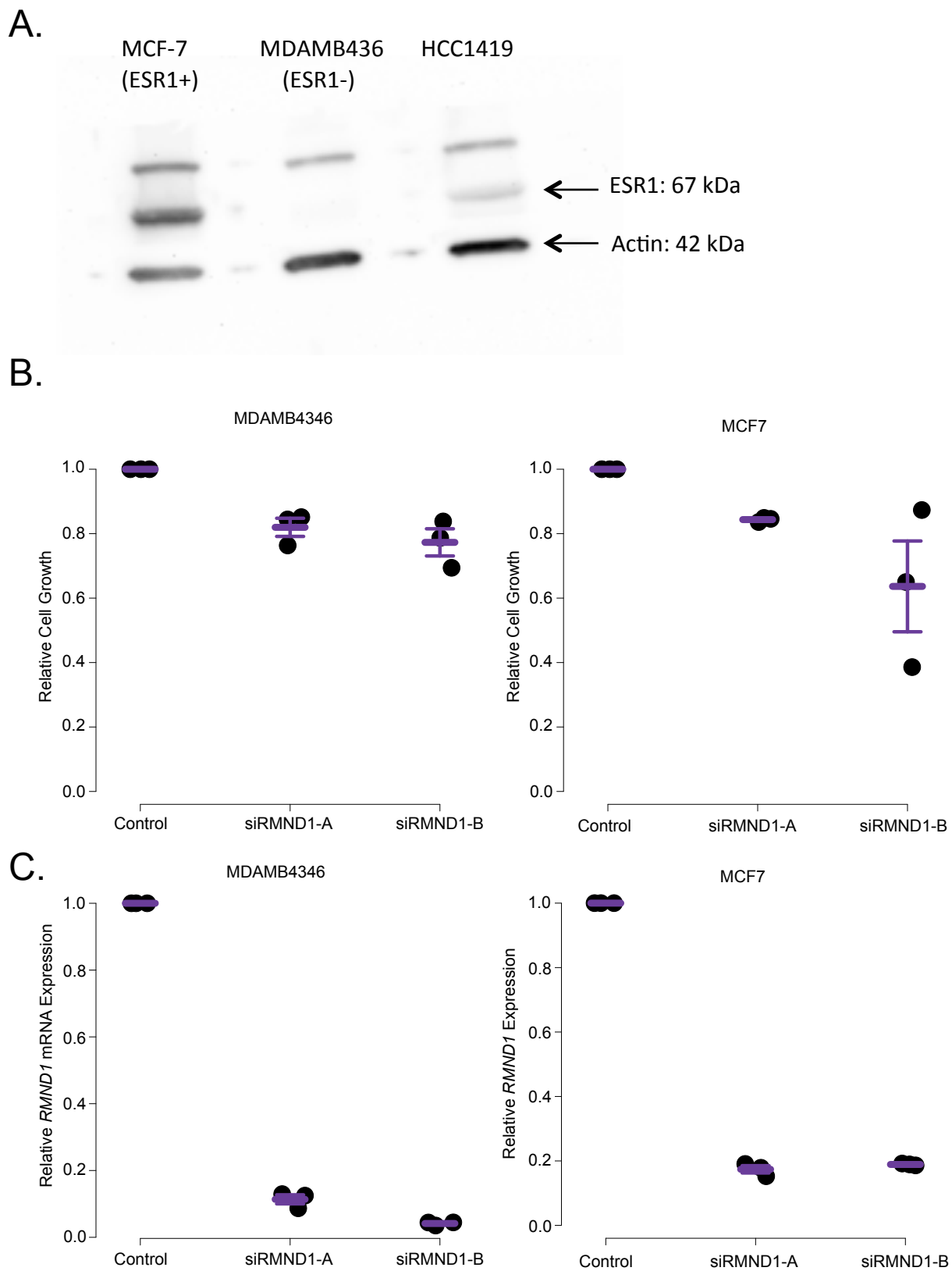
Supplementary Figure 6: The distance to the nearest transcription factor DNA recognition motifs of the mutations within the *ESR1* SRE excluded from Figure 4 for A) the mutations observed in the Alexandrov *et al.*²⁷ dataset and B) the mutations observed in the IMPACT/COMPACT samples. The y-axis is a function (1.05-distance) of the distance to the mutations to emphasize the closest motifs. Each diamond represents the location of a transcription factor motif.



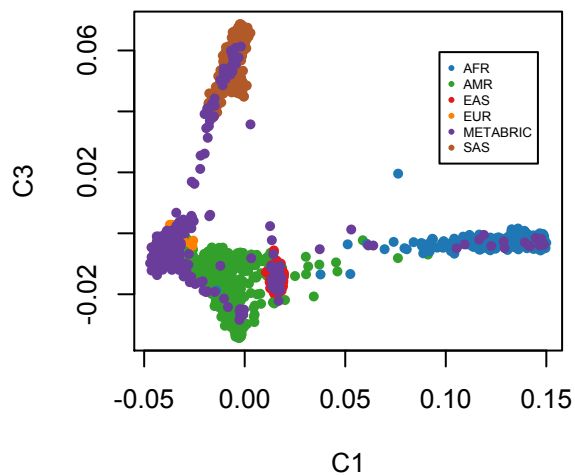
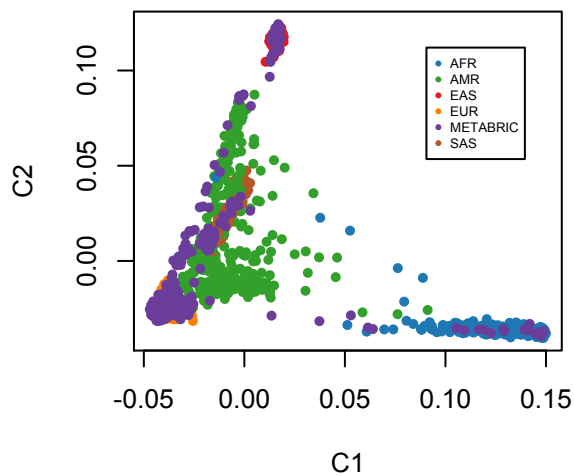
Supplementary Figure 7: Volcano plots of the IGR predicted modulation in chromatin binding intensity for transcription factors caused by the mutations in *ESR1*'s SRE excluded from Figure 4 for A) the mutations observed in the Alexandrov *et al.*²⁷ dataset and B) the mutations observed in the IMPACT/COMPACT samples.



Supplementary Figure 8: Luciferase assays for the mutations in *ESR1*'s SRE excluded from Figure 4 observed in the Alexandrov *et al.*²⁷ dataset.



Supplementary Figure 9: A) Western blot verification of the ESR1 protein expression status of HCC1419 and MDAMB436. MCF-7 are used as positive control. B) Proliferation assays following silencing of *RMND1* in HCC1419 and MDAMB436 cells. C) Verification of the reduced expression of *RMND1* following silencing.



EAS = East Asians

(CHB=Han Chinese in Beijing China, JPT = Japanese in Toyko Japan, CHS = Southern Han Chinese, CDX = Chinese Dai in Xishuangbanna China, KHV = Kinh in Ho Chi Minh City, Vietnam)

EUR = Europeans

(CEU = Utah Residents (CEPH) with Northern and Western European Ancestry, TSI = Toscani in Italia, FIN = Finnish in Finland, GBR = British in England and Scotland, IBS = Iberian Population in Spain)

AFR = Africans

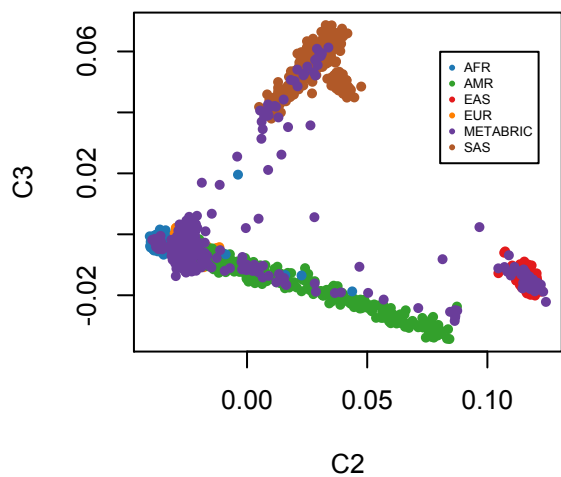
(YRI = Yoruba in Ibadan, Nigeria, LWK = Luhya in Webuye, Kenya , GWD = Gambian in Western Divisions in the Gambia, MSL= Mende in Sierra Leone, ESN = Esan in Nigeria, ASW = Americans of African Ancestry is SW USA, ACB = African Caribbeans in Barbados)

AMR = Americans

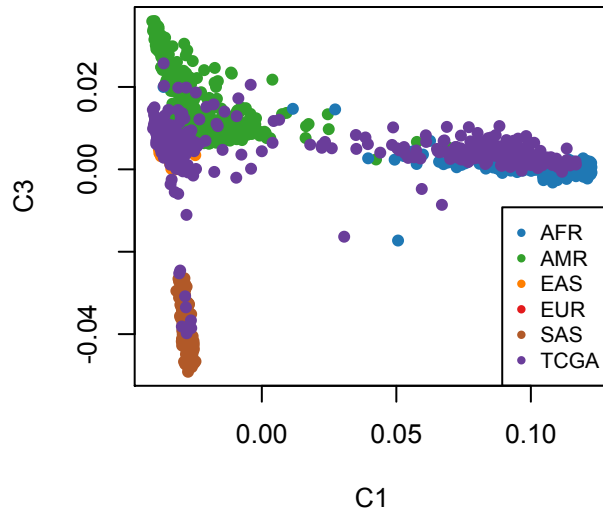
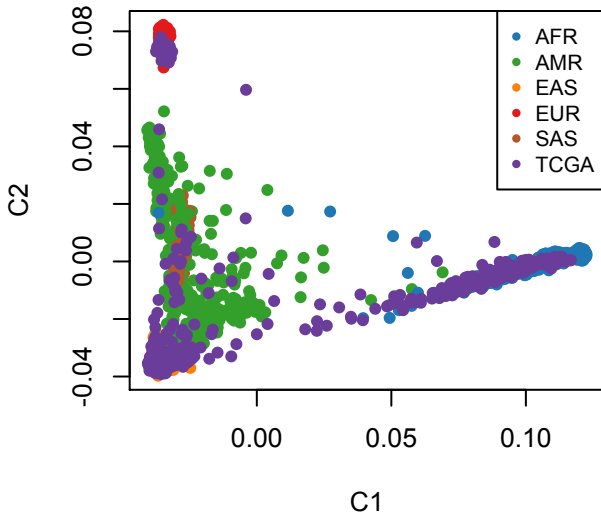
(MXL=Mexican Ancestry from Los Angeles USA, PUR=Puerto Ricans from Puerto Rico, CLM = Colombians from Medellin Colombia, PEL = Peruvians from Lima Peru);

SAS = South Asians

(GIH = Gujarati Indian from Houston Texas, PJI = Punjabi from Lashore, Pakistan, BEB =Bengali from Bengladesh, STU = Sri Lankan Tamil from the UK, ITU = Indian Tegulu from the UK).



Supplementary Figure 10: Verification of the ethnicity of the METABRIC tumour samples through multidimensional scaling of the genotype data. The genotype data of the METABRIC samples was merged with the data generated by the 1,000 genomes project. The METABRIC samples (purple) are shown with respect to the diverse populations samples profiled by the 1,000 genomes project. The 1,000 genomes samples are coloured according to their continental groups. The first three components are shown.



EAS = East Asians

(CHB=Han Chinese in Beijing China, JPT = Japanese in Toyko Japan, CHS = Southern Han Chinese, CDX = Chinese Dai in Xishuangbanna China, KHV = Kinh in Ho Chi Minh City, Vietnam)

EUR = Europeans

(CEU = Utah Residents (CEPH) with Northern and Western European Ancestry, TSI = Toscani in Italia, FIN = Finnish in Finland, GBR = British in England and Scotland, IBS = Iberian Population in Spain)

AFR = Africans

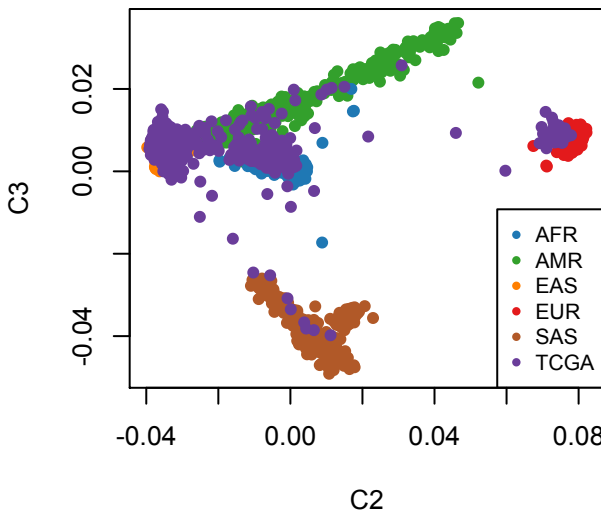
(YRI = Yoruba in Ibadan, Nigeria, LWK = Luhya in Webuye, Kenya , GWD = Gambian in Western Divisions in the Gambia, MSL= Mende in Sierra Leone, ESN = Esan in Nigeria, ASW = Americans of African Ancestry is SW USA, ACB = African Caribbeans in Barbados)

AMR = Americans

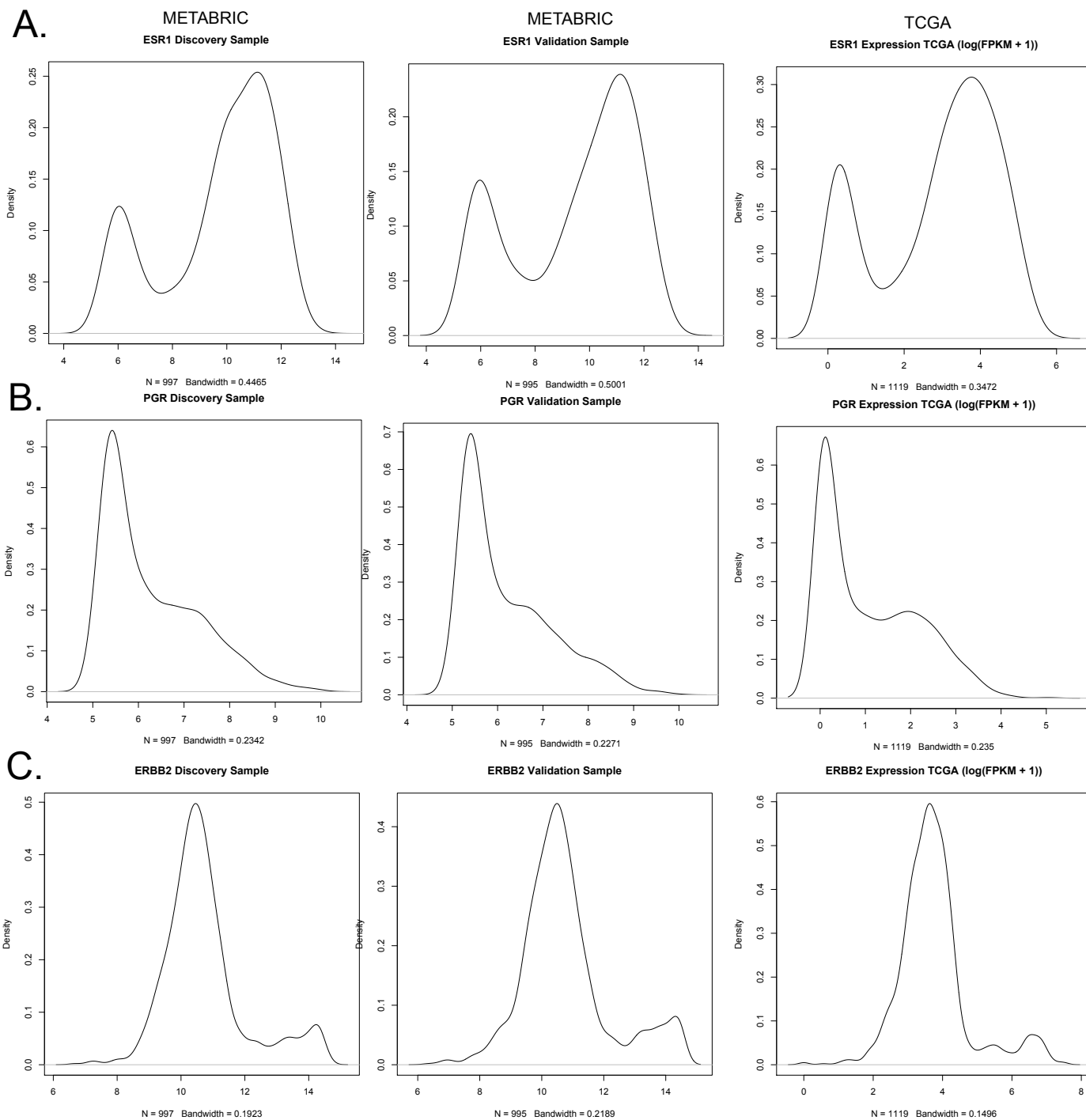
(MXL=Mexican Ancestry from Los Angeles USA, PUR=Puerto Ricans from Puerto Rico, CLM = Colombians from Medellin Colombia, PEL = Peruvians from Lima Peru);

SAS = South Asians

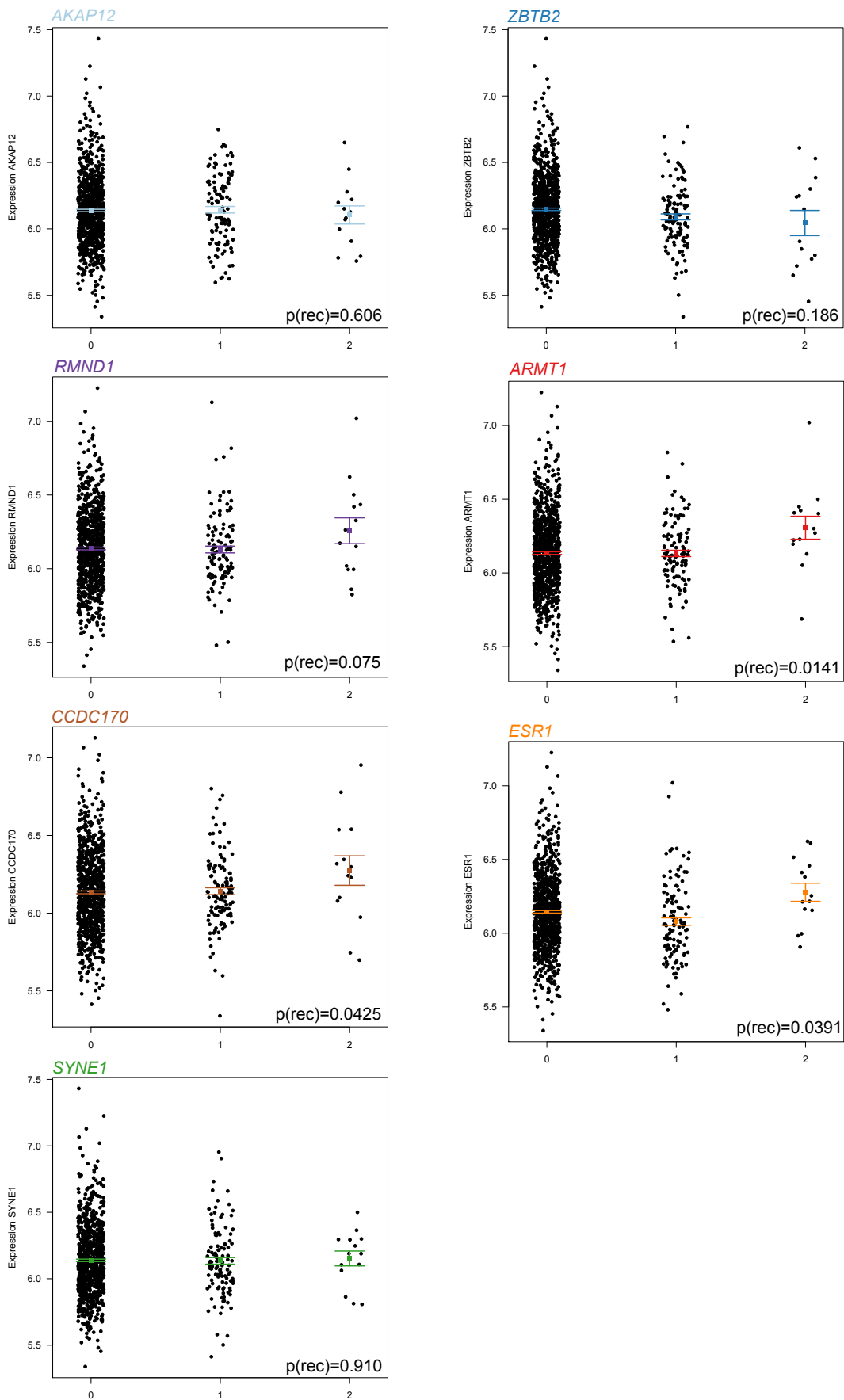
(GIH = Gujarati Indian from Houston Texas, PJI = Punjabi from Lashore, Pakistan, BEB =Bengali from Bengladesh, STU = Sri Lankan Tamil from the UK, ITU = Indian Tegulu from the UK).



Supplementary Figure 11: Verification of the ethnicity of the TCGA tumour samples through multidimensional scaling of the genotype data. The genotype data of the TCGA samples was merged with the data generated by the 1,000 genomes project. The TCGA samples (purple) are shown with respect to the diverse populations samples profiled by the 1,000 genomes project. The 1,000 genomes samples are coloured according to their continental groups. The first three components are shown.

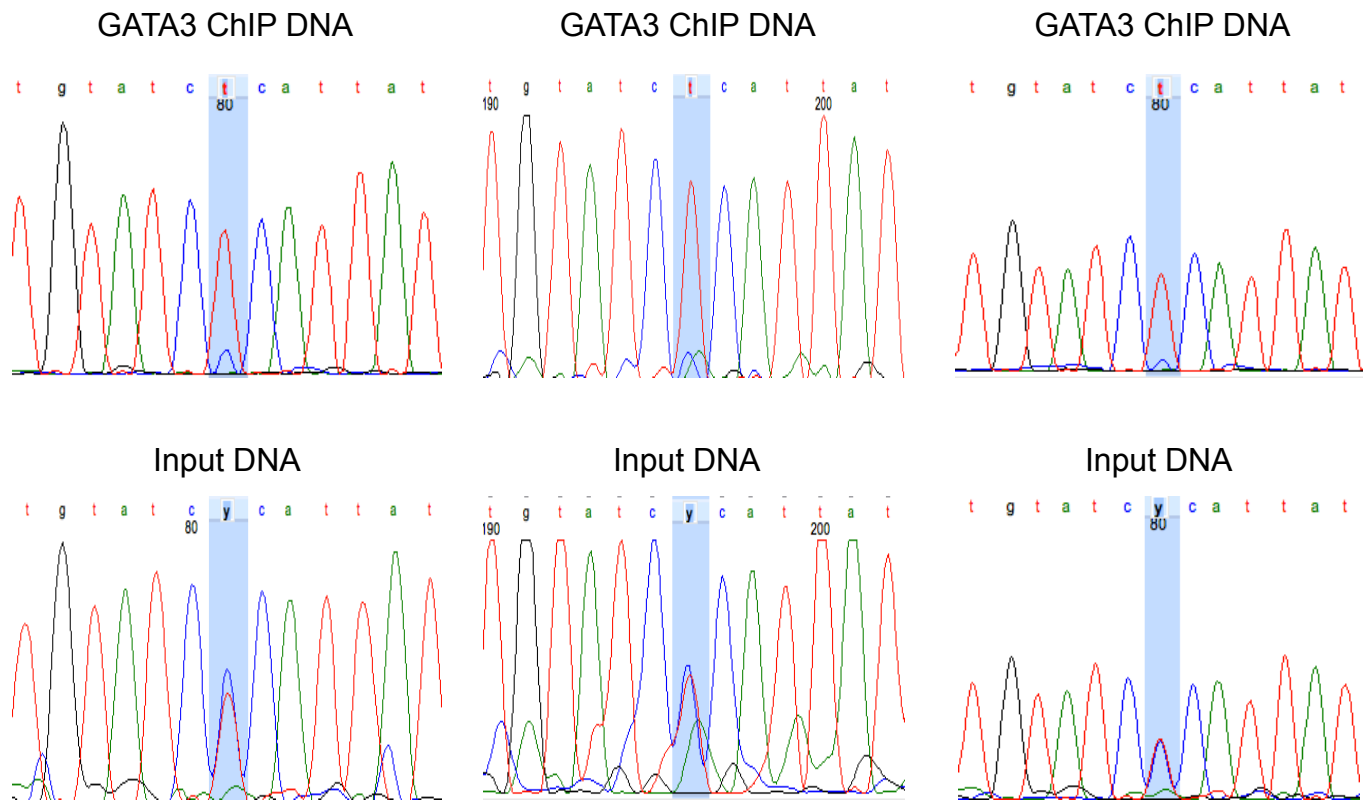


Supplementary Figure 12: The expression of the three breast cancer diagnostic genes (*ESR1*, *PGR*, *ERBB2*) among the METABRIC and TCGA samples. A) The expression of *ESR1* in the METABRIC discovery, METABRIC validation and TCGA samples, respectively. B) The expression of *PGR* in the METABRIC discovery, METABRIC validation and TCGA samples, respectively. The expression of *ERBB2* (HER2) in the METABRIC discovery, METABRIC validation and TCGA samples, respectively.

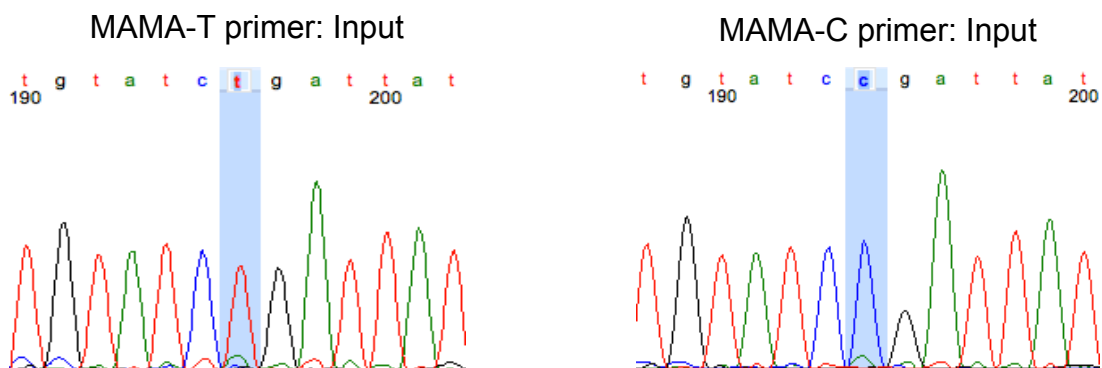


Supplementary Figure 13: Expression quantitative trait loci analysis of the genes at the *ESR1* locus among the METABRIC samples. The analysis is restricted to samples expressing both *ESR1* and *PGR*, but not overexpressing *ERBB2*. The expression value for each gene is plotted by the proxy SNV rs9397437 genotypes. The P-values were calculated using linear regression under a recessive model. The mean and standard error of the mean are shown. The reported p-values are two-sided.

A.

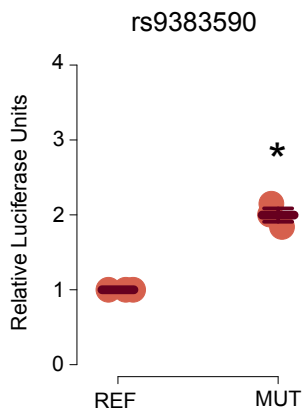


B.

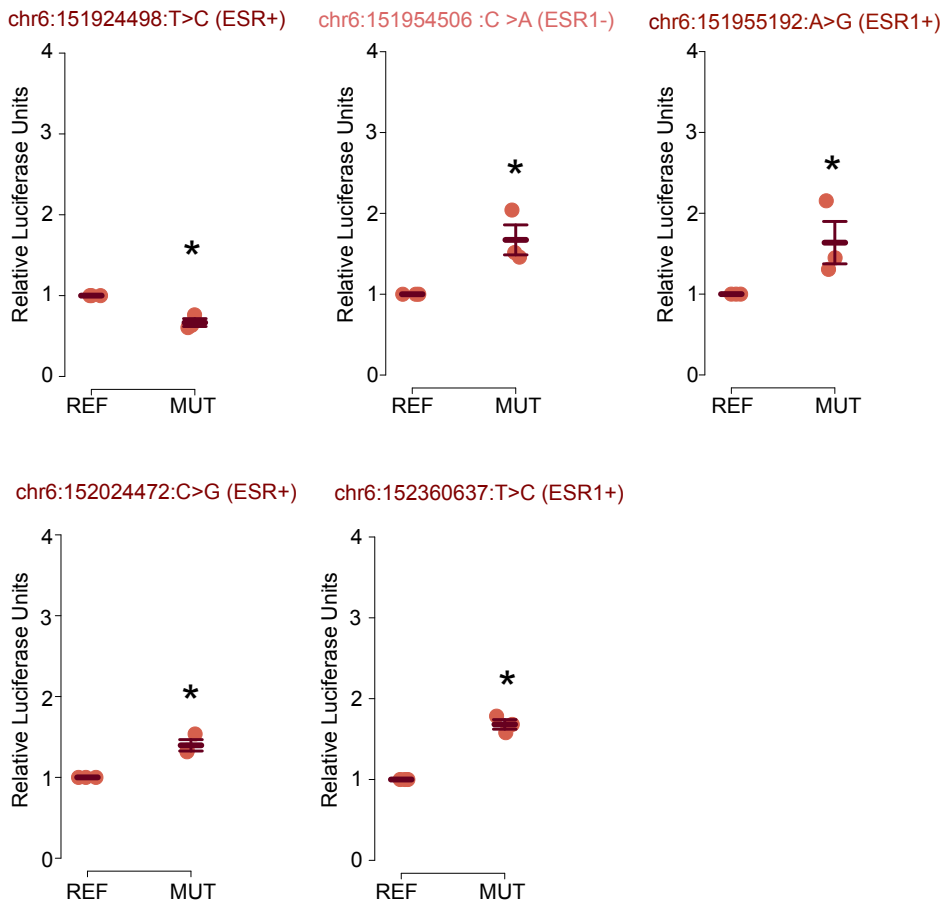


Supplementary Figure 14: A) Sanger sequencing validation of the allele-specific binding of GATA3 caused by the rs9383590 SNV in the heterozygous HCC1419 cells. Sequencing traces of the GATA3 ChIP DNA and input DNA are shown in triplicate. B) Sanger sequencing validation of the specificity of the MAMA primers used to assess the allele-specific binding of GATA3 in HCC1419 cells.

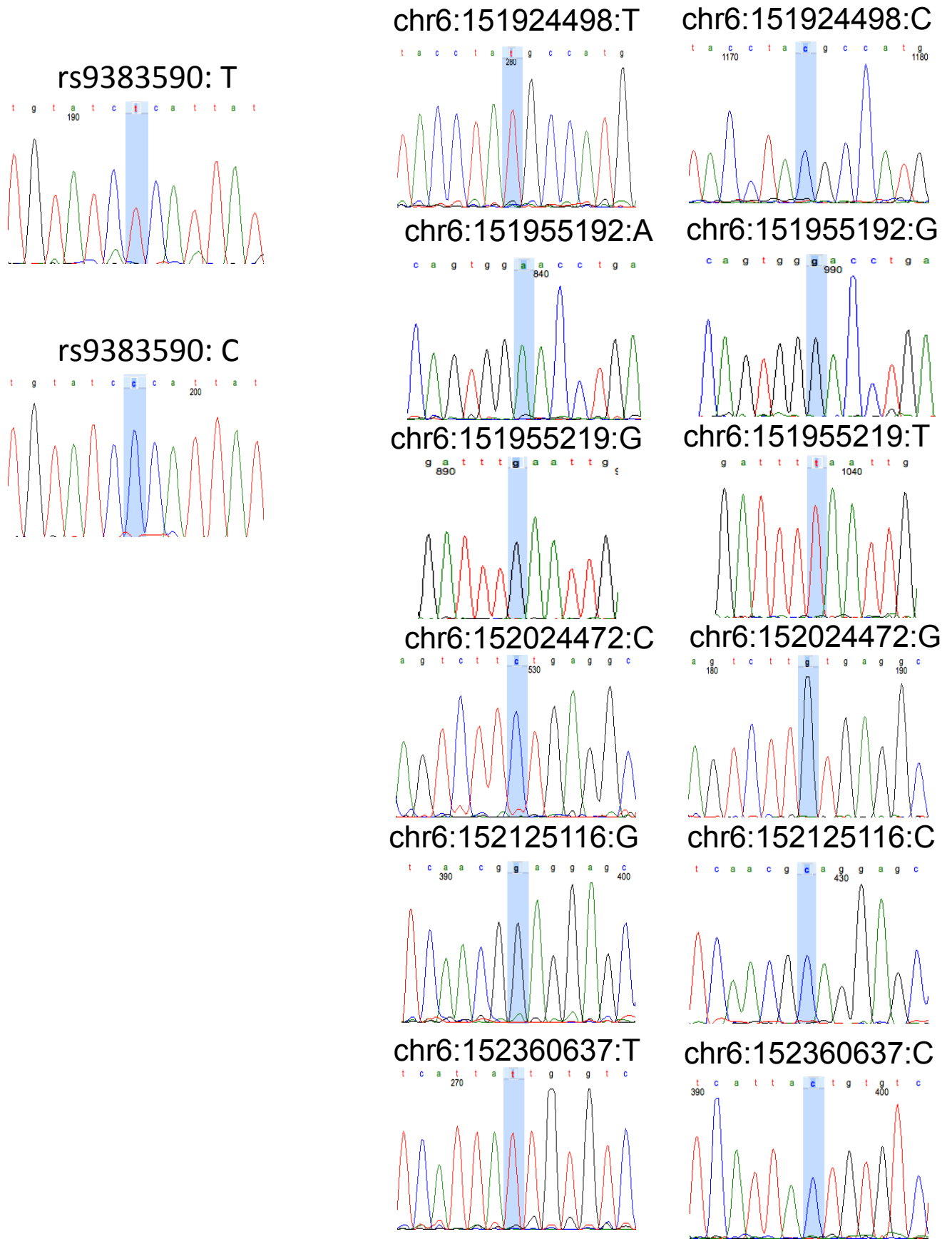
A)



B)

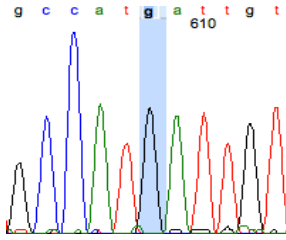


Supplementary Figure 15: Luciferase assays in the sense orientation for the A) rs9383590 and B) the subset of mutations affecting the most correlated enhancer elements.

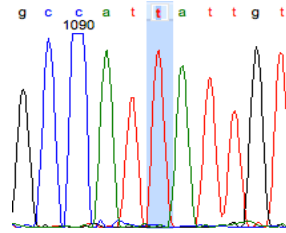


Supplementary Figure 16: Verification of the plasmid sequences used for the luciferase assays. Sequencing traces for each reference and variant allele are shown.

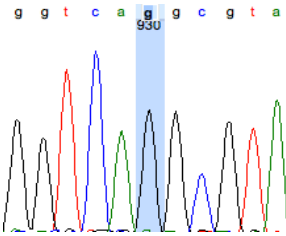
chr6:151923842:G



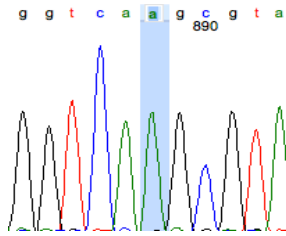
chr6:151923842:T



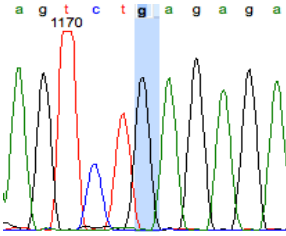
chr6:151937492:G



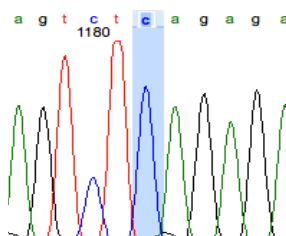
chr6:151937492:A



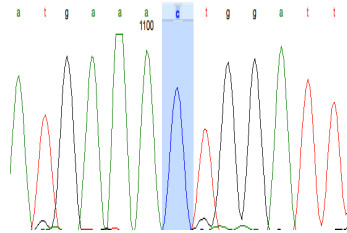
chr6:151937744:G



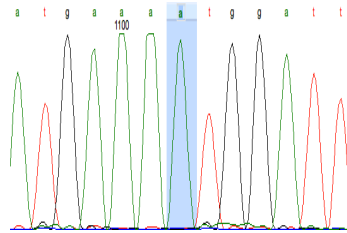
chr6:151937744:C



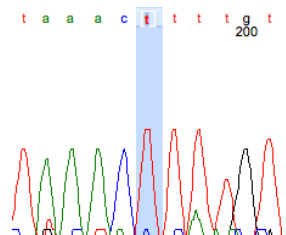
chr6:151954506:C



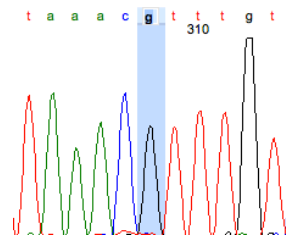
chr6:151954506:A



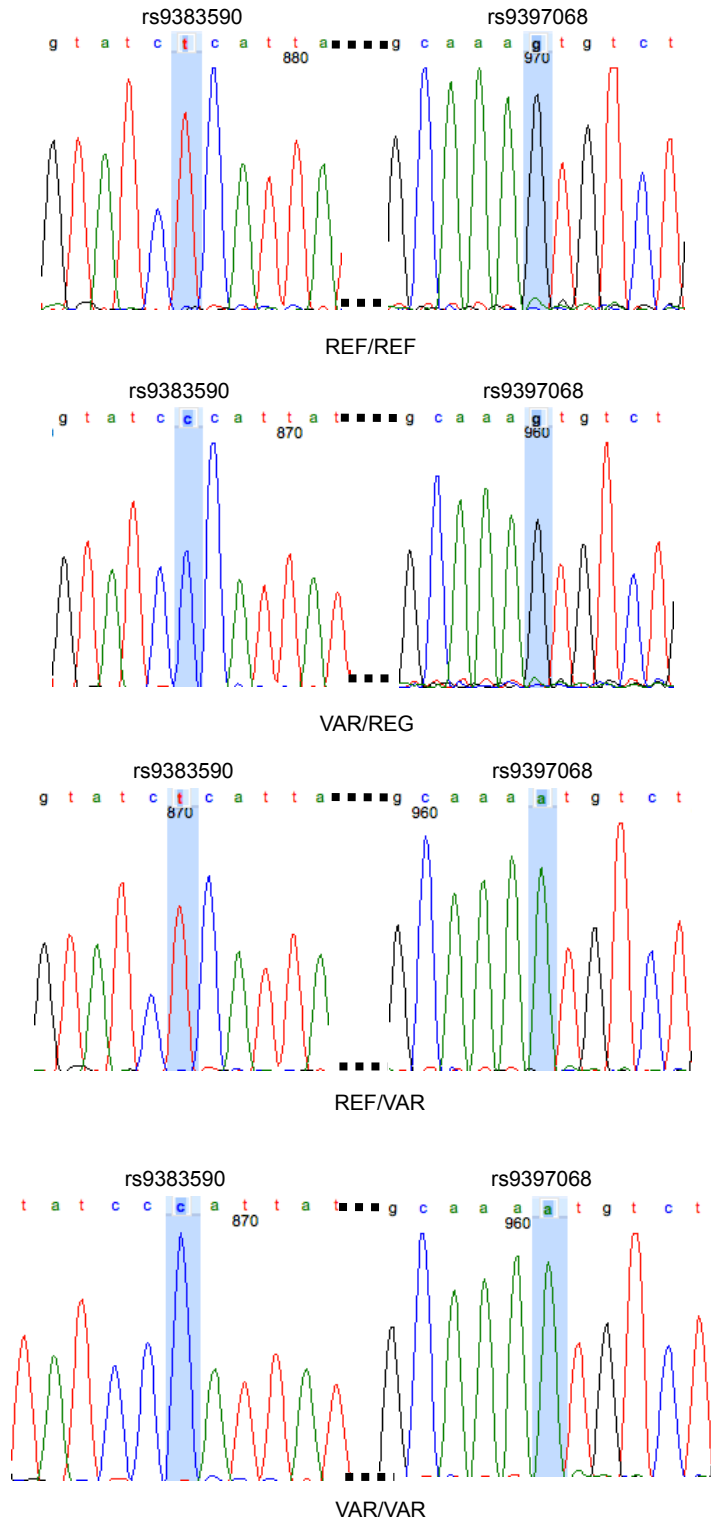
chr6:151979376:T



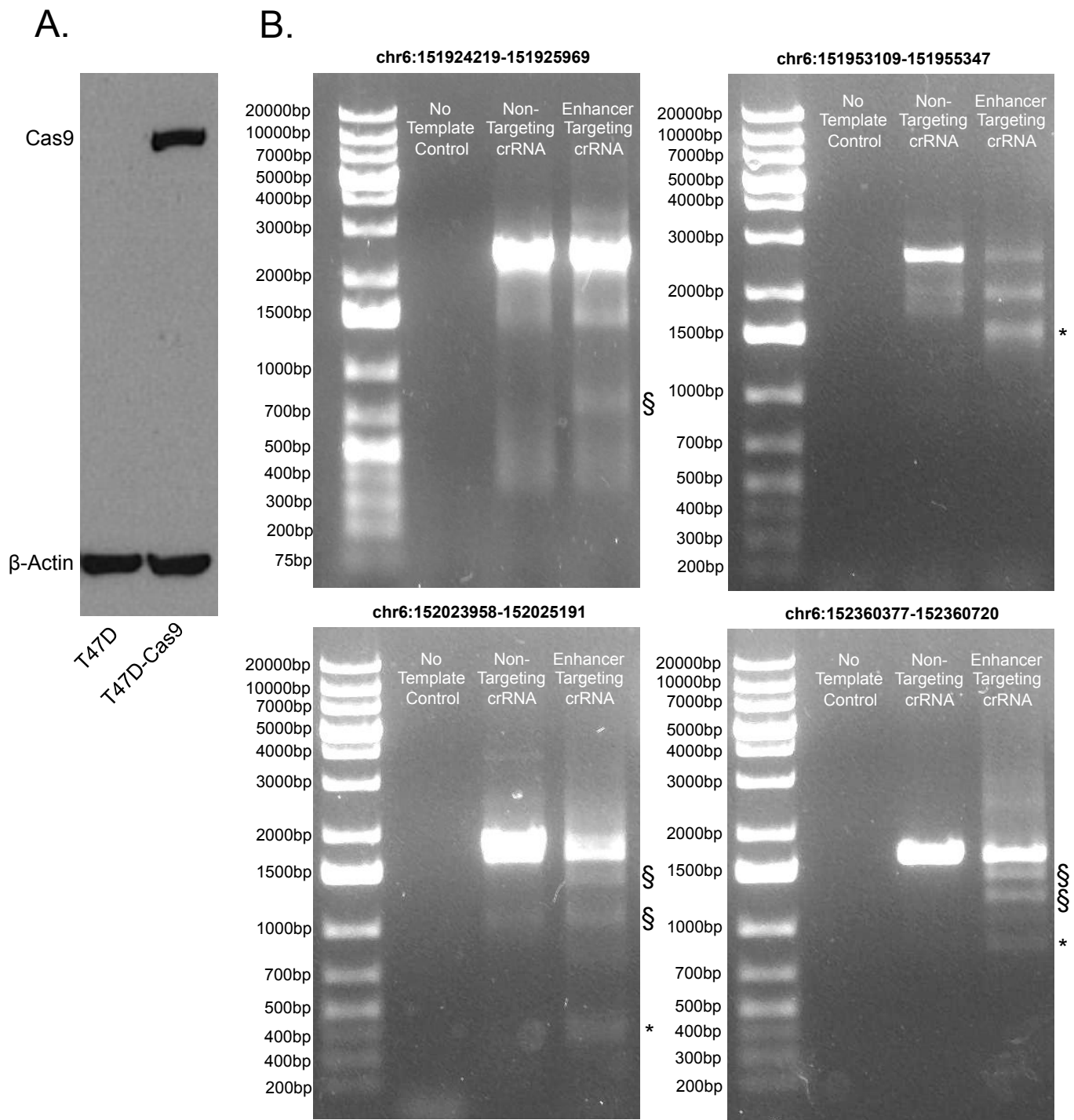
chr6:151979376:G



Supplementary Figure 16 (Continued): Verification of the plasmid sequences used for the luciferase assays. Sequencing traces for each reference and variant allele are shown.



Supplementary Figure 16 (Continued): Verification of the plasmid sequences used for the luciferase assays. Sequencing traces for each reference and variant allele are shown.



Supplementary Figure 17: A) Western blot verification of the T-47D cells stably expressing Cas9. B) Verification the CRISPR/Cas9-based deletions of the enhancers regulating *ESR1* in T-47D cells by gel electrophoresis. * indicates the expected PCR product for the full deletion. § indicates a partial deletions arising from the pool of crRNAs.

Supplementary Table 1: The *ESR1* SRE target regions.

CHROMOSOME	START	STOP	C3D (r)
chr6	151906320	151906705	0.729
chr6	151923646	151923936	0.748
chr6	151924219	151925969	0.903
chr6	151926121	151929145	0.723
chr6	151936846	151939138	0.776
chr6	151943992	151944330	0.855
chr6	151953109	151955347	0.960
chr6	151960883	151961248	0.737
chr6	151978585	151979706	0.895
chr6	151979915	151980209	0.913
chr6	151980470	151980818	0.725
chr6	151986232	151986806	0.700
chr6	151987596	151987862	0.919
chr6	151991285	151991681	0.858
chr6	151996467	151997821	0.857
chr6	152023958	152025191	0.903
chr6	152027481	152027946	0.964
chr6	152074723	152075190	0.948
chr6	152124717	152125407	1.000
chr6	152129784	152130279	0.709
chr6	152180749	152181134	0.844
chr6	152198633	152198868	0.814
chr6	152221688	152223084	0.805
chr6	152360377	152360720	0.913

Supplementary Table 2: The mutational significance among the set of regulatory elements using the breast cancer predicted *ESR1* SRE based on various values for variable parameters, namely window around the *ESR1* promoter and correlation coefficient for C3D used in the MuSE analysis.

Mutation Source	SRE Cancer - type	Window Size (+/-)	DCCC Threshold (r)	Number of Elements	Length of SRE (bp)	Number of Mutations	gBMR P-value	IBMR P-value
BrCa	BrCa	250kb	0.7	23	20,744	10	8.59x10⁻³	7.63x10⁻³
BrCa	BrCa	250kb	0.8	15	12,971	7	3.42x10⁻³	5.79x10⁻³
BrCa	BrCa	250kb	0.9	8	7,746	6	3.31x10⁻⁴	3.15x10⁻³
BrCa	BrCa	500kb	0.7	24	21,044	10	9.32x10⁻³	8.06x10⁻³
BrCa	BrCa	500kb	0.8	15	12,971	7	3.42x10⁻³	3.50x10⁻³
BrCa	BrCa	500kb	0.9	8	7,746	6	3.31x10⁻⁴	2.57x10⁻⁴
ESR1+ BrCa	BrCa	250kb	0.7	23	20,744	7	1.45x10⁻³	2.18x10⁻³
ESR1+ BrCa	BrCa	250kb	0.8	15	12,971	5	2.79x10⁻⁴	6.88x10⁻⁴
ESR1+ BrCa	BrCa	250kb	0.9	8	7,746	5	2.94x10⁻⁵	6.46x10⁻⁵
ESR1+ BrCa	BrCa	500kb	0.7	24	21,044	7	1.55x10⁻³	3.03x10⁻³
ESR1+ BrCa	BrCa	500kb	0.8	15	12,971	5	2.79x10⁻⁴	7.02x10⁻⁴
ESR1+ BrCa	BrCa	500kb	0.9	8	7,746	5	2.94x10⁻⁵	7.02x10⁻⁵
TNBC BrCa	BrCa	250kb	0.7	23	20,744	3	1.17x10⁻²	0.052
TNBC BrCa	BrCa	250kb	0.8	15	12,971	2	0.055	0.265
TNBC BrCa	BrCa	250kb	0.9	8	7,746	1	0.076	0.154
TNBC BrCa	BrCa	500kb	0.7	24	21,044	3	1.75x10⁻²	3.96x10⁻²
TNBC BrCa	BrCa	500kb	0.8	15	12,971	2	0.055	0.159
TNBC BrCa	BrCa	500kb	0.9	8	7,746	1	0.076	0.111
LiHc	BrCa	250kb	0.7	23	20,744	5	0.705	0.692
LiHc	BrCa	250kb	0.8	15	12,971	4	0.257	0.311
LiHc	BrCa	250kb	0.9	8	7,746	3	0.235	0.280
LiHc	BrCa	500kb	0.7	24	21,044	5	0.715	0.790
LiHc	BrCa	250kb	0.8	15	12,971	4	0.257	0.323
LiHc	BrCa	500kb	0.9	8	7,746	3	0.235	0.235

Supplementary Table 3: IMPACT/COMPACT mutation information.

Centre	Build	Chrom	Start_Position	End_Position	Strand	Variant_Classification	Variant_Type	Reference_Allele	Tumour_Seq	dbSNP_RS	Tumor_Sample_ID	Matched_Normal_ID	Genome_Change	Tumour_Fraction
PM	hg19	chr6	151955219	151955219	+	Non Coding	SNV	G	T	.	RCT_15	RCT_8	g.chr6:151955219 G>T	0.422
PM	hg19	chr6	151979547	151979547	+	Non Coding	SNV	A	G	.	RCT_41	RCT_42	g.chr6:151979547 A>G	0.322
PM	hg19	chr6	152075097	152075097	+	Non Coding	SNV	G	C	.	RCT_51	RCT_46	g.chr6:152075097 G>C	0.033

PM=Princess Margaret Cancer Centre

Supplementary Table 4: SNVs in strong LD with the population-specific lead SNVs and their overlapping chromatin annotations.

SNP	R2_with_rs2046210_East_Asians	R2_with_rs3734805_Europeans	H3K27ac	H3K9me3	H3K36me3	H3K4me3	DHS	GATA3	TCF7L2	FOXM1	FOSL2	CEBPB	HDAC2	ZNF217	P300	JUND	MAX	NR2F2	PML	SIN3A	TCF12	TEAD	ERRA	ESR1	FOXA1	
rs58893329	0.557405	0.819071	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	
rs12662670	0.563189	0.819071	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs79475117	0.584369	0.819071	0	1	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs1110776	0.580779	0.819071	0	1	0	0	1	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	1	1	
rs79416523	0.580779	0.819071	0	1	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs17081488	0.584369	0.819071	0	1	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs77334499	0.567719	0.819071	1	1	1	0	1	1	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0
rs12663501	0.567719	0.819071	1	1	1	0	1	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0
rs12663827	0.57097	0.819071	1	1	1	0	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0
rs3734805	0.772357	1	1	0	1	0	0	0	0	0	0	0	0	1	0	0	0	0	0	1	0	0	0	0	0	0
rs9383935	0.776507	1	1	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9383589	0.77284	1	1	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9383936	0.788566	0.986435	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs12665607	0.80445	0.986435	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs74295874	0.80445	0.986435	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9397435	0.800747	0.851507	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9397436	0.800747	0.864325	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9397437	0.800747	0.877499	1	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs58343273	0.800747	0.792285	1	0	0	0	1	1	0	0	0	0	0	1	1	0	0	0	0	0	0	0	0	0	0	0
rs9383590	0.800747	0.877499	1	0	0	1	1	1	1	1	0	0	1	1	1	0	0	1	1	1	1	1	1	0	1	1
rs9397068	0.800747	0.877499	1	0	0	0	1	1	1	1	0	0	1	1	1	0	0	1	1	1	1	1	1	0	1	1
rs60954078	0.800747	0.78133	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9383937	0.800747	0.877499	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs12173562	0.800747	0.877499	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs12173570	0.876896	0.594654	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs6912323	0.800747	0.78133	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs17081533	0.876896	0.601375	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs77275268	0.7407	0.864325	1	0	0	0	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
rs9371545	0.7407	0.864325	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0

1="yes"
0="no"

Supplementary Table 5: Transcription Factor ChIP-Seq files used in the IGR analysis

ChIP-Seq files used for the IGR analysis
wgEncodeHaibTfbsMcf7Cebpbsc150V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7CtfcV0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Egr1V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Elf1V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Fosl2V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Foxm1sc502V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7GabpV0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Gata3V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Hdac2sc6296V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7JundV0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7MaxV0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Nr2f2sc271940V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7NrsfV0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7P300V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Pmlsc71910V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Rad21V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Sin3ak20V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7SrfV0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Taf1V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Tcf12V0422111AlnRep1.bam
wgEncodeHaibTfbsMcf7Tead4sc101184V0422111AlnRep1.bam
wgEncodeSydhTfbsMcf7Hae2f1UcdAlnRep1.bam
wgEncodeSydhTfbsMcf7Tcf7I2UcdAlnRep1.bam
wgEncodeSydhTfbsMcf7Znf217UcdAlnRep1.bam
wgEncodeHaibTfbsT47dEraaV0416102Bpa1hAlnRep1.bam
wgEncodeHaibTfbsT47dEralphaaV0416102Est10nm1hAlnRep1.bam
wgEncodeHaibTfbsT47dFoxa1sc6553V0416102Dm002p1hAlnRep1.bam
wgEncodeHaibTfbsT47dJundV0422111AlnRep1.bam

Supplementary Table 6: The identifiers for the ESR1+ and TNBC tumours used in the MuSE analysis

ESR1+		TNBC
PD3989a	PD4986a	PD4086a
PD4069a	PD5928a	PD4107a
PD4072a	PD5936a	PD4109a
PD4080a	PD5947a	PD4248a
PD4224a	PD5951a	PD4255a
PD4225a	PD5956a	PD4833a
PD4261a	PD6041a	PD4836a
PD4266a	PD6042a	PD4841a
PD4267a	PD6043a	PD4847a
PD4315a	PD6044a	PD4955a
PD4604a	PD6045a	PD4975a
PD4605a	PD6049a	PD4980a
PD4606a	PD6417a	PD5934a
PD4607a	PD6418a	PD5935a
PD4608a	PD6422a	PD5942a
PD4613a	PD6466b	PD5944a
PD4951a	PD6719a	PD6018a
PD4952a	PD6720a	PD6046a
PD4953a	PD6721a	PD6409a
PD4954a	PD7199a	PD6410a
PD4957a	PD7201a	PD6411a
PD4958a	PD7207a	PD6413a
PD4959a	PD7208a	PD6722a
PD4962a	PD7209a	PD7217a
PD4963a	PD7210a	PD7321a
PD4965a	PD7212a	
PD4966a	PD7214a	
PD4967a	PD7215a	
PD4968a	PD7216a	
PD4970a	PD7218a	
PD4971a	PD7219a	
PD4972a	PD7221a	
PD4976a	PD7433a	
PD4981a	PD8618a	
PD4982a	PD8622a	
PD4983a	PD8623a	
PD4985a		

Supplementary Table 7: Capture Probe Sequences

Chr.	Start	Stop	Sequence
6	151906302	151906422	+GGGAGCATTGGGCAAATCAGGTGGGTTGTGGAGAGCTGGAGGACTTAAACATGGGACAGTACTCAGATTCCCCTGCTCTTGTACATTGACACTGACCTTATTAGAGTACCAATAT
6	151906482	151906602	+AGTCAGCATCCCTGGAGTTTAGGGCATAGGTAACCAATCTCAACAGAAAGCTGAAAAAGTTGAAAAAGCATATATCTGTACATGAATCCCTGTAATACCTCAATGTIACCTACCAA
6	151906662	151906782	+TGACTCTTTGTAGATTTTACAGTACTTTTGTCTTAATCTTTTCTACTAACTTTGAAACTCTTAGCAATAGTTTGGGGTAAGTAAATGTATCCATAAATTTTATTGATTAAC
6	151924390	151924510	+TATTATCTTTGAGTTGGCATGTTTAAAGTTGTTTCCCTAAGGATAGGAGTATTGTACCATTGTTTATTTAATACTGCAGATACTTATTGTACCTATGCCATGACATAG
6	151924561	151924681	+ACCATTCTTATTAGCCAGGATACTACTAATTTGGCACAACATAGCTTATACACAAATGCAATGCCAAGTACTGAAATCATGGGGCAGAATGGCAGAAGATGAAGAAAGTGTCTT
6	151925061	151925181	+AGAAATGGGCTTTTCTGTACAGTAAGAAATAAGTAAGAAATAGCTATTGGTGGAGAGCCATCAGTACTAGCTATATCATGTGGCCCACTTAGTAAGGAGTATGGCCGAAGA
6	151925229	151925349	+GCACCTTTCAAGGAAACACCAAAAGGATTAACATCTTCAGCCACCTGTGAAGTCTCTCCCACTTTCTAACTCTCTTTTATATCTCCACACAAGACAGCTCTCCCCACATCTCTTC
6	151925397	151925517	+TTTTCCCTGAGCTCCATCTCATACTAGCTCCGATGATGATGATCATTGGATTTCTGGGACTGCTTTGAGCTTTCCCACTCTGAACGTATACAGCAAGCAGATCTCCAGCAAGA
6	151925789	151925909	+GCTCTACGGTAGGGAAGGAGGGGAGGTAGGCCAGACTGCAAACTGCAATCTCACTTTGATTTCTGTACTCTGCTTAGTTACACTTTTCTTTCATTTTAGTTTACATAIA
6	151928274	151928394	+GTTTGTGGTGGCAGTAGAGCAGGCAATTGAGACAGAAAAATCCCTACTAAGTGTGGGTACGGGGGTGACCTGGAGAGAAATGATTTTTCAGTATATTTTAAATGAG
6	151936750	151936870	+GGTAACAGTGAATGAAGACACTAAAAATCTCTGGAAGAAGCAGAAAAGAGAGAAAAGCAGGTGGGTCTAAATCTGGTATGAAACCTGTTTAGGAAATGACAGGCAATCAAGA
6	151937051	151937171	+TCTATCTTTTCAACATATCCACTGCCCTCCAGCACTTTTGGCATCAAGTAACCACTATGATTTACCCCGCTAAGAAAAATATTCACATTTCCATGGCACAAATGTAGGAAA
6	151937222	151937342	+TTCTGGTCTGGGGAGGTTTGTATGTAGGTGATGCCTCTGAAGTTGCTATCATAGAGCTCTTAAAGTAAACAGGGCTTTGGCCCTCAGGAGGGAAGCTTATCTGTAGCAGA
6	151937393	151937513	+ATGGCCAGCCAGGCACAGATGAAGCCATAAAAGTACTCAATTTGCTGTGGCTACAGAAGGAGCAATTTATCTAGTGGTCCATCAGGAGGTCAGGCTAAAGAAACATGTACCCG
6	151937564	151937684	+TCAAAAGGGCATAGAGTGGACTGTATCTAGGATTTTTTCCCTGCTTGGCTGCCTCTGTTTGGTTCATGTGTCAGCAGAGCAGGGAAGCAACACACAATGAGCGTCT
6	151938163	151938283	+GGAAGCGGTTGCAGAGCAAAATGAGCTGGGGTGGGACTGTTGGGTGGCACTCTCTGGATTAACCTGATTCAGTTTGGATCATCCCTTTCTAAATGGAGGGTGCCTTTAATGAGG
6	151938331	151938451	+ACAGAAACAGCCTAAGAGAAATGAAGGGATATGAGGCATATTAACCAATTTGCCAGCCCTACTTTCTTTCTCAATGGACAAATGAGAAATCCAGAGCAGTCCCATGGAATAT
6	151938862	151938982	+CATCAGAAAAGTATGTGAAATCTGAAAGTTGGCAGTTGATGTGACTTATCTGGAATCTGGTATTTAFAAGTGAGAGAACTAGGAAGAGAGTAGATAATTTCCCTACCTCATGCAA
6	151939042	151939162	+TTGTCTCCAGTAAATGAGTTCTGTTTATATGATGACAGCTGGCAGACTCAGGGAGGTTGGTCCGAGATGCTAGGCTGGAACGTGACCAAGCTTGTCTCTCTGATTGAAATCATC
6	151943940	151944060	+AACCAAAAAACCCACCATGAGTCGAGAAAAAGAGACACCCCTGTCTCAAGTTCATGCAAAAATATAGCCGAGAGGCTGATCATCTTCCAGGTCAATACGATCTTGAAC
6	151944252	151944365	+ACACACACACACACACACTCTGTGACACGTGGCTGTGAGCACTAACCACTAGAATATGATCTCTGAAATCTAACTAAAAAGCTTCAACCTCTTACCATCCCAA
6	151953971	151954091	+CTGCAAAATTTCTCCAGCAAGTTTTAAGGTAATAATGTTGAGAGATTAATAATTAATCTGAGCAATGTGAAATAAAAAATAAGTTAAAAATAAACAGAAATAATATAGCTAT
6	151954127	151954247	+TGATGCTTAAACAAATCTGTTTTTATGTAGGAGGAGCATATACAAAATTTCTTGTGAGTTGCAAAATGTAGTAGGAAAAATACCAAGAAATACATGATCTAAATAAAGGCTGA
6	151954467	151954587	+GCATACCAGTTAAAAATATCTCAAAATTTCTATGAACTGGATTTGCAGAAACAATCTAAGTGTGTAGATGACCAGCTCGTGCCACTAACACATTTGGGCCATATATGATGAT
6	151954804	151954924	+AACAAATGTTGCCATTATACATCAAAACAATGACAGCAAGCAATAAGCTGTGTTGTCATAAGCAAGGGAGGAATGCCAGTCTCCCTGGTAGAGGCAGACTAACCTTGGAACTAGA
6	151954984	151955104	+GTGCACCTGGGTAGTATCTTCTGCTGATATCTTCTCTGTACTGTGGCTGACCTTTGACCAAGGACGCTGCTTAAAGGTTCTGGTGGGACAGGCATATTTGAGCATACCCA
6	151955164	151955284	+TATAGACCTTACACAGGCTCAGTGGAACTGAATCACAACCTCTGGGATTTGAATGATTCAAATCAGTGAATCACTCTAGTTGTGAGTACAGCCATAGAACCACACACTAA
6	151955344	151955464	+GAAAGAGGTGCTAAGGCTGACAAGATGTTAACTATGAGTTATTAAGTTTTTCAGGGAAGTCAAAAACATGGACTCTGTGCAATAGGAATGATGTGACCTCAATAATATAAAGAC
6	151960855	151960975	+CCATGTATGATAIACATTAATAAATCAITTTGTTATTTGGTAAACATGAGGCTGTTTCTACTTTTGGTTATATGAGTAAATGCTGCTAAAAACATGTAIACAAGTCTTGTG
6	151961035	151961155	+AGTAATGCTATGTTTACCTTTGAGACATGCAAAATTTTCCAAAGCAGCTGCACATATAAATCCACCAGCAATGATGAGGATCAATTTCCACATGTTGTGCAACACTT
6	151961215	151961335	+CATGCTCTAATAACTATAATGTTGAGCGTATTTTCAATGCTACTGGCCATTTGTACATCTCTTTGGAAATATAIATCACTCCCTTTTGGCCATTTAAATGTTGATATATTT
6	151978583	151978703	+TAAGGAAATGAGTCCCTTAGGGAGAAAATGTTCTAAATTTATTTGGTAAATGTTGCTGACTAACATGTTTACTGAAATGGATCACCATGGGTGTTTAGGTTTTGTTCTTT
6	151978763	151978883	+GTCCCTTAGTTAAATATATGCAATTTTCTGCAAACTAICTGAACTGAAAAGGCCAGAAAGGAAATAGCGTGGTCCAGAGTAGATGGACTGCAAAATGTTACACATAATTTCAA
6	151979023	151979143	+GGCAAAAAAAGATGATGACCAAGTGTGCTACTTTTAAAGCTTATGTTTCAAAATATTTTAAATTTAAACCAATCAAGTGCATCCCAAAAAACATGGAAAAATTTGAGGC
6	151979310	151979430	+CTCAGTAAATATTTATGATGGTCTGATATGGATGATGAAATATGCTAAAGGTAACCTAACTTTGTCATAAACCAGGTTTCCAGTTAGTTGACCTTTCCGTAICTCTAAAT
6	151979490	151979610	+CTTTCCCTCTGGTATCTGAGCTAICTCACTCACACAGCACCAGTGTATGTTGCTGTTTGTGTTAAATCACTCATATAATTTCTCAITCAATAAATAAGTATGAT
6	151979670	151979790	+TCATTTGTTTCCATCCACATAAATGTTGATGCAATATGAAGTGAATATAAATAAATATGCTTTTATTTATTTGTTTATTTTACTTTAAGTCTGGGATATAAT
6	151980046	151980166	+GGCATAGATGAGCAAGAACAGGTTTTCTGAAAGTGGCCAGTCTAAGGCTTAGAAGAAGAAAGCTCGGGAGCCTAAACCAAGATATGCTAAGGCAAGTTTTTGTGCTGTTGTTTTG
6	151980427	151980547	+GTGAGCAGTGTGCCCGGCTGTAAGGCAAGTATGACCATTAGAGGAGATGACTAGTCCAGTGGCAGGCTTCTACTGGGAGTCAGCAAGAGCTCCCACTCTCTGGTCCAT
6	151980607	151980727	+AATCCAGTCTTTTAGCACTGAGTTTCTCATAGCTTTTACCATTGAGTTTCTCATGGCTAATGAATGCTAATGAATGGCATTCAGATTTCACTATAGGATCCATGAGTCAGCT
6	151980787	151980907	+GAGTAATCAGCAGGCAATGTTCTCTAATAACACTCCAGTCTACTGAGAGCTTCTAATCAAAACAACATTAAGGCTTACAGTGTGACTTCACTAACTAACTGAAAAAGAAATAGA
6	151986219	151986339	+CAAGTTTTGTGAACACCAAAATCCATTTGCTGTGTGTGGAGGGTGGTTAGGATGGTAGAATAAATCTTTGCAACGGCTGCACCTAGCATGGTCCCTGAAAGCTTGGAGT
6	151986399	151986519	+AGAGGATTCACAGGGTAACAGGAACCTGGACTGAACTGAGCTCTGCACTCCGAGATTTTTTAAAGTGGTTGTTTTTAGAAGTGTGTCACAGCTCTAAATCATGTAAATTC
6	151986579	151986699	+GATCTGGAATCAGAAAGCCAGAACTCAAGATGGAAGCCAGAGCTTAGCAAGTGAAGCTCTGCAATCTGGAGCCGAGATTTGACCAGCTGTGCACAGCTTCCACTGTGGC
6	151986759	151986879	+TTGCTGAAATCATCACTCCCTTCCATGCGGAGACTTAAAGGTTTCAAACTTCAAAATCAAGTATTTCAATCACTCAGATGACTTACATACAATGTTTATACATTT
6	151987494	151987614	+TCTGCTAATAATCCCAACATTTCTACAGGAAAAGTGCATGTTCTGCTGTTGTTGAGAGGGCCACCAAGGAATCAGGTTCTGGTGGTAAAGATGACAGAAATGACCAAGCT
6	151987674	151987794	+TCTTCTGATTAACTATGAGCTCTGTTCTTCAAAAGTACTGGAGTCTGGCTTAGCACATCTGTTTCTGAGCCAGAGCACTTCACTGAGAAGCAGACCTTACTTGGCTGG
6	151987861	151987981	+CCTCTCTCTCTCCAGGAAACATATGTTGGCAGCAAGGGTGCAGAAAGTAAAGGCAATGAAGGCAATGATGATGCAATGTTGAGGAGCAAGGTTCTGTTTTACTCTCAGTGCACAAA
6	151991638	151991751	+CGTTTGTGAATAAATAATGATACATCTTCTGTAATATGAGAAAGTGGGAGGCAAGCTTAGGTGAAGAAGCACTTAAAAAATTTTTTTTTGAGACAGCTTGC
6	151996502	151996622	+ATGGTAGCTCAGTGTCTCGATGCTGGAATTTGTTCTAATAAGTTCTGAGCTGTTTATAAACCAAGAGGGTAGGGGAACTAGTAGTGAATCTCTTCTCTCTCTCTCTCC
6	151996682	151996802	+AAAAAATCAGTGTACAAAAATGTTGACATTTGGAATACTTACACTTACATAGTGTCTTAGTATAGAAACAAAAAGCCATCATTTAGGCTAGATTTTCAAGATGATCAC
6	151996862	151996982	+TTGCTGTAGCATAATTTGGCCATTAACATAAGCATTGTTGAGACCCGTAAGTCAAGCCAGGCTATGGTGAACAAAAATAGCAAAATCGAATGATGCGGAAACTGGTGAAG

6	151997042	151997162	+	GGTGGGGCAGCAGAGAGCTGGCTGTGGTCTCCAGAAGGGAAGGCCAGCACCTGCCTGCAGATTCCAAGGCACAGGTGGAGGTGGTAATGAGTACGAGGTAGAGTCAAGGTTGGAGCTG
6	151997222	151997342	+	AAGCCAGAATGGAAGTAGGACAAAGAACTGGTGAAGTCAAAGAGCAAAGGTGCAGGGGTACAGGGAGGAGAGGTTGGGTGGGCAGACAGGAGGTGCCGAAGGAGGCCAGCCTGCTTC
6	151997402	151997522	+	TGGTCTTCCAAGGGCTATGAAGGGACAGGATGTGAGAGACACCAGCCATCTCTCTCTGAGCTATTAATAATGAAAGTTCCTGTTTTATGAACAAATAGTAAATGAATGGAGAACA
6	151997582	151997702	+	GGTTCCTGCAAAAGGACTAAGTCTCAGCAGCCTGTGCCATCTTTTCGGAGATTTTCCACAATCACCAGGAAATATAGTTCAGTGCCTGTTTACTTAATGGTCGGTACGCTTGAA
6	151997762	151997882	+	ACCCCAACACATGGGCAGCCTTCTTCCAGACTTGGCACCCTCAGTGCCTGATTTAGCACCCCTCATGGGAAGGCATCCTGGAGATCCCTATCTGCACCCTGTCTCCATCACCCT
6	152023884	152024004	+	AAGTCCATTTCTATTCTCCATAGAACATTTAAACTAGTTCACAACTCTTGAAGTCAATGCTCTAGAAAGTGAATGCAAAATGTCACCATTGCCTAGAACCAACTTGAGAGGCTG
6	152024064	152024184	+	ACCCCTGATTTGTGAATATCTAGCTTCAAATGTTCAAAAGCACAGGAAGTTCGACTTTAAAAATATAACCTGCCAAATGCAAGGGAGGGGTTGGGTGAGGACTAGGTC
6	152024244	152024364	+	GGCAAAATTAAGTACAGCTGGTAATTTCTAATGTGAAGTGGCTGTTGGTGAAGTATTCAGGAAAATTTAGCTGGACCTTTCTACCTACTGCATAAGAAATGAAGGTTTCT
6	152024424	152024544	+	TTAGTACTGTCTCAAACAGCTCAGGACTGGTACGGCAGTCTCTGAGGCTGGAGGTCATGGAACCCCAAGGCTTGTCTGCAAGGGCATGGTACTGCAGGCTGTTTAGA
6	152024604	152024724	+	TCTCTGCTCAGAGCTGAGCAGCTGTAATGAGCTCTCTGTGAATCAGAAAATGTTAATGGGTGAGTGTCTAGCTTCTTACTTCAAGTGAAGGCGCTGTAGCTTTTGGCAGC
6	152024784	152024904	+	AAAAAGATTTAATATAGAAACGAGTCTTCCATCTGGCTGAGGCTGAACAAAGTAGAAGAACAGTGAAGTACTGGGGTATGGATCTATACCAATTTACAGATATACCTCCCTTC
6	152024964	152025084	+	GAAGTCTTAGATCCAAACTTTGATCCAGAAAGTCTAAGTGTCTTGTCTGAGATTTGAAGAGAGCTGAGACAAACTGTGGGATCTAGACGCTATTGCGCACTGCCTGGGTAA
6	152025144	152025264	+	TTATAACAACACAGAACATCAGTGGTCTCCCAATAATCAATATAATTTGGCTCTATAATATAAAATTTGACTTGTAGTAGAAGATTTCAAGGCTAAAAATAAAGATGATAAT
6	152027473	152027593	+	TTGGCAACAGGAGGATGACTAGTGGGCTGAAAGTATCCACTGCTGTTTCTGTTAACTTCTCTAATCTACTGGGCTTGGGAAGAAGAGTACGGGCACTGTTTGCCTCA
6	152027653	152027773	+	GCTCCACAGGGCATGTTTACTTGTGGCGGTGAGTCTCTCAGGTAAGGTTCTGGGATAAGAGCAAAGCTTATGGTTTTCCTGCTGTCACGAGACACACAGATCACC
6	152027833	152027953	+	TTACTCAGGAGGAGAAACACAGAACACATGGAGGAAGTGTCCAGTGTAAAGCTTTAGACCAAACTATAGAAATCTGTTACTGGAACCTGAAGAAATAATCATGACTGCT
6	152074792	152074912	+	AGTTTGGTGCACAGATATGCGGTAGTGTCTATCTAATGTGTCAAATTAATAGTAAAGATTTGGCTTAACCTTGTGACATGGAAATTTATATCTCCAGTTTCTGGTTTAACT
6	152074972	152075092	+	TCCTTACCTAATTTGACTTGCCACTTTTCTCTGTGGGCTGTCTCTAGAACTCAATGATTTCTGTTGGGTGACATCAGCTGCTACTCGGGGAAGAAATAGAATTTATGGTGC
6	152075152	152075272	+	AGGGGATAAAAGAGAGAAATCAGAGATGCTCCCAACACAGAGAAACCTCAGAAAGCAGCAGGAAAGACAGAGTAAATGGAGAAACAAAACCTAAGTCAAATAGTCAATGAAATAA
6	152124642	152124762	+	GGGCAGAGAGGAAGCCCTGAACCTATGGGGGAAGCTAATGTTGGGTAAAGACAGAGCTGGGTCATGTAAGATGGATCTGAAGCAGTAAATAATCAAGAAAGTAAACAGGTTG
6	152124822	152124942	+	CAGGTAAGGTAATGGTCTCAAAGGAGTGGCCAAATGCAATGGAAGAAAGAGAGATGTAAGCTAGAAAGCTTAGGAATTCCTTGTATAGGTGGGAAGGCAAGGAAATCAGC
6	152125002	152125122	+	AGACACGGGAAGTTGAGAGAACACCATGTTGAGAAATGGTACTCATATTTGAACAAGCTGCAATGCCAGCAGACCGCTGGAAAAGTGGGGTGGAGACACATCAACGGAGGAGC
6	152125182	152125302	+	TGTCCAGCAGGAGGGGTAGGAGGGAAAGCAAGAGAGCTGGGAGCCGAGTGAAGTTTTGCCAAAGCAGAGAGGAAAGTCGGCTAGCACAGTATACTTTCCACCCATGCTCACC
6	152125362	152125482	+	TAAGAAAAGTCTTACTGAACCTTCTGGGCTAAGTTGATGTAAGTCTCTGAACAATCAAAGCTGTGAGGAGACAGTAACTTCTTATCTTCCATGTAATGAAACAATG
6	152129702	152129822	+	CCGGGCAGTGAAAAAAGTACTCTCCACCCACTTACCCTCCGTCGAGAGGACAGCCGAAAGCCGGGCTCTTAACAAAACACACGTTGAAAACAGACAAAGCAGCAGTATTT
6	152129882	152130002	+	TCGCTCTGGCATTAAAGTTGGGGTGTGGAGTGTAGCAGCTCAGCAGAGTTTATTTATCTTTTAAAGTTTTGTTTAAATGTCCTCCCAAAATTTCTTCTCAGACTATTT
6	152130062	152130182	+	TCCCTCTCCCAACCCACGTCCTGGGGCTTAGAGAGCGAATGGGAGTTGAATGGGTGATTTCCGGAGTGTAGCTGGCTGAGTCCCGCTGGAGCGGATGCTGGCATGTACTCTG
6	152130242	152130362	+	CTTAAAAATAATCTCTGCCAGCCAGTGAACAAGCTGTCCACCCGGGAGAAATGCCCGGAGTGGGTGCGGGTCAAGCAGGCTGCCTCGCAGCAGCTGGAAGGAGCGCGGC
6	152180661	152180781	+	ATCACAGACCAGGACTCAGACCTGTGAGGACCTTAGCCTTACCCTCCTCTTCCATGTGTCAGCTTCGCGTCTCTGTCTGCAAGTATGCAATTTCCCTAAGTGGGAGAGCAGAG
6	152180841	152180961	+	AAGGACTCTGTTGGCCAGCTTGGGACACACTTGGACCAACTAGTGTCTCAGGAGGAGTAAATGATACAGAGGTGATCCCTGGGGTGCCTGTGAAATCATGTGAGGCCAA
6	152181126	152181246	+	ACCTCTGTTGGCTCTATCCCATCCCTATAGATATCTTCTGCTGAGACTAAGCAGAGAAAATAATTAACATTAATTAATGATGCTTAAATCCACTTTTACTCTTCTTCTT
6	152198631	152198751	+	ATGATCTGTGTACATATTTCCATTTTGTAAAGAGACGAATCAATGTGAAAAATCTAGTACTTTTATTCATTAACATGTTAGTTCAGGAATTCACCTGGTGTACAAAAGATA
6	152198811	152198931	+	TTAGTCTCTCTGCTCTTCTGGCTTACAAAGTAACTGGCTGGGGAGAAAGTGAATCAAAGTAAATTTGGTTTGAATGGTTTATGACTTATTTCTGTAACCTTAATGTAATA
6	152223083	152223203	+	TGCCTCTTCCACCATTTTTTGGCCTTGACGATTAATTAATTTGATTTGAAAGTTTGGCAGTTTATTTTGAATAATTTACATAACAAATGCCAAGCAAATCCAACTTCTTGG
6	152360309	152360429	+	GCAGGTCATGTTCTTACCTGGTGGAGTGTCCCAACCTTCACTCTGAAGGCTGTGGCCATTTGATGCTCTGCTGGATCAGATGTTGTCATTTTATGACCTTAATCAGAGG
6	152360489	152360609	+	CCTTCATATGGAGTAGTAGACTGATCTATCTGTATAGCTAAGTCACTCAGTCAACCCAGCAACACTAATCTTCTTAGCCTGTGACTTAGAGGTAGGAGGAGCCGAGATTGCAATC
6	152360669	152360789	+	GAACCTGAGACTTCTAGCCAGCAGAACGTAATGCTGTGGAACAAGAAGCACAAATTTACTAGCGGGTCTTAGGGGTGATGGTGGTGGCTCAATTTCCACCCCTGATTTCT

Supplementary Table 8: Somatic mutations identified within the *ESR1* SRE.

Mutation	Sample ID	ESR1/ER α Status
chr6:152125116:G>C	PD6417a	Positive (ESR1+)
chr6:151979376:T>G	PD4847a	Negative (TNBC)
chr6:151937492:G>A	PD4966a	Positive (ESR1+)
chr6:151937744:G>C	PD4072a	Positive (ESR1+)
chr6:151954506:C>A	PD5942a	Negative (TNBC)
chr6:151955192:A>G	PD7214a	Positive (ESR1+)
chr6:151924498:T>C	PD4983a	Positive (ESR1+)
chr6:152024472:C>G	PD6719a	Positive (ESR1+)
chr6:152360637:T>C	PD7219a	Positive (ESR1+)
chr6:151923842:G>T	PD6722a	Negative (TNBC)
chr6:151979547:A>G	RCT_41	Positive (ESR1+)
chr6:151955219:G>T	RCT_15	Positive (ESR1+)
chr6:152075097:G>C	RCT_51	Positive (ESR1+)

Supplementary Table 9: Experimental primers.

Experiment	No.	Sequence	Description
ChIP	ChIP_01_F	TTCTGTGCTTTTGTGTGGAC	rs9383590 forward
	ChIP_02_R	TGAACAGACACTTTGCCAGA	rs9383590 reverse
	ChIP_03_R	GGAAACTGAAAATTAAGCAATAATCA	Reference MAMA primer (T)
	ChIP_04_R	GGAAACTGAAAATTAAGCAATAATCG	Variant MAMA primer (C)
Cloning	Luc_01_F	AATATAGGATCCAGACAACATGCCAGCTGGGTAT	Chr6:151924498 mutation-containing enhancer
	Luc_02_R	ACAATAGGATCCATTATCCATTGTATACATGCATCAAAATATCACTCTGT	Chr6:151924498 mutation-containing enhancer
	Luc_03_F	ACGATAGGATCCGGTTCACCTTTAATGTTGGA	Chr6:151954506, chr:151955192 mutations and chr:151953765 genetic variant-containing enhancer
	Luc_04_R	ACGATAGGATCCTTCCATATTAGATCATAGAGCAA	Chr6:151954506, chr:151955192 mutations and chr:151953765 genetic variant-containing enhancer
	Luc_05_F	AATATAGGATCCGAAAGTGCAAATGTCACCA	Chr:152024472 mutation-containing enhancer
	Luc_06_R	ACGATAGGATCCATATATTTGATTATGGGAGAACCAC	Chr:152024472 mutation-containing enhancer
	Luc_07_F	ACGATAGGATCCTGGATCCTGAAGCAGTAAAAAAAAT	Chr:152125116 mutation-containing enhancer
	Luc_08_R	ACGATAGGATCCACAATAAAGTAGCCCAAGAGT	Chr:152125116 mutation-containing enhancer
	Luc_09_F	ACAATAGGATCCTAGTCTGCCTGGATCGAGT	Chr:152360637 mutation-containing enhancer
	Luc_10_R	AATATAGGATCCGTGCTTCTTGTCCACAGCATT	Chr:152360637 mutation-containing enhancer
Mutagenesis	Mut_01_F	ATACTGCAGATACTTATTGTACTACGCCATGACATAGTTGG	Chr:151924498 (T -> C) sense
	Mut_02_R	CCAAACTATGTATGCGGTAGGTACAAATAAAGTATCTGCAGTAT	Chr:151924498 (T -> C) antisense
	Mut_03_F	TGCATACCAGTTAAAAATATCTCAAAATTTCTATGAAAATGGATTGTGCAGAAACAATC	Chr:151954506 (C -> A) sense
	Mut_04_R	GATTTGTTCTGACAAATCCATTTTCATAGAAAATTTGAGAATATTTTAAACTGGTATGCA	Chr:151954506 (C -> A) antisense
	Mut_05_F	CACCAGGCTTCAGTGGGACCTGAATCACCAACT	Chr:151955192 (A -> G) sense
	Mut_06_R	AGTTGGTGATTAGTCCCACTGAAGCCTGGTG	Chr:151955192 (A -> G) antisense
	Mut_07_F	GTCAGGCCAGTCTTGTGAGGCTGGAGG	Chr:152024472 (C -> G) sense
	Mut_08_R	CCTCCAGCCTCACAAGACTGGCCTGAC	Chr:152024472 (C -> G) antisense
	Mut_09_F	GGAGACACATTCACCGAGGAGCCAGATCAATC	Chr:152125116 (G -> C) sense
	Mut_10_R	GATGATCTGGCTCCTGCGTTGAATGTGTCTCC	Chr:152125116 (G -> C) antisense
	Mut_11_F	AATCTTAATTTCCAGTTAATGAAATCATTACTGTGCTCCTGGTGGC	Chr:152360637 (T -> C) sense
	Mut_12_R	GCCACCAGGAGACACAGTAATGATTTTCATTAACCTGAAAATTAAGATT	Chr:152360637 (T -> C) antisense
	Mut_13_F	GGAAACTGAAAATTAAGCAATAATGGGATACAAACACATACCTATTAGAATG	Chr:151953765 (rs9383590) (T -> C) sense
	Mut_14_R	CATCTAATAGGTATGTGTTGTATCCCAATATGTCTTAAATTTTCAGTTTCC	Chr:151953765 (rs9383590) (T -> C) antisense
CRISPR	RT_01	TCCAGATGCCTAATGTTGGA	ESR1_F
	RT_02	AGCAAAATTAGCTGCCCTGA	ESR1_R
	RT_03	GCACCCGAACATTAGGAGAA	CCDC170_F
	RT_04	ACATACCGGCTCATTTTGG	CCDC170_R
	RT_05	TAGCTTCTGGTGCCCATCT	ARMT1_F
	RT_06	AACCCAGCTATCCCCAAAAC	ARMT1_R
	RT_07	CTCGTCCCAGCTCAAACATT	RMND1_F
	RT_08	CTTCAGCACTGCATGGAAT	RMND1_R
	RT_09	TGGCAGCTGCTAGAAAATGG	ZBTB2_F
	RT_010	TTGCTGGCTTCTTTCCAAGT	ZBTB2_R
	RT_11	TTCCCCGTCACCTGTTTATC	SYNE1_F
	RT_12	GAGGATCGGGTCATAAGCAA	SYNE1_R
	RT_13	GGACTTCGAGCAAGAGATGG	Actin_F
	RT_14	AGCACTGTGTGGGCTACAG	Actin_R
Viability assay	RT_15	CTCGTCCCAGCTCAAACATT	RMND1_F
	RT_16	CTTCAGCACTGCATGGAAT	RMND1_R
Deletion Validation			
chr6:151924219-151925969		GAGATGGAGTCCGCTCTTGT	primers to verify deletion of the enhancer chr6:151924219-151925969
		CAACATGGATGAGCTAGAGGA	
chr6:151953109-151955347		TATTTGGAGCGGTTTTGG	primers to verify the deletion of the enhancer chr6:151953109-151955347
		CCACTCAAGCAGCTCACTT	
chr6:152023958-152025191		AGCAAAATGTGAGTGTGCTTTT	primers to verify the deletion of the enhancer chr6:152023958-152025191
		AAAATCGCTTGAACCGGGGA	
chr6:152360377-152360720		ATGCACCAATCCCAACCAA	primers to verify the deletion of the enhancer chr6:152360377-152360720

		CTGGCTACAGCCACAACCTGA	
CRISPR (T-47D Stable Cas9)			
chr6:151924219-151925969			
	CRISPR_crRNA_1	CUUCAGCCUCUUGAGUUGCGUUUUAGAGCUAUGCU	crRNAs targeting the enhancer chr6:151924219-151925969
	CRISPR_crRNA_2	AUCCUGGGCUAAUAAGAAAAGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_3	GUGAAAGCCAACUAUGUCAGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_4	UUAGCGUUCUAAAGCACUAUGUUUUAGAGCUAUGCU	
chr6:151953109-151955347			
	CRISPR_crRNA_5	UGCCAGAGAAGAUAUACAGAGUUUUAGAGCUAUGCU	crRNAs targeting the enhancer chr6:151953109-151955347
	CRISPR_crRNA_6	AGCAACACCAAUUGCUGCCAAGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_7	GGUAGAGGCAGACUAACCCUGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_8	AGUCGUGUUUGCAUAAAGCAGUUUUAGAGCUAUGCU	
chr6:152023958-152025191			
	CRISPR_crRNA_9	AGCCAUGCCACUUGUAUUUGUUUUAGAGCUAUGCU	crRNAs targeting the enhancer chr6:152023958-152025191
	CRISPR_crRNA_10	CCCCAGAGCCUUGUCUGCAAAGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_11	CUGGUCAGGCCAGUCUUCUGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_12	UUAGUGUAGAAGCCACCCUAGUUUUAGAGCUAUGCU	
chr6:152360377-152360720			
	CRISPR_crRNA_13	UCUUAGUACAAGUGUAUACGUUUUAGAGCUAUGCU	crRNAs targeting the enhancer chr6:152360377-152360720
	CRISPR_crRNA_14	AAGGUAAAGGAAGAACAUGUGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_15	GCUAAGAAAGGAGUUAGUGUUUUAGAGCUAUGCU	
	CRISPR_crRNA_16	AUUCACCCGUUCUAUCAUUCGUUUUUAGAGCUAUGCU	
3C			
	3C_001	CATGAAGGTTTGCTGTGTTCAA	Anchor promoter chr6:152124717-152125407
	3C_002	TTTGGTTGTCATTCAGCA	Test region 1: Enhancers chr6:151923646-151923936 and chr6:151924219-151925969
	3C_003	GGAAAGCCAAACGACACAAT	Test region 2: Enhancer chr6:151936846-151939138
	3C_004	TTCCATGGCATCTTGTGTGT	NC region 1
	3C_005	CAAGAGTGCCTCCTGCTATG	Test region 3: Enhancer chr6:151953109-151955347
	3C_006	ACCGGAACCTTGGAGAGGTA	NC region 2
	3C_007	GCATAATTTCCCGAAAGCA	NC region 3
	3C_008	CCACATGAAATTTGGGTTTTT	Test region 4: Enhancer chr6:151978585-151979706
	3C_009	GGACACAAGAAACATCTGTGA	NC region 4
	3C_010	GGGTAGGGTTCCTGGGATAA	Test region 5: Enhancer chr6:152023958-152025191
	3C_011	CCCACAGAACTTCCCTGAAA	NC region 5
	3C_012	CCCTGAACAAGGCATCGTAT	NC region 6
	3C_013	CAATCCTGCGTGTGCTATGT	NC region 7
	3C_014	TGGGGCTACAITTAAGTGCTTT	NC region 8
	3C_015	ACCTGATGGAGCTGACAACC	NC region 9
	3C_016	CCCCCTCTCCCTCAITTA	Test region 6: Enhancer chr6:152360377-152360720
	3C_017	TTTTGGGATGCACTTAGATGG	Control template generation
	3C_018	TTGGCATTTCATATCATCCA	
	3C_019	TTTTGGGATGCACTTAGATGG	
	3C_020	AGGCATCAAGGTCTCAATCA	
	3C_021	CACCGACCTAGTCTCCCTGA	
	3C_022	GAAAACGGCAGTTCCAAAA	
	3C_023	CAAACACACCTGGTGCATA	
	3C_024	TGGGTGAGGTTTCTTTGCAT	
	3C_025	TGGTAAAGCTCTGAGGGAAAA	
	3C_026	TCCAGGAGCTAATCAAAAACC	
	3C_027	AGTGCCAATTGCTGATGAGA	
	3C_028	TTTGAGGGTTGCCTTGGTTA	
	3C_029	TAACATCCCAGCCCTACCAG	
	3C_030	ATTAGATAGGCGGGCAGA	
	3C_031	TCAGTCTGGCATGGTAGGAA	
	3C_032	TTACATATGGGTGGCAAAG	
	3C_033	TGGAGAATTTGGAGCACTGA	
	3C_034	GCCCCAGGTTCAAGAAGAAG	
	3C_035	TCATTTCCCTGGAACAGGAC	
	3C_036	GCCTGGCACATAGTTTACAAATC	

3C_037	GAGCCACTGAAGCTACACAGC
3C_038	AGGAGGGGACCATCTCATT
3C_039	GTCACCGTGGCAGCTTTTAT
3C_040	ACGCCCTAAAACCTTGTC
3C_041	GCAAGACCTGGCAAATTCAC
3C_042	CCTTTTGTGCCTCTTGCTC
3C_043	TTGTCCTCAATGACTGGCTTA
3C_044	TGTGCAGACAGATGAATGG
3C_045	GCCTGGATTGCAATGATGT
3C_046	ACCTCAAGTTATCCGTTGC
3C_047	TAACACATGTGCTCCCGAAG
3C_048	TGAATGCCAAAAGATGCTG
3C_049	AAAAAGCATAACAAAGGAACAAA
3C_050	TTTCTCTGACTGCTTGAACC
3C_051	GATGGGACTGTGCCACTGAT
3C_052	TTTGTACGGCTGTCTGGTAG
3C_053	CAGCTGAAACCCTAGGCAAG
3C_054	AGAGAGGGCTTCTGGAAAGG
3C_055	GGAACCTTCAATGTGACCAA
3C_056	TTGGCTTCAGCTACATTTGC
3C_057	ATCCAAACACCCAACAGAG
3C_058	GCAATTGGAGTCACCGCTTA
3C_059	CACATCCCGTTCAACTTTC
3C_060	TCCATCCCTTCCCTATATTGC
3C_061	GCAAGGAAGAAAAGAAGACACA
3C_062	CTCCACCTTCATGGTGCAAA
3C_063	GCATCACACTCAATGCATCA
3C_064	TTGGAAAAGGCTTAACTTCAA
3C_065	CAAGGCTTTATCTTGGGGITT
3C_066	GTAATGCGAGCAATGGGAGT
3C_067	CCCAACCCTGGTTAGGAACT
3C_068	TGGTTCCTCACTAAGCCAATC
3C_069	GGATCTGCCACTGTGTTCTT
3C_070	AAAAACTAAGGAAAAATGGAATGAA
3C_071	GCTGCCTCACTGAACACAAA
3C_072	CTTTCAGTGTTTCGGCCAGT
3C_073	CCAGCAAGGCAAGGAATAA
3C_074	GGCTCGATGTGAATCATGTG
3C_075	ATTCTGAGAGCATGGGGGTA
3C_076	CAGCACTGCATCAGGAAAAA
3C_077	AAATCGATGCATAAAAACATCTCA
3C_078	CATGAAATGCTTCCCCAGTT
3C_079	GCGGTACCTCCTTCTCTCT
3C_080	TTGTATTCGGCCATTCTTCC
3C_081	TGCTCAACGGAGACTGCATA
3C_082	CTGTGACAGAACCAGTAGCTT
3C_083	CCTTTATCAGATGGGCTGGA
3C_084	GGTGTA CTGCCTGCCTCTCT
3C_085	ITGCCTCCCACTTTTCTCAC
3C_086	TAITGGGGGTTGGAITTCA
3C_087	GGAGGAGACCGGTTTCATTT
3C_088	CGGGGTCTTCTACATTGCT
3C_089	TTCGACAACGGAGCCATAAT
3C_090	ATATTTCCTTGCCCTGCTT
3C_091	CCAGGCCCGAATCTAATTT
3C_092	TGCTGCAAACCTTTGTGCTTC
3C_093	TTCTCCAAGTGTATCCCAAAGAA
3C_094	GTGGAACATACCTGCCTGCT
3C_095	CTCCCTAAGCCAGGCTCTTT
3C_096	TGGACATGAAATAGAGGCAAG

3C_097	CCACTTTTCACCCTTTTCCA
3C_098	ACTGGTGAGCAAGGAGCCTA
3C_099	TTGCACATTGATGGCAAAA
3C_100	GCTGGAGGAGGAGATCCAAT
3C_101	CCGTGCTGTAAATATAGCCTCA
3C_102	CCTTGTGTTGAGGGAAGCTC
3C_103	GGCAATCATGCCAGATACT
3C_104	CGGTATCAGCTATCCACCATT
3C_105	AGCCACCCAGTTTGTGGTAG
3C_106	AATTGTGACATGTGGAGCCTA
3C_107	GGTCCACAACTAAGCAGAGG
3C_108	GTTGCTGTCTGGTCTCTCT
3C_109	CTTGCCTTCAAATCCCTTG
3C_110	TTGGTGACATCAACTCACC
3C_111	GGGGATAGAGGGGAGGTCT
3C_112	CCATCACTGATAACCTTTCTTCC
3C_113	GATAGGCACTGTGGTCTCAGC
3C_114	GCTGCTGCTGGTAGAGTTCC
3C_115	GGGTTGAATCTGGGGAGACT
3C_116	CCATGGGCACTGTCAGTAAA
3C_117	TGCACCATGACTAGCACACA
3C_118	AGAAATTTCCAGGCAGGGATT
3C_119	TTTTGGCCATTCTAACAGGTG
3C_120	CCTCACACAGAAGACACACA