

**Genome-wide association study in Chinese men identifies two new prostate cancer risk loci  
at 9q31.2 and 19q13.4**

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Supplementary Table 1. Characteristics of subjects in the GWAS and subsequent Replications

Characteristics	GWAS		Replication I		Replication II		Replication III		CAPS	
	Cases (N=1,417)	Controls (N=1,008)	Cases (N=782)	Controls (N=1,792)	Cases (N=1,102)	Controls (N=4,501)	Cases (N=1,183)	Controls (N=1,633)	Cases (N= 2919)	Controls (N=1612)
Age (year) <sup>a</sup>										
Mean (SD)	71.3(8.1)	62.1(10.0)	71.9(8.2)	65.5(7.4)	71.2(7.8)	59.7(16.6)	72.9(8.7)	55.1(11.3)	66.5(7.2)	67.2(7.5)
PSA levels (ng/ml) <sup>b</sup>										
0-3.99	54(4.0)	965(95.9)	20(5.2)	1750(98.8)	33(3.2)	N/A	29(3.4)	N/A	146(5.3)	N/A
4-9.99	187(14.0)	32(3.2)	71(18.5)	16(0.9)	191(18.4)	N/A	119(13.8)	N/A	977(35.5)	N/A
10-19.99	305(22.8)	6(0.6)	85(22.2)	4(0.2)	205(19.8)	N/A	169(19.7)	N/A	640(23.2)	N/A
20-49.99	312(23.3)	3(0.3)	88(23.0)	0	202(19.5)	N/A	195(22.7)	N/A	453(16.5)	N/A
50-99.99	187(14.0)	0	47(12.2)	1(0.06)	150(14.5)	N/A	95(11.0)	N/A	226(8.2)	N/A
≥ 100	292(21.9)	0	72(18.8)	0	257(24.8)	N/A	253(29.4)	N/A	312(11.3)	N/A
Missing	80	2	399	21	64	N/A	323	N/A	165	N/A
T-stage										
T1	180(14.7)	N/A	59(17.9)	N/A	115(20.8)	N/A	59(10.5)	N/A	1057(38.4)	N/A
T2	547(44.7)	N/A	145(44.1)	N/A	305(55.1)	N/A	270(48.2)	N/A	885(32.2)	N/A
T3	359(29.4)	N/A	75(22.8)	N/A	98(17.7)	N/A	172(30.7)	N/A	108(25.7)	N/A
T4	137(11.2)	N/A	50(15.2)	N/A	36(6.5)	N/A	59(10.5)	N/A	102(3.7)	N/A
TX	194	N/A	453	N/A	548	N/A	623	N/A	167	N/A
N-stage										
N0	786(68.1)	N/A	246(74.5)	N/A	152(30.3)	N/A	447(79.1)	N/A	504(84.9)	N/A
N1	369(31.9)	N/A	84(25.5)	N/A	350(69.7)	N/A	118(20.9)	N/A	90(15.1)	N/A
NX	262	N/A	452	N/A	600	N/A	618	N/A	2325	N/A
M-stage										
M0	832(65.4)	N/A	231(68.5)	N/A	167(27.1)	N/A	421(65.3)	N/A	1214(82.0)	N/A
M1	440(34.6)	N/A	106(31.5)	N/A	450(62.9)	N/A	224(34.7)	N/A	267(18.0)	N/A
MX	145	N/A	445	N/A	485	N/A	538	N/A	1438	N/A
Gleason score										
<7	355(26.3)	N/A	162(49.2)	N/A	304(34.4)	N/A	119(19.3)	N/A	1350(52.3)	N/A
≥7	993(73.7)	N/A	167(50.8)	N/A	579(65.6)	N/A	496(80.7)	N/A	1232(47.7)	N/A
Missing	69	N/A	453	N/A	219	N/A	568	N/A	337	N/A

<sup>a</sup> Age at diagnosis for cases or at recruitment for controls;<sup>b</sup> Serum PSA levels were measured at diagnosis for cases or at recruitment for controls.

Supplementary Table 2. Association results in GWAS stage for prostate cancer susceptibility loci identified in populations of European, Japanese or African ancestry.

Chr	SNP (alleles)	Region	Gene	Status	Risk allele <sup>a</sup>	RAF <sup>b</sup>		OR (95%CI) <sup>c</sup>	P <sup>c</sup>
						Case	Control		
Loci identified in populations of European ancestry									
2	rs10187424 (A/G)	2p11	<i>GGCX</i>	Genotyped	A	0.63	0.61	1.05(0.93-1.18)	0.41
2	rs1465618 (A/G)	2p21	<i>THADA</i>	Imputed	A	0.74	0.71	1.16(1.02-1.32)	0.02
2	rs721048 (G/A)	2p15	<i>EHBP1</i>	Genotyped	A	0.04	0.03	1.35(0.97-1.89)	0.07
2	rs7584330 (T/C)	2q37	<i>MLPH</i>	Genotyped	C	0.74	0.73	1.08(0.95-1.23)	0.26
2	rs12621278 (A/G)	2q31	<i>ITGA6</i>	Imputed	A	0.75	0.72	1.13(1.00-1.30)	0.05
3	rs2660753 (C/T)	3p12		Genotyped	T	0.31	0.28	1.12(0.99-1.27)	0.08
3	rs10934853 (C/A)	3q21		Genotyped	A	0.44	0.43	1.06(0.94-1.19)	0.34
3	rs6763931 (C/T)	3q23	<i>ZBTB38</i>	Genotyped	T	0.37	0.34	1.15(1.02-1.30)	0.02
3	rs10936632 (A/C)	3q26		Imputed	A	0.27	0.25	1.09(0.94-1.26)	0.25
4	rs17021918 (C/T)	4q22	<i>PDLIM5</i>	Imputed	C	0.66	0.65	1.04(0.92-1.17)	0.55
4	rs7679673 (A/G)	4q24	<i>TET2</i>	Genotyped	C	0.21	0.19	1.16(1.01-1.34)	0.04
5	rs2121875 (T/G)	5p12	<i>FGF10</i>	Genotyped	G	0.52	0.53	0.96(0.86-1.08)	0.53
6	rs130067 (T/G)	6q21	<i>CCHCR1</i>	Genotyped	G	0.33	0.31	0.98(0.86-1.12)	0.79
6	rs9364554 (C/T)	6q25	<i>SLC22A3</i>	Genotyped	T	0.32	0.34	0.90(0.80-1.02)	0.09
7	rs10486567 (T/C)	7p15	<i>JAZF1</i>	Imputed	C	0.15	0.13	1.24(1.04-1.47)	0.01
7	rs6465657 (C/T)	7q21	<i>LMTK2</i>	Genotyped	C	0.87	0.86	1.14(0.97-1.36)	0.12
8	rs2928679 (G/A)	8p21	<i>NKX3.1</i>	Genotyped	A	0.13	0.12	1.14(0.95-1.35)	0.16
8	rs1512268 (G/A)	8p21	<i>NKX3.1</i>	Genotyped	A	0.32	0.28	1.26(1.11-1.43)	4.82E-4
8	rs10086908 (T/C)	8q24 (Region 5)		Genotyped	T	0.83	0.82	1.08(0.93-1.26)	0.31
8	rs16901979 (C/A)	8q24 (Region 2)		Imputed	A	0.34	0.26	1.41(1.24-1.60)	1.44E-7
8	rs16902094 (A/G)	8q24.21		N/A	G				
8	rs620861 (G/A)	8q24 (Region 4)		N/A	G				
8	rs6983267 (T/G)	8q24 (Region 3)		Genotyped	G	0.47	0.41	1.22(1.09-1.37)	7.27E-4
8	rs1447295 (C/A)	8q24 (Region 1)		Genotyped	A	0.20	0.15	1.41(1.20-1.64)	1.77E-5
9	rs1571801 (G/T)	9q33	<i>DAB2IP</i>	Genotyped	T	0.05	0.05	1.07(0.82-1.38)	0.63
10	rs10993994 (T/C)	10q11	<i>MSMB</i>	Genotyped	T	0.51	0.48	1.11(0.99-1.25)	0.07
10	rs4962416 (A/G)	10q26	<i>CTBP2</i>	N/A	G				
11	rs7127900 (G/A)	11p15	<i>IGF2/IGF2AS</i>	Genotyped	A	0.11	0.12	0.87(0.72-1.03)	0.11
11	rs12418451 (G/A)	11q13		Imputed	A	0.05	0.05	1.11(0.85-1.46)	0.44
11	rs10896449 (A/G)	11q13		Genotyped	G	0.10	0.08	1.19(0.97-1.45)	0.10
12	rs10875943 (T/C)	12q13		Genotyped	C	0.85	0.85	1.03(0.88-1.21)	0.70
17	rs11649743 (C/T)	17q12	<i>HNF1B</i>	Genotyped	C	0.69	0.67	1.07(0.95-1.22)	0.26
17	rs4430796 (T/C)	17q12	<i>HNF1B</i>	Genotyped	T	0.74	0.72	1.09(0.96-1.25)	0.18
17	rs1859962 (T/G)	17q24		Imputed	G	0.43	0.40	1.12(1.00-1.26)	0.06
19	rs8102476 (A/G)	19q13		Genotyped	G	0.37	0.36	1.05(0.94-1.19)	0.35
19	rs887391 (T/C)	19q13		Imputed	T	0.61	0.59	1.03(0.92-1.16)	0.56
19	rs2735839 (G/A)	19q13	<i>KLK2/KLK3</i>	Genotyped	G	0.61	0.62	0.98(0.87-1.10)	0.73
22	rs9623117 (T/C)	22q13	<i>TNRC6B</i>	Imputed	C	0.04	0.03	1.15(0.85-1.56)	0.37
22	rs5759167 (G/T)	22q13	<i>TLL1/BIK</i>	Genotyped	G	0.73	0.71	1.11(0.98-1.27)	0.11
X	rs5945619 (A/G)	Xp11	<i>NUDT10/NUDT11</i>	N/A	G				
Loci identified in populations of Japanese ancestry									
2	rs13385191 (G/A)	2p24	<i>C2orf43</i>	Imputed	G	0.46	0.44	1.10(0.97-1.23)	0.13
3	rs2055109 (T/C)	3p11		Genotyped	C	0.08	0.07	1.20(0.96-1.49)	0.11
5	rs12653946 (C/T)	5p15	<i>IRX4</i>	Genotyped	T	0.41	0.36	1.24(1.10-1.39)	3.86E-4
6	rs1983891 (C/T)	6p21	<i>FOXP4</i>	Imputed	T	0.34	0.33	1.04(0.91-1.18)	0.55
6	rs339331 (T/C)	6q22	<i>GPRC6A/RFX6</i>	Genotyped	T	0.69	0.64	1.27(1.12-1.43)	1.52E-4
10	rs2252004 (G/T)	10q26		Imputed	G	0.77	0.74	1.20(1.04-1.41)	0.01
11	rs1938781 (T/C)	11q12	<i>FAM111A</i>	Imputed	C	0.33	0.31	1.11(0.98-1.26)	0.11
13	rs9600079 (G/T)	13q22		Genotyped	T	0.49	0.44	1.19(1.06-1.34)	3.75E-3
Locus identified in populations of African ancestry									
17	rs7210100 (G/A)	17q21	<i>ZNF652</i>	N/A	A				

<sup>a</sup>Reported risk allele; <sup>b</sup>Risk allele frequency; <sup>c</sup>Derived from additive model with adjustment for age and top eigen.

Supplementary Table 3. Selected SNPs suggestively associated with prostate cancer risk in the GWAS stage and from Replication I.

Chr.	SNP	Position	Alleles <sup>a</sup>	GWAS (N <sub>case/control</sub> =1,417/1,008)			Replication I (N <sub>case/control</sub> =782/1,792)			Combined	
				MAF <sub>case/control/ref.</sub> <sup>b</sup>	OR <sub>add</sub> (95%CI) <sup>c</sup>	P <sub>add</sub> <sup>c</sup>	MAF <sub>case/control</sub> <sup>d</sup>	OR <sub>add</sub> (95%CI) <sup>c</sup>	P <sub>add</sub> <sup>c</sup>	OR <sub>add</sub> (95%CI) <sup>e</sup>	P <sub>add</sub> <sup>e</sup>
1	rs4650310	69218513	A/G	0.161/0.206/0.204	0.75(0.65 - 0.87)	1.43E-04	0.175/0.184	0.94(0.80 - 1.10)	0.434	0.84(0.75 - 0.93)	9.64E-04
1	rs1465132	104260408	C/T	0.269/0.317/0.313	0.80(0.70 - 0.90)	4.06E-04	0.302/0.284	1.09(0.96 - 1.24)	0.190	0.93(0.85 - 1.02)	0.103
1	rs12130030	161465956	C/T	0.068/0.099/0.086	0.68(0.55 - 0.83)	2.20E-04	0.086/0.066	1.32(1.06 - 1.65)	0.013	0.93(0.80 - 1.08)	0.332
1	rs12029534	219622780	A/G	0.196/0.236/0.224	0.79(0.69 - 0.91)	7.97E-04	0.197/0.208	0.93(0.80 - 1.08)	0.354	0.85(0.77 - 0.94)	2.06E-03
1	rs12023401	227400917	A/G	0.120/0.160/0.141	0.70(0.60 - 0.83)	4.22E-05	0.128/0.134	0.95(0.79 - 1.13)	0.533	0.81(0.72 - 0.91)	6.73E-04
1	rs6685619	235198820	G/T	0.344/0.392/0.389	0.81(0.72 - 0.92)	8.63E-04	0.404/0.374	1.14(1.01 - 1.28)	0.041	0.96(0.88 - 1.05)	0.374
1	rs6429451	242394714	G/A	0.381/0.446/0.429	0.75(0.66 - 0.84)	1.50E-06	0.405/0.397	1.04(0.92 - 1.17)	0.574	0.88(0.81 - 0.96)	2.46E-03
2	rs992618	45241278	T/C	0.337/0.391/0.379	0.80(0.71 - 0.91)	3.08E-04	0.335/0.367	0.87(0.77 - 0.99)	0.031	0.84(0.77 - 0.91)	4.06E-05
2	rs7580494	62635003	C/T	0.435/0.379/0.398	1.27(1.13 - 1.43)	7.82E-05	0.411/0.407	1.02(0.90 - 1.15)	0.784	1.14(1.05 - 1.24)	2.67E-03
2	rs264927	103647704	C/T	0.381/0.332/0.329	1.23(1.09 - 1.39)	6.89E-04	0.371/0.360	1.05(0.93 - 1.19)	0.447	1.14(1.04 - 1.24)	3.16E-03
2	rs16823914	183567718	T/C	0.169/0.211/0.200	0.77(0.66 - 0.89)	3.28E-04	0.195/0.174	1.15(0.99 - 1.34)	0.067	0.93(0.84 - 1.03)	0.186
3	rs6796957	66889884	A/G	0.442/0.383/0.402	1.28(1.13 - 1.44)	5.21E-05	0.424/0.406	1.08(0.95 - 1.21)	0.239	1.17(1.08 - 1.28)	2.07E-04
3	rs9836404	87254695	G/A	0.153/0.119/0.122	1.34(1.13 - 1.59)	6.01E-04	0.148/0.127	1.20(1.01 - 1.42)	0.041	1.27(1.12 - 1.43)	1.05E-04
3	rs4280673	123008797	C/T	0.091/0.061/0.051	1.55(1.23 - 1.95)	2.01E-04	0.024/0.025	0.96(0.65 - 1.41)	0.819	1.37(1.12 - 1.67)	2.00E-03
3	rs2335052	129687641	C/T	0.345/0.401/0.415	0.79(0.70 - 0.89)	8.38E-05	0.382/0.411	0.89(0.79 - 1.00)	0.058	0.84(0.77 - 0.91)	3.40E-05
3	rs9824588	154505470	C/T	0.131/0.086/0.099	1.61(1.33 - 1.96)	1.39E-06	0.107/0.104	1.04(0.85 - 1.26)	0.718	1.30(1.13 - 1.49)	2.19E-04
4	rs753972	106573233	G/A	0.297/0.355/0.339	1.27(0.66 - 0.86)	1.43E-05	0.347/0.330	1.02(0.90 - 1.22)	0.239	0.91(0.83 - 0.99)	0.029
4	rs11728665	146451823	T/C	0.446/0.403/0.409	1.22(1.09 - 1.38)	7.95E-04	0.433/0.428	1.02(0.91 - 1.15)	0.723	1.12(1.03 - 1.22)	8.14E-03
4	rs7692951	166636093	A/G	0.317/0.260/0.280	1.31(1.15 - 1.49)	4.03E-05	0.298/0.291	1.03(0.91 - 1.18)	0.616	1.17(1.06 - 1.28)	1.05E-03
5	rs6881843	31116520	T/G	0.186/0.141/0.155	1.41(1.20 - 1.66)	2.55E-05	0.167/0.160	1.05(0.90 - 1.23)	0.543	1.22(1.09 - 1.36)	6.70E-04
6	rs898165	21316408	G/A	0.198/0.244/0.229	0.77(0.67 - 0.89)	3.69E-04	0.051/0.047	1.10(0.84 - 1.45)	0.484	0.83(0.74 - 0.94)	4.41E-03
6	rs13218904	153997386	A/G	0.310/0.357/0.375	0.82(0.72 - 0.92)	9.93E-04	0.338/0.330	1.04(0.91 - 1.17)	0.594	0.91(0.84 - 1.00)	0.045
7	rs9691259	46213105	A/G	0.233/0.292/0.272	0.75(0.66 - 0.86)	2.07E-05	0.238/0.260	0.89(0.77 - 1.02)	0.093	0.81(0.74 - 0.90)	2.08E-05
8	rs2980945	6703161	A/G	0.190/0.155/0.163	1.31(1.12 - 1.53)	6.28E-04	0.181/0.162	1.14(0.98 - 1.34)	0.098	1.22(1.10 - 1.37)	3.25E-04
8	rs2241881	70661462	T/C	0.181/0.144/0.149	1.33(1.13 - 1.56)	4.90E-04	0.161/0.159	1.02(0.86 - 1.19)	0.849	1.17(1.04 - 1.31)	8.50E-03
9	rs817826	109196121	T/C	0.107/0.075/0.084	1.43(1.16 - 1.77)	6.82E-04	0.110/0.082	1.39(1.14 - 1.70)	1.05E-03	1.41(1.22 - 1.63)	2.40E-06
11	rs1919302	14089869	A/G	0.189/0.148/0.157	1.30(1.11 - 1.52)	8.93E-04	0.179/0.152	1.22(1.04 - 1.42)	0.016	1.26(1.13 - 1.41)	4.95E-05
11	rs10742620	42253844	G/T	0.214/0.255/0.245	0.79(0.69 - 0.91)	6.85E-04	0.238/0.250	0.94(0.82 - 1.08)	0.371	0.86(0.78 - 0.95)	2.24E-03
11	rs7944576	88738911	C/T	0.149/0.198/0.184	0.72(0.62 - 0.84)	3.65E-05	0.167/0.182	0.90(0.77 - 1.05)	0.190	0.80(0.72 - 0.90)	1.07E-04
12	rs7980991	6415678	A/C	0.450/0.503/0.511	0.80(0.71 - 0.90)	2.18E-04	0.486/0.485	1.00(0.89 - 1.13)	0.962	0.89(0.82 - 0.97)	8.13E-03
12	rs17124060	59366306	T/G	0.278/0.329/0.317	0.80(0.70 - 0.90)	3.47E-04	0.288/0.312	0.89(0.78 - 1.01)	0.079	0.84(0.77 - 0.92)	1.47E-04
14	rs3742943	104679609	C/T	0.145/0.189/0.180	0.74(0.63 - 0.86)	1.32E-04	0.185/0.170	1.11(0.95 - 1.30)	0.180	0.90(0.81 - 1.01)	0.075
15	rs1563894	66422829	A/G	0.114/0.150/0.144	0.74(0.62 - 0.87)	3.87E-04	0.124/0.128	0.96(0.81 - 1.15)	0.689	0.84(0.74 - 0.95)	4.29E-03
19	rs7260240	6271963	G/A	0.139/0.101/0.121	1.43(1.20 - 1.72)	9.73E-05	0.136/0.122	1.13(0.95 - 1.35)	0.160	1.27(1.12 - 1.44)	1.98E-04
19	rs103294	59489660	T/C	0.301/0.242/0.249	1.28(1.12 - 1.45)	2.94E-04	0.304/0.237	1.40(1.23 - 1.60)	4.98E-07	1.34(1.22 - 1.47)	9.88E-10
X	rs5978499	12116360	G/A	0.428/0.351/--	1.38(1.16 - 1.63)	1.83E-04	0.345/0.384	0.84(0.71 - 1.01)	0.056	1.09(0.97 - 1.23)	0.166
X	rs1476499	12124778	G/A	0.279/0.205/--	1.48(1.22 - 1.80)	6.45E-05	0.212/0.240	0.85(0.69 - 1.06)	0.155	1.16(1.01 - 1.34)	0.042
X	rs3008916	39609316	A/G	0.201/0.265/--	0.70(0.58 - 0.85)	3.09E-04	0.253/0.242	1.06(0.87 - 1.29)	0.545	0.86(0.75 - 0.99)	0.034
X	rs12688535	62663781	A/G	0.298/0.376/--	0.70(0.59 - 0.84)	7.03E-05	0.322/0.337	0.93(0.78 - 1.12)	0.463	0.81(0.71 - 0.91)	7.35E-04
X	rs17279546	87285589	A/G	0.131/0.186/--	0.67(0.54 - 0.84)	4.72E-04	0.124/0.146	0.82(0.64 - 1.07)	0.139	0.73(0.62 - 0.87)	3.07E-04
X	rs11093017	87378976	G/A	0.067/0.110/--	0.58(0.44 - 0.78)	2.40E-04	0.091/0.078	1.19(0.88 - 1.60)	0.256	0.82(0.67 - 1.01)	0.064
X	rs12014732	91407032	G/A	0.291/0.358/--	0.74(0.62 - 0.88)	7.62E-04	0.322/0.341	0.92(0.77 - 1.10)	0.339	0.82(0.73 - 0.93)	2.06E-03
X	rs3924343	102527323	G/A	0.286/0.361/--	0.71(0.60 - 0.84)	1.09E-04	0.317/0.338	0.91(0.76 - 1.09)	0.309	0.80(0.71 - 0.91)	4.81E-04

<sup>a</sup> Major/Minor allele; <sup>b</sup> Minor allele frequency (MAF) among cases and controls in GWAS stage and reported GWAS controls (case/control/ref.); <sup>c</sup> Odds ratios (ORs), 95% confidence intervals (95%CI) and corresponding *P* values were estimated in an additive model; <sup>d</sup> Minor allele frequency (MAF) among cases and controls in Replication I stage; <sup>e</sup> Results from GWAS and Replication I were combined by meta-analysis.

Supplementary Table 4. SNPs at 9q31.2 associated with prostate cancer risk at  $P < 0.01$  in the GWAS stage

SNP	Position	Status	Alleles <sup>a</sup>	MAF <sup>b</sup> <sub>case/control</sub>	OR <sub>add</sub> (95%CI) <sup>c</sup>	$P_{add}$ <sup>c</sup>	LD <sup>d</sup>	$P_{conditioned}$ <sup>e</sup>
rs817864	109182469	Imputed	G/A	0.346/0.307	1.20(1.06 - 1.36)	4.24E-03	0.166	0.090
rs817865	109183520	Imputed	G/A	0.346/0.305	1.21(1.07 - 1.37)	2.37E-03	0.165	0.069
rs817866	109183552	Imputed	C/T	0.346/0.305	1.21(1.07 - 1.37)	2.37E-03	0.165	0.069
rs28414294	109183798	Imputed	A/G	0.044/0.027	1.57(1.13 - 2.19)	7.73E-03	0.393	0.710
rs817867	109183892	Imputed	T/G	0.346/0.305	1.21(1.07 - 1.37)	2.40E-03	0.165	0.069
rs1746814	109184113	Imputed	C/T	0.346/0.305	1.21(1.07 - 1.37)	2.40E-03	0.165	0.069
rs817868	109184208	Imputed	A/T	0.347/0.306	1.21(1.07 - 1.37)	2.50E-03	0.167	0.072
rs817869	109184256	Imputed	T/C	0.347/0.306	1.21(1.07 - 1.37)	2.34E-03	0.167	0.069
rs817870	109184509	Genotyped	T/C	0.348/0.308	1.20(1.06 - 1.36)	3.27E-03	0.165	0.086
rs7870307	109184767	Imputed	T/C	0.351/0.311	1.20(1.06 - 1.36)	4.33E-03	0.166	0.097
rs4366138	109184882	Imputed	T/C	0.347/0.306	1.21(1.07 - 1.37)	2.34E-03	0.167	0.069
rs2417788	109184918	Imputed	C/T	0.347/0.306	1.21(1.07 - 1.37)	2.31E-03	0.167	0.068
rs1746815	109185022	Imputed	C/T	0.347/0.306	1.21(1.07 - 1.37)	2.31E-03	0.167	0.068
rs1746816	109185147	Imputed	C/A	0.347/0.305	1.21(1.07 - 1.37)	2.23E-03	0.167	0.066
rs1746817	109185202	Imputed	G/A	0.347/0.306	1.21(1.07 - 1.37)	2.31E-03	0.167	0.068
rs1771727	109185554	Imputed	G/C	0.347/0.306	1.21(1.07 - 1.37)	2.31E-03	0.167	0.068
rs1323804	109186054	Imputed	G/A	0.347/0.306	1.21(1.07 - 1.37)	2.48E-03	0.167	0.071
rs1323805	109186118	Imputed	G/T	0.347/0.306	1.21(1.07 - 1.37)	2.48E-03	0.167	0.071
rs12348903	109186290	Imputed	C/T	0.347/0.306	1.21(1.07 - 1.37)	2.48E-03	0.167	0.071
rs12336356	109186300	Imputed	T/C	0.347/0.306	1.21(1.07 - 1.37)	2.48E-03	0.167	0.071
rs10978820	109186310	Imputed	G/A	0.348/0.307	1.20(1.07 - 1.36)	3.05E-03	0.167	0.079
rs4978922	109186451	Imputed	G/A	0.347/0.305	1.21(1.07 - 1.37)	2.52E-03	0.170	0.080
rs4978923	109186599	Imputed	A/C	0.346/0.305	1.21(1.07 - 1.37)	2.60E-03	0.170	0.079
rs4978924	109186659	Imputed	G/A	0.347/0.305	1.21(1.07 - 1.37)	2.64E-03	0.170	0.082
rs10114711	109186875	Imputed	T/C	0.347/0.305	1.21(1.07 - 1.37)	2.57E-03	0.170	0.079
rs10123735	109187061	Imputed	G/A	0.345/0.305	1.20(1.06 - 1.36)	3.80E-03	0.170	0.093
rs28649013	109187212	Imputed	A/G	0.34/0.3	1.20(1.06 - 1.36)	4.06E-03	0.175	0.105
rs12156485	109187268	Imputed	C/A	0.34/0.3	1.20(1.06 - 1.36)	4.17E-03	0.175	0.108
rs12156453	109187323	Imputed	C/T	0.34/0.3	1.20(1.06 - 1.36)	4.17E-03	0.175	0.108
rs11794415	109187420	Imputed	C/G	0.346/0.305	1.20(1.06 - 1.36)	3.59E-03	0.171	0.089
rs1617227	109187446	Imputed	T/C	0.335/0.296	1.20(1.06 - 1.36)	5.36E-03	0.181	0.140
rs1746821	109187726	Imputed	G/A	0.34/0.3	1.20(1.06 - 1.36)	4.17E-03	0.175	0.108
rs1746822	109188006	Imputed	G/A	0.335/0.295	1.21(1.06 - 1.37)	3.89E-03	0.166	0.128
rs1746823	109188131	Imputed	C/T	0.337/0.296	1.21(1.07 - 1.38)	2.86E-03	0.176	0.083
rs1746824	109188193	Imputed	T/C	0.34/0.3	1.20(1.06 - 1.36)	4.17E-03	0.175	0.108
rs1746825	109188377	Imputed	C/T	0.34/0.3	1.20(1.06 - 1.36)	3.97E-03	0.175	0.105
rs1771726	109188412	Imputed	A/G	0.34/0.3	1.20(1.06 - 1.36)	3.97E-03	0.175	0.105
rs1771725	109188634	Imputed	T/A	0.34/0.3	1.20(1.06 - 1.36)	3.97E-03	0.175	0.105
rs1746826	109188712	Imputed	G/A	0.34/0.3	1.20(1.06 - 1.36)	3.97E-03	0.175	0.105
rs1746827	109188989	Imputed	G/A	0.339/0.3	1.20(1.06 - 1.36)	5.05E-03	0.180	0.145
rs1746828	109189133	Imputed	T/C	0.343/0.304	1.20(1.06 - 1.35)	4.82E-03	0.175	0.131
rs1746829	109189163	Imputed	T/A	0.343/0.301	1.21(1.07 - 1.38)	2.79E-03	0.180	0.099
rs1622267	109190480	Genotyped	G/A	0.197/0.161	1.25(1.08 - 1.45)	3.62E-03	0.345	0.256
rs1771716	109191394	Imputed	G/C	0.196/0.157	1.28(1.10 - 1.49)	1.55E-03	0.350	0.191
rs1746831	109191435	Imputed	G/T	0.3/0.257	1.24(1.09 - 1.42)	1.58E-03	0.231	0.095
rs1771715	109191489	Imputed	G/A	0.191/0.153	1.28(1.10 - 1.49)	1.91E-03	0.407	0.248
rs817834	109191592	Imputed	C/T	0.118/0.091	1.32(1.09 - 1.60)	4.84E-03	0.107	0.076
rs843865	109192688	Imputed	G/T	0.043/0.028	1.57(1.12 - 2.18)	8.00E-03	0.384	0.474
rs817831	109193494	Imputed	T/G	0.1/0.07	1.44(1.16 - 1.79)	9.59E-04	0.995	N/A
rs1746832	109195006	Imputed	T/C	0.282/0.236	1.26(1.10 - 1.44)	8.96E-04	0.268	0.072
rs1771714	109195009	Imputed	A/G	0.042/0.026	1.62(1.15 - 2.27)	5.42E-03	0.378	0.391
rs817829	109195179	Genotyped	A/G	0.23/0.198	1.22(1.06 - 1.40)	6.67E-03	0.029	0.029
rs817827	109196100	Genotyped	T/C	0.473/0.518	0.83(0.74 - 0.94)	2.14E-03	0.106	0.037
rs817826	109196121	Genotyped	T/C	0.107/0.075	1.43(1.17 - 1.77)	6.82E-04	N/A	N/A
rs817823	109199999	Imputed	A/G	0.043/0.026	1.67(1.19 - 2.35)	2.90E-03	0.388	0.332

<sup>a</sup> Major/Minor allele; <sup>b</sup> Minor allele frequency (MAF) between cases and controls; <sup>c</sup> Odds ratios (ORs), 95% confidence intervals (95%CI) and corresponding  $P$  values were estimated using a logistic regression model with adjustment for age and the top eigen in an additive model; <sup>d</sup> Linkage disequilibrium ( $r$ -square) between each SNP at 9q31.2 and the index SNP of rs817826; <sup>e</sup>  $P$  values were estimated using a logistic regression model after conditioning on rs817826.

Supplementary Table 5. Summary of fine-mapping study at 19q13.4 and association results with prostate cancer risk in all of the samples in the GWAS stage.

SNP	Position	Alleles <sup>a</sup>	MAF <sub>case/control</sub> <sup>b</sup>	OR <sub>add</sub> (95%CI) <sup>c</sup>	<i>P</i> <sub>add</sub> <sup>c</sup>	LD <sup>d</sup>	<i>P</i> <sub>conditioned</sub> <sup>e</sup>
rs6509859	59454220	C/T	0.067/0.054	1.21(0.95-1.54)	0.126	0.10/0.08	0.62/0.55
rs595872	59455781	A/G	0.260/0.220	1.19(1.04-1.37)	0.014	0.30/0.25	0.60/0.41
rs402071	59459691	C/T	0.152/0.136	1.08(0.91-1.28)	0.390	0.23/0.19	0.27/0.41
rs17273295	59466404	C/T	0.155/0.142	1.05(0.89-1.24)	0.574	0.22/0.22	0.28/0.24
rs7254645	59467201	C/T	0.168/0.152	1.06(0.90-1.25)	0.478	0.27/0.22	0.13/0.22
rs444004	59471267	A/G	0.027/0.026	0.92(0.64-1.32)	0.643	0.06/0.05	0.20/0.18
rs4022330	59473655	G/A	0.177/0.151	1.17(0.99-1.38)	0.068	0.29/0.28	0.95/0.96
rs3848607	59480959	G/A	0.151/0.156	0.96(0.82-1.13)	0.640	0.04/0.05	0.80/0.54
rs8100863	59481078	G/T	0.047/0.035	1.26(0.93-1.71)	0.135	0.11/0.08	0.52/0.51
rs397600	59483600	C/T	0.079/0.068	1.11(0.88-1.39)	0.394	0.18/0.18	0.55/0.51
rs6509860	59484170	A/G	0.070/0.061	1.09(0.86-1.38)	0.498	0.17/0.14	0.49/0.57
rs798887	59485000	G/A	0.150/0.132	1.12(0.93-1.34)	0.236	0.56/0.47	0.80/0.67
rs117311656	59485734	T/G	0.053/0.050	1.09(0.83-1.42)	0.550	0.13/0.12	0.49/0.58
rs427366	59487111	C/T	0.096/0.085	1.11(0.91-1.36)	0.303	0.26/0.22	0.33/0.45
rs17305346	59487148	G/A	0.115/0.090	1.25(1.03-1.52)	0.022	0.31/0.25	0.88/0.69
rs103294	59489660	T/C	0.301/0.242	1.28(1.12-1.45)	2.94E-04	N/A /0.83	N/A/0.67
rs368178	59492034	C/T	0.119/0.087	1.29(1.06-1.57)	0.012	0.27/0.28	0.32/0.34
rs410852	59492183	C/T	0.119/0.089	1.26(1.04-1.53)	0.021	0.30/0.26	0.52/0.54
Deletion	59492668	Non-del/Del	0.307/0.252	1.25(1.10-1.43)	9.60E-04	0.83/ N/A	0.31/N/A
rs453755	59501155	C/A	0.215/0.165	1.30(1.12-1.52)	8.43E-04	0.56/0.59	0.19/0.25
rs117372111	59501324	T/C	0.060/0.064	0.93(0.72-1.18)	0.541	0.01/0.02	0.83/0.80

<sup>a</sup> Major/Minor allele; <sup>b</sup> Minor allele frequency (MAF) between cases and controls; <sup>c</sup> Odds ratios (ORs), 95% confidence intervals (95%CI) and corresponding *P* values were estimated using a logistic regression model in an additive model; <sup>d</sup> Linkage disequilibrium (r-square) between each variant at 19q13.4 and variants of rs103294/deletion, respectively; <sup>e</sup> *P* values were derived from logistic regression with an additive model conditioned by rs103294/deletion, respectively.

Supplementary Table 6. The relationship between rs817826 at 9q31.2 and rs103294 at 19q13.4 and prostate cancer clinical features

Marker (alleles <sup>a</sup> )	Phenotype	MAF <sup>b</sup>				OR <sub>combined</sub> (95%CI) <sup>c</sup>	P <sub>combined</sub> <sup>c</sup>
		GWAS	Replication I	Replication II	Replication III		
rs817826 (T/C)	PSA levels						
	<10 ng/ml	0.095	0.077	0.121	0.103	Ref.	
	≥10 ng/ml	0.107	0.115	0.117	0.115	1.10(0.91-1.34)	0.31
	Gleason score						
	<7	0.116	0.093	0.122	0.097	Ref.	
	≥7	0.105	0.111	0.117	0.110	0.98(0.83-1.17)	0.84
	T-stage						
	T1-2	0.111	0.093	0.118	0.108	Ref.	
	T3-4	0.100	0.120	0.090	0.109	0.94(0.78-1.13)	0.49
	N-stage						
	N0	0.102	0.087	0.115	0.112	Ref.	
	N1	0.108	0.131	0.113	0.107	1.08(0.89-1.32)	0.44
	M-stage						
	M0	0.106	0.093	0.105	1.115	Ref.	
M1	0.105	0.118	0.11	0.105	1.01(0.85-1.21)	0.88	
Aggressiveness <sup>e</sup>							
Nonaggressive	0.113	0.103	0.120	0.097	Ref.		
Aggressive	0.106	0.113	0.115	0.115	1.01(0.84-1.21)	0.92	
rs103294 (T/C)	PSA levels						
	<10 ng/ml	0.295	0.330	0.255	0.277	Ref.	
	≥10 ng/ml	0.297	0.272	0.279	0.288	1.02(0.89-1.16)	0.80
	Gleason score						
	<7	0.320	0.283	0.252	0.298	Ref.	
	≥7	0.294	0.314	0.274	0.294	1.00(0.88-1.13)	0.97
	T-stage						
	T1-2	0.306	0.298	0.261	0.297	Ref.	
	T3-4	0.278	0.292	0.301	0.270	0.94(0.83-1.06)	0.31
	N-stage						
	N0	0.298	0.306	0.319	0.295	Ref.	
	N1	0.313	0.262	0.254	0.256	0.92(0.80-1.05)	0.20
	M-stage						
	M0	0.297	0.294	0.299	0.282	Ref.	
M1	0.301	0.278	0.260	0.280	0.96(0.85-1.09)	0.52	
Aggressiveness <sup>e</sup>							
Nonaggressive	0.311	0.317	0.268	0.279	Ref.		
Aggressive	0.298	0.284	0.271	0.286	0.96(0.85-1.09)	0.54	

<sup>a</sup> Major alleles/minor alleles; <sup>b</sup> Minor allele frequency (MAF); <sup>c</sup> Combined results from GWAS and replications in additive models by meta-analysis; <sup>d</sup> Odds ratios (ORs) and 95% confidence intervals (95%CI) were estimated using a logistic regression model based on additive model; <sup>e</sup> Patients with a Gleason score of 7 or higher, a stage of T3/T4, N+, or M+, were classified as having aggressive disease, whereas all other patients were classified as having nonaggressive disease.

Supplementary Table 7. Summary results of association with prostate cancer risk at 9q31.2 and 19q13.4 in Populations of European ancestry

Marker (alleles)	Location (gene)	Study	TT/TC/CC genotypes		C allele frequency		OR (95%CI) <sup>a</sup>	P <sup>a</sup>
			Cases	Controls	Cases	Controls		
rs817826 (T/C)	9q31.2 ( <i>RAD23B/KLF4</i> )	CAPS	2160/667/56	1241/315/32	0.135	0.119	1.15(1.01-1.31)	0.036
		CGEMS	855/299/22	816/271/14	0.146	0.136	1.09(0.92-1.29)	0.323
		Combined <sup>b</sup>					1.13(1.02-1.25)	0.023
rs103294 (T/C)	19q13.4 ( <i>LILRA3</i> )	CAPS	177/1117/1590	113/580/897	0.745	0.747	0.99(0.90-1.10)	0.871
		CGEMS	47/397/732	40/370/690	0.791	0.795	0.97(0.84-1.13)	0.722
		Combined <sup>b</sup>					0.99(0.91-1.07)	0.738

<sup>a</sup> Odds ratios (ORs), 95% confidence intervals (95%CIs) and corresponding *P* value in additive model (CC versus TC versus TT) were estimated using logistic regression model for additive model; <sup>b</sup> Results in additive model from two studies were combined by meta-analysis using a fixed-effect model under the absence of heterogeneity among studies for both loci (rs817826: *P* = 0.63, *I*<sup>2</sup> = 0; rs103294: *P* = 0.84, *I*<sup>2</sup> = 0).



Supplementary Table 8. Expression quantitative trait loci (eQTL) analysis for rs817826 at 9q31.2 and rs103294 at 19q13.4<sup>a</sup>

Probe ID	Gene	Probe Start	Cell types			Tissue types (Twin1)			Tissue types (Twin2)			
			Fibroblast	Lymphoblastoid	T	Fat	Lymphoblastoid	Skin	Fat	Lymphoblastoid	Skin	
rs817826 at 9q31.2												
ILMN_1686679	<i>ZNF462</i>	108673302	0.296	0.588	0.005	0.833	0.276	0.339	0.176	0.203	0.333	
ILMN_1722662	<i>RAD23B</i>	109089103	0.670	0.308	0.192	0.964	0.467	0.658	0.009	0.051	0.451	
ILMN_2137789	<i>KLF4</i>	109289352	0.826	0.529	0.473	0.729	0.141	0.088	0.999	0.440	0.914	
ILMN_1779857	<i>KLF4</i>	109289583	0.717	0.340	0.410	0.043	0.157	0.718	0.702	0.558	0.426	
rs103294 at 19q13.4												
ILMN_2078404	<i>ZNF765</i>	58594190				0.445	0.376	0.160	0.353	0.462	0.170	
ILMN_1764854	<i>ZNF761</i>	58652225	0.424	0.681	0.672	0.776	0.127	0.693	0.509	0.426	0.784	
ILMN_1749809	<i>ZNF813</i>	58666798				0.591	0.013	0.442	0.430	0.356	0.314	
ILMN_2269193	<i>ZNF331</i>	58716841	0.210	0.844	0.702	0.332	0.008	0.263	0.995	0.484	0.806	
ILMN_1711199	<i>ZNF331</i>	58719957	0.522	0.866	0.730	0.596	0.276	0.214	0.568	0.071	0.809	
ILMN_2362232	<i>ZNF331</i>	58720480	0.957	0.735	0.748	0.914	0.983	0.653	0.099	0.743	0.354	
ILMN_2051483	<i>DPRXP4</i>	58827413				0.122	0.754	0.677	0.753	0.748	0.968	
ILMN_1739532	<i>DPRXP4</i>	58827508				0.906	0.011	0.295	0.380	0.458	0.118	
ILMN_1653158	<i>DPRXP4</i>	58827689	0.745	0.599	0.153	0.592	0.678	0.287	0.006	0.315	0.640	
ILMN_1716105	<i>NLRP12</i>	59016112	0.991	0.188	0.919	0.024	0.179	0.697	0.824	0.147	0.867	
ILMN_1758735	<i>NLRP12</i>	59016511	0.637	0.263	0.542	0.994	0.933	0.407	0.626	0.397	0.178	
ILMN_1739815	<i>NLRP12</i>	59018880	0.760	0.180	0.892	0.087	0.185	0.778	0.737	0.372	0.678	
ILMN_1675345	<i>MYADM</i>	59061425	0.062	0.350	0.232	0.122	0.187	0.916	0.187	0.416	0.163	
ILMN_2350574	<i>MYADM</i>	59064085	0.400	0.723	0.645	0.875	0.614	0.737	0.534	0.650	0.369	
ILMN_2308849	<i>MYADM</i>	59064217	0.234	0.161	0.110	0.383	0.323	0.845	0.112	0.069	0.548	
ILMN_1739812	<i>PRKCG</i>	59080125	0.006	0.801	0.142	0.529	0.858	0.402	0.367	0.872	0.872	
ILMN_1669816	<i>CACNG7</i>	59105872	0.669	0.598	0.698	0.338	0.049	0.928	0.453	0.669	0.913	
ILMN_1776079	<i>CACNG8</i>	59158853	0.490	0.779	0.056	0.996	0.828	0.664	0.056	0.666	0.818	
ILMN_2106874	<i>CACNG8</i>	59159437	0.425	0.944	0.018	0.555	0.385	0.882	0.191	0.746	0.915	
ILMN_1809676	<i>CACNG6</i>	59188437	0.230	0.917	0.706	0.741	0.698	0.379	0.967	0.172	0.091	
ILMN_2356441	<i>CACNG6</i>	59188533	0.589	0.939	0.281	0.879	0.080	0.240	0.347	0.593	0.621	
ILMN_1779043	<i>CACNG6</i>	59188874	0.483	0.092	0.636	0.947	0.432	0.919	0.095	0.585	0.832	
ILMN_1763455	<i>VSTM1</i>	59258264	0.364	0.009	0.359	0.824	0.676	0.556	0.146	0.152	0.354	
ILMN_1784641	<i>NDUFA3</i>	59282252	0.793	0.458	0.363	0.085	0.185	0.218	0.888	0.402	0.866	
ILMN_2367418	<i>OSCAR</i>	59296737	0.992	0.431	0.719	0.698	0.742	0.949	0.041	0.435	0.483	
ILMN_1741917	<i>OSCAR</i>	59296829	0.241	0.115	0.875	0.965	0.341	0.736	0.372	0.896	0.928	
ILMN_1709451	<i>TFPT</i>	59309986	0.378	0.167	0.106	0.244	0.269	0.448	0.786	0.621	0.608	
ILMN_1719204	<i>PRPF31</i>	59312451	0.848	0.490	0.917	0.418	0.431	0.342	0.852	0.682	0.846	
ILMN_2207393	<i>CNOT3</i>	59335678	0.417	0.693	0.753	0.086	0.931	0.563	0.399	0.765	0.952	
ILMN_1679238	<i>LENG1</i>	59354714	0.709	0.295	0.128	0.346	0.370	0.399	0.063	0.804	0.890	
ILMN_1803219	<i>TMC4</i>	59366497	0.600	0.312	0.993	0.544	0.171	0.419	0.891	0.864	0.110	
ILMN_1722218	<i>MBOAT7</i>	59383288	0.946	0.780	0.827	0.059	0.789	0.907	0.345	0.638	0.293	
ILMN_1673111	<i>TSEN34</i>	59387047	0.980	0.580	0.713	0.144	0.496	0.507	0.436	0.965	0.378	
ILMN_2368292	<i>TSEN34</i>	59387266	0.414	0.666	0.341	0.249	0.599	0.965	0.326	0.994	0.546	
ILMN_2038772	<i>RPS9</i>	59396730	0.344	0.438	0.414	0.216	0.280	0.565	0.692	0.395	0.740	

ILMN_1749447	<i>RPS9</i>	59396734	0.329	0.731	0.279	0.340	0.411	0.822	0.421	0.351	0.609
ILMN_2406132	<i>LILRB3</i>	59416845	0.333	0.867	0.849	0.553	0.586	0.866	0.182	0.145	0.497
ILMN_1784884	<i>LILRB3</i>	59417065	0.506	0.592	0.992	0.740	0.239	0.964	0.905	0.217	0.738
ILMN_2058841	<i>LILRA6</i>	59435687	0.288	0.665	0.915	0.680	0.382	0.876	0.937	0.421	0.331
ILMN_1694243	<i>LILRA6</i>	59437530	0.103	0.961	0.429	0.664	0.612	0.750	0.054	0.526	0.743
ILMN_1651404	<i>LILRA6</i>	59437708	0.641	0.159	0.483	0.151	0.732	0.124	0.274	0.672	0.634
ILMN_2339294	<i>LILRB5</i>	59450972	0.144	0.049	0.247	0.882	0.696	0.665	0.342	0.283	0.235
ILMN_1732919	<i>LILRB5</i>	59451098	0.223	0.823	0.963	0.685	0.812	0.691	0.549	0.847	0.700
ILMN_2312340	<i>LILRB2</i>	59474228	0.515	0.397	0.898	0.520	0.704	0.549	0.550	0.664	0.072
ILMN_1734234	<i>LILRB2</i>	59474679	0.256	0.524	0.417	0.488	0.732	0.300	0.301	0.676	0.931
ILMN_1695744	<i>LILRB2</i>	59476835	0.751	0.262	0.525	0.943	0.435	0.652	0.387	0.673	0.738
ILMN_1661631	<i>LILRA3</i>	59500385	0.915	0.105	2.3E-5	0.009	0.393	0.291	5.4E-5	0.450	0.065
ILMN_1786303	<i>LILRA3</i>	59500609	0.023	0.100	2.5E-5	0.341	0.936	0.776	7.1E-3	0.131	0.984
ILMN_1668588	<i>LILRA3</i>	59501708				0.366	0.715	0.092	0.338	0.185	0.409
ILMN_1726545	<i>LILRA5</i>	59514620				0.840	0.904	0.724	0.645	0.386	0.951
ILMN_2266595	<i>LILRA5</i>	59514912	0.429	0.167	0.940	0.334	0.313	0.800	0.306	0.196	0.092
ILMN_2357419	<i>LILRA5</i>	59515439	0.052	0.204	0.859	0.429	0.261	0.657	0.971	0.249	0.672
ILMN_1702385	<i>LILRA4</i>	59540651	0.021	0.734	0.986	0.970	0.699	0.979	0.127	0.111	0.474
ILMN_2389211	<i>LAIR1</i>	59571384	0.889	0.580	0.032	0.151	0.853	0.216	0.110	0.252	0.923
ILMN_1768598	<i>LAIR1</i>	59573180	0.949	0.902	0.958	0.721	0.590	0.795	0.494	0.571	0.970
ILMN_2351269	<i>TTYH1</i>	59619779	0.641	0.901	0.057	0.176	0.967	0.561	0.466	0.880	0.767
ILMN_1758497	<i>TTYH1</i>	59620110	0.965	0.819	0.005	0.192	0.320	0.933	0.464	0.587	0.619
ILMN_1654890	<i>LENG8</i>	59655481	0.499	0.308	0.190	0.355	0.270	0.913	0.892	0.262	0.858
ILMN_1732720	<i>LENG9</i>	59665067	0.675	0.544	0.416	0.233	0.958	0.237	0.071	0.684	0.409
ILMN_1774982	<i>CDC42EP5</i>	59675413	0.690	0.382	0.197	0.142	0.716	0.705	0.950	0.863	0.820
ILMN_2323933	<i>LAIR2</i>	59706332	0.117	0.592	0.538	0.338	0.830	0.717	0.846	0.877	0.447
ILMN_1807491	<i>LAIR2</i>	59706378	0.975	0.560	0.525	0.254	0.644	0.443	0.249	0.857	0.699
ILMN_1768819	<i>KIR3DX1</i>	59748248	0.451	0.683	0.227	0.936	0.066	0.589	0.777	0.459	0.743
ILMN_1716983	<i>LILRA2</i>	59778160	0.491	0.032	0.189	0.648	0.865	0.311	0.274	0.401	0.813
ILMN_1734748	<i>LILRA1</i>	59798409	0.146	0.384	0.005	0.526	0.911	0.540	0.492	0.470	0.055
ILMN_1708248	<i>LILRB1</i>	59822729	0.396	0.111	0.463	0.527	0.643	0.579	0.057	0.442	0.614
ILMN_2316974	<i>LILRB1</i>	59823125	0.584	0.218	0.553	0.415	0.969	0.988	0.107	0.939	0.656
ILMN_2355953	<i>LILRB4</i>	59867107	0.473	0.297	0.869	0.346	0.963	0.194	0.741	0.361	0.920
ILMN_1778974	<i>LILRB4</i>	59868316	0.289	0.814	0.161	0.363	0.631	0.776	0.363	0.266	0.231
ILMN_1733426	<i>KIR3DL3</i>	59928720	0.517	0.640	1.000	0.821	0.863	0.559	0.906	0.436	0.032
ILMN_2082593	<i>KIR3DL3</i>	59929273	0.008	0.102	0.480	0.700	0.842	0.340	0.587	0.619	0.652
ILMN_2402106	<i>KIR2DL3</i>	59942155				0.324	0.866	0.095	0.105	0.459	0.689
ILMN_1667232	<i>KIR2DL3</i>	59942937				0.019	0.059	0.995	0.129	0.349	0.228
ILMN_1678882	<i>KIR2DL1</i>	59974324	0.733	0.817	0.814	0.462	0.240	0.276	0.203	0.370	0.922
ILMN_1786810	<i>KIR2DL1</i>	59974626	0.896	0.452	0.731	0.270	0.203	0.269	0.408	0.482	0.340
ILMN_2390197	<i>KIR2DL4</i>	60007796	0.813	0.119	0.075	0.745	0.543	0.530	0.231	0.456	0.037
ILMN_1693207	<i>KIR2DL4</i>	60008101				0.760	0.653	0.735	0.377	0.530	0.101
ILMN_1739756	<i>KIR2DL4</i>	60008300				0.012	0.117	0.701	0.252	0.647	0.954
ILMN_1733245	<i>KIR3DL1</i>	60020792	0.876	0.355	0.886	0.946	0.518	0.083	0.277	0.029	0.244
ILMN_2131828	<i>KIR3DL1</i>	60021307	0.213	0.309	0.516	0.843	0.892	0.442	0.821	0.449	0.137

ILMN_2190842	<i>KIR3DL2</i>	60055232	0.670	0.261	0.411	0.448	0.679	0.171	0.105	0.678	0.472
ILMN_2379967	<i>FCAR</i>	60077519	0.295	0.370	0.187	0.359	0.443	0.089	0.642	0.911	0.507
ILMN_2365091	<i>FCAR</i>	60077541				0.198	0.008	0.904	0.511	0.990	0.888
ILMN_2279367	<i>FCAR</i>	60078816				0.800	0.985	0.678	0.509	0.043	0.896
ILMN_1750761	<i>NCR1</i>	60110290	0.021	0.133	0.100	0.516	0.417	0.067	0.047	0.804	0.982
ILMN_1652366	<i>NLRP7</i>	60147578	0.746	0.391	0.366	0.161	0.368	0.736	0.340	0.851	0.233
ILMN_1798063	<i>NLRP7</i>	60147841	0.866	0.659	0.689	0.652	0.289	0.117	0.745	0.908	0.394
ILMN_1658632	<i>NLRP7</i>	60148662	0.315	0.357	0.135	0.312	0.498	0.178	0.820	0.505	0.503
ILMN_1664878	<i>NLRP2</i>	60172608	0.240	0.745	0.195	0.122	0.809	0.035	0.136	0.500	0.879
ILMN_2223772	<i>GP6</i>	60239341				0.022	0.505	0.817	0.108	0.562	0.729
ILMN_1732269	<i>GP6</i>	60239966	0.470	0.955	0.983	0.066	0.503	0.249	0.104	0.257	0.847
ILMN_1660723	<i>RDH13</i>	60270915	0.891	0.868	0.303	0.384	0.099	0.404	0.425	0.760	0.624
ILMN_1681193	<i>EPS8L1</i>	60275218	0.416	0.447	0.299	0.951	0.218	0.357	0.532	0.681	0.762
ILMN_1662950	<i>EPS8L1</i>	60275322	0.943	0.241	0.763	0.981	0.393	0.317	0.304	0.775	0.634
ILMN_1671600	<i>EPS8L1</i>	60277429	0.651	0.306	0.912	0.503	0.595	0.075	0.676	0.079	0.152
ILMN_1659522	<i>EPS8L1</i>	60286742				0.701	0.156	0.305	0.505	0.670	0.924
ILMN_1685286	<i>LENG3</i>	60318143	0.861	0.367	0.229	0.700	0.956	0.579	0.575	0.877	0.688
ILMN_2115991	<i>TNNT1</i>	60351761	0.820	0.373	0.302	0.031	0.736	0.304	0.746	0.138	0.051
ILMN_1768637	<i>TNNI3</i>	60360285	0.540	0.706	0.168	0.851	0.370	0.430	0.517	0.525	0.669
ILMN_1772074	<i>C19orf51</i>	60367793	0.662	0.678	0.632	0.592	0.427	0.531	0.766	0.538	0.177
ILMN_1755571	<i>SYT5</i>	60381988	0.503	0.181	0.012	0.337	0.631	0.127	0.405	0.487	0.855
ILMN_1741465	<i>PTPRH</i>	60409125	0.049	0.532	0.020	0.521	0.279	0.233	0.851	0.970	0.177
ILMN_2049417	<i>TMEM86B</i>	60430902	0.875	0.845	0.786	0.562	0.814	0.899	0.738	0.935	0.483
ILMN_1794085	<i>SAPS1</i>	60458773				0.549	0.156	0.628	0.347	0.754	0.892
ILMN_1678949	<i>HSPBP1</i>	60482754	0.442	0.733	0.791	0.518	0.758	0.251	0.389	0.825	0.661
ILMN_1769902	<i>HSPBP1</i>	60482756	0.462	0.742	0.690	0.327	0.506	0.345	0.532	0.208	0.299
ILMN_2185845	<i>BRSK1</i>	60490150	0.951	0.773	0.983	0.230	0.243	0.731	0.332	0.535	0.041

<sup>a</sup>eQTL analysis for rs817826 and rs103294 with mRNA expression of nearby genes within 1 Mb of either the 5' or 3' end of the specified SNPs in three cell types (T-cell, fibroblast and lymphoblastoid cell line) of 75 Geneva individuals and three tissue types (166 adipose, 156 LCL and 160 skin samples) of healthy female twins (Twins 1 and 2).

Supplementary Table 9. *LILRA3* deletion distribution and association with prostate cancer risk among Chinese men

Status <sup>a</sup>	Cases (n=1,304)		Controls (n=991)		OR(95%CI) <sup>b</sup>	P <sup>b</sup>
	n	%	n	%		
Del/Del	629	48.2	558	56.3	Ref.	
Del/Non-del	550	42.2	367	37.0	1.26(1.07-1.51)	8.68×10 <sup>-3</sup>
Non-del/Non-del	125	9.6	66	6.7	1.57(1.14-2.18)	6.04×10 <sup>-3</sup>
Non-del allele		30.7		25.2	1.25(1.10-1.43)	9.60×10 <sup>-4</sup>

<sup>a</sup> “Del” represents deleted allele of *LILRA3*; “Non-del” represents non-deleted allele of *LILRA3*; <sup>b</sup> Odds ratios (ORs), 95% confidence intervals (95% CIs) and corresponding *P* values were estimated using a logistic regression model.

Supplementary Table 10. *LILRA3* mRNA expression in prostate tissues

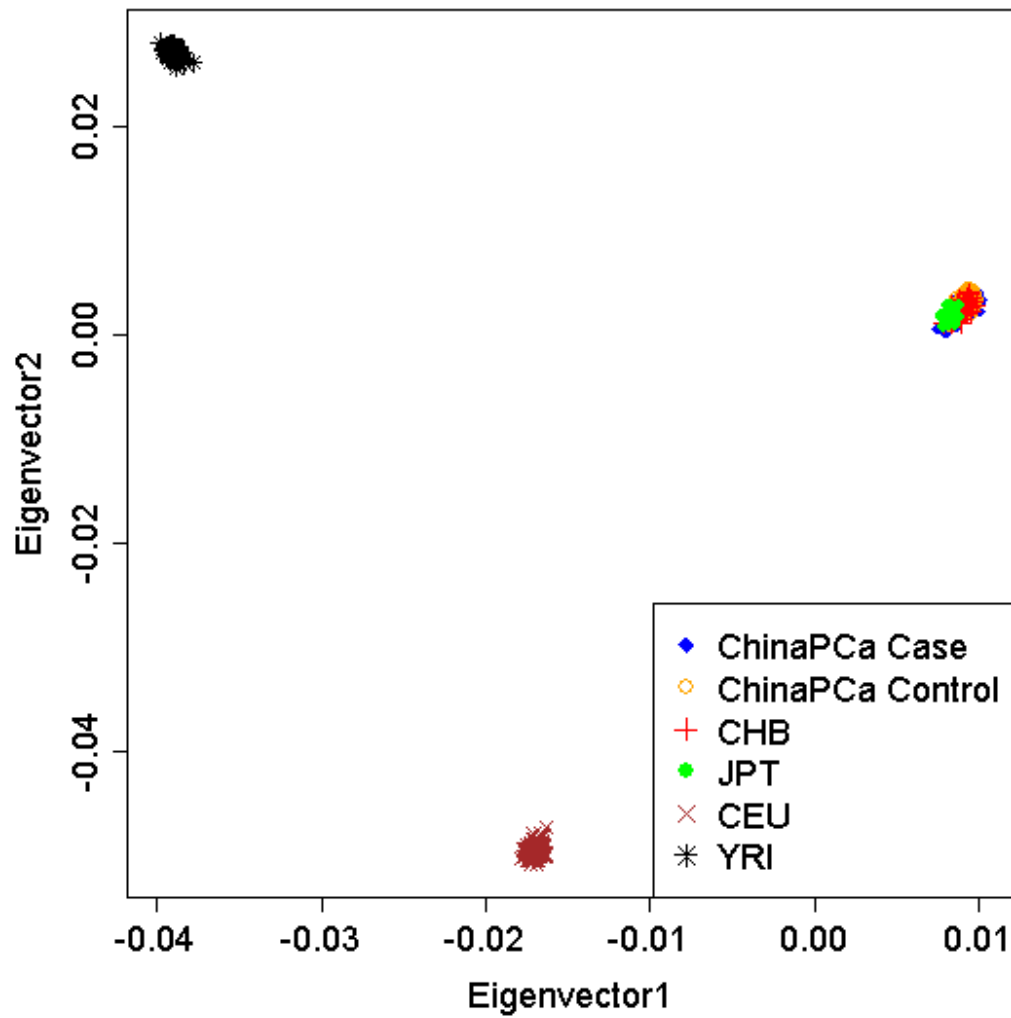
	Mean and standard deviation (SD) of Ct number <sup>a</sup>		
	Del/Del	Del/Non-Del	Non-Del/Non-Del
Number of samples	36	35	9
<i>LILRA3</i>	36.34 (0.26)	34.20 (0.49)	34.97 (0.63)
<i>β-ACTIN</i>	21.09 (0.29)	21.06 (0.29)	21.70 (0.53)

<sup>a</sup> Among the total of 36 samples that harbored a homozygous deletion (Del/Del), expression of the *LILRA3* mRNA was detected in 6 samples with Ct (cycle threshold) values >36 after 45 PCR cycles. For 35 samples that were heterozygous for the deletion (Del/Non-Del), mRNA was detectable among 26 with Ct values of  $34.20 \pm 0.49$ . Among the 9 samples that carried no deletions, mRNA was detected in all samples with Ct values at  $34.97 \pm 0.63$ . The mRNA for the reference gene of *β-ACTIN* was successfully detected; the mean Ct numbers were  $21.09 \pm 0.29$ ,  $21.06 \pm 0.29$ , and  $21.70 \pm 0.53$ , respectively, for samples with homozygous deletion, heterozygous deletion, and homozygous non-deletion type.

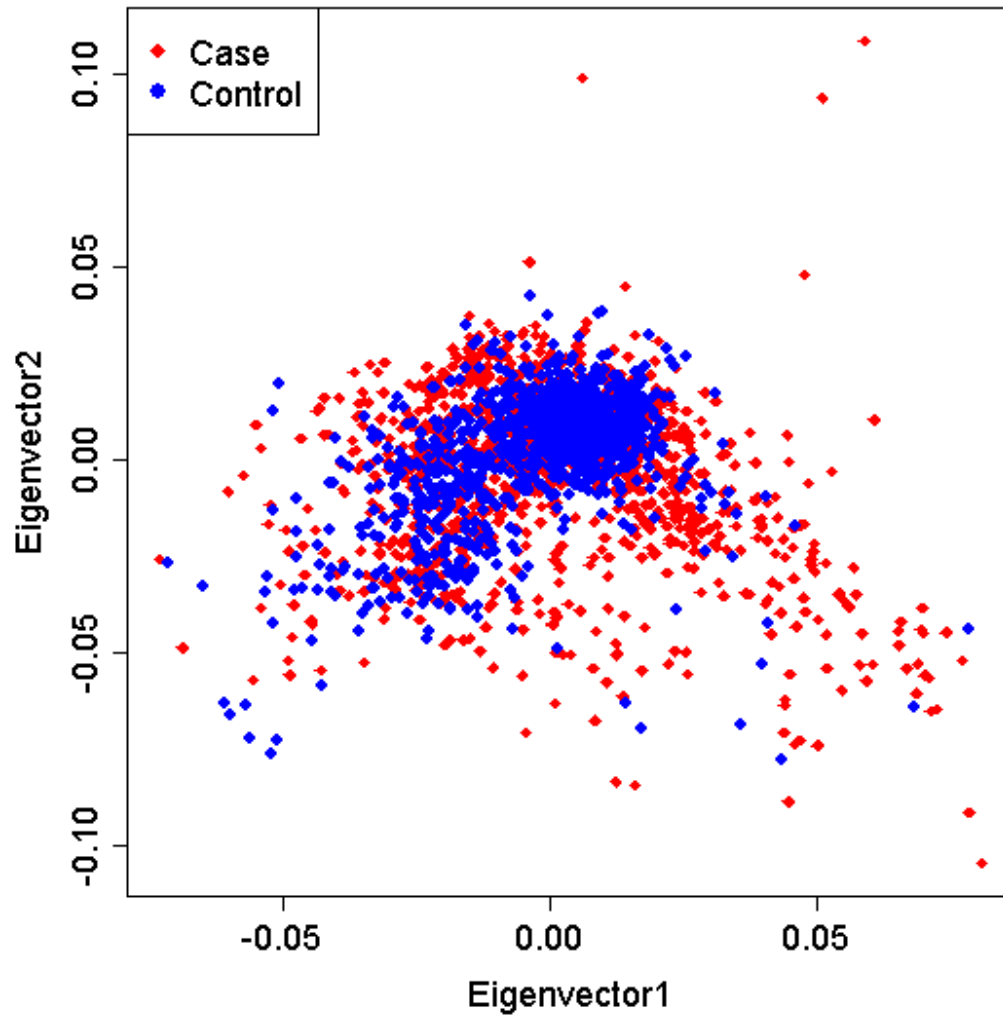
Supplementary Table 11. Primers for mRNA expression measurement of *LILRA3* using a real-time quantitative PCR assay

Primer	Oligo sequence
Forward	5' - GCGCCAATCTCATAAGTACCAGGCTG-3'
Reverse	5' - CCAGCCTTGGAGTCGGA CTGTT-3'

Supplementary Fig. 1. Principal components analysis determination of ethnicity. The first two principal components for each individual were plotted: the relatedness between the 1,417 cases and 1,008 controls, along with the European (CEU), African (YRI), Chinese (CHB), and Japanese (JPT) data from the HapMap project was analyzed.

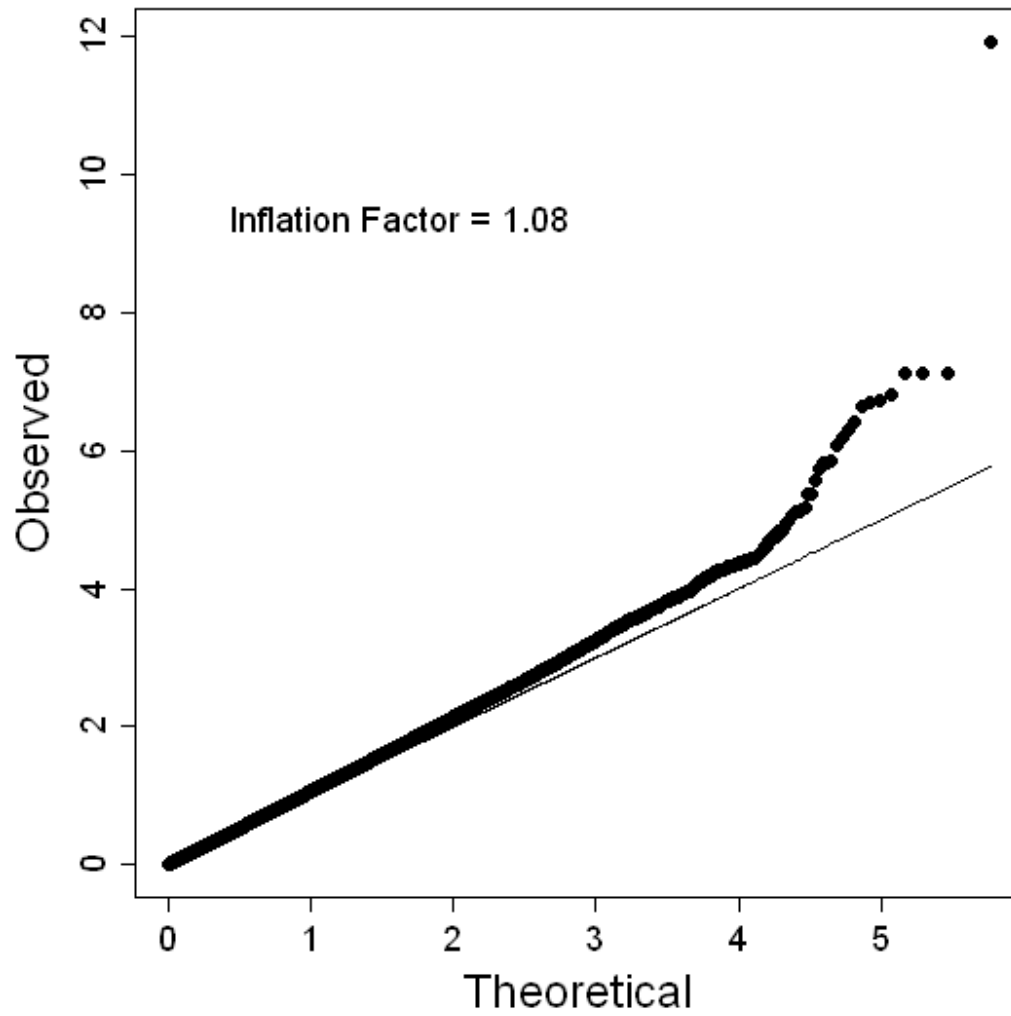


Supplementary Fig. 2. Principal component analysis between cases and controls.

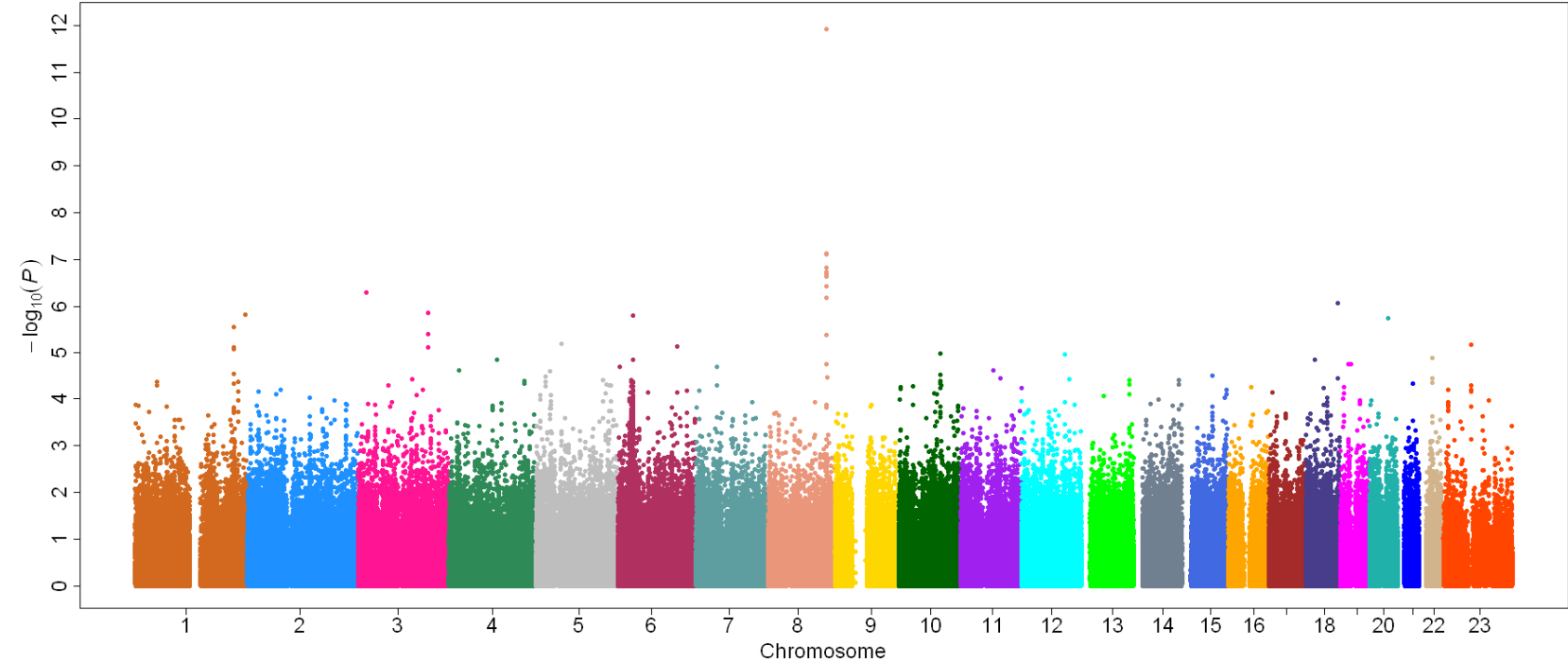




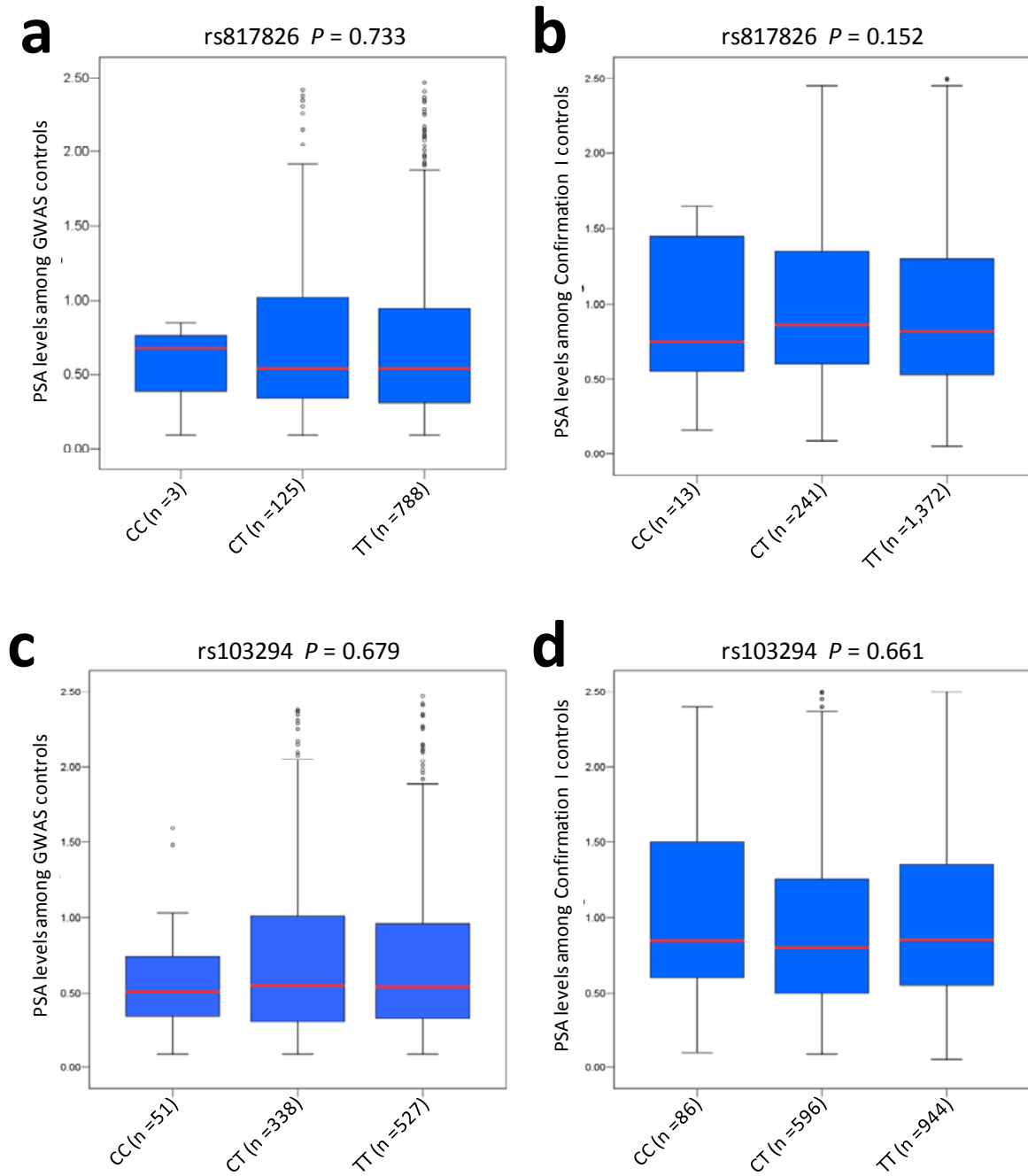
Supplementary Fig. 3. Quantile-quantile plots and genomic inflation factors ( $\lambda$ ) for associations with prostate cancer risk, with observed  $P$  values plotted as a function of theoretical  $P$  values.



Supplementary Fig. 4. Manhattan plot of the strength of association ( $-\log_{10}(P)$  values; Y-axis) between SNPs (X-axis by chromosome and chromosomal position) and prostate cancer risk.

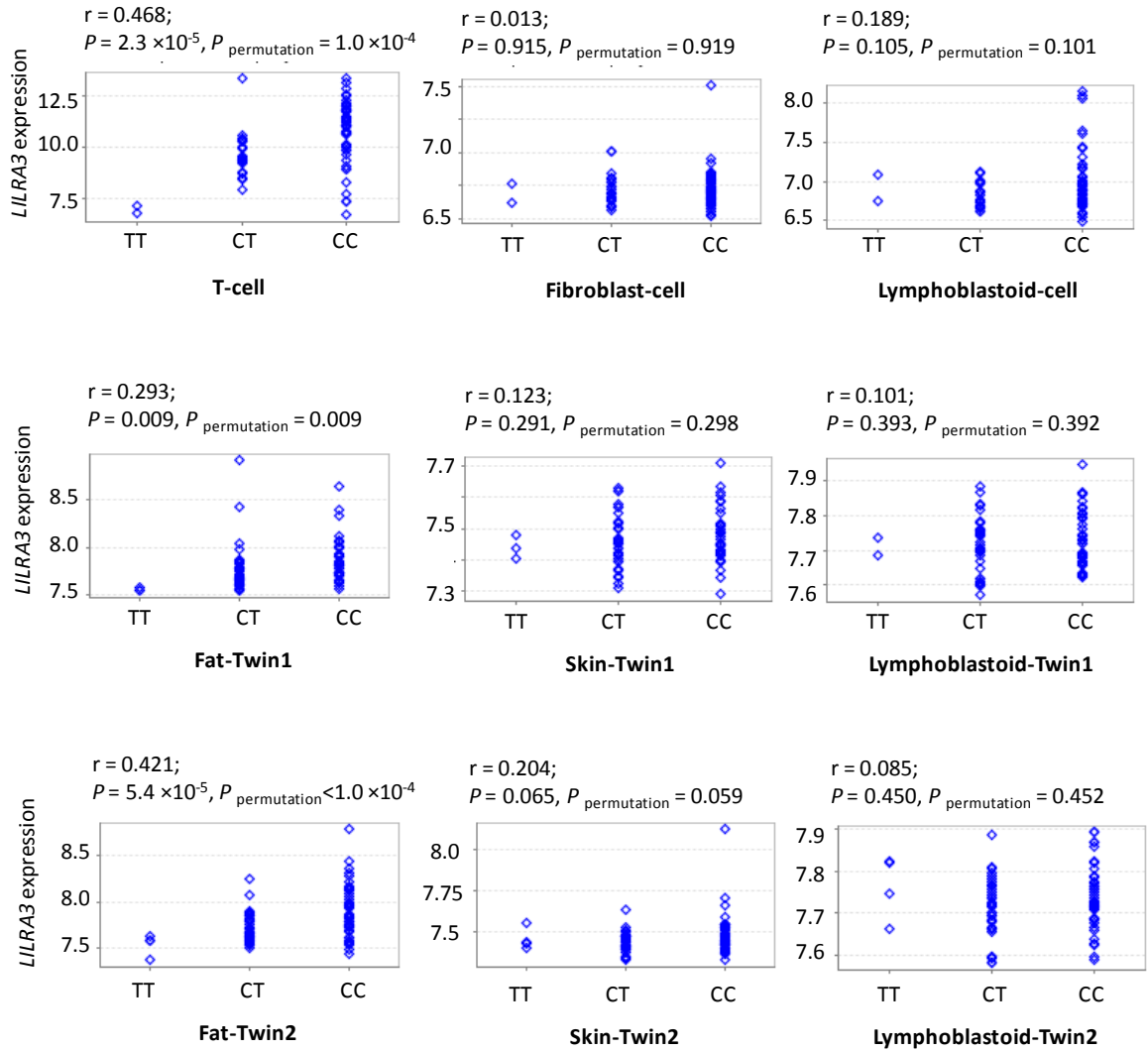


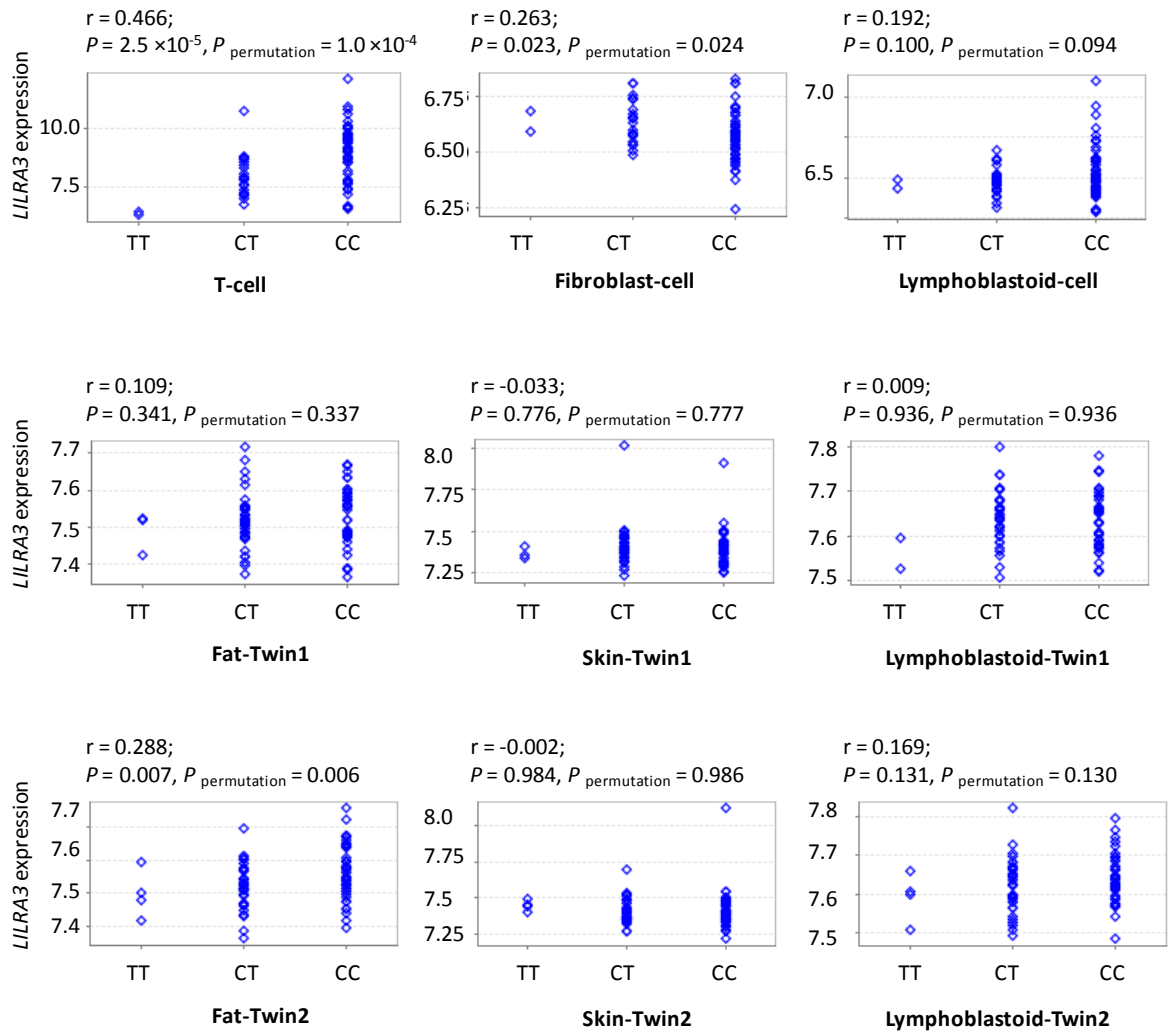
Supplementary Fig. 5. Distribution of PSA levels among GWAS and Replication I controls by genotypes of rs817826 (a, GWAS; b, Replication I) and rs103294 (c, GWAS; d, Replication I).



Supplementary Fig. 6. Expression quantitative trait loci (eQTL) analysis for rs103294 at 19q13.4 with mRNA expression of *LILRA3*. The mRNA expression of *LILRA3* was measured in three cell types (T-cell, fibroblast and lymphoblastoid cell line) of 75 Geneva individuals and three tissue types (166 adipose, 156 LCL and 160 skin samples) of healthy female twins (Twins 1 and 2) by two array probes, ILMN\_166631 (a) and ILMN\_1786303 (b) on exons 7 and 6, respectively.

**a**



**b**

Supplementary Figure 7. Functional annotation of the genetic locus on 9q31 based on UCSC genome browser.

