Meta-analysis identifies four new loci associated with testicular germ cell tumor

SUPPLEMENTARY MATERIAL

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1. The US Servicemen's Testicular Tumor Environmental and Endocrine Determinants Study (STEED)

The study description has been published in detail previously[1]. Briefly, between April 2002 and January 2005 Servicemen aged 18-45 years with at least one serum sample stored in the U.S. Department of Defense Serum Repository (DoDSR, Silver Spring, Maryland) were eligible for enrollment. By use of a person-specific identifier, the specimens in the DoDSR computerized database were linked to the Defense Medical Surveillance System (DMSS)[2] and to other military medical databases in order to determine which military personnel had developed medical conditions. All men with a sample in the DoDSR who subsequently developed testicular germ cell tumor (TGCT) while on active duty were eligible to participate as cases. Men with a sample in the DoDSR who did not subsequently develop TGCT were eligible to participate as controls. TGCT diagnoses were based on the original pathology reports or on review (6.5 %) of the pathology slides. The study was designed as a pair-matched, case-control study. Year of birth (within 1 year), race/ethnicity (White, Black, other), and date of available serum sample (within 30 days) were the characteristics used for matching. Of a total of 1,302 DNA specimens obtained from the study subjects[3], 553 TGCT cases and 676 controls were genotyped on the Illumina 660K microarray SNP chip. Standard protocols for genotyping quality control (QC) were implemented. Individuals with missing rate > 0.06 and/or autosomal heterozygosity <0.25 or >0.35 (N=61) and SNPs with missing rate >0.1 and/or Hardy-Weinberg equilibrium p-value $<1\times10^{-7}$ (N=113,525) were excluded. No individuals with cryptic relatedness (IBD greater than 45%) or unexpected duplicate was detected. Genotype concordance rate from 41 expected duplicates was greater than 99.9%. After further excluding ancestral outliers (N=134), in total, we included 479 TGCT cases (86.6%) and 555 controls (82.1%)[4].

2. NCI Familial Testicular Cancer Study (FTCS)

The NCI FTCS Study (NCI protocol 02-C-0178; NCT-00039598) consisted of 354 individuals drawn from 110 multiplex families, including 164 men with TGCT, both probands and affected male relatives[5-8]. Participants provided written informed consent, blood samples, and detailed questionnaire information. Genotypes were determined using the Illumina 660K microarray SNP chip. Standard protocols for genotyping QC were implemented. Individuals with missing rate >0.05 (N=9) and SNPs with missing rate >0.1 (N=99,032) were excluded. No individual was excluded due to autosomal heterozygosity <0.25 or >0.35. Twelve ancestral outliers were excluded. After QC, one case per each family was selected (51 unilateral cases and 13 bilateral cases from multi-case families, and 39 sporadic bilateral cases; total N=103) and compared with 501 controls from the Prostate, Lung, Colon and Ovarian (PLCO) Cancer Screening Trial study[9].

3. University of Pennsylvania (UPENN)

A total of 349 TGCT cases recruited from the University of Pennsylvania (Abramson Cancer Center) and Fox Chase Cancer Center were genotyped and compared to a control group of 919 men enrolled in the University of Pennsylvania Catherterization Study (PennCATH)[10]. All genotyping was completed on the Affymetrix SNP 6.0 array. Detailed description of study samples and QC were reported previously[11].

4. UK Testicular Cancer Consortium (UKTCC)

A total of 979 TGCT cases including unrelated TGCT cases identified from a UK study of familial testicular cancer and a UK-wide hospital-based collection of TGCT cases were genotyped on the Illumina HumanCNV370-Duo bead array. These genotypes were compared to those of 4947 controls from the 1958 Birth Cohort and the UK National Blood Service, that were genotyped by the Wellcome

Trust Case Control Consortium (WTCCC2) using the Illumina 1.2M array. Details of QC and study description were reported previously[12].

5. University of Southern California (USC)

Individuals analyzed are part of a population-based study at the University of Southern California based in the California and the California Cancer Registry (CCR). Among men diagnosed from the inception of each of the 10 regional registries through 2006 and more recently referred by collaborating clinicians and survivor groups, 5,792 men provided family and personal medical history information, 548 reporting two primary TGCTs, 642 reporting personal history of cryptorchidism, and 886 reporting family history of TGCT and/or cryptorchidism. Families oversampled among these sets were invited to participate by providing biospecimens from the index case, affected family members, and additional relatives informative for genetic linkage of association, 3247 individuals in all. The present analysis was limited to non-Hispanic white participants for whom genomic DNA was available at the time of genotypic analysis: Genotypes were determined using the Illumina 610K platform for 376 index TGCT cases – primarily those without participating family members[4] – and – and 258 non-Hispanic white TGCT controls from the Multiethnic Cohort (MEC) study. Our control sample has been described elsewhere[13]. Briefly, we utilized cancer-free controls from the MEC contribution to the NCI Breast & Prostate Cancer Cohort Consortium genome-wide study of aggressive prostate cancer. Both our TGCT case and control series were genotyped using the Illumina 610K platform at the USC Epigenome Center. Standard protocols for genotyping quality control (QC) were implemented. Individuals and SNPs were excluded where call rates were <98% (N=6 individuals; N=39,874 SNPs), autosomal heterozygosity <0.25 or >0.35 (N=0), high heterozygosity on the X chromosome (N=1), unexpected related subjects (N=2), and ancestral outliers (N=11). In total, we included 358 TGCT cases (95.2%) and 258 controls (99.2%).

6. Oslo University Hospital-Radium Hospital, Norway (OUHRH)

Since 1980, men with TGCT have been recruited at the Norwegian Radium Hospital (Rikshospitalet), which has now merged with the Oslo University Hospital. This cohort of men with TGCT (along with other TGCT patients collected through other Norwegian oncology hospitals) has been extensively studied in previous studies of genetic polymorphisms in candidate genes[14,15] and in the context of survivorship issues[14,15]. A total of 865 TGCT cases and 392 controls were genotyped using optimized TaqMan assays (ABI 7900HT Real-Time PCR System, Applied Biosystems by Life Technologies, Foster City, CA) and compared with 392 controls of the Cohort of Norway (CONOR) who were also analyzed by TaqMan. These controls were male, cancer free controls males who were military recruits or employees in a Norwegian company were analyzed previously in the third stage of the prostate cancer GWAS as part of the Cancer Genetic Markers of Susceptibility (CGEMS)[16].

7. The Testicular Cancer in Philadelphia Area Counties study (TestPAC)

This study is an NCI funded population-based case-control study set in Philadelphia, Pennsylvania and its seven neighboring counties. Cases and controls complete a self-administered questionnaire eliciting information on established and putative TGCT risk factors, and each gives a blood or saliva sample for isolation of DNA. A total of 331 cases and 683 controls, distinct from the PENN set were genotyped using iPlex (Sequenom, Inc.).

8. The Adult Testicular Lifestyle and Blood Specimen study (ATLAS)

This study is an NCI-funded population-based case-control and case-parent triad/dyad study conducted in western Washington State[17,18]. Cases were identified through the Seattle-Puget Sound SEER registry, and controls through random-digit dialing. All study participants completed an in-person

interview to ascertain known and suspected TGCT risk factors, and provided a blood sample (90%) or oral buccal cell sample (10%) from which DNA was extracted by a standard method. A total of 439 TGCT cases and 960 controls were genotyped using iPlex (Sequenom, Inc.).

9. M.D. Anderson Cancer Center (MDA)

Cases of TGCT were recruited through clinics at the University of Texas MD Anderson Cancer Center. Cases were over 18 years of age at the time of recruitment. Each patient completed a questionnaire on demographics, education, medical history, smoking behavior, alcohol use, work history, medication use, and family history. Controls were genotyped using the Illumina HumanHap610 Beadchip previously as part of a GWAS for bladder cancer[19]. Briefly, controls were of European descent and recruited from Kelsey Seybold Clinics in the Houston, Texas metropolitan area. All participants provided written informed consent prior to providing blood specimens for processing and genotyping at MD Anderson Cancer Center. Study approval was granted by the Institutional Review Board of both MD Anderson Cancer Center and Kelsey Seybold Clinics. A total of 304 TGCT cases were genotyped using the optimized TaqMan assays and 239 non-admixed European cases were compared with 351 controls drawn from the previous bladder cancer GWAS.

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Supplementary Tables

Supplementary Table 1 Meta-analysis of reported TGCT susceptibility loci with two NCI GWAS

SNP[1]	Nearby genes	Study[2]	Allelic OR (95% CI)	*P* value	*P* for heterogeneity
rs4657482	*UCK2*	UK1	1.39 (1.21-1.59)	1.60E-06	
G|A	1q24.1	UK2	1.14 (0.99-1.31)	0.07	
		NCI	1.43 (1.22-1.67)	5.97E-06	
		Combined	1.31 (1.20-1.42)	2.17E-10	0.059
rs4699052	*CENPE*	UK1	0.72 (0.63-0.83)	3.00E-06	
C|T	4q24	UK2	0.83 (0.72-0.95)	8.00E-03	
		NCI	0.82 (0.70-0.96)	0.015	
		Combined	0.79 (0.72-0.85)	1.03E-08	0.328
rs4624820	*SPRY4*	UK1	0.68 (0.60-0.77)	4.60E-09	
A|G	5q31.3	UK2	0.73 (0.63-0.84)	6.70E-06	
		NCI	0.61 (0.53-0.72)	8.36E-10	
		Combined	0.68 (0.62-0.74)	8.04E-21	0.282
rs210138	*BAK1*	UK1	1.5 (1.30-1.74)	4.50E-08	
A|G	6p21.31	UK2	1.5 (1.28-1.75)	6.60E-07	
		NCI	1.62 (1.35-1.95)	2.83E-07	
		Combined	1.53 (1.39-1.68)	2.28E-19	0.778
rs995030	*KITLG*	UK1	0.44 (0.36-0.53)	1.90E-18	
G|A	12q21.32	UK2	0.39 (0.31-0.49)	1.20E-19	
		NCI	0.43 (0.34-0.54)	3.17E-13	
		Combined	0.42 (0.37-0.48)	9.59E-43	0.748
rs1508595	*KITLG*	UK1	0.39 (0.31-0.49)	1.90E-19	
C|T	12q21.32	UK2	0.37 (0.29-0.48)	4.90E-18	
		NCI	0.42 (0.33-0.54)	1.29E-12	
		Combined	0.39 (0.34-0.45)	8.40E-41	0.796
rs6897876	*SPRY4*	UPENN1	1.59 (1.30-1.94)	3.57E-06	
T|C	5q31.3	UPENN2	1.37 (1.14-1.64)	6.77E-04	
		Combined	1.47 (1.28-1.68)	2.00E-08	0.277
rs4324715	*SPRY4*	UPENN1	0.63 (0.52-0.77)	2.96E-06	
T|C	5q31.3	UPENN2	0.72 (0.60-0.86)	3.67E-04	
		NCI	0.63 (0.53-0.75)	1.42E-07	
		Combined	0.66 (0.59-0.73)	8.57E-15	0.492
rs3782179	*KITLG*	UPENN1	2.45 (1.79-3.35)	7.34E-09	
G|A	12q21.32	UPENN2	3.07 (2.29-4.12)	5.88E-15	
		Combined	2.76 (2.23-3.42)	1.85E-20	0.304
rs4474514	*KITLG*	UPENN1	2.36 (1.73-3.21)	1.95E-08	
G|A	12q21.32	UPENN2	3.08 (2.29-4.14)	5.88E-15	
		Combined	2.71 (2.19-3.36)	4.78E-20	0.222
rs2736100	*TERT*	UK3	0.74 (0.67-0.81)	2.80E-10	
T|G	5p15.33	UK4	0.75 (0.67-0.85)	4.90E-06	
		NCI	0.73 (0.63-0.85)	4.82E-05	
		Combined	0.74 (0.69-0.79)	1.32E-18	0.952

rs4635969	*TERT-CLPTM1L*	UK3	1.65 (1.47-1.86)	9.60E-17	
C|T	5p15.33	UK4	1.54 (1.33-1.79)	1.40E-08	
		NCI	1.4 (1.17-1.68)	2.22E-04	
		Combined	1.56 (1.44-1.69)	1.87E-26	0.319
rs755383	*DMRT1*	UK3	0.64 (0.58-0.71)	8.10E-19	
T|C	9p24.3	UK4	0.73 (0.64-0.83)	5.70E-07	
		NCI	0.66 (0.56-0.77)	4.25E-07	
		Combined	0.67 (0.62-0.72)	1.46E-28	0.242
rs2900333	*ATF7IP*	UK3	0.78 (0.70-0.86)	7.60E-07	
C|T	12p13.1	UK4	0.79 (0.69-0.89)	2.00E-04	
		NCI	0.93 (0.79-1.09)	0.344	
		Combined	0.81 (0.75-0.87)	2.08E-09	0.162

[1] SNP genotype depicted as reference allele|effect allele

[2] UK1 and UK2 data were taken from Rapley et al., UPENN1 and UPENN2 data were taken from Kanetsky et al., and UK3 and UK4 data were taken from Turnbull et al.

NCI depicts combined analysis results of the two GWAS scans STEED and FTCS performed at NCI

Supplementary Table 2 Summary of 40 SNPs with discovery meta-analysis *P* value < 10^{-4} in association with testicular germ cell tumor

Locus Information								NCI+Penn Meta-analysis					1000 Genomes Feb2012 EUR (n=379)				
Cytoband	Gene neighborhood	annotation	Study	SNP	Location	ref|eff	EAF	Allelic OR	95% CI	*P* value	*P* for heterogeneity	Meta.*P* rank	MA	MAF	r^2	*D'*	haplotype
3p26.1	*AF279782*		NCI	rs17288317	6625357	T|C	0.381	1.32	(1.13-1.54)	3.54E-04			C	0.3404			
			UPENN	rs2323813	6679798	T|C	0.415	1.19	(0.99-1.41)	0.0568			G	0.4116			
			NCI+UPENN					**1.26**	**(1.12-1.42)**	**8.45E-05**	**0.3578**	**39**			**0.578**	**0.885**	**CG/TA**
			NCI	rs9811958	6678484	A|C	0.494	1.4	(1.21-1.63)	1.08E-05			C	0.4406			
			UPENN	rs4686074	6682834	G|C	0.435	1.12	(0.94-1.33)	0.2254			G	0.4406			
			NCI+UPENN					**1.27**	**(1.13-1.43)**	**3.80E-05**	**0.0547**	**29**			**1**	**1**	**CG/AC**
4q22.3	*SMARCAD1 HPGDS*	intron	NCI	rs11097408	95165662	C|T	0.474	1.33	(1.14-1.55)	2.42E-04			T	0.4472			
		intron	UPENN	rs2632412	95149319	A|C	0.433	1.28	(1.07-1.53)	6.67E-03			G	0.4367			
			NCI+UPENN					**1.31**	**(1.16-1.47)**	**5.54E-06**	**0.7423**	**19**			**0.958**	**1**	**TG/CT**
		intron	NCI	rs6532476	95192461	A|C	0.474	1.31	(1.13-1.53)	4.25E-04			C	0.4499			
		intron	UPENN	rs2632412	95149319	A|C	0.433	1.28	(1.07-1.53)	6.67E-03			G	0.4367			
			NCI+UPENN					**1.3**	**(1.16-1.46)**	**9.29E-06**	**0.8172**	**24**			**0.948**	**1**	**CG/AT**
		intron	NCI	rs17021463	95224812	G|T	0.475	1.33	(1.14-1.55)	2.12E-04			T	0.4485			
		intron	UPENN	rs2632412	95149319	A|C	0.433	1.28	(1.07-1.53)	6.67E-03			G	0.4367			
			NCI+UPENN					**1.31**	**(1.17-1.47)**	**4.91E-06**	**0.7302**	**15**			**0.953**	**1**	**TG/GT**
		intron	NCI	rs10516950	95254730	G|A	0.499	1.3	(1.12-1.52)	5.85E-04							
			UPENN	rs10516950	95254730	C|T	0.473	1.27	(1.06-1.52)	9.28E-03							
			NCI+UPENN	**rs10516950**	**95254730**			**1.29**	**(1.15-1.45)**	**1.70E-05**	**0.8365**	**26**					
6q24.2			NCI	rs7760281	144446518	T|C	0.448	1.3	(1.12-1.51)	6.72E-04							
			UPENN	rs7760281	144446518	A|G	0.409	1.21	(1.01-1.45)	0.0358							
			NCI+UPENN	**rs7760281**	**144446518**			**1.26**	**(1.12-1.42)**	**8.11E-05**	**0.5513**	**38**					
7p22.3	*MAD1L1*	intron	NCI	rs10275045	1920826	C|T	0.48	1.36	(1.17-1.57)	5.19E-05							
			UPENN	rs10275045	1920826	C|T	0.440	1.39	(1.17-1.65)	1.92E-04							
			NCI+UPENN	**rs10275045**	**1920826**			**1.37**	**(1.22-1.53)**	**3.94E-08**	**0.8372**	**3**					
		intron	NCI	rs6952727	1947958	A|G	0.482	1.35	(1.16-1.56)	9.41E-05			G	0.4222			
		intron	UPENN	rs4721190	1954732	G|A	0.444	1.4	(1.17-1.66)	1.64E-04			A	0.4208			
			NCI+UPENN					**1.37**	**(1.22-1.53)**	**6.34E-08**	**0.7481**	**6**			**0.962**	**0.984**	**GA/AG**
		intron	NCI	rs4721184	1950784	T|C	0.443	1.34	(1.15-1.56)	1.36E-04			C	0.3905			
		intron	UPENN	rs2056480	1954301	G|A	0.407	1.37	(1.15-1.63)	4.25E-04			A	0.3879			
			NCI+UPENN					**1.35**	**(1.21-1.52)**	**2.21E-07**	**0.8557**	**10**			**0.978**	**0.994**	**CA/TG**
		intron	NCI	rs11772205	1951236	C|T	0.483	1.35	(1.16-1.56)	8.89E-05			T	0.4248			
		intron	UPENN	rs4721190	1954732	G|A	0.444	1.4	(1.17-1.66)	1.64E-04			A	0.4208			
			NCI+UPENN					**1.37**	**(1.22-1.53)**	**5.99E-08**	**0.7574**	**4**			**0.973**	**0.995**	**TA/CG**
		intron	NCI	rs12699477	1968953	T|C	0.412	1.31	(1.13-1.53)	4.64E-04			C	0.3654			
		intron	UPENN	rs2056480	1954301	G|A	0.407	1.37	(1.15-1.63)	4.25E-04			A	0.3879			
			NCI+UPENN					**1.34**	**(1.19-1.50)**	**7.54E-07**	**0.7139**	**12**			**0.791**	**0.933**	**CA/TG**
		intron	NCI	rs12666575	2004421	C|T	0.43	1.35	(1.16-1.57)	1.19E-04			T	0.3747			
		intron	UPENN	rs4721295	2036669	T|G	0.418	1.38	(1.16-1.65)	2.78E-04			G	0.3799			
			NCI+UPENN					**1.36**	**(1.22-1.53)**	**1.28E-07**	**0.8245**	**8**			**0.901**	**0.96**	**TG/CT**
		intron	NCI	rs3778969	2139990	G|A	0.445	1.37	(1.17-1.59)	6.17E-05							
			UPENN	rs3778969	2139990	G|A	0.398	1.4	(1.17-1.67)	2.50E-04							
			NCI+UPENN	**rs3778969**	**2139990**			**1.38**	**(1.23-1.55)**	**6.24E-08**	**0.8538**	**5**					
		intron	NCI	rs3800913	2163237	A|G	0.473	1.35	(1.16-1.57)	9.34E-05			G	0.4142			

		intron	UPENN	rs10224497	2149967	A|G	0.443	1.42 (1.19-1.69)	8.89E-05			G	0.4261			
			NCI+UPENN					**1.38 (1.23-1.55)**	**3.66E-08**	**0.6662**	**2**			**0.921**	**0.983**	**GG/AA**
		intron	NCI	rs10256077	2171450	T|C	0.493	1.31 (1.13-1.52)	4.28E-04			C	0.4314			
		intron	UPENN	rs10224497	2149967	A|G	0.443	1.42 (1.19-1.69)	8.89E-05			G	0.4261			
			NCI+UPENN					**1.36 (1.21-1.52)**	**1.83E-07**	**0.4865**	**9**			**0.894**	**0.956**	**CG/TA**
		intron	NCI	rs3778991	2172455	G|A	0.425	1.38 (1.18-1.61)	4.76E-05			A	0.3628			
		intron	UPENN	rs3779003	2184902	C|T	0.365	1.43 (1.19-1.71)	1.02E-04			T	0.3641			
			NCI+UPENN					**1.4 (1.24-1.57)**	**2.10E-08**	**0.7661**	**1**			**0.983**	**0.994**	**AT/GC**
		intron	NCI	rs3778994	2175855	C|A	0.454	1.33 (1.14-1.55)	1.96E-04							
			UPENN	rs3778994	2175855	G|T	0.409	1.35 (1.14-1.60)	6.22E-04							
			NCI+UPENN	**rs3778994**	**2175855**			**1.34 (1.20-1.50)**	**4.40E-07**	**0.8947**	**11**					
		intron	NCI	rs3800924	2188249	C|T	0.359	1.35 (1.15-1.58)	2.38E-04			T	0.3285			
		intron	UPENN	rs4721441	2184060	A|G	0.389	1.28 (1.07-1.53)	5.92E-03			C	0.3641			
			NCI+UPENN					**1.32 (1.17-1.49)**	**4.98E-06**	**0.6909**	**16**			**0.821**	**0.98**	**TC/CT**
			NCI	rs7204293	50028105	G|A	0.05	0.54 (0.40-0.75)	1.59E-04			A	0.05805			
			UPENN	rs12445755	50034945	A|G	0.073	0.6 (0.41-0.88)	9.75E-03			C	0.06992			
			NCI+UPENN					**0.57 (0.44-0.72)**	**6.11E-06**	**0.7060**	**21**			**0.78**	**0.975**	**AC/GT**
			NCI	rs8046148	50142944	G|A	0.177	0.7 (0.58-0.85)	2.12E-04			A	0.1926			
16q12.1	*HEATR3*	intron	UPENN	rs2058813	50129454	C|T	0.191	0.82 (0.65-1.03)	0.0923			T	0.1715			
	AF086132		**NCI+UPENN**					**0.75 (0.65-0.86)**	**8.93E-05**	**0.2997**	**42**			**0.868**	**1**	**AT/GC**
			NCI	rs1496430	51096349	C|T	0.243	1.44 (1.19-1.74)	1.32E-04			T	0.1715			
			UPENN	rs1566043	51113337	G|A	0.147	1.2 (0.94-1.54)	0.1411			A	0.1319			
			NCI+UPENN					**1.35 (1.16-1.56)**	**9.08E-05**	**0.2493**	**43**			**0.734**	**1**	**TA/CG**
		missense	NCI	rs7193541	74664743	T|C	0.359	0.76 (0.65-0.88)	4.24E-04							
		Ile>Val	UPENN	rs7193541	74664743	T|C	0.396	0.83 (0.69-1.00)	0.0503							
			NCI+UPENN	**rs7193541**	**74664743**			**0.79 (0.70-0.89)**	**7.86E-05**	**0.4414**	**37**					
		coding-synon	NCI	rs4888262	74670458	T|C	0.552	1.36 (1.16-1.58)	9.12E-05							
16q23.1	*RFWD3*	Thr>Thr	UPENN	rs4888262	74670458	T|C	0.498	1.39 (1.16-1.67)	3.12E-04							
			NCI+UPENN	**rs4888262**	**74670458**			**1.37 (1.22-1.54)**	**1.12E-07**	**0.8255**	**7**					
		intron	NCI	rs4888265	74683737	C|T	0.553	1.35 (1.16-1.57)	1.26E-04			C	0.4789			
		intron	UPENN	rs8062783	74691264	C|T	0.482	1.31 (1.09-1.57)	3.35E-03			C	0.4763			
			NCI+UPENN					**1.33 (1.19-1.50)**	**1.51E-06**	**0.8069**	**13**			**0.989**	**1**	**CC/TT**
			NCI	rs9891129	56630474	C|T	0.282	0.74 (0.63-0.87)	2.59E-04			T	0.3272			
		intron	UPENN	rs302867	56752819	T|C	0.336	0.78 (0.65-0.93)	6.93E-03			C	0.3259			
			NCI+UPENN					**0.76 (0.67-0.85)**	**6.30E-06**	**0.7010**	**22**			**0.994**	**1**	**TC/CT**
			NCI	rs9905704	56632543	T|G	0.281	0.73 (0.62-0.86)	1.88E-04			G	0.3272			
		intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
			NCI+UPENN					**0.75 (0.66-0.85)**	**3.66E-06**	**0.7231**	**14**			**0.994**	**1**	**GT/TC**
		intron	NCI	rs1267543	56656624	G|A	0.184	0.7 (0.58-0.84)	1.81E-04			A	0.2375			
		intron	UPENN	rs2003537	56611871	G|C	0.211	0.82 (0.67-1.02)	0.0701			G	0.2137			
			NCI+UPENN					**0.75 (0.65-0.86)**	**6.57E-05**	**0.2665**	**34**			**0.858**	**0.992**	**AG/GC**
		intron	NCI	rs302858	56692800	C|A	0.282	0.74 (0.63-0.87)	2.77E-04			T	0.3259			
		intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
	C17orf47		**NCI+UPENN**					**0.75 (0.67-0.85)**	**5.21E-06**	**0.7736**	**18**			**1**	**1**	**TT/GC**
	TEX14	intron	NCI	rs304283	56785725	C|T	0.281	0.74 (0.63-0.87)	2.72E-04			T	0.3245			
17q22	*RAD51C*	intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
	PPM1E		**NCI+UPENN**					**0.75 (0.67-0.85)**	**5.11E-06**	**0.7719**	**17**			**0.994**	**1**	**TT/CC**

		intron	UPENN	rs10224497	2149967	A|G	0.443	1.42 (1.19-1.69)	8.89E-05			G	0.4261			
			NCI+UPENN					**1.38 (1.23-1.55)**	**3.66E-08**	**0.6662**	**2**			**0.921**	**0.983**	**GG/AA**
		intron	NCI	rs10256077	2171450	T|C	0.493	1.31 (1.13-1.52)	4.28E-04			C	0.4314			
		intron	UPENN	rs10224497	2149967	A|G	0.443	1.42 (1.19-1.69)	8.89E-05			G	0.4261			
			NCI+UPENN					**1.36 (1.21-1.52)**	**1.83E-07**	**0.4865**	**9**			**0.894**	**0.956**	**CG/TA**
		intron	NCI	rs3778991	2172455	G|A	0.425	1.38 (1.18-1.61)	4.76E-05			A	0.3628			
		intron	UPENN	rs3779003	2184902	C|T	0.365	1.43 (1.19-1.71)	1.02E-04			T	0.3641			
			NCI+UPENN					**1.4 (1.24-1.57)**	**2.10E-08**	**0.7661**	**1**			**0.983**	**0.994**	**AT/GC**
		intron	NCI	rs3778994	2175855	C|A	0.454	1.33 (1.14-1.55)	1.96E-04							
			UPENN	rs3778994	2175855	G|T	0.409	1.35 (1.14-1.60)	6.22E-04							
			NCI+UPENN	**rs3778994**	**2175855**			**1.34 (1.20-1.50)**	**4.40E-07**	**0.8947**	**11**					
		intron	NCI	rs3800924	2188249	C|T	0.359	1.35 (1.15-1.58)	2.38E-04			T	0.3285			
		intron	UPENN	rs4721441	2184060	A|G	0.389	1.28 (1.07-1.53)	5.92E-03			C	0.3641			
			NCI+UPENN					**1.32 (1.17-1.49)**	**4.98E-06**	**0.6909**	**16**			**0.821**	**0.98**	**TC/CT**
			NCI	rs7204293	50028105	G|A	0.05	0.54 (0.40-0.75)	1.59E-04			A	0.05805			
			UPENN	rs12445755	50034945	A|G	0.073	0.6 (0.41-0.88)	9.75E-03			C	0.06992			
			NCI+UPENN					**0.57 (0.44-0.72)**	**6.11E-06**	**0.7060**	**21**			**0.78**	**0.975**	**AC/GT**
			NCI	rs8046148	50142944	G|A	0.177	0.7 (0.58-0.85)	2.12E-04			A	0.1926			
16q12.1	*HEATR3*	intron	UPENN	rs2058813	50129454	C|T	0.191	0.82 (0.65-1.03)	0.0923			T	0.1715			
	AF086132		**NCI+UPENN**					**0.75 (0.65-0.86)**	**8.93E-05**	**0.2997**	**42**			**0.868**	**1**	**AT/GC**
			NCI	rs1496430	51096349	C|T	0.243	1.44 (1.19-1.74)	1.32E-04			T	0.1715			
			UPENN	rs1566043	51113337	G|A	0.147	1.2 (0.94-1.54)	0.1411			A	0.1319			
			NCI+UPENN					**1.35 (1.16-1.56)**	**9.08E-05**	**0.2493**	**43**			**0.734**	**1**	**TA/CG**
		missense	NCI	rs7193541	74664743	T|C	0.359	0.76 (0.65-0.88)	4.24E-04							
		Ile>Val	UPENN	rs7193541	74664743	T|C	0.396	0.83 (0.69-1.00)	0.0503							
			NCI+UPENN	**rs7193541**	**74664743**			**0.79 (0.70-0.89)**	**7.86E-05**	**0.4414**	**37**					
		coding-synon	NCI	rs4888262	74670458	T|C	0.552	1.36 (1.16-1.58)	9.12E-05							
16q23.1	*RFWD3*	Thr>Thr	UPENN	rs4888262	74670458	T|C	0.498	1.39 (1.16-1.67)	3.12E-04							
			NCI+UPENN	**rs4888262**	**74670458**			**1.37 (1.22-1.54)**	**1.12E-07**	**0.8255**	**7**					
		intron	NCI	rs4888265	74683737	C|T	0.553	1.35 (1.16-1.57)	1.26E-04			C	0.4789			
		intron	UPENN	rs8062783	74691264	C|T	0.482	1.31 (1.09-1.57)	3.35E-03			C	0.4763			
			NCI+UPENN					**1.33 (1.19-1.50)**	**1.51E-06**	**0.8069**	**13**			**0.989**	**1**	**CC/TT**
			NCI	rs9891129	56630474	C|T	0.282	0.74 (0.63-0.87)	2.59E-04			T	0.3272			
		intron	UPENN	rs302867	56752819	T|C	0.336	0.78 (0.65-0.93)	6.93E-03			C	0.3259			
			NCI+UPENN					**0.76 (0.67-0.85)**	**6.30E-06**	**0.7010**	**22**			**0.994**	**1**	**TC/CT**
			NCI	rs9905704	56632543	T|G	0.281	0.73 (0.62-0.86)	1.88E-04			G	0.3272			
		intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
			NCI+UPENN					**0.75 (0.66-0.85)**	**3.66E-06**	**0.7231**	**14**			**0.994**	**1**	**GT/TC**
		intron	NCI	rs1267543	56656624	G|A	0.184	0.7 (0.58-0.84)	1.81E-04			A	0.2375			
		intron	UPENN	rs2003537	56611871	G|C	0.211	0.82 (0.67-1.02)	0.0701			G	0.2137			
			NCI+UPENN					**0.75 (0.65-0.86)**	**6.57E-05**	**0.2665**	**34**			**0.858**	**0.992**	**AG/GC**
		intron	NCI	rs302858	56692800	C|A	0.282	0.74 (0.63-0.87)	2.77E-04			T	0.3259			
		intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
	C17orf47		**NCI+UPENN**					**0.75 (0.67-0.85)**	**5.21E-06**	**0.7736**	**18**			**1**	**1**	**TT/GC**
	TEX14	intron	NCI	rs304283	56785725	C|T	0.281	0.74 (0.63-0.87)	2.72E-04			T	0.3245			
17q22	*RAD51C*	intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
	PPM1E		**NCI+UPENN**					**0.75 (0.67-0.85)**	**5.11E-06**	**0.7719**	**17**			**0.994**	**1**	**TT/CC**

	TRIM37	intron	NCI	rs304271	56802458	G|A	0.282	0.75 (0.63-0.88)	3.78E-04			T	0.3259			
		intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
			NCI+UPENN					**0.76 (0.67-0.85)**	**6.89E-06**	**0.8132**	**23**			**1**	**1**	**TT/CC**
		intron	NCI	rs304272	56803028	G|T	0.282	0.74 (0.63-0.87)	3.17E-04			A	0.3259			
		intron	UPENN	rs302868	56752123	C|T	0.337	0.77 (0.64-0.93)	5.46E-03			T	0.3259			
			NCI+UPENN					**0.75 (0.67-0.85)**	**5.89E-06**	**0.7917**	**20**			**1**	**1**	**AT/CC**
		intron	NCI	rs7221274	57008128	A|G	0.34	0.72 (0.62-0.84)	3.56E-05			G	0.405			
		intron	UPENN	rs9898048	57108543	C|T	0.402	0.84 (0.70-1.00)	0.0527			A	0.4063			
			NCI+UPENN					**0.77 (0.69-0.87)**	**1.22E-05**	**0.2132**	**25**			**0.962**	**0.983**	**GA/AG**
		intron	NCI	rs11869052	57143404	T|G	0.34	0.74 (0.63-0.86)	1.20E-04			G	0.4063			
		intron	UPENN	rs9898048	57108543	C|T	0.402	0.84 (0.70-1.00)	0.0527			A	0.4063			
			NCI+UPENN					**0.78 (0.69-0.88)**	**3.18E-05**	**0.2856**	**27**			**1**	**1**	**GA/TG**
18q12.1	*MCART2*		NCI	rs1482516	29331752	G|T	0.313	0.74 (0.63-0.87)	2.72E-04			T	0.343			
			UPENN	rs4799597	29326919	A|C	0.350	0.83 (0.69-1.01)	0.0616			G	0.3404			
			NCI+UPENN					**0.78 (0.69-0.88)**	**6.79E-05**	**0.3775**	**35**			**0.977**	**0.994**	**TG/GT**
18q21.1	*DYM LIPG*	intron	NCI	rs7232791	46717796	T|C	0.119	0.67 (0.54-0.84)	3.45E-04			C	0.153			
		intron	UPENN	rs357897	46584184	T|C	0.159	0.75 (0.58-0.97)	0.0275			C	0.1425			
			NCI+UPENN					**0.7 (0.60-0.83)**	**3.55E-05**	**0.5227**	**28**			**0.879**	**0.978**	**CC/TT**
		intron	NCI	rs12458268	46928736	C|T	0.051	0.54 (0.39-0.74)	9.88E-05			T	0.09103			
		intron	UPENN	rs3794825	46806580	T|C	0.079	0.75 (0.53-1.07)	0.1130			G	0.08971			
			NCI+UPENN					**0.62 (0.49-0.79)**	**8.78E-05**	**0.1730**	**40**			**0.984**	**1**	**TG/CA**
		intron	NCI	rs12456319	46943301	G|A	0.052	0.54 (0.39-0.74)	1.01E-04			A	0.08839			
		intron	UPENN	rs3794825	46806580	T|C	0.079	0.75 (0.53-1.07)	0.1130			G	0.08971			
			NCI+UPENN					**0.62 (0.49-0.79)**	**8.91E-05**	**0.1740**	**41**			**0.952**	**0.984**	**AG/GA**
		intron	NCI	rs2276269	47102002	C|T	0.458	1.34 (1.15-1.56)	1.31E-04			T	0.4499			
		intron	UPENN	rs3786249	47097489	A|G	0.497	1.18 (0.99-1.41)	0.0696			T	0.4815			
			NCI+UPENN					**1.27 (1.13-1.43)**	**4.43E-05**	**0.2827**	**32**			**0.73**	**0.981**	**TC/CT**
		intron	NCI	rs9951026	47109693	A|G	0.525	1.34 (1.15-1.56)	1.18E-04			A	0.4855			
		intron	UPENN	rs3786249	47097489	A|G	0.497	1.18 (0.99-1.41)	0.0696			T	0.4815			
			NCI+UPENN					**1.27 (1.13-1.43)**	**3.95E-05**	**0.2854**	**31**			**0.942**	**0.979**	**AT/GC**

Supplementary Table 3 Description of the cohorts in TGCT meta-analysis

Label	Study	Design	Genotyping	Case	Control
STEED	The US Servicemen's Testicular Tumor Environmental and Endocrine Determinants Study	GWAS	Illumina 660K	479	555
FTCS	The NCI's Familial Testicular Cancer Study	GWAS	Illumina 660K	103	501
UPENN	University of Pennsylvania Health System/Fox Chase Cancer Center	GWAS	Affymetrix 6.0	349	919
			Discovery Meta	931	1,975
UKTCC	UK Testicular Cancer Collaboration	GWAS	Illumina HumanCNV370-Duo (case)/1.2M (control)	979	4,947
USC	University of Southern California	GWAS	Illumina 610K	358	258
TestPAC	The Testicular Cancer in Philadelphia Area Counties	follow-up	iPLEX Mass Array platform	331	683
ATLAS	The Adult Testicular Lifestyle and Blood Specimen Study	follow-up	iPLEX Mass Array platform	439	960
OUHRH	Oslo University Hospital-Radium Hospital, Norway	follow-up	TaqMan	865	392
MDA	M.D. Anderson Cancer Center	follow-up	TaqMan	239	351
			Replication	3,211	7,591
			Total	**4,142**	**9,566**

Supplementary Table 4 Meta-analysis results for 17 SNPs in association with TGCT

Gene nearby	cytoband	SNP	NCI	UPENN	USC	UKTCC	ATLAS	TestPAC	OUHRH	MDA	#study	Final Meta-analysis results for 17 SNPs					
												case	control	Allelic OR	95% CI (L-U)	*P* value	*P* for heterogeneity
C17orf47,TEX14	17q22	rs9905704	●	○	●	●	●	●	●	●	8	3,863	9,152	1.27	(1.18-1.33)	4.32E-13	0.6678
PPM1E	17q22	rs7221274	●	○	●	●	●	●	●	●	8	3,829	9,120	1.20	(1.12-1.28)	4.04E-09	0.1303
MAD1L1	7p22.3	rs12699477	●	○	●	●	●	●			6	2,832	8,424	1.21	(1.14-1.29)	5.59E-09	0.3177
RFWD3	16q22.3	rs4888262	●	●	●	○	●	●			5	2,801	8,393	1.26	(1.18-1.34)	5.15E-12	0.3968
HPGDS	4q22.2	rs17021463	●	○	●	●	●	●	●	●	8	3,866	9,146	1.19	(1.12-1.26)	1.11E-08	0.5827
MAD1L1	7p22.3	rs10275045	●	●	●	○	●	●	●	●	7	3,839	9,080	1.20	(1.13-1.27)	3.78E-10	0.0016
MAD1L1	7p22.3	rs3778991	●	○	●	○	●	●	●	●	7	3,839	9,123	1.21	(1.14-1.28)	6.73E-10	0.0037
TEX14	17q22	rs1267543	●	○	●	○	●	●			5	2,852	8,432	0.82	(0.76-0.89)	5.05E-07	0.3924
HEATR3	16q12.1	rs8046148	●	○	●	●	●	●		●	7	3,060	8,770	0.83	(0.77-0.90)	2.55E-06	2.45E-04
LIPG	18q21.1	rs9951026	●	○	●		●	●			5	1,845	3,459	1.22	(1.12-1.33)	4.66E-06	0.6348
	16q12.1	rs7204293	●	○	●	●	●	●			6	2,850	8,428	0.78	(0.69-0.87)	1.42E-05	0.0792
MRPL42P3	6q24.2	rs7760281	●	●	●	●	●	●			6	2,836	8,426	1.12	(1.05-1.19)	7.98E-04	0.0791
DYM	18q21.1	rs7232791	●	○	●	●	●	●			6	2,831	8,407	0.92	(0.84-1.00)	0.0478	0.0060
	3p26.1	rs9811958	●	○	●	●	●	●			6	2,832	8,398	1.06	(1.00-1.14)	0.0550	0.0012
	16q12.1	rs1496430	●	○	●	●	●	●			6	2,708	8,180	1.08	(1.00-1.17)	0.0569	0.0199
MCART2	18q12.1	rs1482516	●	○	●	●	●	●			6	2,830	8,409	0.94	(0.88-1.00)	0.0697	0.0082
DYM	18q21.1	rs12458268	●	○	●	●	●	●			6	2,810	8,301	0.99	(0.88-1.10)	0.8069	3.89E-07
	# data available per study		17	17	17	16	17	17	5	6							

open circle depicts indirect meta-analysis using surrogate and solid circle depicts direct match of SNP

Supplementary Table 5 Summary of genomic annotation by HaploReg and RegulomeDB

chr	pos(hg19)	LD*	variant	Ref	Alt	Alternative allele frequency			Conservation		Histone marks		DNase	Proteins bound	Motifs changed	RefSeq genes	dbSNP func annot	RegulomeDB score†
						ASN	CEU	YRI	GERP	SiPhy	Promoter	Enhancer						
4	95129238	0.87	rs2276910	T	C	0.7	0.59	0.97			9 cell types		55 cell types	8 bound proteins	Hic1 (>2.6)	*SMARCAD1*	intronic	4
4	95130025	0.87	rs183993	G	A	0.72	0.59	0.36			6 cell types	Huvec, K562, NHEK	Jurkat	FOXA1	SEF-1	*SMARCAD1*	intronic	3a
4	95137542	0.87	rs2632417	G	C	0.72	0.59	0.36								*SMARCAD1*	intronic	7
4	95139598	0.90	rs282450	G	C	0.72	0.58	0.32							NRSF,Klf4	*SMARCAD1*	intronic	7
4	95143484	0.87	rs2664881	A	G	0.72	0.59	0.34							Foxc1,Pou2f1	*SMARCAD1*	intronic	5
4	95144177	0.87	rs2664880	T	A	0.71	0.59	0.36								*SMARCAD1*	intronic	7
4	95145469	0.87	rs4693003	A	G	0.72	0.59	0.38								*SMARCAD1*	intronic	7
4	95146952	0.90	rs2664872	G	A	0.72	0.6	0.36								*SMARCAD1*	intronic	7
4	95147827	0.93	rs3113863	G	C	0.72	0.59	0.36								*SMARCAD1*	intronic	7
4	95148042	0.93	rs4558832	G	A	0.72	0.59	0.36								*SMARCAD1*	intronic	7
4	95149319	0.93	rs2632412	G	T	0.8	0.59	0.97							Mef2	*SMARCAD1*	intronic	7
4	95149434	0.90	rs2632411	G	A	0.72	0.6	0.38							Pax-4	*SMARCAD1*	intronic	7
4	95149507	0.90	rs2632410	A	T	0.72	0.6	0.36							Hoxb6,Pou2f1	*SMARCAD1*	intronic	6
4	95150805	0.90	rs10030586	A	C	0.72	0.6	0.36							Hsf	*SMARCAD1*	intronic	6
4	95152215	0.84	rs2632405	T	G	0.72	0.57	0.36								*SMARCAD1*	intronic	6
4	95153831	0.90	rs2664894	T	C	0.72	0.6	0.35								*SMARCAD1*	intronic	5
4	95153987	0.93	4:95373010	T	TAGCC	0.39	0.37	0.07							Mef2	*SMARCAD1*	intronic	NA
4	95155505	0.84	rs17309887	C	T	0.18	0.37	0								*SMARCAD1*	intronic	6
4	95156565	0.90	rs2865345	A	G	0.74	0.6	0.42							Foxj2,Foxa	*SMARCAD1*	intronic	6
4	95156664	0.93	rs115166545	T	C	0	0.03	0							CEBPG	*SMARCAD1*	intronic	NA
4	95157156	0.90	rs4235050	A	G	0.74	0.6	0.36					AG04449,HAEpiC,HIPEpiC			*SMARCAD1*	intronic	5
4	95157240	0.90	rs4693376	A	G	0.73	0.6	0.95					AG04449,HAEpiC			*SMARCAD1*	intronic	5
4	95158495	0.90	rs10016806	G	T	0.72	0.6	0.36						MAFK		*SMARCAD1*	intronic	5
4	95158610	0.90	rs2883	A	T	0.72	0.6	0.36						MAFF,MAFK	Arid3a,CEBPG,Dbx1	*SMARCAD1*	intronic	5
4	95159361	0.84	rs62320427	A	G	0.19	0.37	0							LBP-9	*SMARCAD1*	intronic	6
4	95160081	0.93	rs35833098	C	CAA	0.66	0.59	0.49								*SMARCAD1*	intronic	7
4	95162425	0.93	rs12511433	G	A	0.71	0.59	0.59								*SMARCAD1*	intronic	5
4	95165662	0.93	rs11097408	T	C	0.72	0.59	0.6							Irx	*SMARCAD1*	intronic	6
4	95167087	0.93	rs28522293	A	G	0.73	0.59	0.4								*SMARCAD1*	intronic	7
4	95167088	0.93	rs28701470	A	C	0.72	0.59	0.41								*SMARCAD1*	intronic	7
4	95167751	0.93	rs13435702	G	A	0.72	0.59	0.42								*SMARCAD1*	intronic	7
4	95168019	0.93	rs13434570	T	C	0.72	0.59	0.36								*SMARCAD1*	intronic	7
4	95168269	1.00	rs34755772	A	ATTG	0.64	0.5	0.26							Foxd3,Sox,Zfp105	*SMARCAD1*	intronic	6
4	95168552	0.93	rs4693378	A	T	0.72	0.59	0.36								*SMARCAD1*	intronic	7
4	95169682	0.93	rs10034781	G	T	0.72	0.59	0.92							CEBPG,CEBPB	*SMARCAD1*	intronic	6
4	95173779	0.93	rs7439869	T	C	0.72	0.59	0.95							Pou5f1,Sox	*SMARCAD1*	missense	7
4	95175682	0.80	rs7685121	G	A	0.72	0.59	0.33			NHEK		HRPEpiC			*SMARCAD1*	intronic	5
4	95178589	0.90	rs7657789	C	T	0.72	0.6	0.31								*SMARCAD1*	intronic	7
4	95179295	0.93	rs10024642	A	G	0.7	0.59	0.92								*SMARCAD1*	intronic	7
4	95183000	0.90	rs1565062	G	A	0.72	0.58	0.6				HMEC			NR3C	*SMARCAD1*	intronic	7
4	95184748	1.00	rs7697827	G	A	0.72	0.59	0.92							11 altered motifs	*SMARCAD1*	intronic	6
4	95188231	1.00	rs10640840	T	7-mer	0.66	0.48	0.27							PLZF	*SMARCAD1*	intronic	7
4	95192461	1.00	rs6532476	C	A	0.72	0.59	0.36							4 altered motifs	*SMARCAD1*	intronic	6
4	95197520	1.00	rs6823404	C	T	0.72	0.59	0.33						CEBPB,MAFK		*SMARCAD1*	synonymous	5
4	95203023	0.93	rs34933276	GCAA	G	0.6	0.43	0.2								*SMARCAD1*	intronic	6
4	95206252	0.93	rs61050998	TTATC	T	0.66	0.57	0.27							7 altered motifs	*SMARCAD1*	intronic	6
4	95206855	1.00	rs4693381	A	T	0.72	0.59	0.35							9 altered motifs	*SMARCAD1*	intronic	6
4	95211610	1.00	rs8336	T	C	0.72	0.59	0.42					5 cell types	CTCF		*SMARCAD1*	3'-UTR	4
4	95212813	1.00	rs2865350	A	T	0.78	0.59	0.96								369bp 3' of *SMARCAD1*		7
4	95213539	1.00	rs6814486	A	G	0.72	0.59	0.99							Foxj2	1.1kb 3' of *SMARCAD1*		6
4	95214407	0.87	4:95433430	T	TTGTG	0.57	0.49	0								2kb 3' of *SMARCAD1*		NA
4	95215887	0.87	rs28838299	T	G	0	0.56	0							Irf	3.4kb 3' of *SMARCAD1*		6
4	95215892	1.00	rs6840913	A	G	0	0.59	0								3.4kb 3' of *SMARCAD1*		7
4	95216802	1.00	rs114312933	C	T	0.72	0.59	0.36						MAFK	RXR::LXR,Irf	2.9kb 3' of *HPGDS*		5
4	95222755	0.97	rs10856908	G	T	0.72	0.58	0.39								*HPGDS*	intronic	7
4	95222995	1.00	rs6841121	G	A	0.72	0.59	0.43								*HPGDS*	intronic	7
4	95223698	1.00	rs10028613	T	C	0.8	0.59	0.97								*HPGDS*	intronic	7

4	95224812	1.00	**rs17021463**	T	G	0.72	0.59	0.41				8 cell types			*HPGDS*	intronic	5
4	95229039	0.90	rs2016483	A	T	0.7	0.57	0.36						Prrx2	*HPGDS*	intronic	5
4	95237611	0.84	4:95456634	A	12-mer	0	0.3	0.44						Pou2f1	*HPGDS*	intronic	NA
4	95239783	0.90	rs36099342	T	C	0.3	0.43	0.03						Pou2f1	*HPGDS*	intronic	7
4	95245527	0.90	rs13102926	C	T	0.3	0.43	0.03						EWSR1-FLI1	*HPGDS*	intronic	7
4	95248721	0.90	rs62320439	G	C	0.3	0.43	0.08							*HPGDS*	intronic	7
4	95249286	0.90	rs11097413	C	T	0.3	0.43	0.55						PPARG,PPAR	*HPGDS*	intronic	6
4	95253263	0.90	rs2865352	G	A	0.3	0.43	0.6							*HPGDS*	intronic	6
4	95254730	0.90	rs10516950	G	A	0.3	0.43	0.37					SP1		*HPGDS*	intronic	5
4	95256649	0.87	rs35744894	G	A	0.19	0.38	0.01						DMRT2	*HPGDS*	intronic	7
4	95257554	0.90	rs34465979	C	A	0.3	0.43	0.43						Bcl6b,STAT	*HPGDS*	intronic	6
4	95257753	0.87	rs28403447	G	A	0.29	0.43	0.54						Foxp1,Foxj2,HNF1	*HPGDS*	intronic	6
4	95268071	0.81	rs11097417	A	G	0.3	0.44	0.26						5 altered motifs	4kb 5' of *HPGDS*		5
4	95268748	0.81	rs10856909	C	G	0.3	0.44	0.2						Foxp1	4.7kb 5' of *HPGDS*		5
4	95275157	0.81	rs34181160	T	G	0.28	0.44	0.19						GATA,Evi-1	11kb 5' of *HPGDS*		6
7	1968953	1.00	**rs12699477**	T	C	0.44	0.29	0				16 cell types	ZNF263		*MAD1L1*	intronic	4
7	1966492	0.79	rs10950448	G	C	0.44	0.35	0				22 cell types			*MAD1L1*	intronic	4
7	1923695	0.77	rs11761270	C	T	0.38	0.34	0.01		K562		32 cell types	CTCF,GABP,RAD21		*MAD1L1*	intronic	4
7	1935273	0.75	rs11761818	C	T	0.38	0.33	0				7 cell types			*MAD1L1*	intronic	5
7	1932151	0.75	rs11768206	T	G	0.39	0.34	0				6 cell types		Pou1f1,Pou3f2	*MAD1L1*	intronic	5
7	1945678	0.75	rs11771451	T	C	0.38	0.34	0						Foxm1	*MAD1L1*	intronic	6
7	1950440	0.75	rs11771973	C	G	0.41	0.33	0				16 cell types			*MAD1L1*	intronic	5
7	1953650	0.75	rs12531315	C	G	0.41	0.33	0		NHLF		24 cell types		KROX	*MAD1L1*	intronic	5
7	1925454	0.72	rs12532128	T	C	0.38	0.34	0				14 cell types			*MAD1L1*	intronic	5
7	1958672	0.72	rs12536062	G	A	0.4	0.33	0				29 cell types			*MAD1L1*	intronic	5
7	1953571	0.72	rs12536261	G	A	0.41	0.33	0.01		NHLF		27 cell types		Spz1	*MAD1L1*	intronic	5
7	1948359	0.72	rs12537914	C	T	0.41	0.33	0				6 cell types			*MAD1L1*	intronic	5
7	1931030	0.72	rs12667688	C	T	0	0.34	0						NRSF	*MAD1L1*	intronic	7
7	1939765	0.72	rs12669937	C	T	0.38	0.33	0.01				22 cell types			*MAD1L1*	intronic	5
7	1965255	0.72	rs12671113	G	A	0.42	0.34	0.01				28 cell types	4 bound proteins		*MAD1L1*	intronic	4
7	1975624	0.72	rs12699483	C	G	0.44	0.34	0				20 cell types			*MAD1L1*	intronic	5
7	1953766	0.72	rs13224989	C	G	0.41	0.33	0.01		NHLF		21 cell types			*MAD1L1*	intronic	5
7	1976457	0.72	rs1801368	C	T	0.44	0.34	0				17 cell types	ERALPHA_A	Nrf-1	*MAD1L1*	missense	2b
7	1953897	0.72	rs2056478	C	T	0.38	0.33	0.01				21 cell types			*MAD1L1*	intronic	5
7	1954301	0.72	rs2056480	G	A	0.41	0.33	0				Chorion,GM12891,Hepatocy			*MAD1L1*	intronic	5
7	1955435	0.72	rs2056481	A	G	0.41	0.33	0				17 cell types			*MAD1L1*	intronic	5
7	1976863	0.71	rs2280546	G	A	0.44	0.34	0				24 cell types			*MAD1L1*	intronic	5
7	1976667	0.71	rs2280548	G	T	0.44	0.34	0.01				25 cell types	ERALPHA_A,CMYC		*MAD1L1*	intronic	3a
7	1976556	0.71	rs2280550	G	A	0.44	0.34	0				22 cell types	ERALPHA_A,CMYC	Esr2,Esr1,YY1	*MAD1L1*	intronic	2a
7	1926237	0.71	rs34145223	G	A	0.38	0.34	0				17 cell types		KROX	*MAD1L1*	intronic	5
7	1948712	0.71	rs34269264	C	T	0.41	0.33	0.01				13 cell types	GATA2	Spz1	*MAD1L1*	intronic	2b
7	1935245	0.71	rs34296663	C	T	0.38	0.33	0				7 cell types			*MAD1L1*	intronic	5
7	1938655	0.71	rs34922657	C	T	0.38	0.33	0		NHLF		24 cell types		AP-2	*MAD1L1*	intronic	2b
7	1948756	0.71	rs35582663	G	A	0.41	0.33	0				14 cell types	GATA2		*MAD1L1*	intronic	4
7	1954914	0.71	rs35797753	G	A	0.41	0.31	0				21 cell types		PU.1,Brachyury,GABP	*MAD1L1*	intronic	5
7	1942270	0.71	rs4236277	C	T	0.38	0.33	0				5 cell types			*MAD1L1*	intronic	5
7	1955152	0.71	rs4421257	T	C	0.41	0.33	0.38				13 cell types			*MAD1L1*	intronic	5
7	1977906	0.71	rs4719366	G	A	0.35	0.34	0		GM12878		9 cell types	ERALPHA_A	RREB-1	*MAD1L1*	intronic	2b
7	1933224	0.71	rs4721167	C	T	0.35	0.33	0				5 cell types		PPARG,HNF4	*MAD1L1*	intronic	5
7	1950784	0.71	rs4721184	T	C	0.41	0.33	0.01				19 cell types			*MAD1L1*	intronic	5
7	1973579	0.70	rs4721217	C	T	0.45	0.34	0				17 cell types		AP-2	*MAD1L1*	intronic	5
16	74657485	0.90	rs8058133	A	T	0.83	0.47	0.82				10 cell types	CTCF		*RFWD3*	3'-UTR	3a
16	74658867	0.90	rs8052367	G	A	0.83	0.47	0.82				RPTEC			*RFWD3*	intronic	5
16	74664810	0.97	rs7188880	A	T	0.83	0.45	0.83						Smad3,AP-4	*RFWD3*	synonymous	6
16	74668473	1.00	rs9929496	G	A	0.83	0.46	0.83						Mef2	*RFWD3*	intronic	6
16	74670458	1.00	**rs4888262**	C	T	0.83	0.46	0.82						CREB,Gmeb1,E4F1	*RFWD3*	synonymous	6
16	74672900	0.84	rs77736938	C	A	0.8	0.42	0.81						6 altered motifs	*RFWD3*	intronic	NA
16	74672901	0.84	rs7188581	T	A	0.79	0.42	0.81						DMRT3,Mef2	*RFWD3*	intronic	6
16	74675367	0.93	rs56032517	GATTA	G	0.73	0.21	0.62						OTX	*RFWD3*		6
16	74680801	0.93	rs57231731	13-mer	T	0.68	0.33	0.73							*RFWD3*	intronic	NA

16	74681632	1.00	rs4888264	T	G	0	0.46	0								*RFWD3*	intronic	7
16	74682496	0.84	rs28681530	C	T	0.85	0.42	0.82								*RFWD3*	intronic	7
16	74683737	1.00	rs4888265	T	C	0.84	0.46	0.82				HeLa-S3		HNF6		*RFWD3*	intronic	5
16	74684896	0.90	rs11414908	G	GT	0.81	0.45	0.76								*RFWD3*	intronic	NA
16	74686166	1.00	rs9923145	G	A	0.83	0.46	0.82						Lmo2-complex,GATA		*RFWD3*	intronic	6
16	74687906	0.97	rs4888267	A	G	0.87	0.45	0.82						AP-3,Pax-4		*RFWD3*	intronic	7
16	74688948	1.00	rs7191665	T	G	0.84	0.46	0.82								*RFWD3*	intronic	7
16	74691264	1.00	rs8062783	T	C	0.83	0.46	0.82	GM12878							*RFWD3*	intronic	6
16	74691362	0.90	rs8061942	C	T	0.84	0.43	0.82								*RFWD3*	intronic	7
16	74693499	0.93	rs58273376	C	CA	0.74	0.34	0.66	GM12878					Zfp105		*RFWD3*	intronic	7
16	74694058	0.93	rs7201320	T	G	0.85	0.46	0.82	GM12878, NHEK							*RFWD3*	intronic	7
16	74699293	1.00	rs9931225	T	C	0.87	0.46	0	HepG2							*RFWD3*	intronic	7
16	74700717	0.90	rs11276299	C	25-mer	0.69	0.37	0.6	9 cell types			46 cell types	32 bound proteins			*RFWD3*	5'-UTR	4
16	74700743	0.93	rs4887783	G	C	0.88	0.46	0	9 cell types			46 cell types	33 bound proteins			*RFWD3*	5'-UTR	4
16	74703907	0.93	rs5817922	TAA	T	0.66	0.32	0.62						8 altered motifs		1.8kb 3' of *MLKL*		7
16	74706298	0.81	rs4888274	A	T	0.83	0.46	0.81								*MLKL*	3'-UTR	7
16	74706736	0.87	rs4072222	A	C	0.87	0.48	0.81								*MLKL*	intronic	6
17	56630474	0.84	rs9891129	T	C	1	0.73	0.98								3.6kb 3' of *TEX14*		7
17	56632543	1.00	**rs9905704**	G	T	1	0.74	0.98				Melano		Pou1f1		1.5kb 3' of *TEX14*		5
17	56637770	0.80	rs2013182	G	A	1	0.72	0.79								*TEX14*	intronic	7
17	56674173	0.92	rs390524	A	G	0	0.28	0.08								*TEX14*	intronic	7
17	56685214	0.92	rs302852	C	T	0	0.28	0.09					HAE2F1	HEB		*TEX14*	intronic	5
17	56685424	0.85	rs302851	G	A	0	0.29	0.22						TFE		*TEX14*	intronic	7
17	56687481	0.80	rs302847	C	T	0	0.28	0.23						NF-I		*TEX14*	intronic	6
17	56687536	0.80	rs302846	T	A	0	0.28	0.23								*TEX14*	intronic	7
17	56692387	0.92	rs302857	C	G	0	0.28	0.08		GM12878		GM19238,HMVEC-LBl,NB4	PU1	NRSF		*TEX14*	intronic	3a
17	56692800	0.92	rs302858	G	T	0	0.28	0		GM12878		GM19240				*TEX14*	intronic	5
17	56695967	0.92	rs186221	C	T	0	0.28	0.08		GM12878						*TEX14*	intronic	7
17	56697040	0.85	rs302861	G	C	0	0.29	0.2						Pitx2		*TEX14*	intronic	6
17	56697444	0.92	rs302862	G	T	0	0.28	0						NF-AT,Nkx6-1		*TEX14*	intronic	6
17	56698827	0.85	rs302863	G	A	0	0.29	0.23				7 cell types	NFKB	PPARG,PLAG1,PPAR		*TEX14*	intronic	2b
17	56707245	0.83	rs12949598	C	T	0	0.26	0.08								*TEX14*	intronic	7
17	56707603	0.83	rs34232744	GAGGC	G	0	0.13	0.04						Sp1		*TEX14*		6
17	56708019	0.92	rs694720	T	C	0	0.28	0.02								*TEX14*	intronic	7
17	56709086	0.92	rs369184	C	T	0	0.28	0.08	4 cell types			105 cell types	79 bound proteins	Pax-8		*TEX14*	intronic	2c
17	56709206	0.92	rs430973	T	G	0	0.28	0.08	9 cell types			36 cell types	49 bound proteins	Srf		*TEX14*	intronic	4
17	56709222	0.92	rs444393	A	G	0	0.28	0.08	9 cell types			33 cell types	42 bound proteins	Myc		*TEX14*	intronic	4
17	56717249	0.92	rs68139131	A	G	0	0.28	0.08				Huh-7.5,HCM,MCF-7	ERALPHA_A			*TEX14*	intronic	3a
17	56719197	0.92	rs581903	C	T	0	0.28	0.08		Huvec		Medullo		Evi-1		*TEX14*	intronic	5
17	56730593	0.92	rs385793	G	A	0	0.28	0.02								*TEX14*	intronic	7
17	56732304	0.80	rs2611774	G	A	0	0.28	0.08						Lmo2-complex,AREB6		*TEX14*	intronic	7
17	56736490	0.92	rs654743	T	C	0	0.28	0.08	GM12878, HepG2, NHEK			100 cell types	67 bound proteins			*TEX14*	intronic	4
17	56736785	0.85	rs372648	A	C	0	0.29	0.23	9 cell types			89 cell types	19 bound proteins			*TEX14*	intronic	4
17	56737765	0.88	rs2643126	A	G	0	0.27	0						4 altered motifs		*TEX14*	intronic	6
17	56749298	0.88	rs304265	C	T	0	0.27	0.08						Elf1,Ets,GATA		*TEX14*	intronic	6
17	56752123	0.92	rs302868	C	T	0	0.28	0.06								*TEX14*	intronic	6
17	56752819	0.92	rs302867	T	C	0	0.28	0.08								*TEX14*	intronic	6
17	56756797	0.92	17:54111796	C	CTCG	0	0.27	0.09				34 cell types	50 bound proteins			*TEX14*	intronic	NA
17	56757442	0.92	rs5821242	CT	C	0	0.15	0.05	NHEK	GM12878, K562		5 cell types	EBF1,NFKB	Evi-1		*TEX14*	intronic	2b
17	56761411	0.92	rs302880	C	T	0	0.28	0.08								*TEX14*	intronic	7
17	56762964	0.85	rs302878	G	A	0	0.29	0.22								*TEX14*	intronic	7
17	56764018	0.85	rs302877	G	A	0	0.29	0.23								*TEX14*	intronic	7
17	56765478	0.85	rs80030779	T	G	0	0.29	0.22						RREB-1,Zfp105		*TEX14*	intronic	NA
17	56767874	0.84	rs302875	G	A	0	0.28	0.08								*TEX14*	intronic	6
17	56776663	0.88	rs695137	G	A	0	0.28	0.11						Sox		*RAD51C*	intronic	6
17	56783255	0.92	rs2567901	C	T	0	0.28	0								*RAD51C*	intronic	7
17	56785571	0.92	rs304282	T	C	0	0.28	0.14	GM12878							*RAD51C*	intronic	7
17	56785725	0.92	rs304283	C	T	0	0.28	0.08								*RAD51C*	intronic	7
17	56786062	0.92	17:54141061	T	TG	0	0.15	0.06				WI-38	CMYC,CTCF	MAZR,Zbtb7b		*RAD51C*	intronic	NA
17	56791832	0.92	rs2611779	A	G	0	0.28	0						9 altered motifs		*RAD51C*	intronic	6

17	56794115	0.85	rs2567894	G	A	0	0.29	0.23						*RAD51C*	intronic	7
17	56798207	0.85	rs28363318	T	C	0	0.29	0.23						*RAD51C*	intronic	5
17	56799380	0.85	rs304267	A	G	0	0.29	0.23						*RAD51C*	intronic	7
17	56799907	0.85	rs304268	T	C	0	0.29	0.23					Zbtb12	*RAD51C*	intronic	6
17	56802059	0.81	rs304269	G	A	0	0.3	0.23					7 altered motifs	*RAD51C*	intronic	6
17	56802458	0.92	rs304271	C	T	0	0.28	0.09						*RAD51C*	intronic	7
17	56803028	0.92	rs304272	C	A	0	0.28	0.13						*RAD51C*	intronic	6
17	56804213	0.80	17:54159212	GCT	G	0	0.09	0.03						*RAD51C*		NA
17	56808530	0.92	rs2611783	A	G	0	0.28	0.13					5 altered motifs	*RAD51C*	intronic	6
17	56814456	0.85	rs304262	A	G	0	0.29	0.23					MIF-1	2.8kb 3' of *RAD51C*		6
17	56823298	0.92	rs304293	T	G	0	0.28	0					Olf-1,LUN-1	9.9kb 5' of *PPM1E*		6
17	56826949	0.91	rs2643120	A	T	0	0.26	0.14						6.3kb 5' of *PPM1E*		7
17	56826953	0.83	rs34556895	G	T	0	0.26	0.14						6.3kb 5' of *PPM1E*		7
17	56826958	0.83	rs2611777	A	G	0	0.26	0.14					Hoxb13	6.3kb 5' of *PPM1E*		6
17	56829516	0.91	rs2643117	G	A	0	0.26	0		K562,LNCaP			RREB-1,Foxp1	3.7kb 5' of *PPM1E*		5
17	56831887	0.84	rs170024	T	C	0	0.28	0.2		Fibrobl,Hepatocytes,Myometr			Pou2f1,Cart1	1.3kb 5' of *PPM1E*		5
17	56839689	0.85	rs304298	G	A	0	0.29	0.23						*PPM1E*	intronic	7
17	56842043	0.92	rs11657676	A	G	0	0.28	0.02		HRCEpiC,HRE,RPTEC	PAX5C20,PAX5N19		ZID	*PPM1E*	intronic	3a
17	56844109	0.88	rs28483505	G	C	0	0.27	0						*PPM1E*	intronic	6
17	56848677	0.83	rs12939500	T	C	0	0.26	0.14					Foxl1	*PPM1E*	intronic	6
17	56854517	0.92	rs11655808	T	C	0	0.28	0.02			CMYC,CTCF			*PPM1E*	intronic	5
17	56861253	0.88	rs12942879	G	A	0	0.28	0.11						*PPM1E*	intronic	7
17	56863336	0.92	17:54218335	A	AT	0	0.09	0.05		K562	SK-N-SH_RA	CEBPB		*PPM1E*	intronic	NA
17	56863918	0.85	rs7212271	C	T	0	0.29	0.22		HSMM, K562, NHEK	80 cell types	23 bound proteins		*PPM1E*	intronic	4
17	56864242	0.85	rs11079356	C	A	0	0.29	0.23		HSMM, K562, NHEK	AoSMC,Huh-7.5		Pou3f3,Foxj2	*PPM1E*	intronic	5
17	56876627	0.92	rs34430710	A	T	0	0.28	0.14					10 altered motifs	*PPM1E*	intronic	6
17	56883236	0.92	rs72828707	A	G	0	0.28	0.14						*PPM1E*	intronic	7
17	56887394	0.92	rs34694297	CT	C	0	0.19	0.08						*PPM1E*	intronic	6
17	56892872	0.92	rs12942969	A	T	0	0.28	0.08					Bcl6b	*PPM1E*	intronic	6
17	56894638	0.92	rs35934668	A	G	0	0.28	0.01						*PPM1E*	intronic	6
17	56900870	0.92	rs12952362	A	G	0	0.28	0.14					Pou3f3	*PPM1E*	intronic	5
17	56902444	0.92	rs12947196	G	A	0	0.28	0.12					SREBP	*PPM1E*	intronic	7
17	56914643	0.80	rs57058929	AG	A	0	0.32	0.09					5 altered motifs	*PPM1E*		6
17	56914687	0.85	17:54269469	C	CAT	0	0.16	0.25						*PPM1E*		NA
17	56916804	0.92	rs36071323	C	T	0	0.28	0.13						*PPM1E*	intronic	6
17	56919549	0.92	rs11650343	G	A	0	0.28	0.02		MCF-7				*PPM1E*	intronic	5
17	56943950	0.92	rs57543170	16-mer	C	0	0.15	0.15						*PPM1E*	intronic	7
17	56647528	0.86	rs11289066	CA	C	0.66	0.73	0.14					Zfp105,Pou2f1	*TEX14*	intronic	6
17	56950920	0.96	rs4337355	C	T	0.23	0.34	0.42						*PPM1E*	intronic	7
17	56950926	0.93	17:54305708	13-mer	G	0.27	0.58	0.45						*PPM1E*	intronic	5
17	56954655	1.00	rs12940492	C	G	0.22	0.35	0.42						*PPM1E*	intronic	7
17	56957968	0.93	rs35028478	A	AGTTT	0.26	0.21	0.29						*PPM1E*	intronic	5
17	56966052	1.00	rs11659035	C	T	0.22	0.35	0.41						*PPM1E*	intronic	6
17	56972741	1.00	rs11650809	A	G	0.22	0.35	0.4						*PPM1E*	intronic	7
17	56984757	0.93	17:54339539	TTTA	T	0.11	0.14	0.22		Urothelia			Pou3f3,Hoxb9,Cdx2	*PPM1E*		7
17	56986095	0.96	rs11652761	C	T	0.21	0.36	0.41						*PPM1E*	intronic	7
17	56987142	1.00	rs9901655	G	A	0.21	0.35	0.15						*PPM1E*	intronic	6
17	57008128	1.00	**rs7221274**	A	G	0.22	0.35	0.42						*PPM1E*	intronic	7
17	57008912	0.96	rs28709536	C	A	0.2	0.34	0.42						*PPM1E*	intronic	7
17	57009024	1.00	rs11079367	A	G	0.22	0.35	0.42						*PPM1E*	intronic	7
17	57015777	0.93	rs10541417	GTC	G	0.15	0.25	0.33						*PPM1E*	intronic	7
17	57028126	1.00	rs6503888	T	C	0.21	0.35	0.38		Monocytes-CD14+	KAP1,SETDB1		Pax-5	*PPM1E*	intronic	4
17	57033816	0.93	rs79776482	A	C	0	0.18	0					5 altered motifs	*PPM1E*	intronic	NA
17	57038282	1.00	rs2036730	A	G	0.22	0.35	0.41					Sox,Foxp1	*PPM1E*	intronic	6
17	57041293	1.00	rs12944274	G	C	0.2	0.35	0.14						*PPM1E*	intronic	6
17	57043367	1.00	rs2286299	T	G	0.23	0.35	0.4					PPAR	*PPM1E*	intronic	5
17	57047885	1.00	rs10221208	T	C	0.22	0.35	0.42						*PPM1E*	intronic	7
17	57061978	1.00	rs3809723	T	G	0.21	0.35	0.41		HAEpiC				*PPM1E*	3'-UTR	5
17	57065924	0.96	rs6503891	A	C	0.2	0.36	0.42					UF1H3BETA,MAZ,Ik-1	*TRIM37*	intronic	6
17	57067086	0.93	rs6503892	G	A	0.22	0.35	0.42					NR3C	*TRIM37*	intronic	7

17	57072461	0.96	rs34718875	G	A	0.17	0.36	0.31			HSMM					*TRIM37*	intronic	7
17	57074153	0.96	rs10595343	ATG	A	0	0.24	0.3								*TRIM37*	intronic	6
17	57079742	1.00	rs11652713	C	T	0.19	0.35	0.06			GM12878, Huvec	GM06990,GM12864,Monoc			Esr1,Six	*TRIM37*	intronic	1f
17	57093825	0.96	rs8080071	C	T	0.8	0.64	0.85								*TRIM37*	intronic	7
17	57094617	0.96	rs4932691	T	C	0.82	0.64	0.86							Cdx2,Hoxb8,Hoxa10	*TRIM37*	intronic	6
17	57097047	1.00	rs12943705	T	C	0.8	0.65	0								*TRIM37*	intronic	7
17	57102133	1.00	rs2108435	T	C	0.8	0.65	0.64					CTCF			*TRIM37*	intronic	5
17	57105223	1.00	rs8077332	C	T	0.8	0.65	0.85			GM12878	6 cell types			EWSR1-FLI1	*TRIM37*	intronic	1b
17	57108543	1.00	rs9898048	A	G	0.8	0.65	0.85				T-47D	FOXA1			*TRIM37*	intronic	1f
17	57114148	1.00	rs33922512	C	G	0.8	0.65	0.86								*TRIM37*	intronic	7
17	57123931	0.93	17:54478713	A	ACT	0.43	0.26	0.46					GATA1			*TRIM37*	intronic	6
17	57131294	0.93	rs10634008	T	TAAACA	0.67	0.67	0.56								*TRIM37*	intronic	6
17	57133010	1.00	rs7218103	T	G	0	0.65	0								*TRIM37*	intronic	7
17	57134059	0.90	rs9906653	A	G	0.8	0.63	0.84								*TRIM37*	intronic	7
17	57139293	1.00	rs2877926	G	A	0.8	0.65	0.64				NHEK	JUND			*TRIM37*	intronic	4
17	57140304	0.93	rs972849	A	T	0.8	0.63	0.81							23 altered motifs	*TRIM37*	intronic	6
17	57143404	1.00	rs11869052	T	G	0.2	0.35	0.15				A549,Caco-2,MCF-7				*TRIM37*	intronic	4
17	57144355	1.00	rs35180305	C	T	0.23	0.35	0.33								*TRIM37*	intronic	7
17	57149904	1.00	rs9904621	C	T	0.2	0.35	0.19							Hoxb3,Pax	*TRIM37*	intronic	7
17	57151579	1.00	rs11650456	T	C	0.2	0.35	0.19			Huvec	5 cell types				*TRIM37*	intronic	5
17	57158345	1.00	rs7214402	T	C	0.19	0.35	0.19							Pou2f1	*TRIM37*	intronic	6
17	57159268	0.89	rs34817776	A	C	0.2	0.34	0.33							Zbtb7b,Zfp740	*TRIM37*	intronic	6
17	57159990	1.00	rs7503179	A	T	0.2	0.35	0.19								*TRIM37*	intronic	6
17	57162209	1.00	rs12103622	A	G	0.21	0.35	0.23				WERI-Rb-1				*TRIM37*	intronic	5
17	57176106	1.00	rs9892666	A	T	0.2	0.35	0.36								*TRIM37*	intronic	6
17	57177770	1.00	rs8078926	T	C	0.18	0.35	0.11							Hoxa9,Hoxa10	*TRIM37*	intronic	7
17	57183266	0.86	rs10590674	CATAT	C	0.16	0.27	0.07		8 cell types	Huvec					*TRIM37*	intronic	5
17	57186832	0.93	rs61092888	G	GT	0	0.15	0								474bp 3' of *SKA2*		NA
17	57187795	0.89	rs7208282	C	T	0.14	0.33	0.12							7 altered motifs	*SKA2*	3'-UTR	6
17	57201717	0.89	rs2333445	C	G	0	0.33	0			GM12878					*SKA2*	intronic	6

*LD threshold was set to r2>=0.8 and for rs12699477, due to no surrogates passed 0.8 threshold, set to r2>=0.7

†RegulomeDB scores: 1a, eQTL + TF binding + matched TF motif + matched DNase Footprint + DNase peak;1b, eQTL + TF binding + any motif + DNase Footprint + DNase peak;1c, eQTL + TF binding + matched TF motif + DNase peak;binding + matched TF motif; 1d, eQTL + TF binding + any motif + DNase peak;1e, eQTL + TF;1f, eQTL + TF binding / DNase peak;2a, TF binding + matched TF motif + matched DNase Footprint + DNase peak;2b,TF binding + any motif + DNase Footprint + DNase peak;+ any motif + DNase peak; 2c, TF binding + matched TF motif + DNase peak;3a, TF binding; 3b, TF binding + matched TF motif;4, TF binding + DNase peak;5, TF binding or DNase peak;6, other

Supplementary Table 6 Prediction of potential regulatory SNPs

variant	Motifs changed	RefSeq genes	dbSNP func annot	Motif of interest 1	LOD score 1[#]	Motif of interest 2	LOD score 2[#]	Histone marks in H1-ESC
rs2276910	Hic1	*SMARCAD1*	intronic	Hic1	>2.6			Active Promoter
rs183993	SEF-1	*SMARCAD1*	intronic	None [$]	NA [$]			Weak/poised Promoter
rs7439869	Pou5f1, Sox	*SMARCAD1*	missense	Pou5f1 (OCT4)	6.9	Sox4	>2.7	Transcriptional elongation
rs35744894	DMRT2	*HPGDS*	intronic	DMRT2	>6.9			Heterochromatin; Low/no signal
rs11768206	Pou1f1, Pou3f2	*MAD1L1*	intronic	Pou3f2	<-10.5	Pou1f1	<-2.4	Weak transcribed
rs12531315	KROX	*MAD1L1*	intronic	KROX	<-2.7			Weak transcribed
rs12536261	Spz1	*MAD1L1*	intronic	Spz1	-0.6			Weak transcribed
rs1801368	Nrf-1	*MAD1L1*	missense	Nrf-1	<-0.9			Heterochromatin; Low/no signal
rs2280550	Esr2, Esr1, YY1	*MAD1L1*	intronic	ESR2	-3.7	ESR1	<-1.9	Heterochromatin; Low/no signal
rs34145223	KROX	*MAD1L1*	intronic	KROX	<-2.4			Weak transcribed
rs34269264	Spz1	*MAD1L1*	intronic	Spz1	1.4			Weak transcribed
rs34922657	AP-2	*MAD1L1*	intronic	AP-2	>2.3			Weak transcribed
rs35797753	PU.1, Brachyury, GABP	*MAD1L1*	intronic	GABP	<-5			Weak transcribed
rs4719366	RREB-1	*MAD1L1*	intronic	RREB	2			Heterochromatin; Low/no signal
rs4721167	PPARG, HNF4	*MAD1L1*	intronic	PPARG	-4.1	HNF4 (RXR-alpha)	<-2.1	Weak transcribed
rs4721217	AP-2	*MAD1L1*	intronic	AP-2	<-1.6			Heterochromatin; Low/no signal
rs7188880	Smad3, AP-4	*RFWD3*	synonymous	Smad3	<-2.5	AP-4	-1	Transcriptional elongation
rs4888262	CREB, Gmeb1, E4F1	*RFWD3*	synonymous	E4F1	-10.7			Transcriptional elongation
rs58273376	Zfp105	*RFWD3*	intronic	Zfp105	1.2			Transcriptional initiation
rs302857	NRSF	*TEX14*	intronic	NRSF (REST)	-3.7			Heterochromatin; Low/no signal
rs302863	PPARG, PLAG1, PPAR	*TEX14*	intronic	PPARG	<-6.8			Heterochromatin; Low/no signal
rs369184	Pax-8	*TEX14*	intronic	EGR1	NA [%]			Active promotor
rs430973	Srf	*TEX14*	intronic	Srf	<-2.5			Active Promoter
rs444393	Myc	*TEX14*	intronic	Myc	>3			Active Promoter
rs5821242	Evi-1	*TEX14*	intronic	Evi-1	-2.9			Heterochromatin; Low/no signal
rs304269	7 altered motifs	*RAD51C*	intronic	multiple forkhead box motifs	12 (Foxc1)			Heterochromatin; Low/no signal
rs11657676	ZID	*PPM1E*	intronic	ZID	<-2.6			Weak transcribed
rs11652713	Esr1, Six	*TRIM37*	intronic	Esr1	-2	Six	-0.3	Weak transcribed
rs8077332	EWSR1-FLI1	*TRIM37*	intronic	None [@]	NA [@]			Weak transcribed
rs8336	miRNA binding	*SMARCAD1*	3'-UTR	hsa-miR-573				Weak transcribed
rs4888274	miRNA binding	*MLKL*	3'-UTR	hsa-miR-483-3p, hsa-miR-483-3p, hsa-miR-502-5p, hsa-miR-502-5p, hsa-miR-99a, hsa-miR-9				Heterochromatin; Low/no signal

[#] LOD score = LOD(alt)-LOD(ref) [change in log-odds score] of the motif, calculated by HaploReg

[$] Motif for non human TF

[@] Motif for fusion protein

[%] only predicted by Regulome (no LOD score available)

Supplementary Table 7 Conditional analysis on the two multi-SNP loci identified by meta-analysis using NCI data

cytoband	SNP	Location(hg19)	distance(bps)	LD with conditioned SNP (r^2)	NCI *P* value	conditional *P* in NCI	meta *P* value	*P* for heterogeneity
7p22.3	rs10275045	1,920,826	48,127	0.66	5.19E-05	0.040062	4.96E-08	8.92E-04
7p22.3	rs12699477	1,968,953	0	1	4.64E-04	conditioned SNP	5.59E-09	0.3177
7p22.3	rs3778991	2,172,455	251,629	0.496	4.76E-05	0.0171688	6.21E-06	0.0023
17q22	rs9905704	56,632,543	0	1	1.88E-04	conditioned SNP	4.32E-13	0.6678
17q22	rs7221274	57,008,128	375,585	0.707	3.56E-05	0.0613351	4.04E-09	0.1303