

# Analysis Summary: Male Pattern Baldness

## Phenotype Description

The male pattern baldness phenotype was defined for use in a recent meta-analysis (Li *et al.*, *PLoS Genet* 8: e1002746, 2012). All subjects are male, and participants provided responses to the "Hair Loss in Men and Women" survey.

- "Please choose the image that best captures your hair's pattern and density. If your head is shaved, please answer for how your hair looks when grown out. If none of these images are similar to your hair's pattern and density, choose none of the above." (images a to s, corresponding to Hamilton scale)
- "Have you experienced hair loss or thinning?" (Yes, No, I'm not sure, Decline to state)
- "How old were you when you first started to notice hair loss?" (under 18, 18-24, 25-29, 30-34, 35-39, 40-44, 45-49, 50-54, 55-59, 60-64, 65-69, 70-74, 75-79, 80 or older, I'm not sure)

Cases reported having experienced hair loss or thinning, with onset before age 40, and current hair loss of Hamilton grade III or higher. Controls are at least 30 years old, and report not having experienced hair loss or thinning, and at most Hamilton grade I, or if age 50 or older, Hamilton grade II.

## Phenotype Statistics

The following table shows demographics of unrelated, European individuals included in the GWAS.

Phenotype	Group	Total	M	F	(0,30]	(30,45]	(45,60]	(60,Inf]
male_pattern_meta	case	9009	9009	0	317	2677	3064	2951
	control	8491	8491	0	283	3476	2717	2015

The following table shows the phenotypic distribution across 23andMe genotyping platforms for individuals included in the GWAS.

Phenotype	Group	Total	v1/v2	v3	v4
male_pattern_meta	case	9009	1752	6685	572
	control	8491	1770	6250	471

## Null Model with Covariates

The following table shows results of fitting a model for the trait based on just the covariates. Principal coordinates have been standardized, so these effect sizes are in units of standard deviations.

	Estimate	Std. Error	z value	Pr(> z )	LRT	Pr(>Chi)
age	0.01999	0.00113	17.8	$1.4 \times 10^{-70}$	322.2	$4.7 \times 10^{-72}$
pc.0	-0.09092	0.01551	-5.9	$4.6 \times 10^{-9}$	34.8	$3.7 \times 10^{-9}$
pc.1	-0.00984	0.01545	-0.6	0.52	0.4	0.52
pc.2	0.00853	0.01543	0.6	0.58	0.3	0.58
pc.3	-0.00627	0.01540	-0.4	0.68	0.2	0.68
pc.4	0.03646	0.01557	2.3	0.019	5.5	0.019

## SNP-level QC information

The following table shows results for QC filters on the genotyped data:

	failed	passed
no filters	0	1030430
not V1-only, chrM, chrY	4790	1025640
parent-offspring test	2129	1023511
MAF > 0%	3203	1020494
HWE > 1e-20	48225	972832
gt.rate > 90%	30775	952826
batch effects	28267	945446

The following table shows results for QC filters on the imputed dosage data:

	failed	passed
no filters	0	13733809
MAF > 0%	0	13733809

imputation quality      0 13733809  
 batch effects            2168 13731641

The following table shows results for QC filters on the merged association test results:

	passed	total
imputed only	12833621	12833621
both passed	898002	13731623
genotyped only	47444	13779067
no test result	-34089	13744978
failed to converge	-308337	13436641

## Genetic Association Tests

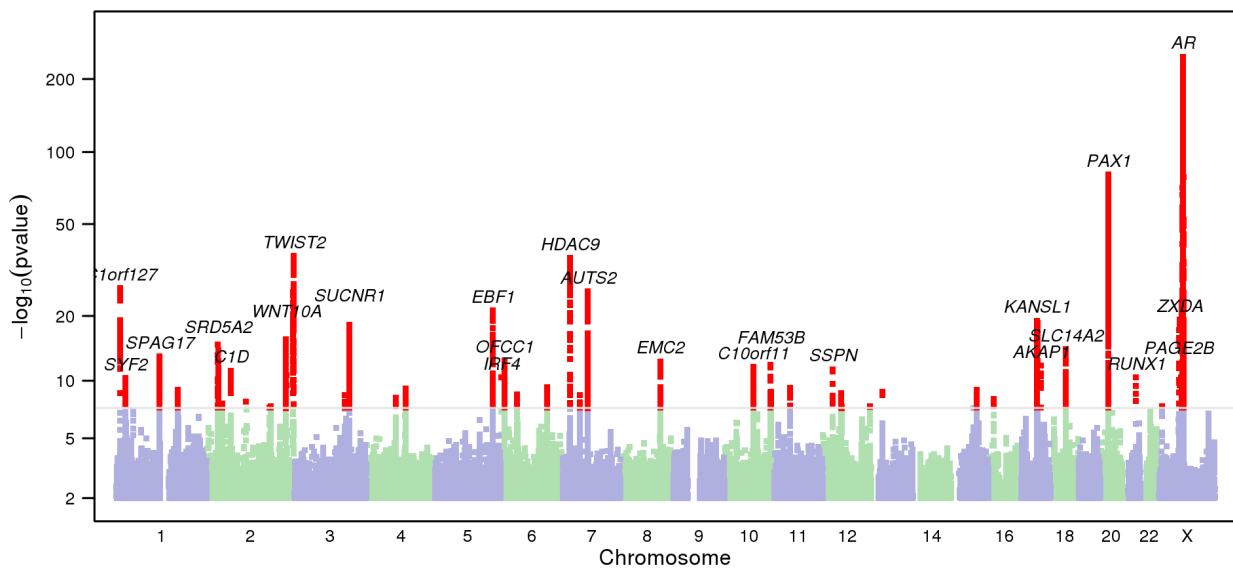
We performed logistic regression assuming an additive model for allelic effects, using the model:

*male\_pattern\_meta* ~ age + pc.0 + pc.1 + pc.2 + pc.3 + pc.4 + genotype

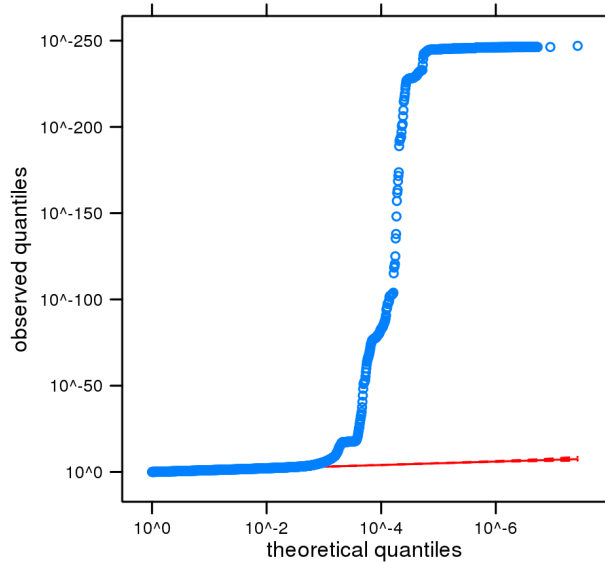
This genome-wide association analysis includes data from 9009 cases and 8491 controls of European ancestry, filtered to remove close relatives.

The results in this report have been adjusted for a genomic control inflation factor  $\lambda=1.065$ . The equivalent inflation factor for 1000 cases and 1000 controls  $\lambda_{1000} = 1.007$ , and for 10000,  $\lambda_{10000} = 1.074$ .

## Manhattan Plot



## Q-Q Plot of GWAS Results



## Index SNPs for Strongest Associations

cytoband	assay.name	scaffold	position	alleles	src	pvalue	OR	95% CI	gene.context
Xq12	rs200644307	chrX	66300914	D/I	I	$1.0 \times 10^{-247}$	2.277	[2.160,2.400]	EDA2R---[---AR
20p11.22	rs2015163	chr20	22000281	C/T	I	$2.7 \times 10^{-81}$	1.547	[1.479,1.619]	PAX1---[---FOXA2
2q37.3	rs11684254	chr2	239695893	C/G	I	$2.3 \times 10^{-37}$	1.350	[1.289,1.414]	ASB1---[---TWIST2
7p21.1	rs7801037	chr7	18897511	A/C	I	$1.3 \times 10^{-36}$	0.750	[0.717,0.785]	[HDAC9]
1p36.22	rs2095921	chr1	11033322	C/G	I	$3.1 \times 10^{-27}$	0.744	[0.705,0.785]	[C1orf127]
7q11.22	rs34991987	chr7	68595226	D/I	I	$2.3 \times 10^{-26}$	1.269	[1.214,1.326]	[---AUTS2
5q33.3	rs62385385	chr5	158367249	A/T	I	$7.4 \times 10^{-22}$	1.247	[1.192,1.304]	[EBF1]
Xp11.21	rs111763724	chrX	58002334	A/G	I	$5.6 \times 10^{-20}$	0.597	[0.532,0.670]	ZXDA--[
17q21.31	rs201408539	chr17	44166078	D/I	I	$8.4 \times 10^{-20}$	1.338	[1.257,1.425]	[KANSL1]
3q25.1	rs4679956	chr3	151654862	C/T	I	$5.3 \times 10^{-19}$	1.223	[1.170,1.279]	SUCNR1--[---MBNL1
Xp11.21	rs143149578	chrX	55950016	C/T	I	$6.0 \times 10^{-18}$	0.584	[0.514,0.664]	RRAGB---[---KLF8
2q35	rs74333950	chr2	219746292	G/T	I	$2.2 \times 10^{-16}$	0.764	[0.716,0.815]	[WNT10A]
2p23.1	rs9282858	chr2	31805826	C/T	I	$1.5 \times 10^{-15}$	0.601	[0.529,0.682]	[SRD5A2]
18q12.3	rs34800162	chr18	42808059	G/T	I	$8.2 \times 10^{-15}$	1.217	[1.158,1.278]	[SLC14A2]
1p12	rs12083887	chr1	118881689	A/G	I	$9.1 \times 10^{-14}$	0.843	[0.806,0.882]	SPAG17---[---TBX15
6p24.3	rs9357047	chr6	9327556	C/T	I	$3.0 \times 10^{-13}$	0.847	[0.811,0.886]	SLC35B3---[---OFCC1
8q23.1	rs79437808	chr8	109597801	C/T	I	$4.1 \times 10^{-13}$	2.153	[1.738,2.668]	EMC2--[---TMEM74
Xp11.21	rs185597083	chrX	55133845	A/C	I	$7.5 \times 10^{-13}$	1.388	[1.267,1.520]	PAGE2B--[---FAM104B
10q26.13	rs3781458	chr10	126343931	C/T	I	$1.2 \times 10^{-12}$	0.849	[0.812,0.889]	[FAM53B]
17q22	rs62060349	chr17	55231168	C/T	I	$1.5 \times 10^{-12}$	0.853	[0.816,0.891]	AKAP1--[---MSI2
10q22.3	rs11593840	chr10	78196612	A/G	I	$2.2 \times 10^{-12}$	0.852	[0.815,0.891]	[C10orf11]
12p12.1	rs9300169	chr12	26426671	A/G	I	$4.4 \times 10^{-12}$	0.839	[0.798,0.882]	SSPN--[---ITPR2
2p14	rs6546334	chr2	68078849	C/T	I	$6.4 \times 10^{-12}$	0.851	[0.813,0.891]	ETAA1---[---C1D
6p25.3	rs12203592	chr6	396321	C/T	I	$4.0 \times 10^{-11}$	1.220	[1.150,1.294]	[IRF4]
21q22.12	rs68088846	chr21	36208167	A/G	I	$4.4 \times 10^{-11}$	0.828	[0.783,0.876]	[RUNX1]
1p36.11	rs7534070	chr1	25498175	G/T	I	$5.3 \times 10^{-11}$	0.832	[0.787,0.879]	RUNX3---[---SYF2
6q22.32	rs1262557	chr6	127054588	C/T	I	$4.8 \times 10^{-10}$	1.150	[1.100,1.202]	CENPW---[---RSPO3
11p11.2	rs11037975	chr11	44410862	C/G	I	$6.3 \times 10^{-10}$	0.854	[0.812,0.898]	ALX4--[---CD82
4q25	rs78311490	chr4	107883049	A/G	I	$6.7 \times 10^{-10}$	0.757	[0.692,0.827]	[DKK2]
1q24.2	rs78003935	chr1	170341522	A/T	I	$9.9 \times 10^{-10}$	0.861	[0.820,0.903]	METTL11B---[---GORAB
15q23	rs7177657	chr15	70041693	C/T	I	$1.0 \times 10^{-9}$	0.793	[0.736,0.855]	RPLP1---[---TLE3
13q12.3	rs9314998	chr13	30746969	A/G	I	$1.5 \times 10^{-9}$	1.146	[1.097,1.198]	UBL3---[---KATNAL1
12q13.12	rs7974517	chr12	51145082	A/T	I	$2.0 \times 10^{-9}$	0.868	[0.829,0.909]	DIP2B--[---ATF1
6p21.1	rs227808	chr6	44666915	C/T	I	$2.9 \times 10^{-9}$	0.846	[0.800,0.894]	CDC5L---[---SUPT3H
3q23	rs7642536	chr3	139032333	C/T	I	$3.1 \times 10^{-9}$	0.807	[0.751,0.866]	PRR23C---[---MRPS22
7p12.3	rs12702271	chr7	46943759	C/T	I	$3.2 \times 10^{-9}$	1.171	[1.111,1.233]	IGFBP3---[---TNS3
4q21.21	rs4690116	chr4	81206377	A/T	I	$5.5 \times 10^{-9}$	0.876	[0.838,0.916]	[FGF5]
16p13.12	rs246180	chr16	14391923	A/C	I	$8.0 \times 10^{-9}$	1.152	[1.098,1.209]	MKL2--[---PARN
2q12.3	rs3827760	chr2	109513601	A/G	I	$1.3 \times 10^{-8}$	0.447	[0.335,0.595]	[EDAR]
2p21	rs11694173	chr2	43590899	A/G	I	$2.2 \times 10^{-8}$	0.852	[0.805,0.901]	[THADA]
12q24.33	rs76972608	chr12	130563363	A/T	I	$3.6 \times 10^{-8}$	0.834	[0.781,0.889]	TMEM132D---[---FZD10
2q31.1	rs1819008	chr2	177697882	C/T	I	$3.7 \times 10^{-8}$	0.884	[0.846,0.924]	MTX2---[---HNRNPA3
Xp22.31	rs5933688	chrX	8880680	A/G	I	$3.7 \times 10^{-8}$	1.102	[1.065,1.141]	FAM9A---[---FAM9B
2q31.1	rs13405699	chr2	174605633	A/C	I	$4.7 \times 10^{-8}$	0.672	[0.582,0.776]	CDCA7---[---SP3

## Quality Statistics for Index SNPs

assay.name	is.v2	is.v3	is.v4	gt.rate	hw.p.value	p.date	freq.b	avg.rsqr	min.rsqr	p.batch	dose.b	qc.mask
rs200644307	FALSE	FALSE	FALSE					0.9106	0.8966	3.9×10 <sup>-5</sup>	0.8400	v2v3v4
rs201563	FALSE	FALSE	FALSE					0.9882	0.9596	0.0029	0.5423	v2v3v4
rs11684254	FALSE	FALSE	FALSE					0.9938	0.9899	0.26	0.3402	v2v3v4
rs7801037	FALSE	FALSE	FALSE					0.9915	0.9845	0.033	0.5851	v2v3v4
rs2095921	FALSE	FALSE	FALSE					0.9525	0.9394	0.36	0.2293	v2v3v4
rs34991987	FALSE	FALSE	FALSE					0.9978	0.9890	0.92	0.4602	v2v3v4
rs62385385	FALSE	FALSE	FALSE					0.9967	0.9933	0.75	0.6070	v2v3v4
rs111763724	FALSE	FALSE	FALSE					0.8177	0.7634	0.00023	0.0256	v2v3v4
rs201408539	FALSE	FALSE	FALSE					0.8363	0.8254	0.33	0.8170	v2v3v4
rs4679956	FALSE	FALSE	FALSE					0.9965	0.9863	0.68	0.4109	v2v3v4
rs143149578	FALSE	FALSE	FALSE					0.7889	0.7642	1.2×10 <sup>-5</sup>	0.0208	v2v3v4
rs74333950	FALSE	FALSE	FALSE					0.9812	0.9630	0.037	0.8628	v2v3v4
rs9282858	FALSE	TRUE	TRUE	0.9998	0.032	0.45	0.0325	0.9735	0.7250	0.51	0.0321	v2v3v4
rs34800162	FALSE	FALSE	FALSE					0.9963	0.9946	0.11	0.7284	v2v3v4
rs12083887	FALSE	FALSE	FALSE					0.9961	0.9945	0.25	0.6052	v2v3v4
rs9357047	FALSE	FALSE	FALSE					0.9977	0.9967	0.15	0.5787	v2v3v4
rs79437808	FALSE	FALSE	FALSE					0.9273	0.7854	0.76	0.0116	v2v3v4
rs185597083	FALSE	FALSE	FALSE					0.7349	0.7007	5.5×10 <sup>-6</sup>	0.9587	v2v3v4
rs3781458	FALSE	TRUE	FALSE	0.9951	0.00064	0.14	0.6178	0.9980	0.9906	0.0013	0.6169	v2v3v4
rs62060349	FALSE	FALSE	FALSE					0.9938	0.9916	0.50	0.5467	v2v3v4
rs11593840	FALSE	FALSE	FALSE					0.9973	0.9914	7.2×10 <sup>-7</sup>	0.4148	v2v3v4
rs9300169	FALSE	FALSE	FALSE					0.9952	0.9710	0.54	0.7337	v2v3v4
rs6546334	FALSE	FALSE	FALSE					0.9945	0.9873	0.077	0.3509	v2v3v4
rs12203592	TRUE	TRUE	TRUE	1.0000	9.7×10 <sup>-37</sup>	0.014	0.1728	0.9923	0.9819	4.2×10 <sup>-6</sup>	0.1705	v2v3v4
rs68088846	FALSE	FALSE	FALSE					0.9074	0.8766	0.18	0.7914	v2v3v4
rs7534070	FALSE	FALSE	FALSE					0.6340	0.5370	1.4×10 <sup>-10</sup>	0.6458	v2v3v4
rs1262557	FALSE	FALSE	FALSE					0.9888	0.9871	0.0017	0.4688	v2v3v4
rs11037975	FALSE	FALSE	FALSE					0.9094	0.8896	0.047	0.3036	v2v3v4
rs78311490	FALSE	FALSE	FALSE					0.9500	0.8998	0.026	0.0713	v2v3v4
rs78003935	FALSE	FALSE	FALSE					0.9318	0.9242	0.0079	0.6752	v2v3v4
rs7177657	FALSE	FALSE	FALSE					0.9933	0.9895	0.041	0.9074	v2v3v4
rs9314998	FALSE	FALSE	FALSE					0.9943	0.9930	0.0043	0.5683	v2v3v4
rs7974517	FALSE	FALSE	FALSE					0.9629	0.9491	0.53	0.6201	v2v3v4
rs227808	TRUE	TRUE	TRUE	0.9974	0.023	0.83	0.1963	0.9988	0.9968	0.72	0.1956	v2v3v4
rs7642536	FALSE	FALSE	FALSE					0.8209	0.7172	0.19	0.8708	v2v3v4
rs12702271	FALSE	FALSE	FALSE					0.9689	0.9527	0.39	0.2402	v2v3v4
rs4690116	FALSE	FALSE	FALSE					0.9964	0.9911	0.52	0.5776	v2v3v4
rs246180	FALSE	FALSE	FALSE					0.9068	0.6514	0.046	0.6706	v2v3v4
rs3827760	TRUE	TRUE	TRUE	1.0000	0.13	0.0089	0.0077	0.9883	0.9711	0.013	0.0081	v2v3v4
rs11694173	FALSE	TRUE	FALSE	0.9995	0.48	0.52	0.8115	0.9993	0.9973	0.26	0.8129	v2v3v4
rs76972608	FALSE	FALSE	FALSE					0.9852	0.9606	0.57	0.1378	v2v3v4
rs1819008	FALSE	FALSE	FALSE					0.9946	0.9877	0.14	0.5413	v2v3v4
rs5933688	FALSE	FALSE	FALSE					0.9954	0.9899	9.6×10 <sup>-5</sup>	0.2853	v2v3v4
rs13405699	FALSE	FALSE	FALSE					0.6476	0.4863	1.5×10 <sup>-13</sup>	0.9631	v2v3v4

## SNP Statistics in the GWAS Sample

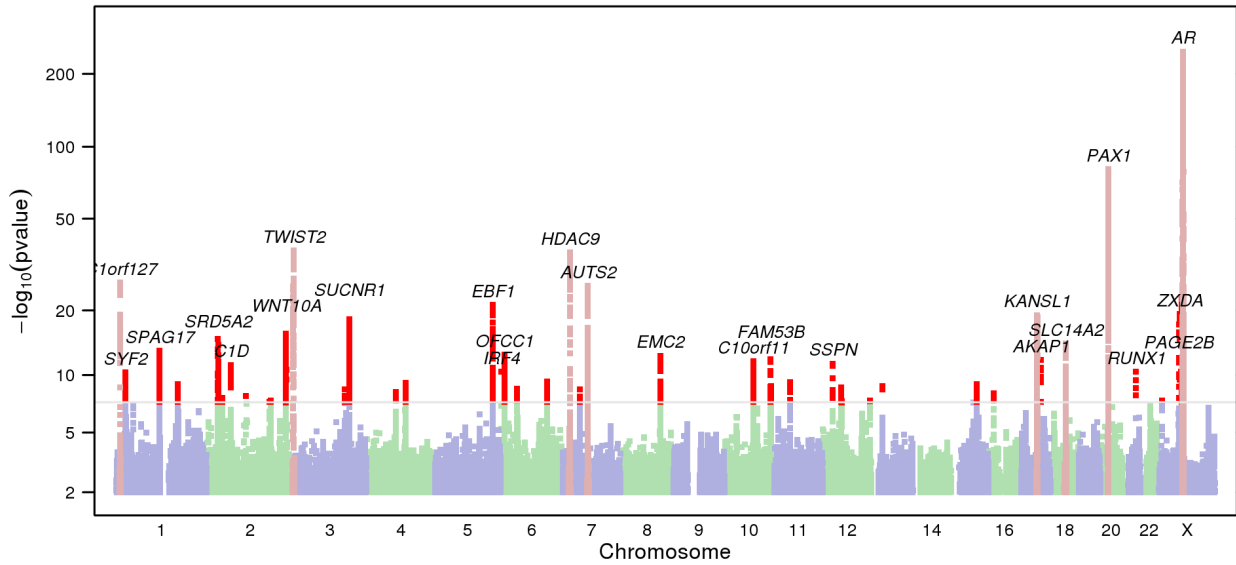
assay.name	AA.0	AB.0	BB.0	im.num.0	dose.b.0	AA.1	AB.1	BB.1	im.num.1	dose.b.1
rs200644307				8491	0.7467				9009	0.9246
rs201563				8491	0.5067				9009	0.6077
rs11684254				8491	0.3132				9009	0.3803
rs7801037				8491	0.6136				9009	0.5437
rs2095921				8491	0.2515				9009	0.2046
rs34991987				8491	0.4357				9009	0.4948
rs62385385				8491	0.5843				9009	0.6362
rs111763724				8491	0.0363				9009	0.0164
rs201408539				8491	0.8007				9009	0.8353
rs4679956				8491	0.3904				9009	0.4376
rs143149578				8491	0.0308				9009	0.0138
rs74333950				8491	0.8767				9009	0.8462
rs9282858	6173	521	25	8491	0.0422	6881	375	0	9009	0.0259
rs34800162				8491	0.7136				9009	0.7497
rs12083887				8491	0.6251				9009	0.5853
rs9357047				8491	0.5895				9009	0.5520
rs79437808				8491	0.0087				9009	0.0171
rs185597083				8491	0.9473				9009	0.9663
rs3781458	795	2899	2537	8491	0.6378	1128	3110	2428	9009	0.5963
rs62060349				8491	0.5636				9009	0.5237
rs11593840				8491	0.4402				9009	0.3997
rs9300169				8491	0.7532				9009	0.7197
rs6546334				8491	0.3718				9009	0.3353
rs12203592	5970	2289	232	8491	0.1627	5948	2695	366	9009	0.1905
rs68088846				8491	0.8031				9009	0.7749
rs7534070				8491	0.6576				9009	0.6275
rs1262557				8491	0.4552				9009	0.4861
rs11037975				8491	0.3138				9009	0.2849
rs78311490				8491	0.0785				9009	0.0620
rs78003935				8491	0.6874				9009	0.6581
rs7177657				8491	0.9133				9009	0.8940
rs9314998				8491	0.5542				9009	0.5872
rs7974517				8491	0.6344				9009	0.6052
rs227808	5299	2812	374	8491	0.2086	6009	2696	299	9009	0.1822
rs7642536				8491	0.8799				9009	0.8593
rs12702271				8491	0.2273				9009	0.2548
rs4690116				8491	0.5927				9009	0.5624
rs246180				8491	0.6602				9009	0.6908
rs3827760	8299	163	0	8491	0.0097	8910	76	0	9009	0.0043
rs11694173	182	1824	4239	8491	0.8226	280	2127	4274	9009	0.7980
rs76972608				8491	0.1458				9009	0.1247
rs1819008				8491	0.5508				9009	0.5204
rs5933688				8491	0.2587				9009	0.3011

## Annotations from NHGRI GWAS Catalog

The following table shows, for each index SNP, all entries in the NHGRI GWAS Catalog that are within 500kb and in at least moderate linkage disequilibrium ( $r^2 > 0.5$ ).

region	position	our.name	our.pval	dist	rsqr	assay.name	pvalue	pubmed.id	trait	genes
Xq12	66300914	rs200644307	$1.0 \times 10^{-247}$	210070	0.684	rs6625163	$5.0 \times 10^{-11}$	18849991	Male-pattern baldness	AR
Xq12	66300914	rs200644307	$1.0 \times 10^{-247}$	262104	0.625	rs2497938	$2.0 \times 10^{-91}$	22693459	Male-pattern baldness	AR
Xq12	66300914	rs200644307	$1.0 \times 10^{-247}$	262104	0.625	rs2497938	$3.0 \times 10^{-22}$	22032556	Male-pattern baldness	AR, EDA2R
20p11.22	22000281	rs201563	$2.7 \times 10^{-81}$	-147181	0.602	rs2180439	$4.0 \times 10^{-17}$	22032556	Male-pattern baldness	Intergenic
20p11.22	22000281	rs201563	$2.7 \times 10^{-81}$	-147181	0.602	rs2180439	$3.0 \times 10^{-15}$	18849994	Male-pattern baldness	PAX1, BQ013595, BE789145
20p11.22	22000281	rs201563	$2.7 \times 10^{-81}$	37294	0.995	rs6047844	$2.0 \times 10^{-39}$	22693459	Male-pattern baldness	PAX1, FOXA2
20p11.22	22000281	rs201563	$2.7 \times 10^{-81}$	50222	0.902	rs1160312	$1.0 \times 10^{-14}$	18849991	Male-pattern baldness	PAX1
2q37.3	239695893	rs11684254	$2.3 \times 10^{-37}$	-1262	0.890	rs9287638	$1.0 \times 10^{-12}$	22693459	Male-pattern baldness	HDAC4
7p21.1	18897511	rs7801037	$1.3 \times 10^{-36}$	-19637	0.663	rs2073963	$1.0 \times 10^{-12}$	22693459	Male-pattern baldness	HDAC9
1p36.22	11033322	rs2095921	$3.1 \times 10^{-27}$	-240	0.915	rs12565727	$9.0 \times 10^{-11}$	22693459	Male-pattern baldness	TARDBP
1p36.22	11033322	rs2095921	$3.1 \times 10^{-27}$	13533	0.628	rs9430161	$1.0 \times 10^{-20}$	22327514	Ewing sarcoma	TARDBP
7q11.22	68595226	rs34991987	$2.3 \times 10^{-26}$	16734	0.868	rs6945541	$2.0 \times 10^{-9}$	22693459	Male-pattern baldness	AUTS2
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-451228	0.702	rs2942168	$1.0 \times 10^{-28}$	21292315	Parkinson's disease	MAPT
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-446935	0.697	rs393152	$2.0 \times 10^{-16}$	19915575	Parkinson's disease	MAPT, C17orf69, ...
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-242395	0.702	rs12185268	$3.0 \times 10^{-14}$	21738487	Parkinson's disease	MAPT
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-241859	0.693	rs12373124	$5.0 \times 10^{-10}$	22693459	Male-pattern baldness	Intergenic
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-188251	0.702	rs1864325	$5.0 \times 10^{-11}$	22504420	Bone mineral density	MAPT
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-109311	0.702	rs1981997	$9.0 \times 10^{-14}$	23583980	Interstitial lung disease	MAPT
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-85014	0.693	rs8070723	$2.0 \times 10^{-118}$	21685912	Progressive supranuclear palsy	MAPT
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-85014	0.693	rs8070723	$7.0 \times 10^{-12}$	21044948	Parkinson's disease	MAPT
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	21179	0.682	rs9303525	$8.0 \times 10^{-15}$	22504418	Intracranial volume	MAPT, GRN, ...
2q35	219746292	rs74333950	$2.2 \times 10^{-16}$	10091	0.975	rs7349332	$3.0 \times 10^{-14}$	20585627	Common traits (Other)	WNT10A
2q35	219746292	rs74333950	$2.2 \times 10^{-16}$	10091	0.975	rs7349332	$1.0 \times 10^{-6}$	19896111	Hair morphology	WNT10A
18q12.3	42808059	rs34800162	$8.2 \times 10^{-15}$	-7911	1.000	rs10502861	$3.0 \times 10^{-9}$	22693459	Male-pattern baldness	SETBP1
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$3.0 \times 10^{-23}$	23548203	Tanning	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$2.0 \times 10^{-6}$	23548203	Sunburns	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$7.0 \times 10^{-14}$	23548203	Non-melanoma skin cancer	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$1.0 \times 10^{-28}$	23548203	Hair color	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$6.0 \times 10^{-15}$	21685912	Progressive supranuclear palsy	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$4.0 \times 10^{-7}$	20585627	Hair color	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$2.0 \times 10^{-91}$	20585627	Freckling	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$2.0 \times 10^{-15}$	20585627	Eye color	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$9.0 \times 10^{-28}$	18483556	Black vs. red hair color	IRF4
6p25.3	396321	rs12203592	$4.0 \times 10^{-11}$	0	1.000	rs12203592	$7.0 \times 10^{-127}$	18483556	Black vs. blond hair color	IRF4
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-355869	0.686	rs9388489	$4.0 \times 10^{-13}$	19430480	Type 1 diabetes	C6orf173
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-286988	0.698	rs1361108	$9.0 \times 10^{-6}$	21998595	Height	Intergenic
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-286988	0.698	rs1361108	$2.0 \times 10^{-8}$	21102462	Menarche (age at onset)	C6orf173, TRMT11
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-218933	0.694	rs1490388	$6.0 \times 10^{-7}$	18391951	Height	C6orf173
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-203428	0.802	rs1490384	$1.0 \times 10^{-16}$	23563607	Height	C6orf173
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-203428	0.802	rs1490384	$4.0 \times 10^{-21}$	20881960	Height	C6orf173
6q22.32	127054588	rs1262557	$4.8 \times 10^{-10}$	-88280	0.875	rs4549631	$5.0 \times 10^{-13}$	18391952	Height	LOC387103
15q23	70041693	rs7177657	$1.0 \times 10^{-9}$	6464	1.000	rs10152591	$3.0 \times 10^{-10}$	20881960	Height	TLE3
16p13.12	14391923	rs246180	$8.0 \times 10^{-9}$	-3618	0.916	rs1659127	$4.0 \times 10^{-9}$	21102462	Menarche (age at onset)	MKL2
16p13.12	14391923	rs246180	$8.0 \times 10^{-9}$	-3618	0.916	rs1659127	$1.0 \times 10^{-11}$	20881960	Height	MKL2
Xp22.31	8880680	rs5933688	$3.7 \times 10^{-8}$	33146	0.948	rs5934505	$2.0 \times 10^{-8}$	22936694	Androgen levels	FAM9B

## Replication of GWAS Catalog Results



The following table shows, for each GWAS Catalog result for similar traits, our association test result for our best available proxy (distance < 100kb,  $r^2 > 0.8$ ).

region	position	our.name	our.pval	dist	rsqr	assay.name	pvalue	pubmed.id	trait	genes
1p36.22	11033082	rs12565727	$9.9 \times 10^{-27}$	0	1.000	rs12565727	$9.0 \times 10^{-11}$	22693459	Male-pattern baldness	TARDBP
2q37.3	239694631	rs9287638	$4.7 \times 10^{-35}$	0	1.000	rs9287638	$1.0 \times 10^{-12}$	22693459	Male-pattern baldness	HDAC4
7p21.1	18877874	rs2073963	$1.2 \times 10^{-26}$	0	1.000	rs2073963	$1.0 \times 10^{-12}$	22693459	Male-pattern baldness	HDAC9
7q11.22	68611960	rs6945541	$4.1 \times 10^{-22}$	0	1.000	rs6945541	$2.0 \times 10^{-9}$	22693459	Male-pattern baldness	AUTS2
17q21.31	43924219	rs12373124	$3.4 \times 10^{-18}$	0	1.000	rs12373124	$5.0 \times 10^{-10}$	22693459	Male-pattern baldness	Intergenic
18q12.3	42800148	rs10502861	$1.0 \times 10^{-14}$	0	1.000	rs10502861	$3.0 \times 10^{-9}$	22693459	Male-pattern baldness	SETBP1
20p11.22	21853100	rs2180439	$3.2 \times 10^{-78}$	0	1.000	rs2180439	$4.0 \times 10^{-17}$	22032556	Male-pattern baldness	Intergenic
20p11.22	21853100	rs2180439	$3.2 \times 10^{-78}$	0	1.000	rs2180439	$3.0 \times 10^{-15}$	18849994	Male-pattern baldness	PAX1, BQ013595, BE789145
20p11.22	22037575	rs6047844	$9.3 \times 10^{-81}$	0	1.000	rs6047844	$2.0 \times 10^{-39}$	22693459	Male-pattern baldness	PAX1, FOXA2
20p11.22	22050503	rs1160312	$2.8 \times 10^{-75}$	0	1.000	rs1160312	$1.0 \times 10^{-14}$	18849991	Male-pattern baldness	PAX1
Xq12	66510984	rs6625163	$2.5 \times 10^{-232}$	0	1.000	rs6625163	$5.0 \times 10^{-11}$	18849991	Male-pattern baldness	AR
Xq12	66563018	rs2497938	$7.0 \times 10^{-229}$	0	1.000	rs2497938	$2.0 \times 10^{-91}$	22693459	Male-pattern baldness	AR
Xq12	66563018	rs2497938	$7.0 \times 10^{-229}$	0	1.000	rs2497938	$3.0 \times 10^{-22}$	22032556	Male-pattern baldness	AR, EDA2R

## Nearby Nonsynonymous SNPs

region	position	our.name	our.pval	dist	rsqr	assay.name	gene	aa.chg
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-243136	0.702	rs62621252	SPPL2C	S224P
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-242812	0.702	rs62054815	SPPL2C	A332T
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-242424	0.702	rs12185233	SPPL2C	R461P
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-242395	0.702	rs12185268	SPPL2C	I471V
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-242005	0.702	rs12373123	SPPL2C	S601P
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-241948	0.702	rs12373139	SPPL2C	G620R
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-241878	0.693	rs12373142	SPPL2C	P643R
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-241859	0.693	rs12373124	SPPL2C	H649H
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-241847	0.693	rs12373140	SPPL2C	Q653Q
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-105303	0.702	rs63750417	MAPT	P202L
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-105055	0.702	rs62063786	MAPT	D285N
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-105042	0.702	rs62063787	MAPT	V289A
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-104800	0.702	rs17651549	MAPT	R370W
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-98678	0.702	rs10445337	MAPT	S447P
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-61667	0.696	rs199706121	MAPT	?1735?
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-57592	0.570	rs200384907	KANSL1	?1225?
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-57172	0.702	rs34579536	KANSL1	I1085T
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-55546	0.702	rs36076725	KANSL1	F917F
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-55537	0.702	rs35833914	KANSL1	D914D
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-48959	0.702	rs34043286	KANSL1	S718P
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	82759	0.568	rs35643216	KANSL1	N225D
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	83121	0.568	rs17585974	KANSL1	K104T
2p23.1	31805826	rs9282858	$1.5 \times 10^{-15}$	0	1.000	rs9282858	SRD5A2	A48T
4q25	107883049	rs78311490	$6.7 \times 10^{-10}$	-36006	0.503	rs35290077	DKK2	G96R
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-417376	0.611	rs6580741	FAM186A	H2228Q
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-400963	0.614	rs7296291	FAM186A	H2166Y
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-398165	0.610	rs10876023	FAM186A	L1233P
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-390519	0.614	rs12303082	FAM186A	K187Q
2q12.3	109513601	rs3827760	$1.3 \times 10^{-8}$	0	1.000	rs3827760	EDAR	V370A

## Nearby Expression QTLs

region	position	our.name	our.pval	dist	rsqr	assay.name	eql.dist	eql.gene	eql.pval	eql.rsqr	tissue	pubmed.id
20p11.22	22000281	rs201563	$2.7 \times 10^{-81}$	76273	0.617	rs804531	-779453	XRN2	0.00012		Lymphoblastoid	20220756
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-446935	0.697	rs393152	580720	NMT1	0.00045	0.156	Lymphoblastoid	19644074
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	-414165	0.605	rs1635291	175935	HS.554608	0.00060	0.041	B-Cell	22446964
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	168210	0.641	rs142039218	666195	GOSR2	$7.4 \times 10^{-13}$	0.130	Lymphoblastoid	24037378
17q21.31	44166078	rs201408539	$8.4 \times 10^{-20}$	180592	0.557	rs139480590	647403	CRHR1	$2.4 \times 10^{-11}$	0.113	Lymphoblastoid	24037378
2q35	219746292	rs74333950	$2.2 \times 10^{-16}$	20380	0.755	rs10932789	636628	ACCN4	$9.0 \times 10^{-5}$	0.054	Monocyte	22446964
17q22	55231168	rs62060349	$1.5 \times 10^{-12}$	-3498	0.812	rs8081915	821240	VEZF1	$4.6 \times 10^{-5}$	0.204	Fibroblast	19644074
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-439367	0.557	rs6580738	28425	LIMA1	$2.6 \times 10^{-19}$	0.056	Monocyte	20502693
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-406074	0.557	rs4768951	-215091	LASS5	$2.5 \times 10^{-9}$	0.119	Monocyte	22446964
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-406074	0.557	rs4768951	-215091	LASS5	$1.2 \times 10^{-9}$	0.124	B-Cell	22446964
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-390519	0.614	rs12303082	-184541	LIMA1	$9.1 \times 10^{-12}$	0.154	Monocyte	22446964
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-205275	0.749	rs7487429	378662	LASS5	$2.3 \times 10^{-22}$	0.065	Monocyte	20502693
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-71559	0.610	rs11169520	68656	DIP2B	$5.2 \times 10^{-9}$	0.117	Monocyte	22446964
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-70209	0.610	rs7955736	0	DIP2B	$5.1 \times 10^{-26}$	0.075	Monocyte	20502693
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-58151	0.765	rs2090852	-731724	LOC643435	0.00011	0.053	Monocyte	22446964
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	-6220	0.581	rs1047912	240094	DIP2B	$8.4 \times 10^{-24}$	0.239	Lymphoblastoid	24037378
12q13.12	51145082	rs7974517	$2.0 \times 10^{-9}$	35061	0.961	rs10783387	145977	METTL7A	0.00057	0.042	B-Cell	22446964
4q21.21	81206377	rs4690116	$5.5 \times 10^{-9}$	-5732	0.962	rs6827834	773916	BMP3	0.0014	0.036	B-Cell	22446964
2p21	43590899	rs11694173	$2.2 \times 10^{-8}$	0	1.000	rs11694173	-139827	ZFP36L2	0.00034	0.045	Monocyte	22446964

## Nearby Clinical Variants

source	region	our.name	our.pval	dist	rsqr	assay.name	gene	phenotype	accession
clinvar	2p23.1	rs9282858	$1.5 \times 10^{-15}$	0	1.000	rs9282858	SRD5A2	3-Oxo-5 alpha-steroid delta 4-dehydrogenase deficiency	SNOMED CT57514000
clinvar	2q12.3	rs3827760	$1.3 \times 10^{-8}$	0	1.000	rs3827760	EDAR	Autosomal recessive hypohidrotic ectodermal dysplasia syndrome	SNOMED CT27025001
clinvar	2q12.3	rs3827760	$1.3 \times 10^{-8}$	0	1.000	rs3827760	EDAR	Autosomal dominant hypohidrotic ectodermal dysplasia	NCBI curation
clinvar	2q12.3	rs3827760	$1.3 \times 10^{-8}$	0	1.000	rs3827760	EDAR	Hair morphology 1	NCBI curation

## Regional Association Plots

