CHAPTER 8

Appendix.

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Table 8-1. Genetic loci selected for the high-density DNA tiling array. The array contained (A) genetic loci associated with haematological and cardiovascular-related traits, and (B) lineage-specific reference genes. The criteria for selection are described in **Section 2.4**. Genomic coordinates were based on the human reference genome, build hg18 (NCBI build 36). Abbreviations: CAD: coronary artery disease; MI: myocardial infarction; MPV: mean platelet volume; PLT: platelet count; PLS: platelet signalling; WBC: white blood cell count; RBC: red blood cell count; MCV: mean corpuscular volume of erythrocytes; MCH: mean corpuscular haemoglobin; SBP: systolic blood pressure; DBP: diastolic blood pressure; HYP: hypertension. Key: [a]target gene ± 10 kb; [b]biological evidence for association.

(A) Association loci									
#	Gene(s)	GWA index SNP(s)		Trait(s)	Reference(s)	Genomic position			Interval (kb)
		ID	*Position*			*Chromosome*	*Start*	*End*	
1	*PCSK9*	rs11206510	55,268,627	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009	1p32.3	55,240,000	55,300,000	60.00
2	*VAV3*[b]	rs17229705	107,940,484	PLS	Jones et al., 2009	1p13.3	107,890,000	108,055,000	165.00
3	*CELSR2, PSRC1, SORT1*	rs599839	109,623,689	CAD	Samani et al., 2007; Coronary Artery Disease Consortium, 2009	1p13.3	109,489,481	109,739,481	250.00
		rs646776	109,620,053	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009				
4	*PEAR1*[b]	rs3737224	155,146,204	PLS	Jones et al., 2009	1q23.1	155,100,000	155,250,000	150.00
5	*FCER1G*[a,b]	rs3557	159,455,517	PLS	Jones et al., 2009	1q23.3	159,441,711	159,465,662	23.95
6	*DNM3*	rs10914144	170,216,373	MPV	Soranzo, Spector, et al., 2009	1q24.3	170,130,000	170,390,000	260.00
7	*TMCC2*	rs1668873	203,502,613	MPV	Soranzo, Spector, et al., 2009	1q32.1	203,445,000	203,540,000	95.00
8	*MIA3*	rs17465637	220,890,152	CAD/early-onset MI	Samani et al., 2007; Myocardial Infarction Genetics Consortium, 2009	1q41	220,678,228	221,078,228	400.00
		rs3008621	220,870,669	CAD	Coronary Artery Disease Consortium, 2009				
9	*EHD3*	rs647316	31,318,333	MPV	Soranzo, Spector, et al., 2009	2p23.1	31,305,000	31,346,000	41.00
10	*STK39*[b]	rs6749447	168,749,632	SBP/DBP	Wang et al., 2009	2q24.3	168,695,000	168,800,000	105.00
		rs3754777	168,724,160						
11	*WDR12*[a]	rs6725887	203,454,130	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009	2q33.1	203,443,575	203,494,639	51.06
12	*gene desert*	rs2943634	226,776,324	CAD	Samani et al., 2007; Coronary Artery Disease Consortium, 2009	2q36.3	226,532,739	226,982,739	450.00
13	*ITPR1*[b]	rs17786144	4,804,575	PLS	Jones et al., 2009	3p26.2	4,760,000	4,860,000	100.00
14	*RAF1*[a,b]	rs3729931	12,601,516	PLS	Jones et al., 2009	3p25.1	12,590,108	12,690,678	100.57

15	*ULK4*	rs9815354	41,887,655	DBP	Newton-Cheh et al., 2009; Levy et al., 2009	3p22.1	41,710,000	41,970,000	260.00
16	*ARHGEF3*	rs12485738	56,840,816	MPV	Soranzo, Spector, et al., 2009; Meisinger et al., 2009	3p14.3	56,750,000	56,880,000	130.00
17	*MRAS*	rs9818870	139,604,812	CAD/MI	Erdmann et al., 2009	3q22.3	139,550,000	139,613,000	63.00
18	*P2RY12*[b]	rs1472122	152,517,292	PLS	Jones et al., 2009	3q25.1	152,410,000	152,580,000	170.00
19	*ITGA2*[b]	rs41305896	52,311,475	PLS	Jones et al., 2009	5q11.2	52,267,000	52,436,366	169.37
20	*PHACTR1*	rs12526453	13,035,530	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009	6p24.1	12,985,000	13,060,000	75.00
21	*HFE*	rs1800562	26,201,120	MCV	Soranzo, Spector, et al., 2009	6p22.1	26,190,000	26,260,000	70.00
22	*BAK1*	rs210135	33,648,670	PLT	Soranzo, Spector, et al., 2009	6p21.31	33,644,000	33,665,000	21.00
23	*MAPK14*[b]	rs851007	36,172,014	PLS	Jones et al., 2009	6p21.31	36,050,000	36,210,000	160.00
24	*BSYL, CCND3*	rs11970772	42,033,268	MCV	Soranzo, Spector, et al., 2009	6p21.1	41,985,000	42,105,000	120.00
25	*HBS1L, MYB*	rs9402686	135,469,510	MCV	Soranzo, Spector, et al., 2009	6q23.3	135,400,000	135,582,003	182.00
26	*MTHFD1L*	rs6922269	151,294,678	CAD	The Wellcome Trust Case Control Consortium, 2007; Samani et al., 2007; Coronary Artery Disease Consortium, 2009	6q25.1	151,199,579	151,399,579	200.00
27	*SLC22A3, LPAL2, LPA*	rs2048327-rs3127599-rs7767084-rs10755578 haplotype		CAD	Trégouët et al., 2009	6q25.3–q26	160,780,000	161,055,000	275.00
28	*CD36*[b]	rs1049654	80,113,391	PLS	Jones et al., 2009	7q21.11	80,045,000	80,137,000	92.00
29	*TFR2*	rs7385804	100,073,906	RBC	Soranzo, Spector, et al., 2009	7q22.1	100,050,000	100,100,000	50.00
30	*FLJ36031, PIK3CG*	rs342293	106,159,455	MPV	Soranzo, Spector, et al., 2009; Soranzo, Rendon, et al., 2009	7q22.3	106,085,000	106,190,000	105.00
31	*AK3, RCL1, JAK2* (b, for PLS)	rs385893	4,753,176	PLT	Soranzo, Spector, et al., 2009	9p24.1	4,730,000	5,050,000	320.00
		rs10429491	5,040,706	PLS	Jones et al., 2009				
32	*CDKN2A, CDKN2B*	rs1333049	22,115,503	CAD/MI	The Wellcome Trust Case Control Consortium, 2007; Samani et al., 2007; Schunkert et al., 2008; Coronary Artery Disease Consortium, 2009; Myocardial Infarction Genetics Consortium, 2009	9p21.3	21,900,000	22,200,000	300.00
		rs4977574	22,088,574	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009				
33	*CACNB2*	rs11014166	18,748,804	DBP	Newton-Cheh et al., 2009; Levy et al., 2009	10p12.33	18,570,000	18,840,000	270.00

34	*CXCL12/SDF1*	rs501120	44,073,873	CAD	Samani et al., 2007; Coronary Artery Disease Consortium, 2009	10q11.21	43,950,000	44,200,000	250.00
		rs1746048	44,095,830	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009				
35	*JMJD1C*[a]	rs2393967	64,803,162	MPV	Soranzo, Spector, et al., 2009	10q21.2–q21.3	64,586,991	64,905,728	318.74
36	*CYP17A1, C10orf32*	rs1004467	104,584,497	SBP	Newton-Cheh et al., 2009; Levy et al., 2009	10q24.32	104,520,000	104,690,000	170.00
37	*BET1L, SIRT3, PSMD13*	rs11602954	192,856	MPV	Soranzo, Spector, et al., 2009	11p15.5	180,000	240,000	60.00
38	*PLEKHA7*	rs381815	16,858,844	SBP	Newton-Cheh et al., 2009; Levy et al., 2009	11p15.1	16,795,000	16,960,000	165.00
39	*ATP2B1*	rs2681492	88,537,220	SBP	Newton-Cheh et al., 2009; Levy et al., 2009	12q21.33	88,460,000	88,650,000	190.00
		rs2681472	88,533,090	HYP/DBP	Newton-Cheh et al., 2009; Levy et al., 2009				
40	*SH2B3/LNK, ATXN2*	rs3184504	110,368,991	MI; SBP/DBP	Gudbjartsson et al., 2009; Newton-Cheh et al., 2009; Levy et al., 2009	12q24.12	110,310,000	110,570,000	260.00
		rs11065987	110,556,807	PLT; CAD/MI	Soranzo, Spector, et al., 2009				
41	*PTPN11*[a]	rs11066301	111,355,755	PLT; CAD/MI	Soranzo, Spector, et al., 2009	12q24.13	111,330,919	111,442,100	111.18
42	*TBX3, TBX5*	rs2384550	113,837,114	DBP	Newton-Cheh et al., 2009; Levy et al., 2009	12q24.21	113,810,000	113,930,000	120.00
43	*HNF1A/TCF1, C12orf43*	rs2259816	119,919,970	CAD/MI	Erdmann et al., 2009	12q24.31	119,850,000	119,980,000	130.00
44	*WDR66*	rs7961894	120,849,966	MPV	Meisinger et al., 2009; Soranzo, Spector, et al., 2009	12q24.31	120,760,000	120,970,000	210.00
45	*TPM1*	rs11071720	61,129,049	MPV	Soranzo, Spector, et al., 2009	15q22.2	61,080,000	61,160,000	80.00
46	*SMAD3*[b]	rs17228212	65,245,693	CAD	Samani et al., 2007	15q22.33–q23	65,150,000	65,350,000	200.00
47	*CSK, ULK3*	rs6495122	72,912,698	DBP	Newton-Cheh et al., 2009; Levy et al., 2009	15q24.1	72,810,000	73,030,000	220.00
48	*CDH13*	rs11646213	81,200,152	HYP/SBP/DBP	Org et al., 2009	16q23.3	81,190,000	81,240,000	50.00
49	*MAP2K4*[a,b]	rs41307923	11,968,409	PLS	Jones et al., 2009	17p12	11,854,860	11,997,776	142.92
50	*TAOK1*	rs2138852	24,727,475	MPV	Meisinger et al., 2009; Soranzo, Spector, et al., 2009	17q11.2	24,680,000	24,930,000	250.00
51	*GSDMA, ORMDL3*	rs17609240	35,364,215	WBC	Soranzo, Spector, et al., 2009	17q12	35,314,374	35,387,545	73.17
52	*CD226*	rs893001	65,667,825	MPV	Soranzo, Spector, et al., 2009	18q22.2	65,620,000	65,730,000	110.00
53	*MAP2K2*[b]	rs350916	4,045,775	PLS	Jones et al., 2009	19p13.3	4,025,000	4,095,000	70.00
54	*LDLR*	rs1122608	11,024,601	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009	19p13.2	11,010,000	11,120,000	110.00
55	*AKT2*[b]	rs41275750	45,429,935	PLS	Jones et al., 2009	19q13.2	45,380,000	45,520,000	140.00
56	*APOC1, APOC4, APOE*[b]	rs4420638	50,114,786	CAD	The Wellcome Trust Case Control Consortium, 2007	19q13.32	50,000,000	50,230,000	230.00
57	*GP6*[a,b]	rs1613662	60,228,407	PLS	Jones et al., 2009	19q13.42	60,206,885	60,251,444	44.56
58	*SIRPA*	rs6136489	1,871,734	MPV	Soranzo, Spector, et al., 2009	20p13	1,820,000	1,950,000	130.00

59	*SLC5A3, MRPS6, KCNE2*	rs9982601	34,520,998	Early-onset MI	Myocardial Infarction Genetics Consortium, 2009	21q22.11	34,340,000	34,530,000	190.00
60	*GNAZ*[b]	rs3788337	21,742,017	PLS	Jones et al., 2009	22q11.22	21,720,000	21,840,000	120.00
61	*FBXO7*	rs9609565	31,197,528	MCV	Soranzo, Spector, et al., 2009	22q12.3	31,186,000	31,250,000	64.00
62	*TMPRSS6*	rs5756506	35,797,338	MCH	Soranzo, Spector, et al., 2009	22q12.3	35,730,000	35,830,000	100.00

(B) Lineage-specific reference genes					
Lineage	Gene	Genomic position			Interval (kb)
		Chromosome	*Start*	*End*	
monocytic	*ASGR2*	17p13.1	6,943,365	6,960,852	17.49
	CD163	12p13.31	7,512,676	7,549,681	37.01
	FER1L3	10q23.33	95,054,176	95,234,029	179.85
	KLF4	9q31.2	109,284,956	109,293,576	8.62
	PID1	2q36.3	229,594,933	229,846,301	251.37
	RIN2	20p11.23	19,816,210	19,933,100	116.89
	SLC46A2	9q32	114,679,021	114,694,866	15.85
	TMEM176A	7q36.1	150,126,787	150,135,141	8.35
erythroblastoid	*CA1*	8q21.2	86,425,709	86,479,594	53.89
	CALB2	16q22.3	69,948,127	69,983,842	35.72
	EPB42	15q15.2	41,274,720	41,302,773	28.05
	ERAF	16p11.2	31,444,704	31,449,625	4.92
	FAM83A	8q24.13	124,261,933	124,293,499	31.57
	GDF15	19p13.11	18,355,968	18,362,986	7.02
	HBZ	16p13.3	140,854	146,504	5.65
	LOC51252	2q11.2	96,903,349	96,929,558	26.21
megakaryocytic	*ADCY6*	12q13.12	47,444,248	47,471,087	26.84
	CMTM5	14q11.2	22,913,857	22,920,821	6.96
	DDEF2	2p25.1	9,262,345	9,465,257	202.91
	LY6G6D	6p21.33	31,789,112	31,795,560	6.45
	MEIS1	2p14	66,514,036	66,655,395	141.36
	MYLK	3q21.1	124,811,833	125,087,839	276.01
	NFIB	9p23–p22.3	14,069,847	14,305,945	236.10
	SELP	1q24.2	167,822,711	167,868,001	45.29

Table 8-2. Ontology analysis of genes flanking FAIRE peaks using GREAT. I report the ontology of genes flanking ('stringent') FAIRE peaks of both lower (Bin 2) and higher peak score (Bin 4). For these gene sets, the GO biological process and the mouse phenotype enrichment are reported, i.e. the top 15 terms with binominal raw $P<10^{-4}$.

Term name	Binom. raw *P*-val	Binom. FDR Q-val	Binom. fold enrichment	Binom. observed region hits
(A) EB Bin 2				
GO biological process				
erythrocyte differentiation	$3.44x10^{-08}$	$1.30x10^{-05}$	2.727	40
erythrocyte homoeostasis	$1.82x10^{-07}$	$5.02x10^{-05}$	2.488	42
iron ion homoeostasis	$6.99x10^{-05}$	$6.96x10^{-03}$	2.299	28
Mouse phenotype				
anaemia	$9.55x10^{-17}$	$1.55x10^{-13}$	2.063	160
abnormal mean corpuscular volume	$3.69x10^{-10}$	$8.24x10^{-08}$	3.117	42
abnormal erythroid progenitor cell morphology	$8.55x10^{-10}$	$1.68x10^{-07}$	2.818	47
decreased mean corpuscular volume	$1.84x10^{-09}$	$3.50x10^{-07}$	4.118	27
decreased erythrocyte cell number	$2.00x10^{-09}$	$3.71x10^{-07}$	2.143	76
microcytosis	$5.08x10^{-09}$	$8.24x10^{-07}$	6.748	16
increased liver iron level	$5.80x10^{-09}$	$8.95x10^{-07}$	5.169	20
abnormal reticulocyte morphology	$3.45x10^{-08}$	$4.48x10^{-06}$	2.615	43
abnormal liver iron level	$7.14x10^{-08}$	$8.74x10^{-06}$	4.227	21
spherocytosis	$1.35x10^{-07}$	$1.51x10^{-05}$	5.685	15
abnormal pro-erythroblast morphology	$1.43x10^{-07}$	$1.55x10^{-05}$	2.216	54
abnormal mean corpuscular haemoglobin	$2.29x10^{-07}$	$2.18x10^{-05}$	3.323	26
microcytic anaemia	$2.64x10^{-07}$	$2.45x10^{-05}$	5.029	16
abnormal mast cell morphology	$8.17x10^{-06}$	$5.40x10^{-04}$	2.554	29
abnormal erythrocyte physiology	$1.22x10^{-05}$	$7.60x10^{-04}$	3.135	20
(B) EB Bin 4				
GO biological process				
intracellular transport	$4.05x10^{-12}$	$4.84x10^{-09}$	2.425	76
protein catabolic process	$1.97x10^{-11}$	$1.76x10^{-08}$	2.963	51
translation	$4.04x10^{-11}$	$2.89x10^{-08}$	3.448	40
proteolysis involved in cellular protein catabolic process	$4.60x10^{-11}$	$3.00x10^{-08}$	2.968	49
cellular protein catabolic process	$6.04x10^{-11}$	$3.61x10^{-08}$	2.944	49
ubiquitin-dependent protein catabolic process	$2.38x10^{-10}$	$1.14x10^{-07}$	2.986	45
modification-dependent protein catabolic process	$2.58x10^{-10}$	$1.16x10^{-07}$	2.978	45
establishment of localisation in cell	$3.46x10^{-10}$	$1.46x10^{-07}$	2.017	92
DNA metabolic process	$8.33x10^{-09}$	$3.14x10^{-06}$	2.290	59
cell cycle process	$6.47x10^{-08}$	$2.32x10^{-05}$	2.080	64
cellular macromolecule catabolic process	$7.64x10^{-08}$	$2.61x10^{-05}$	2.277	52

DNA replication	9.53x10^{-08}	3.11x10^{-05}	3.190	29
macromolecule catabolic process	1.77x10^{-07}	5.30x10^{-05}	2.158	55
translational elongation	2.67x10^{-07}	7.66x10^{-05}	5.002	16
cell cycle phase	1.03x10^{-06}	2.45x10^{-04}	2.141	49
(C) MK Bin 2				
GO biological process				
lamellipodium assembly	4.01x10^{-05}	7.99x10^{-03}	3.351	16
Mouse phenotype				
abnormal platelet physiology	5.66x10^{-10}	2.04x10^{-07}	2.716	51
increased IgG level	2.61x10^{-08}	5.28x10^{-06}	2.055	72
increased T cell proliferation	4.03x10^{-06}	4.93x10^{-04}	2.128	45
thymus hyperplasia	4.27x10^{-06}	5.13x10^{-04}	2.893	25
(D) MK Bin 4				
GO biological process				
cell cycle	3.14x10^{-12}	5.63x10^{-09}	2.304	84
cell cycle process	2.74x10^{-11}	3.28x10^{-08}	2.501	66
intracellular transport	3.24x10^{-08}	2.32x10^{-05}	2.195	59
protein folding	5.10x10^{-08}	3.32x10^{-05}	4.107	22
regulation of cell cycle	7.24x10^{-07}	3.24x10^{-04}	2.125	51
mitotic cell cycle	1.19x10^{-06}	4.06x10^{-04}	2.341	40
DNA metabolic process	1.90x10^{-06}	5.46x10^{-04}	2.127	47
positive regulation of cell activation	6.26x10^{-05}	6.32x10^{-03}	2.686	21
(E) MO Bin 2				
GO biological process				
immune response	1.05x10^{-51}	3.75x10^{-48}	2.203	451
cell activation	3.13x10^{-29}	3.20x10^{-26}	2.076	288
leukocyte activation	5.29x10^{-29}	4.75x10^{-26}	2.144	265
regulation of cytokine production	9.71x10^{-25}	4.35x10^{-22}	2.286	195
inflammatory response	1.41x10^{-23}	4.40x10^{-21}	2.038	240
lymphocyte activation	1.05x10^{-19}	1.94x10^{-17}	2.026	201
activation of pro-apoptotic gene products	6.50x10^{-17}	8.63x10^{-15}	5.363	40
positive regulation of cytokine production	1.20x10^{-16}	1.48x10^{-14}	2.492	107
regulation of transcription factor import into nucleus	1.21x10^{-14}	1.17x10^{-12}	3.687	51
regulation of vascular endothelial growth factor production	2.67x10^{-13}	2.40x10^{-11}	5.916	28
immune effector process	2.76x10^{-13}	2.44x10^{-11}	2.165	111
leukocyte-mediated immunity	8.98x10^{-13}	7.66x10^{-11}	2.553	76
cytokine-mediated signalling pathway	1.25x10^{-12}	1.03x10^{-10}	2.723	67
myeloid leukocyte activation	7.05x10^{-12}	5.11x10^{-10}	2.684	64
B cell activation	1.20x10^{-11}	8.20x10^{-10}	2.131	99

Mouse phenotype				
abnormal immune cell physiology	5.86x10^{-137}	3.80x10^{-133}	2.345	1,025
abnormal adaptive immunity	6.04x10^{-137}	1.96x10^{-133}	2.342	1,027
abnormal cell-mediated immunity	1.38x10^{-136}	2.99x10^{-133}	2.343	1,024
abnormal leukocyte physiology	2.28x10^{-134}	3.70x10^{-131}	2.334	1,015
abnormal antigen presenting cell physiology	4.01x10^{-111}	4.33x10^{-108}	2.524	736
abnormal lymphocyte physiology	6.53x10^{-107}	4.23x10^{-104}	2.415	767
abnormal immune serum protein physiology	8.74x10^{-103}	5.15x10^{-100}	2.400	747
abnormal lymphocyte morphology	6.48x10^{-98}	3.23x10^{-95}	2.185	857
abnormal mononuclear leukocyte morphology	2.84x10^{-94}	1.08x10^{-91}	2.047	952
abnormal bone marrow cell morphology/development	1.93x10^{-92}	6.96x10^{-90}	2.112	872
abnormal cytokine secretion	5.21x10^{-84}	1.78x10^{-81}	2.698	500
abnormal B cell physiology	2.70x10^{-80}	8.33x10^{-78}	2.627	500
abnormal leukopoiesis	6.81x10^{-79}	2.01x10^{-76}	2.155	717
abnormal myeloblast morphology/development	1.85x10^{-78}	5.20x10^{-76}	2.153	715
abnormal mononuclear leukocyte differentiation	2.01x10^{-78}	5.43x10^{-76}	2.196	685
(F) MO Bin 4				
GO biological process				
response to wounding	4.08x10^{-11}	5.85x10^{-08}	2.138	90
translation	8.91x10^{-09}	5.81x10^{-06}	2.766	42
positive regulation of cytokine production	1.04x10^{-07}	4.67x10^{-05}	3.449	26
wound healing	2.42x10^{-07}	8.66x10^{-05}	2.386	44
positive regulation of cellular protein metabolic process	6.56x10^{-07}	2.24x10^{-04}	2.225	47
response to other organism	1.24x10^{-06}	3.56x10^{-04}	2.095	51
cell activation	2.77x10^{-06}	6.86x10^{-04}	2.052	50
regulation of myeloid cell differentiation	7.24x10^{-06}	1.53x10^{-03}	2.865	24
leukocyte-mediated immunity	9.22x10^{-06}	1.79x10^{-03}	3.443	18
positive regulation vascular endothelial growth factor production	1.12x10^{-05}	2.01x10^{-03}	9.622	7
response to bacterium	1.85x10^{-05}	2.71x10^{-03}	2.158	37
regulation of vascular endothelial growth factor production	2.61x10^{-05}	2.97x10^{-03}	8.422	7
neutrophil chemotaxis	5.71x10^{-05}	4.65x10^{-03}	6.248	8
adaptive immune response based on somatic recombination of immune receptors built from immunoglobulin superfamily domains	6.51x10^{-05}	5.07x10^{-03}	3.360	15
adaptive immune response	7.89x10^{-05}	5.90x10^{-03}	3.301	15
Mouse phenotype				
abnormal cell-mediated immunity	3.72x10^{-28}	2.41x10^{-24}	2.423	186
abnormal immune cell physiology	3.77x10^{-28}	1.22x10^{-24}	2.423	186
abnormal adaptive immunity	5.38x10^{-28}	1.16x10^{-24}	2.415	186
abnormal leukocyte physiology	9.80x10^{-27}	1.59x10^{-23}	2.384	182
abnormal blood cell morphology/development	6.39x10^{-25}	8.29x10^{-22}	2.071	224
abnormal haematopoiesis	1.43x10^{-23}	1.16x10^{-20}	2.041	219

abnormal antigen presenting cell physiology	$1.03x10^{-22}$	$7.40x10^{-20}$	2.597	133
abnormal lymphocyte morphology	$5.19x10^{-22}$	$2.80x10^{-19}$	2.309	159
abnormal immune system cell morphology	$1.24x10^{-21}$	$6.18x10^{-19}$	2.101	189
abnormal leukocyte morphology	$1.74x10^{-20}$	$8.06x10^{-18}$	2.069	185
abnormal mononuclear leukocyte morphology	$3.74x10^{-20}$	$1.62x10^{-17}$	2.118	173
abnormal innate immunity	$5.37x10^{-20}$	$2.18x10^{-17}$	3.020	92
abnormal leukocyte cell number	$6.94x10^{-18}$	$2.50x10^{-15}$	2.107	155
abnormal bone marrow cell morphology/development	$1.00x10^{-17}$	$3.43x10^{-15}$	2.110	153
abnormal lymphocyte cell number	$2.70x10^{-17}$	$8.76x10^{-15}$	2.246	131
(G) CHRF Bin 2				
GO biological process				
amino acid import	$1.39x10^{-06}$	$5.37x10^{-05}$	3.093	25
lymphocyte activation involved in immune response	$5.13x10^{-06}$	$1.61x10^{-04}$	2.573	30
natural killer cell activation	$5.35x10^{-05}$	$1.13x10^{-03}$	2.338	28
endothelial cell proliferation	$8.85x10^{-05}$	$1.71x10^{-03}$	2.165	31
Mouse phenotype				
decreased platelet cell number	$1.07x10^{-18}$	$8.80x10^{-17}$	2.073	181
abnormal platelet physiology	$1.22x10^{-16}$	$8.24x10^{-15}$	2.107	153
decreased haemoglobin content	$1.49x10^{-16}$	$9.87x10^{-15}$	2.192	139
abnormal erythroid progenitor cell morphology	$4.56x10^{-14}$	$2.43x10^{-12}$	2.173	118
abnormal splenic cell ratio	$7.66x10^{-13}$	$3.38x10^{-11}$	2.168	107
abnormal embryonic haematopoiesis	$6.51x10^{-12}$	$2.48x10^{-10}$	2.065	110
abnormal mean corpuscular volume	$6.51x10^{-12}$	$2.47x10^{-10}$	2.189	96
decreased mean corpuscular volume	$1.53x10^{-11}$	$5.63x10^{-10}$	2.765	59
increased IgG2a level	$5.27x10^{-11}$	$1.79x10^{-09}$	2.068	100
increased monocyte cell number	$1.46x10^{-10}$	$4.82x10^{-09}$	2.118	90
abnormal strial intermediate cells	$4.73x10^{-10}$	$1.48x10^{-08}$	2.545	58
increased eosinophil cell number	$5.07x10^{-10}$	$1.58x10^{-08}$	2.234	75
abnormal intraocular pressure	$5.74x10^{-10}$	$1.77x10^{-08}$	2.385	65
decreased erythroid progenitor cell number	$6.16x10^{-10}$	$1.89x10^{-08}$	2.148	81
abnormal prostaglandin level	$9.26x10^{-09}$	$2.40x10^{-07}$	2.299	60
(H) CHRF Bin 4				
GO biological process				
translation	$1.97x10^{-11}$	$4.15x10^{-09}$	2.184	91
regulation of protein kinase B signalling cascade	$9.29x10^{-09}$	$1.11x10^{-06}$	3.691	28
translational elongation	$5.38x10^{-08}$	$5.01x10^{-06}$	2.959	34
protein folding	$6.09x10^{-08}$	$5.53x10^{-06}$	2.317	52
nuclear export	$3.05x10^{-07}$	$2.32x10^{-05}$	3.270	26
positive regulation of protein kinase B signalling cascade	$4.33x10^{-07}$	$3.01x10^{-05}$	3.512	23
regulation of endopeptidase activity	$4.99x10^{-07}$	$3.34x10^{-05}$	2.261	47
regulation of caspase activity	$5.04x10^{-07}$	$3.35x10^{-05}$	2.283	46

release of cytochrome c from mitochondria	8.06x10^{-07}	4.98x10^{-05}	4.611	16
regulation of peptidase activity	8.16x10^{-07}	4.96x10^{-05}	2.220	47
protein mono-ubiquitination	8.32x10^{-07}	5.01x10^{-05}	6.290	12
histone modification	8.94x10^{-07}	5.26x10^{-05}	2.066	55
cofactor biosynthetic process	1.29x10^{-06}	7.02x10^{-05}	2.451	37
covalent chromatin modification	1.74x10^{-06}	9.03x10^{-05}	2.019	55
double-strand break repair	2.21x10^{-06}	1.12x10^{-04}	2.683	30
(I) K562 Bin 2				
GO biological process				
erythrocyte differentiation	1.53x10^{-18}	2.03x10^{-16}	2.561	115
erythrocyte homoeostasis	7.48x10^{-16}	7.05x10^{-14}	2.303	119
peptidyl-tyrosine phosphorylation	8.16x10^{-11}	4.03x10^{-09}	2.034	102
peptidyl-tyrosine modification	1.18x10^{-10}	5.62x10^{-09}	2.005	104
organ regeneration	1.35x10^{-10}	6.40x10^{-09}	2.083	94
regulation of actin filament depolymerisation	2.55x10^{-08}	7.98x10^{-07}	2.763	40
actin filament capping	2.75x10^{-08}	8.50x10^{-07}	2.941	36
negative regulation of actin filament depolymerisation	8.49x10^{-08}	2.44x10^{-06}	2.716	38
Mouse phenotype				
abnormal platelet physiology	9.73x10^{-56}	7.01x10^{-53}	3.427	234
abnormal haematopoietic system physiology	4.98x10^{-51}	1.62x10^{-48}	2.165	471
decreased immature B cell number	1.13x10^{-43}	2.37x10^{-41}	2.496	300
abnormal erythrocyte morphology	1.25x10^{-36}	1.80x10^{-34}	2.053	379
abnormal megakaryocyte progenitor cell morphology	4.15x10^{-36}	5.85x10^{-34}	2.188	322
decreased pre-B cell number	9.74x10^{-35}	1.26x10^{-32}	2.596	220
abnormal pre-B cell morphology	2.47x10^{-32}	2.92x10^{-30}	2.357	245
abnormal erythrocyte cell number	1.38x10^{-31}	1.54x10^{-29}	2.099	308
decreased erythrocyte cell number	9.01x10^{-31}	9.74x10^{-29}	2.285	248
abnormal megakaryocyte morphology	1.04x10^{-29}	1.02x10^{-27}	2.083	294
abnormal erythroid progenitor cell morphology	2.09x10^{-29}	1.97x10^{-27}	2.938	150
extramedullary haematopoiesis	2.68x10^{-29}	2.49x10^{-27}	2.292	234
cortical renal glomerulopathies	5.85x10^{-28}	5.26x10^{-26}	2.326	216
abnormal pro-B cell morphology	2.07x10^{-22}	1.37x10^{-20}	2.123	208
decreased erythroid progenitor cell number	2.70x10^{-22}	1.77x10^{-20}	3.018	107
(J) K562 Bin 4				
cell cycle	1.85x10^{-21}	2.22x10^{-18}	2.009	217
intracellular transport	4.36x10^{-19}	2.24x10^{-16}	2.123	169
cell cycle process	9.49x10^{-17}	3.24x10^{-14}	2.047	160
mitotic cell cycle	2.03x10^{-16}	6.61x10^{-14}	2.331	118
mRNA transport	2.99x10^{-16}	9.33x10^{-14}	4.532	45
nucleobase, nucleoside, nucleotide and nucleic acid transport	2.60x10^{-14}	6.66x10^{-12}	3.596	51

RNA localisation	3.82x10^{-14}	9.45x10^{-12}	3.736	48
RNA transport	3.83x10^{-14}	9.14x10^{-12}	3.801	47
RNA processing	1.91x10^{-13}	3.91x10^{-11}	2.008	132
cell cycle phase	8.83x10^{-13}	1.47x10^{-10}	2.047	119
translation	7.52x10^{-12}	1.10x10^{-09}	2.477	73
ncRNA metabolic process	1.71x10^{-11}	2.27x10^{-09}	2.829	56
protein targeting to mitochondrion	7.29x10^{-11}	8.43x10^{-09}	6.306	21
mitochondrion organisation	1.83x10^{-09}	1.73x10^{-07}	2.815	45
translational elongation	2.71x10^{-09}	2.53x10^{-07}	3.693	30

Table 8-3. SNPs associated with platelet and erythrocyte phenotypes located in open chromatin in primary human MKs, EBs and MOs. Regions of open chromatin overlapping SNPs at (A) platelet (Gieger et al., 2011) and (B) erythrocyte (van der Harst et al., 2012) QTLs were determined using the software F-Seq (Boyle, Guinney, et al., 2008), applying the 'stringent' cut-off for peak calling. I retrieved additional candidate functional variants at (C) platelet and (D) erythrocyte QTLs by applying the 'moderate' cut-off for peak calling, including the SNP rs342293 (highlighted in blue). The functional mechanism of this SNP is described in detail in **Chapter 5**. Allele frequencies and r^2 values were retrieved from the European samples of the 1000 Genomes Project (interim phase I release of June 2011) (The 1000 Genomes Project Consortium, 2010). Genomic coordinates were mapped to the human reference genome, build hg19 (NCBI build 37).

(A) Platelet phenotypes, stringent peak calling														
SNP in open chromatin				GWA sentinel SNP				Proxy↔sentinel		FAIRE peak				
Chr	*Position*	*rs number*	*Allele freq*	*Chr*	*Position*	*rs number*	*Gene locus*	r^2	*Distance (bp)*	*Cell type*	*Start position*	*End position*	*F-Seq score*	*Bin*
1	171,939,290	rs2038479	0.827	1	171,949,750	rs10914144	*DNM3*	0.973	10,460	MK	171,938,981	171,939,669	0.1322	3
1	171,939,641	rs2038480	0.827	1	171,949,750	rs10914144	*DNM3*	0.973	10,109	MK	171,938,981	171,939,669	0.1322	3
1	205,185,104	rs12144980	0.381	1	205,244,953	rs1172130	*TMCC2*	0.833	59,849	EB	205,184,914	205,185,139	0.0541	1
1	205,225,457	rs1172147	0.382	1	205,244,953	rs1172130	*TMCC2*	0.839	19,496	EB	205,224,992	205,225,502	0.1785	3
1	205,225,457	rs1172147	0.382	1	205,244,953	rs1172130	*TMCC2*	0.839	19,496	MO	205,225,437	205,225,541	0.0183	1
1	205,254,238	rs1151787	0.362	1	205,244,953	rs1172130	*TMCC2*	0.928	9,285	MO	205,253,848	205,254,286	0.0303	3
2	43,711,523	rs998768	0.094	2	43,687,879	rs17030845	*THADA*	0.908	23,644	MO	43,711,507	43,711,739	0.0220	2
2	43,713,808	rs17030925	0.087	2	43,687,879	rs17030845	*THADA*	0.983	25,929	MO	43,713,592	43,713,949	0.0258	2
2	43,761,299	rs17031005	0.086	2	43,687,879	rs17030845	*THADA*	1.000	73,420	EB	43,761,285	43,761,352	0.0398	1
2	43,764,442	rs17031016	0.090	2	43,687,879	rs17030845	*THADA*	0.952	76,563	MO	43,764,269	43,764,701	0.0336	3
3	12,267,648	rs7616006	0.442	3	12,267,648	rs7616006	*SYN2*	1.000	0	MO	12,267,593	12,267,780	0.0200	1
3	12,267,780	rs7650082	0.443	3	12,267,648	rs7616006	*SYN2*	0.995	132	MO	12,267,593	12,267,780	0.0200	1
3	18,250,509	rs7618405	0.213	3	18,311,412	rs7641175	*SATB1*	0.909	60,903	MK	18,250,349	18,250,815	0.1633	4
3	18,250,509	rs7618405	0.213	3	18,311,412	rs7641175	*SATB1*	0.909	60,903	MO	18,250,463	18,250,606	0.0187	1
3	122,833,003	rs3804749	0.567	3	122,839,876	rs3792366	*PDIA5*	0.973	6,873	MK	122,832,668	122,833,082	0.1414	4
3	124,339,527	rs6771416	0.481	3	124,340,222	rs10512627	*KALRN*	1.000	695	EB	124,339,467	124,339,671	0.0492	1
3	124,339,527	rs6771416	0.481	3	124,340,222	rs10512627	*KALRN*	1.000	695	MK	124,339,398	124,339,684	0.0598	2
4	6,891,435	rs11734099	0.160	4	6,891,519	rs11734132	*KIAA0232*	0.990	84	MK	6,891,308	6,891,504	0.0457	1
4	6,891,455	rs11731274	0.163	4	6,891,519	rs11734132	*KIAA0232*	0.990	64	MK	6,891,308	6,891,504	0.0457	1

5	158,635,102	rs6556405	0.243	5	158,604,963	rs10076782	*RNF145*	0.951	30,139	MO	158,634,891	158,635,107	0.0208	1
6	25,537,194	rs214060	0.447	6	25,548,288	rs441460	*LRRC16*	0.984	11,094	MK	25,536,943	25,537,478	0.1216	3
6	135,419,631	rs9389268	0.271	6	135,419,018	rs9399137	*HBS1L-MYB*	1.000	613	EB	135,419,430	135,419,712	0.0593	1
6	135,419,636	rs9376091	0.271	6	135,419,018	rs9399137	*HBS1L-MYB*	1.000	618	EB	135,419,430	135,419,712	0.0593	1
6	135,419,688	rs9402685	0.271	6	135,419,018	rs9399137	*HBS1L-MYB*	1.000	670	EB	135,419,430	135,419,712	0.0593	1
7	123,410,525	rs112385417	0.097	7	123,411,223	rs4731120	*WASL*	1.000	698	MO	123,410,517	123,410,916	0.0386	4
9	4,811,553	rs13284787	0.231	9	4,814,948	rs13300663	*RCL1*	0.993	3,395	EB	4,811,442	4,811,611	0.0458	1
10	65,016,174	rs7088799	0.410	10	65,050,659	rs7075195	*JMJD1C*	1.000	34,485	MK	65,016,137	65,016,378	0.0437	1
10	65,027,143	rs7098181	0.407	10	65,050,659	rs7075195	*JMJD1C*	0.989	23,516	MK	65,027,095	65,027,550	0.0489	1
11	268,927	rs72882960	0.251	11	270,715	rs17655730	*PSMD13-NLRP6*	0.979	1,788	MO	268,859	269,026	0.0195	1
11	268,940	rs17655663	0.251	11	270,715	rs17655730	*PSMD13-NLRP6*	0.979	1,775	MO	268,859	269,026	0.0195	1
12	29,435,480	rs2015599	0.437	12	29,435,480	rs2015599	*MLSTD1*	1.000	0	MK	29,435,458	29,435,823	0.0842	2
12	29,435,675	rs1006409	0.437	12	29,435,480	rs2015599	*MLSTD1*	1.000	195	MK	29,435,458	29,435,823	0.0842	2
12	57,030,686	rs1107479	0.368	12	57,055,291	rs2950390	*PTGES3-BAZ2A*	0.933	24,605	EB	57,030,512	57,031,051	0.2790	4
12	57,030,686	rs1107479	0.368	12	57,055,291	rs2950390	*PTGES3-BAZ2A*	0.933	24,605	MK	57,030,568	57,031,065	0.1384	4
12	57,030,686	rs1107479	0.368	12	57,055,291	rs2950390	*PTGES3-BAZ2A*	0.933	24,605	MO	57,030,608	57,030,965	0.0204	1
12	57,119,236	rs3214051	0.611	12	57,055,291	rs2950390	*PTGES3-BAZ2A*	0.842	63,945	EB	57,118,946	57,119,448	0.2389	4
12	57,119,236	rs3214051	0.611	12	57,055,291	rs2950390	*PTGES3-BAZ2A*	0.842	63,945	MK	57,119,042	57,119,471	0.1508	4
12	57,119,236	rs3214051	0.611	12	57,055,291	rs2950390	*PTGES3-BAZ2A*	0.842	63,945	MO	57,119,035	57,119,458	0.0293	3
13	95,895,666	rs4148450	0.907	13	95,898,207	rs4148441	*ABCC4*	1.000	2,541	MK	95,895,547	95,895,837	0.0711	2
14	68,394,054	rs12431697	0.201	14	68,520,906	rs8022206	*RAD51L1*	0.841	126,852	EB	68,393,931	68,394,322	0.0743	2
14	68,394,298	rs7142860	0.192	14	68,520,906	rs8022206	*RAD51L1*	0.891	126,608	EB	68,393,931	68,394,322	0.0743	2
14	68,454,505	rs17192586	0.177	14	68,520,906	rs8022206	*RAD51L1*	0.982	66,401	EB	68,454,306	68,454,658	0.0626	1
14	68,454,505	rs17192586	0.177	14	68,520,906	rs8022206	*RAD51L1*	0.982	66,401	MK	68,454,266	68,454,734	0.1252	3
14	103,031,831	rs11160693	0.742	14	103,040,087	rs11628318	*RCOR1*	1.000	8,256	MO	103,031,713	103,032,081	0.0287	3
14	103,031,845	rs7155171	0.774	14	103,040,087	rs11628318	*RCOR1*	0.841	8,242	MO	103,031,713	103,032,081	0.0287	3
14	105,721,603	rs2735816	0.298	14	105,729,792	rs3000073	*BRF1*	0.889	8,189	MK	105,721,485	105,721,688	0.0507	1
15	65,165,459	rs4776639	0.157	15	65,183,801	rs1719271	*ANKDD1A*	0.970	18,342	MO	65,165,319	65,165,589	0.0257	2
15	65,187,874	rs1522744	0.179	15	65,183,801	rs1719271	*ANKDD1A*	0.833	4,073	MO	65,187,724	65,187,887	0.0185	1
17	27,661,844	rs117772072	0.426	17	27,714,587	rs8076739	*TAOK1*	0.859	52,743	EB	27,661,572	27,662,487	0.1082	2

17	27,661,844	rs117772072	0.426	17	27,714,587	rs8076739	*TAOK1*	0.859	52,743	MK	27,661,749	27,661,943	0.0395	1
17	27,662,271	n/a	0.439	17	27,714,587	rs8076739	*TAOK1*	0.907	52,316	EB	27,661,572	27,662,487	0.1082	2
17	27,662,271	n/a	0.439	17	27,714,587	rs8076739	*TAOK1*	0.907	52,316	MK	27,662,178	27,662,322	0.0380	1
20	1,924,707	rs13042885	0.275	20	1,924,707	rs13042885	*SIRPA*	1.000	0	MO	1,924,320	1,924,729	0.0368	4
20	57,589,995	rs55905547	0.194	20	57,587,771	rs4812048	*CTSZ-TUBB1*	0.925	2,224	MK	57,589,647	57,590,002	0.0878	3

(B) Erythrocyte phenotypes, stringent peak calling														
SNP in open chromatin				GWA sentinel SNP				Proxy↔sentinel		FAIRE peak				
Chr	*Position*	*rs number*	*Allele freq*	*Chr*	*Position*	*rs number*	*Gene locus*	*r^2*	*Distance (bp)*	*Cell type*	*Start position*	*End position*	*F-Seq score*	*Bin*
1	3,691,528	rs1175550	0.206	1	3,691,528	rs1175550	*CCDC27-LRRC48*	1.000	0	EB	3,691,366	3,691,660	0.0608	1
1	47,679,258	rs4926524	0.561	1	47,676,233	rs741959	*TAL1*	0.850	3,025	EB	47,679,193	47,679,296	0.0414	1
1	158,596,438	rs2482963	0.278	1	158,575,729	rs857684	*OR10Z1-SPTA1*	0.941	20,709	MO	158,596,370	158,596,475	0.0177	1
1	199,010,721	rs1434282	0.724	1	199,007,208	rs7529925	*MIR181A1*	0.929	3,513	EB	199,010,573	199,011,066	0.0964	2
1	199,010,721	rs1434282	0.724	1	199,007,208	rs7529925	*MIR181A1*	0.929	3,513	MK	199,010,612	199,011,128	0.1143	3
2	111,843,166	rs2880112	0.435	2	111,849,659	rs10207392	*ACOXL*	0.842	6,493	MO	111,843,101	111,843,381	0.0245	2
3	141,217,954	rs6808837	0.384	3	141,266,493	rs6776003	*RASA2*	0.857	48,539	EB	141,217,874	141,218,028	0.0453	1
3	141,217,954	rs6808837	0.384	3	141,266,493	rs6776003	*RASA2*	0.857	48,539	MO	141,217,713	141,218,095	0.0337	3
3	142,233,990	rs6791816	0.589	3	142,120,786	rs13061823	*XRN1*	0.824	113,204	MK	142,233,859	142,234,023	0.0392	1
4	55,408,875	rs218264	0.258	4	55,395,024	rs218238	*KIT*	0.834	13,851	EB	55,408,759	55,409,035	0.0724	2
4	122,745,038	rs769236	0.368	4	122,751,061	rs13152701	*BBS7-CCNA2*	1.000	6,023	EB	122,744,961	122,745,400	0.1267	3
4	122,750,079	rs13145213	0.369	4	122,751,061	rs13152701	*BBS7-CCNA2*	0.994	982	MK	122,750,001	122,750,254	0.0535	2
4	122,791,601	rs2271176	0.368	4	122,751,061	rs13152701	*BBS7-CCNA2*	1.000	40,540	EB	122,791,573	122,791,906	0.1025	2
6	41,925,159	rs9349205	0.234	6	41,914,378	rs9349204	*CCND3*	0.850	10,781	EB	41,924,850	41,925,202	0.0909	2
6	109,625,879	rs1546723	0.422	6	109,626,965	rs1008084	*CCDC162*	0.989	1,086	EB	109,625,663	109,626,087	0.1237	3
6	109,625,879	rs1546723	0.422	6	109,626,965	rs1008084	*CCDC162*	0.989	1,086	MK	109,625,692	109,625,981	0.0581	2
6	135,419,631	rs9389268	0.271	6	135,427,159	rs9389269	*HBS1L*	0.911	7,528	EB	135,419,430	135,419,712	0.0593	1
6	135,419,636	rs9376091	0.271	6	135,427,159	rs9389269	*HBS1L*	0.911	7,523	EB	135,419,430	135,419,712	0.0593	1
6	135,419,688	rs9402685	0.271	6	135,427,159	rs9389269	*HBS1L*	0.911	7,471	EB	135,419,430	135,419,712	0.0593	1
6	135,431,318	rs6920211	0.258	6	135,427,159	rs9389269	*HBS1L*	0.850	4,159	EB	135,431,304	135,431,674	0.0942	2
6	135,431,640	rs9494142	0.255	6	135,427,159	rs9389269	*HBS1L*	0.863	4,481	EB	135,431,304	135,431,674	0.0942	2

6	139,839,960	rs589235	0.499	6	139,844,429	rs590856	*CITED2*	0.904	4,469	EB	139,839,765	139,840,281	0.1883	3
6	164,463,355	rs4709819	0.446	6	164,482,836	rs736661	*QKI*	1.000	19,481	EB	164,463,287	164,463,672	0.1176	2
6	164,463,355	rs4709819	0.446	6	164,482,836	rs736661	*QKI*	1.000	19,481	MK	164,463,305	164,463,683	0.0787	2
6	164,463,572	rs4709820	0.446	6	164,482,836	rs736661	*QKI*	1.000	19,264	EB	164,463,287	164,463,672	0.1176	2
6	164,463,572	rs4709820	0.446	6	164,482,836	rs736661	*QKI*	1.000	19,264	MK	164,463,305	164,463,683	0.0787	2
8	41,630,405	rs4737009	0.261	8	41,630,405	rs4737009	*ANK1*	1.000	0	EB	41,630,153	41,630,603	0.1498	3
8	41,630,405	rs4737009	0.261	8	41,630,405	rs4737009	*ANK1*	1.000	0	MK	41,630,356	41,630,449	0.0357	1
8	41,630,447	rs4737010	0.250	8	41,630,405	rs4737009	*ANK1*	0.946	42	EB	41,630,153	41,630,603	0.1498	3
8	41,630,447	rs4737010	0.250	8	41,630,405	rs4737009	*ANK1*	0.946	42	MK	41,630,356	41,630,449	0.0357	1
9	4,852,599	rs10758656	0.193	9	4,844,265	rs2236496	*RCL1*	0.950	8,334	EB	4,852,346	4,852,777	0.1514	3
10	45,966,422	rs901683	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	0	EB	45,966,011	45,966,515	0.1390	3
10	46,039,930	rs75595592	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	73,508	EB	46,039,726	46,040,064	0.0550	1
10	46,053,061	rs9422657	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	86,639	MK	46,052,986	46,053,213	0.0528	1
11	9,023,421	rs7479407	0.413	11	8,938,049	rs11042125	*AKIP1-C11orf16-C11orf17-NRIP3-ST5*	0.873	85,372	MO	9,023,307	9,023,742	0.0429	4
11	73,115,314	rs7114009	0.114	11	73,009,084	rs7125949	*ARHGEF17-P2RY6*	0.827	106,230	MO	73,115,131	73,115,357	0.0213	1
14	65,499,909	rs12435835	0.481	14	65,502,239	rs7155454	*FNTB-MAX*	0.989	2,330	MK	65,499,871	65,500,238	0.1006	3
14	65,509,878	rs11628273	0.481	14	65,502,239	rs7155454	*FNTB-MAX*	0.989	7,639	EB	65,509,783	65,510,185	0.1592	3
14	65,509,878	rs11628273	0.481	14	65,502,239	rs7155454	*FNTB-MAX*	0.989	7,639	MK	65,509,766	65,510,128	0.0857	3
15	66,070,693	rs2572207	0.209	15	66,070,693	rs2572207	*DENND4A-PTPLAD1*	1.000	0	EB	66,070,410	66,070,913	0.1031	2
15	75,315,778	rs2304903	0.225	15	75,321,262	rs8028632	*PPCDC-SCAMP5*	1.000	5,484	EB	75,315,650	75,316,026	0.1255	3
15	75,315,778	rs2304903	0.225	15	75,321,262	rs8028632	*PPCDC-SCAMP5*	1.000	5,484	MK	75,315,722	75,315,936	0.0476	1
15	75,322,179	rs35911108	0.225	15	75,321,262	rs8028632	*PPCDC-SCAMP5*	1.000	917	MK	75,322,112	75,322,277	0.0384	1
15	75,354,621	rs35577967	0.212	15	75,321,262	rs8028632	*PPCDC-SCAMP5*	0.881	33,359	MO	75,354,500	75,354,655	0.0201	1
16	163,598	rs11248850	0.487	16	163,598	rs11248850	*C16orf35-HBA1*	1.000	0	MK	163,466	163,821	0.0790	2
16	163,667	rs11865131	0.487	16	163,598	rs11248850	*C16orf35-HBA1*	1.000	69	MK	163,466	163,821	0.0790	2
16	170,044	rs11866877	0.460	16	163,598	rs11248850	*C16orf35-HBA1*	0.850	6,446	EB	169,902	170,268	0.0891	2
16	67,927,124	rs7196789	0.172	16	67,902,326	rs2271294	*CTRL-EDC4-NUTF2*	0.991	24,798	EB	67,926,933	67,927,181	0.0645	1
16	67,927,124	rs7196789	0.172	16	67,902,326	rs2271294	*CTRL-EDC4-NUTF2*	0.991	24,798	MK	67,926,943	67,927,172	0.0514	1
16	88,840,462	rs10445033	0.634	16	88,840,462	rs10445033	*FAM38A*	1.000	0	EB	88,840,369	88,840,702	0.0689	2
16	88,840,462	rs10445033	0.634	16	88,840,462	rs10445033	*FAM38A*	1.000	0	MK	88,840,437	88,840,569	0.0389	1

17	42,323,376	rs7209801	0.272	17	42,294,337	rs2269906	*SLC4A1-UBTF*	0.804	29,039	EB	42,323,033	42,323,732	0.0931	2
17	44,217,112	rs2532314	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	390,475	MK	44,217,038	44,217,183	0.0362	1
17	44,253,364	rs2532259	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	426,727	EB	44,253,293	44,253,384	0.0403	1
17	44,271,430	rs2532236	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	444,793	EB	44,271,327	44,271,746	0.1180	2
17	44,271,430	rs2532236	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	444,793	MK	44,271,426	44,271,848	0.0510	1
17	44,272,000	rs2532235	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	445,363	MO	44,271,954	44,272,323	0.0267	2
17	44,272,266	rs2532234	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	445,629	MO	44,271,954	44,272,323	0.0267	2
17	44,272,552	rs17663792	0.231	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.993	445,915	MK	44,272,445	44,272,713	0.0509	1
17	44,272,552	rs17663792	0.231	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.993	445,915	MO	44,272,408	44,272,673	0.0216	1
17	44,272,679	rs2732660	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	446,042	MK	44,272,445	44,272,713	0.0509	1
17	44,276,330	rs1918785	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	449,693	MK	44,276,225	44,276,411	0.0454	1
17	44,280,188	rs2732675	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	453,551	MK	44,280,017	44,280,226	0.0443	1
17	44,289,101	rs2732629	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.985	462,464	MO	44,289,093	44,289,164	0.0171	1
17	44,289,150	rs2732630	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.985	462,513	MO	44,289,093	44,289,164	0.0171	1
18	43,802,778	rs12607898	0.733	18	43,833,278	rs4890633	*C18orf25*	0.980	30,500	EB	43,802,643	43,803,150	0.1602	3
19	4,458,063	rs11670503	0.254	19	4,366,219	rs732716	*MPND-SH3GL1-UBXD1*	0.839	91,844	EB	4,457,808	4,458,132	0.1203	2
19	13,001,547	rs11085824	0.310	19	13,024,250	rs741702	*CALR-FARSA-SYCE2*	0.840	22,703	MO	13,001,382	13,002,002	0.0332	3

19	13,030,280	rs8113575	0.701	19	13,024,250	rs741702	*CALR-FARSA-SYCE2*	0.921	6,030	EB	13,030,047	13,030,376	0.1119	2
19	13,044,544	rs2974750	0.700	19	13,024,250	rs741702	*CALR-FARSA-SYCE2*	0.903	20,294	MO	13,044,538	13,044,646	0.0180	1
22	32,887,498	rs6518786	0.386	22	32,880,585	rs5749446	*FBXO7*	1.000	6,913	EB	32,887,371	32,887,591	0.0472	1
22	32,887,566	rs5754113	0.386	22	32,880,585	rs5749446	*FBXO7*	1.000	6,981	EB	32,887,371	32,887,591	0.0472	1

(C) Platelet phenotypes, moderate peak calling														
SNP in open chromatin				GWA sentinel SNP				Proxy↔sentinel		FAIRE peak				
Chr	*Position*	*rs number*	*Allele freq*	*Chr*	*Position*	*rs number*	*Gene locus*	r^2	*Distance (bp)*	*Cell type*	*Start position*	*End position*	*F-Seq score*	*Bin*
1	205,236,432	rs1768587	0.361	1	205,244,953	rs1172130	*TMCC2*	0.934	8,521	MK	205,236,432	205,236,777	0.0895	4
1	205,253,839	rs3851296	0.422	1	205,244,953	rs1172130	*TMCC2*	0.829	8,886	MO	205,253,816	205,254,326	0.0303	3
2	43,665,943	rs76684055	0.079	2	43,687,879	rs17030845	*THADA*	0.916	21,936	EB	43,665,917	43,666,222	0.0553	2
2	43,668,169	rs7567685	0.086	2	43,687,879	rs17030845	*THADA*	1.000	19,710	EB	43,668,076	43,668,192	0.0325	1
2	43,668,176	rs7581586	0.086	2	43,687,879	rs17030845	*THADA*	1.000	19,703	EB	43,668,076	43,668,192	0.0325	1
2	43,676,433	rs12105786	0.086	2	43,687,879	rs17030845	*THADA*	1.000	11,446	EB	43,676,386	43,676,580	0.0339	1
2	43,678,617	rs17406646	0.079	2	43,687,879	rs17030845	*THADA*	0.916	9,262	MO	43,678,533	43,678,675	0.0153	1
2	43,683,885	rs6728106	0.093	2	43,687,879	rs17030845	*THADA*	0.922	3,994	MO	43,683,870	43,683,943	0.0147	1
2	43,710,444	rs7600657	0.097	2	43,687,879	rs17030845	*THADA*	0.880	22,565	MO	43,710,150	43,710,524	0.0167	1
2	43,737,465	rs10180005	0.087	2	43,687,879	rs17030845	*THADA*	0.983	49,586	MK	43,737,276	43,737,481	0.0323	1
4	6,891,519	rs11734132	0.161	4	6,891,519	rs11734132	*KIAA0232*	1.000	0	MK	6,891,275	6,891,535	0.0457	2
6	25,536,937	rs214059	0.447	6	25,548,288	rs441460	*LRRC16*	0.984	11,351	MK	25,536,902	25,537,516	0.1216	4
7	106,355,943	rs342272	0.425	7	106,372,219	rs342293	*FLJ36031-PIK3CG*	0.913	16,276	MO	106,355,923	106,356,436	0.0170	1
7	106,356,005	rs342273	0.425	7	106,372,219	rs342293	*FLJ36031-PIK3CG*	0.913	16,214	MO	106,355,923	106,356,436	0.0170	1
7	106,372,219	rs342293	0.447	7	106,372,219	rs342293	*FLJ36031-PIK3CG*	1.000	0	MK	106,372,203	106,372,378	0.0339	1
7	123,407,146	rs12671376	0.097	7	123,411,223	rs4731120	*WASL*	1.000	4,077	MK	123,407,083	123,407,197	0.0280	1
7	123,410,918	rs79415660	0.097	7	123,411,223	rs4731120	*WASL*	1.000	305	MO	123,410,502	123,410,962	0.0386	4
7	123,422,357	rs4727976	0.098	7	123,411,223	rs4731120	*WASL*	0.985	11,134	MO	123,422,338	123,422,610	0.0173	1
7	123,424,583	rs725859	0.097	7	123,411,223	rs4731120	*WASL*	1.000	13,360	MO	123,424,389	123,424,594	0.0155	1
8	106,581,528	rs6993770	0.295	8	106,581,528	rs6993770	*ZFPM2*	1.000	0	EB	106,581,358	106,581,529	0.0371	1
8	106,581,528	rs6993770	0.295	8	106,581,528	rs6993770	*ZFPM2*	1.000	0	MK	106,581,443	106,581,722	0.0359	1
9	332,152	rs1536609	0.627	9	331,490	rs10813766	*DOCK8*	0.983	662	MK	332,133	332,338	0.0362	1

10	65,027,610	rs10761731	0.407	10	65,050,659	rs7075195	*JMJD1C*	0.989	23,049	EB	65,027,412	65,027,689	0.0406	1
10	65,062,820	rs7073753	0.409	10	65,050,659	rs7075195	*JMJD1C*	0.995	12,161	EB	65,062,634	65,062,851	0.0440	1
10	65,071,215	rs10822155	0.407	10	65,050,659	rs7075195	*JMJD1C*	0.989	20,556	EB	65,071,215	65,071,462	0.0482	1
12	122,365,583	rs7961894	0.086	12	122,365,583	rs7961894	*WDR66*	1.000	0	MO	122,365,425	122,365,646	0.0172	1
14	68,413,769	rs7151917	0.177	14	68,520,906	rs8022206	*RAD51L1*	0.982	107,137	EB	68,413,594	68,413,802	0.0369	1
14	68,431,295	rs4902540	0.192	14	68,520,906	rs8022206	*RAD51L1*	0.891	89,611	MO	68,431,223	68,431,394	0.0153	1
14	68,599,145	rs2588834	0.823	14	68,520,906	rs8022206	*RAD51L1*	0.809	78,239	MO	68,599,107	68,599,284	0.0161	1
15	65,185,884	rs1684032	0.173	15	65,183,801	rs1719271	*ANKDD1A*	0.882	2,083	MO	65,185,876	65,186,279	0.0265	3
15	65,185,904	rs1719262	0.173	15	65,183,801	rs1719271	*ANKDD1A*	0.882	2,103	MO	65,185,876	65,186,279	0.0265	3
19	16,197,320	rs2228367	0.015	19	16,185,559	rs8109288	*TPM4*	1.000	11,761	MO	16,197,302	16,197,581	0.0187	2

(D) Erythrocyte phenotypes, moderate peak calling

SNP in open chromatin				GWA sentinel SNP				Proxy↔sentinel		FAIRE peak				
Chr	*Position*	*rs number*	*Allele freq*	*Chr*	*Position*	*rs number*	*Gene locus*	r^2	*Distance (bp)*	*Cell type*	*Start position*	*End position*	*F-Seq score*	*Bin*
1	203,651,824	rs11240734	0.890	1	203,655,121	rs7551442	*ATP2B4*	1.000	3,297	EB	203,651,789	203,651,903	0.0322	1
1	205,253,839	rs3851296	0.422	1	205,249,450	rs9660992	*TMCC2*	1.000	4,389	MO	205,253,816	205,254,326	0.0303	3
3	142,004,728	rs4434160	0.552	3	142,120,786	rs13061823	*XRN1*	0.984	116,058	EB	142,004,626	142,004,732	0.0327	1
3	142,247,052	rs6440087	0.589	3	142,120,786	rs13061823	*XRN1*	0.824	126,266	EB	142,247,013	142,247,180	0.0339	1
3	142,272,314	rs9869842	0.585	3	142,120,786	rs13061823	*XRN1*	0.818	151,528	MO	142,272,275	142,272,335	0.0147	1
3	142,313,987	rs6440092	0.585	3	142,120,786	rs13061823	*XRN1*	0.808	193,201	EB	142,313,850	142,314,018	0.0326	1
3	142,315,074	rs13069307	0.561	3	142,120,786	rs13061823	*XRN1*	0.886	194,288	EB	142,315,064	142,315,310	0.0468	1
4	122,748,996	rs1048433	0.368	4	122,751,061	rs13152701	*BBS7-CCNA2*	1.000	2,065	MK	122,748,903	122,749,066	0.0335	1
6	43,810,974	rs9369425	0.708	6	43,811,430	rs9369427	*VEGFA*	0.994	456	EB	43,810,962	43,811,323	0.0606	2
7	50,427,982	rs6592965	0.452	7	50,428,445	rs12718598	*IKZF1*	0.848	463	EB	50,427,715	50,428,000	0.0573	2
7	100,221,849	rs4729597	0.626	7	100,240,296	rs2075672	*ACTL6B-TFR2*	0.925	18,447	EB	100,221,848	100,221,924	0.0318	1
10	45,972,325	rs71494788	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	5,903	MK	45,972,249	45,972,350	0.0278	1
10	45,991,978	rs12764652	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	25,556	EB	45,991,905	45,992,079	0.0347	1
10	46,013,438	rs12781186	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	47,016	MO	46,013,384	46,013,477	0.0148	1
10	46,023,327	rs12764693	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	56,905	MO	46,023,200	46,023,549	0.0164	1
10	46,024,335	rs35902429	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	57,913	EB	46,024,290	46,024,461	0.0368	1

10	46,051,401	rs9422654	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	84,979	MO	46,051,373	46,051,506	0.0153	1
10	46,051,408	rs9422655	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	84,986	MO	46,051,373	46,051,506	0.0153	1
10	46,074,329	rs71496620	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	107,907	EB	46,073,754	46,074,355	0.0461	1
10	46,081,096	rs12779637	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	114,674	MK	46,081,095	46,081,396	0.0339	1
10	46,081,101	rs12772102	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	114,679	MK	46,081,095	46,081,396	0.0339	1
10	46,081,358	rs35183751	0.079	10	45,966,422	rs901683	*CTGLF1-MARCH8*	1.000	114,936	MK	46,081,095	46,081,396	0.0339	1
11	73,018,413	rs77127734	0.111	11	73,009,084	rs7125949	*ARHGEF17-P2RY6*	0.949	9,329	MO	73,018,380	73,018,518	0.0164	1
11	73,136,261	rs12288753	0.112	11	73,009,084	rs7125949	*ARHGEF17-P2RY6*	0.838	127,177	MO	73,136,189	73,136,489	0.0178	1
12	121,163,518	rs2239760	0.433	12	121,126,438	rs3829290	*MLEC*	0.838	37,080	EB	121,163,444	121,163,621	0.0377	1
14	103,822,762	rs17616316	0.065	14	103,822,762	rs17616316	*EIF5*	1.000	0	EB	103,822,621	103,822,799	0.0333	1
15	75,322,310	rs5020842	0.226	15	75,321,262	rs8028632	*PPCDC-SCAMP5*	0.992	1,048	MK	75,321,852	75,322,327	0.0384	1
15	75,370,232	rs8036197	0.226	15	75,321,262	rs8028632	*PPCDC-SCAMP5*	0.806	48,970	MO	75,370,203	75,370,361	0.0167	1
17	27,077,331	rs3181215	0.194	17	27,075,423	rs2070265	*C17orf63-ERAL1-NEK8-TRAF4*	0.992	1,908	MO	27,077,239	27,077,355	0.0156	1
17	27,088,436	rs7215000	0.194	17	27,075,423	rs2070265	*C17orf63-ERAL1-NEK8-TRAF4*	0.992	13,013	MO	27,088,426	27,088,562	0.0156	1
17	44,047,216	rs62641967	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	220,579	MO	44,047,179	44,047,573	0.0176	1
17	44,080,039	rs62064663	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	253,402	MO	44,080,033	44,080,154	0.0159	1
17	44,147,721	rs56323408	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	321,084	MK	44,147,710	44,148,096	0.0294	1
17	44,170,238	rs112596352	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	343,601	MO	44,170,238	44,170,405	0.0166	1
17	44,177,337	rs62061808	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	350,700	MO	44,177,282	44,177,566	0.0156	1
17	44,178,839	rs62061809	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	352,202	EB	44,178,689	44,178,883	0.0396	1
17	44,184,375	rs79346219	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	357,738	MO	44,184,339	44,184,430	0.0147	1
17	44,184,404	rs73984689	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	357,767	MO	44,184,339	44,184,430	0.0147	1

17	44,184,428	rs111295615	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	357,791	MO	44,184,339	44,184,430	0.0147	1
17	44,188,477	rs17577159	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	361,840	MO	44,188,308	44,188,489	0.0161	1
17	44,195,424	rs62061852	0.231	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.993	368,787	MO	44,195,244	44,195,458	0.0156	1
17	44,214,814	rs740711	0.218	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.934	388,177	EB	44,214,746	44,214,863	0.0340	1
17	44,214,815	rs740710	0.218	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.934	388,178	EB	44,214,746	44,214,863	0.0340	1
17	44,217,226	rs2696599	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	390,589	MK	44,216,973	44,217,266	0.0362	1
17	44,218,044	rs1918801	0.202	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.849	391,407	MK	44,217,957	44,218,117	0.0287	1
17	44,218,138	rs1918800	0.200	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.835	391,501	MO	44,218,100	44,218,510	0.0158	1
17	44,218,242	rs1918799	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	391,605	MO	44,218,100	44,218,510	0.0158	1
17	44,221,836	rs2696589	0.202	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.849	395,199	MK	44,221,735	44,221,885	0.0303	1
17	44,228,169	rs1918793	0.237	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.964	401,532	MK	44,228,125	44,228,177	0.0270	1
17	44,241,664	rs2532286	0.231	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.993	415,027	EB	44,241,495	44,241,680	0.0326	1
17	44,244,397	rs2696684	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	417,760	MK	44,244,122	44,244,665	0.0347	1
17	44,244,581	rs17585644	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	417,944	MK	44,244,122	44,244,665	0.0347	1
17	44,244,896	rs2532282	0.202	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.849	418,259	MK	44,244,809	44,245,065	0.0329	1
17	44,244,926	rs2696657	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-*	1.000	418,289	MK	44,244,809	44,245,065	0.0329	1

							IMP5-KIAA1267-MAPT-STH							
17	44,246,527	rs2532277	0.202	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.849	419,890	MO	44,246,415	44,246,533	0.0150	1
17	44,248,042	rs2532271	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	421,405	MO	44,247,806	44,248,048	0.0169	1
17	44,253,275	rs2532260	0.197	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.821	426,638	EB	44,253,215	44,253,442	0.0403	1
17	44,253,275	rs2532260	0.197	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.821	426,638	MK	44,253,213	44,253,362	0.0295	1
17	44,254,291	rs2732645	0.197	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.821	427,654	MO	44,254,143	44,254,400	0.0167	1
17	44,254,379	rs2732646	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	427,742	MO	44,254,143	44,254,400	0.0167	1
17	44,256,655	rs740708	0.194	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.807	430,018	MO	44,256,325	44,257,117	0.0320	4
17	44,258,354	rs740706	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	431,717	EB	44,258,268	44,258,455	0.0354	1
17	44,258,422	rs758523	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	431,785	EB	44,258,268	44,258,455	0.0354	1
17	44,265,477	rs2696709	0.202	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.849	438,840	EB	44,265,394	44,265,607	0.0374	1
17	44,267,617	rs1918788	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	440,980	EB	44,267,586	44,267,789	0.0370	1
17	44,268,488	rs2141298	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	441,851	EB	44,268,377	44,268,742	0.0392	1
17	44,274,560	rs2696440	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	1.000	447,923	MO	44,274,546	44,274,672	0.0157	1
17	44,289,220	rs2532417	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.985	462,583	MO	44,288,999	44,289,261	0.0171	1
17	44,289,232	rs2732631	0.230	17	43,826,637	rs12150672	*ARHGAP27-ARL17-C17orf69-CRHR1-IMP5-KIAA1267-MAPT-STH*	0.985	462,595	MO	44,288,999	44,289,261	0.0171	1

22	32,870,769	rs2072814	0.366	22	32,880,585	rs5749446	*FBXO7*	0.908	9,816	EB	32,870,586	32,870,772	0.0389	1
22	32,884,381	rs7354804	0.386	22	32,880,585	rs5749446	*FBXO7*	1.000	3,796	EB	32,884,051	32,884,413	0.0464	1
22	32,887,661	rs17772071	0.386	22	32,880,585	rs5749446	*FBXO7*	1.000	7,076	EB	32,887,327	32,887,681	0.0472	1

Table 8-4. *In silico* transcription factor binding site predictions. I report transcription factors of whose DNA binding motif may be altered due to the presence of a SNP. The table is ranked from the highest to the lowest absolute difference log(*P*) between the probes harbouring the reference and alternative allele of the platelet SNPs. Only results with absolute differences of log(*P*)>1 or log(*P*)<-1 are shown. The results with the probes containing rs1107479C>T did not meet these criteria.

#	Rank	log(*P*)	Ref allele	Alt allele	Matrix ID	Matrix name	Binding factor
1			rs1006409-A	rs1006409-G			
	1	5.03	$1.84x10^{-06}$	$1.98x10^{-01}$	M00252	V$TATA_01	TATA
	2	4.80	$1.84x10^{-06}$	$1.15x10^{-01}$	M00216	V$TATA_C	TATA
	3	3.88	$1.84x10^{-06}$	$1.41x10^{-02}$	M00215	V$SRF_C	SRF
	4	3.36	$1.20x10^{-04}$	$2.71x10^{-01}$	M01292	V$HOXA13_01	HOXA13
	5	2.45	$2.91x10^{-05}$	$8.11x10^{-03}$	M01304	V$SRF_03	SRF
	6	2.32	$2.98x10^{-04}$	$6.23x10^{-02}$	M00395	V$HOXA3_01	HOXA3
	7	1.75	$1.19x10^{-05}$	$6.61x10^{-04}$	M00922	V$SRF_Q5_01	SRF
	8	1.59	$2.66x10^{-03}$	$1.04x10^{-01}$	M01375	V$HOXD10_01	HOXD10
	9	1.41	$1.30x10^{-02}$	$3.37x10^{-01}$	M00671	V$TCF4_Q5	TCF4
	10	-1.28	$2.36x10^{-01}$	$1.23x10^{-02}$	M00751	V$AML1_Q6	AML1
	11	1.20	$2.25x10^{-03}$	$3.55x10^{-02}$	M00152	V$SRF_01	SRF
	12	1.15	$1.06x10^{-03}$	$1.50x10^{-02}$	M00810	V$SRF_Q4	SRF
	13	-1.07	$1.36x10^{-01}$	$1.17x10^{-02}$	M00722	V$COREBINDINGFACTOR_Q6	Core-binding factor
	14	-1.06	$1.16x10^{-01}$	$1.01x10^{-02}$	M00731	V$OSF2_Q6	OSF2
	15	-1.06	$7.72x10^{-02}$	$6.70x10^{-03}$	M00984	V$PEBP_Q6	PEBP
	16	-1.00	$5.64x10^{-01}$	$5.64x10^{-02}$	M01658	V$AML1_Q4	AML1
2			rs1107479-C	rs1107479-T			
	–	–	–	–	–	–	–
3			rs11731274-T	rs11731274-G			
	1	2.49	$1.61x10^{-03}$	$4.94x10^{-01}$	M00500	V$STAT6_02	STAT6
	2	1.50	$1.43x10^{-02}$	$4.51x10^{-01}$	M00496	V$STAT1_03	STAT1
	3	-1.29	$3.12x10^{-02}$	$1.61x10^{-03}$	M01177	V$SREBP2_Q6	SREBP2
	4	1.08	$3.36x10^{-02}$	$4.02x10^{-01}$	M01281	V$NFAT1_Q6	NFAT1
4			rs11734099-G	rs11734099-A			
	1	1.27	$3.14x10^{-02}$	$5.92x10^{-01}$	M00272	V$P53_02	P53
5			rs17192586-G	rs17192586-A			
	1	-2.44	$2.08x10^{-01}$	$7.63x10^{-04}$	M00175	V$AP4_Q5	AP4
	2	-2.22	$1.76x10^{-01}$	$1.06x10^{-03}$	M01287	V$NEUROD_01	NEUROD
	3	1.85	$2.74x10^{-03}$	$1.92x10^{-01}$	M00227	V$VMYB_02	VMYB
	4	1.69	$4.86x10^{-03}$	$2.38x10^{-01}$	M00003	V$VMYB_01	VMYB
	5	-1.58	$3.97x10^{-01}$	$1.04x10^{-02}$	M00927	V$AP4_Q6_01	AP4
	6	-1.56	$2.52x10^{-01}$	$6.95x10^{-03}$	M01347	V$RHOX11_01	RHOX11
	7	-1.43	$3.15x10^{-01}$	$1.17x10^{-02}$	M01384	V$RHOX11_02	RHOX11
	8	-1.37	$1.99x10^{-02}$	$8.44x10^{-04}$	M01419	V$MEIS1_02	MEIS1
	9	-1.32	$1.60x10^{-02}$	$7.63x10^{-04}$	M01488	V$MEIS2_01	MEIS2

	10	-1.22	5.43x10^{-01}	3.27x10^{-02}	M00973	V$E2A_Q6	E2A
	11	-1.13	2.02x10^{-02}	1.49x10^{-03}	M01459	V$PREP1_01	PREP1
	12	-1.12	4.12x10^{-01}	3.10x10^{-02}	M00712	V$MYOGENIN_Q6	MYOGENIN
	13	-1.08	2.23x10^{-01}	1.88x10^{-02}	M00176	V$AP4_Q6	AP4
	14	-1.06	2.47x10^{-01}	2.15x10^{-02}	M00122	V$USF_02	USF
	15	-1.03	2.77x10^{-02}	2.61x10^{-03}	M01407	V$TGIF2_01	TGIF2
6			rs2015599-G	rs2015599-A			
	1	-1.40	8.62x10^{-02}	3.45x10^{-03}	M01410	V$IRX4_01	IRX4
	2	-1.39	6.08x10^{-02}	2.51x10^{-03}	M01485	V$IRX3_02	IRX3
	3	-1.17	9.20x10^{-02}	6.29x10^{-03}	M01318	V$IRX3_01	IRX3
7			rs2038479-C	rs2038479-A			
	1	-2.63	2.51x10^{-01}	5.86x10^{-04}	M00630	V$FOXM1_01	FOXM1
	2	-2.55	8.43x10^{-02}	2.35x10^{-04}	M00216	V$TATA_C	TATA
	3	-2.23	1.61x10^{-01}	9.50x10^{-04}	M00252	V$TATA_01	TATA
	4	-1.78	1.43x10^{-02}	2.35x10^{-04}	M00980	V$TBP_Q6	TBP
	5	1.53	1.06x10^{-02}	3.59x10^{-01}	M00462	V$GATA6_01	GATA6
	6	1.50	9.91x10^{-03}	3.16x10^{-01}	M00104	V$CDPCR1_01	CDPCR1
	7	1.34	9.23x10^{-03}	2.00x10^{-01}	M00347	V$GATA1_06	GATA1
	8	-1.19	1.43x10^{-01}	9.29x10^{-03}	M01404	V$HOXD13_01	HOXD13
	9	1.06	2.53x10^{-02}	2.90x10^{-01}	M00348	V$GATA2_02	GATA2
	10	1.03	1.38x10^{-02}	1.49x10^{-01}	M01429	V$HOMEZ_01	HOMEZ
8			rs2038480-A	rs2038480-T			
	1	-3.24	9.25x10^{-02}	5.38x10^{-05}	M00744	V$POU1F1_Q6	POU1F1
	2	-2.98	5.08x10^{-02}	5.38x10^{-05}	M00138	V$OCT1_04	OCT1
	3	2.38	5.38x10^{-05}	1.29x10^{-02}	M00451	V$NKX3A_01	NKX3A
	4	2.23	4.68x10^{-03}	7.94x10^{-01}	M01181	V$NKX32_01	NKX32
	5	-1.22	8.44x10^{-02}	5.10x10^{-03}	M00672	V$TEF_Q6	TEF
	6	-1.03	1.51x10^{-01}	1.42x10^{-02}	M00136	V$OCT1_02	OCT1
9			rs214060-C	rs214060-T			
	1	-2.38	5.24x10^{-01}	2.19x10^{-03}	M01275	V$IPF1_Q6	IPF1
	2	-1.53	7.49x10^{-02}	2.19x10^{-03}	M00436	V$IPF1_Q4	IPF1
	3	-1.30	3.47x10^{-01}	1.74x10^{-02}	M00930	V$OCT1_Q5_01	OCT1
	4	-1.25	6.51x10^{-01}	3.66x10^{-02}	M01307	V$POU5F1_01	POU5F1
	5	-1.20	3.90x10^{-01}	2.46x10^{-02}	M00795	V$OCT_Q6	OCT
	6	-1.01	3.94x10^{-01}	3.84x10^{-02}	M01338	V$HOXD3_01	HOXD3
10			rs2735816-G	rs2735816-C			
	1	-2.59	3.01x10^{-01}	7.78x10^{-04}	M00921	V$GR_Q6_01	GR
	2	-1.41	1.91x10^{-02}	7.48x10^{-04}	M00647	V$LXR_Q3	LXR
11			rs3214051-G	rs3214051-A			
	1	-1.39	2.28x10^{-01}	9.35x10^{-03}	M00097	V$PAX6_01	PAX6
	2	1.08	4.24x10^{-03}	5.15x10^{-02}	M00134	V$HNF4_01	HNF4
	3	1.08	6.96x10^{-02}	8.36x10^{-01}	M01107	V$RUSH1A_02	RUSH1A
	4	-1.01	5.61x10^{-01}	5.48x10^{-02}	M00684	V$XPF1_Q6	XPF1

12			rs3804749-C	rs3804749-T			
	1	-1.74	6.66x10^{-01}	1.22x10^{-02}	M00073	V$DELTAEF1_01	DELTAEF1
	2	-1.58	3.98x10^{-01}	1.04x10^{-02}	M00973	V$E2A_Q6	E2A
	3	-1.29	6.72x10^{-01}	3.42x10^{-02}	M01034	V$EBOX_Q6_01	EBOX
	4	-1.13	3.20x10^{-01}	2.37x10^{-02}	M00821	V$NRF2_Q4	NRF2
	5	-1.11	4.17x10^{-01}	3.25x10^{-02}	M00712	V$MYOGENIN_Q6	MYOGENIN
	6	-1.09	5.18x10^{-01}	4.20x10^{-02}	M01116	V$CLOCKBMAL_Q6	CLOCKBMAL
	7	-1.01	5.98x10^{-01}	5.81x10^{-02}	M00799	V$MYC_Q2	MYC
	8	-1.01	3.74x10^{-01}	3.68x10^{-02}	M01287	V$NEUROD_01	NEUROD
13			rs4148450-C	rs4148450-T			
	1	-1.62	4.14x10^{-01}	9.92x10^{-03}	M00762	V$DR1_Q3	DR1
	2	1.54	1.29x10^{-02}	4.47x10^{-01}	M00655	V$PEA3_Q6	PEA3
	3	1.45	3.36x10^{-02}	9.55x10^{-01}	M01079	V$CBF_01	CBF
	4	-1.42	3.54x10^{-01}	1.34x10^{-02}	M00763	V$PPAR_DR1_Q2	PPAR
	5	-1.30	4.04x10^{-01}	2.01x10^{-02}	M00973	V$E2A_Q6	E2A
	6	-1.22	2.28x10^{-01}	1.39x10^{-02}	M00693	V$E12_Q6	E12
	7	-1.21	2.29x10^{-01}	1.41x10^{-02}	M00764	V$HNF4_DR1_Q3	HNF4
	8	-1.07	4.54x10^{-02}	3.88x10^{-03}	M01263	V$TBX15_01	TBX15
	9	-1.04	3.79x10^{-02}	3.41x10^{-03}	M01195	V$TBX22_01	TBX22
	10	-1.04	1.90x10^{-01}	1.75x10^{-02}	M00765	V$COUP_DR1_Q6	COUP
14			rs55905547-A	rs55905547-G			
	1	1.16	6.61x10^{-02}	9.59x10^{-01}	M01033	V$HNF4_Q6_03	HNF4
15			rs6771416-G	rs6771416-A			
	1	-1.85	2.73x10^{-01}	3.87x10^{-03}	M01334	V$NKX11_01	NKX11
	2	-1.62	5.76x10^{-01}	1.38x10^{-02}	M00313	V$GEN_INI2_B	GEN
	3	-1.57	4.93x10^{-01}	1.34x10^{-02}	M00315	V$GEN_INI_B	GEN
	4	1.55	1.74x10^{-02}	6.12x10^{-01}	M01592	V$LBP9_01	LBP9
	5	-1.48	5.33x10^{-01}	1.78x10^{-02}	M00314	V$GEN_INI3_B	GEN
	6	1.30	4.01x10^{-02}	7.92x10^{-01}	M00077	V$GATA3_01	GATA3
	7	-1.19	2.49x10^{-01}	1.61x10^{-02}	M01386	V$EVX2_01	EVX2
	8	-1.07	2.50x10^{-01}	2.11x10^{-02}	M01331	V$ISX_01	ISX
	9	-1.03	6.84x10^{-01}	6.34x10^{-02}	M00624	V$DBP_Q6	DBP
	10	-1.01	3.64x10^{-01}	3.58x10^{-02}	M01382	V$GBX2_01	GBX2
	11	-1.00	3.36x10^{-01}	3.33x10^{-02}	M01427	V$NKX12_01	NKX12
	12	1.00	1.01x10^{-01}	9.99x10^{-01}	M00075	V$GATA1_01	GATA1
	13	-1.00	3.85x10^{-01}	3.89x10^{-02}	M01461	V$EMX2_01	EMX2
16			rs7618405-C	rs7618405-A			
	1	-2.96	4.70x10^{-01}	5.13x10^{-04}	M01131	V$SOX10_Q6	SOX10
	2	-1.83	8.17x10^{-01}	1.22x10^{-02}	M00137	V$OCT1_03	OCT1
	3	-1.30	3.81x10^{-01}	1.93x10^{-02}	M01016	V$SOX17_01	SOX17

Table 8-5. Investigation of the functional role of platelet volume-associated variants at chromosome 7q22.3. Proxy SNPs to rs342293 ($r^2 \geq 0.8$) were retrieved from the 1000 Genomes Project (Pilot 1, CEU). Genomic coordinates were based on the human reference genome, build hg18 (NCBI build 36). *P*-values for association with mean platelet volume (MPV) were obtained from Soranzo, Spector, et al., 2009. *In silico* transcription binding site predictions were performed as described in **Section 2.10**. Based on the HaemAtlas data, we defined genes as expressed when they exhibit a normalised expression value of at least 8.5.

Proxy to rs342293							SNP overlaps with …		
ID	*Chromosome*	*Position*	*r^2*	*Distance*	*Annotation*	*MPV P-value*	*NDR in MK cells*	*Transcription factor (TF) binding site (MatInspector)*	*Binding site of TF expressed in MK cells*
rs342207	7	106,108,641	0.81	50,814	Intergenic	1.46×10^{-10}	–	–	–
rs342209	7	106,109,208	0.84	50,247	Intergenic	n/a	–	–	–
rs342210	7	106,109,256	0.87	50,199	Intergenic	3.57×10^{-11}	–	–	–
rs342212	7	106,111,150	0.84	48,305	Intergenic	5.47×10^{-11}	–	–	–
rs342213	7	106,111,848	0.84	47,607	Intergenic	6.21×10^{-11}	–	–	–
rs342214	7	106,111,979	0.81	47,476	Intergenic	n/a	–	–	–
rs342236	7	106,122,874	0.84	36,581	Intergenic	1.24×10^{-10}	–	–	–
rs342239	7	106,124,138	0.90	35,317	Intergenic	6.97×10^{-11}	–	–	–
rs342240	7	106,124,486	0.90	34,969	Intergenic	6.97×10^{-11}	–	HMX2	–
rs342241	7	106,124,587	0.90	34,868	Intergenic	n/a	–	–	–
rs342242	7	106,126,225	0.90	33,230	Intergenic	6.43×10^{-11}	–	–	–
rs342244	7	106,128,061	0.81	31,394	Intergenic	1.43×10^{-10}	–	–	–
rs342247	7	106,130,427	0.84	29,028	Intergenic	4.23×10^{-11}	–	HHEX, HOXC4, LBX2, MSX	HHEX, MSX
rs342248	7	106,130,541	0.84	28,914	Intergenic	n/a	–	–	–
rs342251	7	106,132,045	0.90	27,410	Intergenic	n/a	–	–	–
rs342252	7	106,133,666	0.90	25,789	Intergenic	n/a	–	–	–
rs342254	7	106,135,492	0.90	23,963	Intergenic	3.84×10^{-11}	–	–	–
rs342257	7	106,137,089	0.84	22,366	Intergenic	6.09×10^{-11}	–	–	–
rs342271	7	106,143,111	0.90	16,344	Intergenic	n/a	–	–	–
rs342275	7	106,146,452	0.90	13,003	Intergenic	1.33×10^{-11}	–	–	–

rs342281	7	106,149,079	0.84	10,376	Intergenic	$5.36x10^{-12}$	–	–	–
rs342284	7	106,149,446	0.87	10,009	Intergenic	$4.62x10^{-12}$	–	–	–
rs342286	7	106,151,835	0.94	7,620	Intergenic	$4.90x10^{-12}$	–	–	–
rs342290	7	106,154,840	1.00	4,615	Intergenic	n/a	–	–	–
rs342292	7	106,157,880	1.00	1,575	Intergenic	$8.64x10^{-13}$	–	MEIS1, MEIS1A/HOXA9	MEIS1, MEIS1A/HOXA9
rs342293	7	106,159,455	1.00	0	Intergenic	$6.75x10^{-13}$	+	GATA1, EVI1	GATA1, EVI1
rs342294	7	106,159,858	1.00	403	Intergenic	n/a	+	–	–
rs342295	7	106,159,996	1.00	541	Intergenic	$4.14x10^{-13}$	–	–	–
rs342296	7	106,160,139	1.00	684	Intergenic	$7.68x10^{-13}$	–	–	–
rs342298	7	106,160,882	0.97	1,427	Intergenic	$7.55x10^{-13}$	–	–	–
rs342299	7	106,160,954	0.97	1,499	Intergenic	$7.55x10^{-13}$	–	–	–
rs386805	7	106,125,700	0.97	33,755	Intergenic	n/a	–	–	–
rs67036916	7	106,154,872	0.97	4,583	Intergenic	n/a	–	–	–
rs77655772	7	106,125,699	0.97	33,756	Intergenic	n/a	–	–	–

Table 8-6. Expression QTL associations at the *PIK3CG* gene locus in platelets, macrophages, monocytes, B cells (LCLs), adipose and skin. The strength of the relationship between alleles and gene expression intensities was estimated with the Spearman's rank correlation coefficient using the software Genevar (**Section 2.12**). In macrophages and monocytes (as part of the data generated by the Cardiogenics Consortium), the rs342293 proxy SNP rs342275 was used for the eQTL analysis. For LCLs, adipose and skin (as part of the data generated by the MuTHER Consortium), the proxy SNPs rs342296 and rs342275 were used for analysis. Values for r^2 were obtained from Phase II HapMap, CEU.

Cell type/tissue	SNP tested			eQTL nominal *P*-value	SNP with the strongest association with *PIK3CG* expression in 1-Mb window		
	ID	*r^2*	*Distance*		*ID*	*r^2 with rs342293*	*eQTL nominal P-value*
Platelets	rs342293	1.000	0 bp	0.0542	rs342293	1.000	–
Macrophages	rs342275	0.935	13,003 bp	0.0018	rs342275	0.935	–
Monocytes	rs342275	0.935	13,003 bp	0.4348	rs10953522	0.002	0.0005
LCLs	rs342296	1.000	684 bp	0.5983	rs7788626	0.032	0.0018
	rs342275	0.935	13,003 bp	0.9999	–	–	–
Adipose	rs342296	1.000	684 bp	0.5308	rs849375	0.059	0.0022
	rs342275	0.935	13,003 bp	0.6917	–	–	–
Skin	rs342296	1.000	684 bp	0.2091	rs13246564	0.045	0.0006
	rs342275	0.935	13,003 bp	0.2537	–	–	–

Table 8-7. Functional ontology classification of differentially expressed genes between *Pik3cg*$^{-/-}$ and wild type mice.

#	GO term	Biological process	*P*-value	Sample frequency	Background frequency	Genes
1	0009987	Cellular process	3.86×10^{-16}	123/187 (65.8%)	11382/33954 (33.5%)	*Tsc1, Rgs10, Tmsb4x, Cst3, Ybx1, Psmd4, Ywhah, Snx15, Fech, Myo6, Map2k2, Acp1, Hist2h2ac, Smox, Atpif1, Ccng2, Gng11, Hist1h1c, Cdkn2c, Glrx5, Bicd2, Clic4, Cela1, C3, Lyz2, Cul4a, Gpx4, Rnf10, Cd81, Sh3bgrl3, Ifit2, Csda, Atp2a3, Car2, Cap1, Bcl2l1, Stx7, Fzr1, Skap2, Dstn, Sorl1, Cmas, Ctnna1, Prdx3, Chka, Acsl1, Ilk, Sytl4, Msi2, Alox12, Nptn, Arhgef3, Vwf, Atox1, Birc2, Actb, F2rl2, Serpine2, Rnf11, Dusp23, Slc44a1, Sdpr, Itpr2, Itgb5, Rffl, Ptp4a3, Pabpc1, Ywhaz, Ifi30, Zyx, Mmd, Plp1, St3gal5, Prdx5, Tpi1, Ehd4, Fancl, Lyz1, Sort1, Gnaz, Fis1, Litaf, Prkar2b, Cux1, Treml1, Lgals3bp, Gp9, Nusap1, Fhl1, F5, Pros1, Ranbp10, Emb, P2ry12, Urod, Gnas, St6galnac2, Itga6, Cdc42ep5, Cd9, Trim10, Arf5, Gp1bb, Snx3, Pygb, Mylk, Dap, Bin1, Epb4.1, Stx11, Spnb1, Ptpn11, Agtrap, Slc2a3, G3bp2, Mast2, Epb4.9, Plek, Ndrg1, Gp5, Pnpo, E2f2, Vcl*
2	0065008	Regulation of biological quality	2.64×10^{-14}	41/187 (21.9%)	1503/33954 (4.4%)	*Tsc1, Tmsb4x, Fech, Myo6, Glrx5, C3, Rnf10, Cd81, Sh3bgrl3, Car2, Dstn, Prdx3, Ilk, Sytl4, Alox12, Nptn, Gp6, Vwf, Atox1, F2rl2, Serpine2, Ywhaz, Ifi30, Plp1, Prdx5, Treml1, Gp9, F5, Pros1, P2ry12, Cdc42ep5, Cd9, Trim10, Gp1bb, Epb4.1, Spnb1, Ptpn11, Agtrap, Epb4.9, Plek, Gp5*
3	0007596	Blood coagulation	7.84×10^{-12}	13/187 (7.0%)	91/33954 (0.3%)	*C3, Gp6, Vwf, F2rl2, Serpine2, Treml1, Gp9, F5, Pros1, P2ry12, Gp1bb, Plek, Gp5*
4	0007599	Haemostasis	9.09×10^{-12}	13/187 (7.0%)	92/33954 (0.3%)	*C3, Gp6, Vwf, F2rl2, Serpine2, Treml1, Gp9, F5, Pros1, P2ry12, Gp1bb, Plek, Gp5*
5	0050817	Coagulation	1.21×10^{-11}	13/187 (7.0%)	94/33954 (0.3%)	*C3, Gp6, Vwf, F2rl2, Serpine2, Treml1, Gp9, F5, Pros1, P2ry12, Gp1bb, Plek, Gp5*

6	0065007	Biological regulation	3.71x10^{-10}	84/187 (44.9%)	7128/33954 (21.0%)	*Tsc1, Rgs10, Tmsb4x, Cst3, Ybx1, Psmd4, Ywhah, Fech, Myo6, Map2k2, Atpif1, Ccng2, Gchfr, Gng11, Cdkn2c, Glrx5, Cela1, C3, Cul4a, Gpx4, Rnf10, Cd81, Sh3bgrl3, Csda, Car2, Bcl2l1, Fzr1, Skap2, Dstn, Ctnna1, Prdx3, Ilk, Sytl4, Alox12, Nptn, Gp6, Arhgef3, Vwf, Atox1, Birc2, F2rl2, Serpine2, Sdpr, Itpr2, Ywhaz, Ifi30, Plp1, Prdx5, Ehd4, Fancl, Gnaz, Litaf, Prkar2b, Cux1, Treml1, Lgals3bp, Gp9, Nusap1, F5, Pros1, Ranbp10, P2ry12, Gnas, B2m, Itga6, Cdc42ep5, Cd9, Trim10, Arf5, Gp1bb, Ctla2b, Bin1, Epb4.1, Spnb1, Ptpn11, Agtrap, Slc2a3, G3bp2, Mast2, Epb4.9, Plek, Gp5, E2f2, Vcl*
7	0042060	Wound healing	3.06x10^{-09}	13/187 (7.0%)	143/33954 (0.4%)	*C3, Gp6, Vwf, F2rl2, Serpine2, Treml1, Gp9, F5, Pros1, P2ry12, Gp1bb, Plek, Gp5*
8	0050878	Regulation of body fluid levels	1.01x10^{-08}	13/187 (7.0%)	157/33954 (0.5%)	*C3, Gp6, Vwf, F2rl2, Serpine2, Treml1, Gp9, F5, Pros1, P2ry12, Gp1bb, Plek, Gp5*
9	0071840	Cellular component organisation or biogenesis	3.22x10^{-08}	42/187 (22.5%)	2396/33954 (7.1%)	*Tsc1, Tmsb4x, Ywhah, Fech, Myo6, Hist2h2ac, Ccng2, Gchfr, Hist1h1c, C3, Lyz2, Gpx4, Cd81, Cap1, Bcl2l1, Fzr1, Dstn, Sorl1, Ctnna1, Ilk, Alox12, Nptn, Birc2, Actb, Serpine2, Pf4, Ywhaz, Ehd4, Lyz1, Sort1, Nusap1, Pros1, Ranbp10, Itga6, Cd9, Bin1, Epb4.1, Spnb1, Ptpn11, Epb4.9, Plek, Vcl*
10	0006950	Response to stress	5.03x10^{-08}	31/187 (16.6%)	1371/33954 (4.0%)	*Cst3, Psmd4, Chi3l3, Fech, Cela1, C3, Lyz2, Cul4a, Gpx4, Fzr1, Prdx3, Gp6, Vwf, Atox1, Birc2, F2rl2, Serpine2, Ywhaz, Fancl, Lyz1, Treml1, Gp9, F5, Pros1, P2ry12, B2m, Cdc42ep5, Gp1bb, Ptpn11, Plek, Gp5*
11	0050896	Response to stimulus	5.20x10^{-08}	45/187 (24.1%)	2752/33954 (8.1%)	*Ifi27l2a, Cst3, Ybx1, Psmd4, Chi3l3, Fech, Myo6, S100a8, Cela1, C3, Lyz2, Cul4a, Gpx4, Ifit2, Bcl2l1, Fzr1, Prdx3, Acsl1, Alox12, Gp6, Vwf, Atox1, Birc2, F2rl2, Serpine2, Pf4, Ywhaz, Oasl2, Fancl, Lyz1, Sort1, Prkar2b, Treml1, Gp9, F5, Pros1, P2ry12, Gnas, B2m, Itga6, Cdc42ep5, Gp1bb, Ptpn11, Plek, Gp5*

Table 8-8. Genotype and phenotype information for TAR cases and unaffected parents. Healthy individuals are on dark grey background. Abbreviations: Unkn.: Unknown; het: heterozygous; del: deletion; M: male; F: female.

Unique Case Number (UCN)	TAR diagnosed	Heterozygous 1q21 deletion	1q21 deletion origin	Genotype 5'-UTR SNP	Genotype intronic SNP	Sex	Age (years)	Gestation/delivery (weeks)	BW (g)	Neonatal problems	Lowest platelet count (x10^9/L)	Highest platelet count (x10^9/L)	Upper limb abnormality	Lower limb abnormality	Cardio-vascular abnormality	Cow's milk intolerance
1	Yes	Yes	*De novo*	A/Del	G/Del	M	31	Unkn.	Unkn.	Diarrhoea	110	111	Radii absent or hypoplastic	Yes	No	Unkn.
105 (mother of 1)	No	No		G/G	G/G											
106 (mother of 1)	No	No		G/A	G/G											
2	Yes	Yes	*De novo*	A/Del	G/Del	F	6 months	41	3,549	Bruising	7	20	Radii absent or hypoplastic	Yes	No	Unkn.
109 (mother of 2)	No	No		G/G	G/G											
110 (father of 2)	No	No		G/A	G/G											
3	Yes	Yes	Maternal	G/Del	C/Del	F	14	Unkn.	3,232	Bruising	90	140	Radii absent or hypoplastic	Yes	Unkn.	Yes
103 (mother of 3)	No	Yes		G/Del	G/Del											
104 (father of 3)	No	No		G/G	G/C											
4	Yes	Yes	Paternal	A/Del	G/Del	F	14	Unkn.	Unkn.	Unkn.	112	Unkn.	Radii absent or hypoplastic	Unkn.	Unkn.	Unkn.
107 (mother of 4)	No	No		G/A	G/G											
108 (father of 4)	No	Yes		G/Del	G/Del											
5	Yes	Yes	Paternal	A/Del	G/Del	F	29	40	3,175	Tube fed	11	78	Radii absent or hypoplastic	No	No	No
111 (mother of 5)	No	No		G/A	G/G											
112 (father of 5)	No	Yes		G/Del	G/Del											
6	Yes	Yes	Paternal	A/Del	G/Del	F	28.5	40	2,900	Unkn.	101	142	Radii absent or hypoplastic	Yes	No	Yes
81 (mother of 6)	No	No		A/A	G/G											
82 (father of 6)	No	Yes		G/Del	G/Del											
7	Yes	Yes	Maternal	A/Del	G/Del	F	22	Unkn.	Unkn.	Unkn.	12	59	Radii absent or hypoplastic	Yes	Yes	No
90 (mother of 7)	No	No		G/A	G/G											
91 (father of 7)	No	Yes		G/Del	G/Del											
8	Yes	Yes	*De novo*	A/Del	G/Del	F	2 days	Unkn.	Unkn.	Unkn.	Unkn.	20	Radii absent or hypoplastic	Yes	No	No

99 (mother of 8)	No	No		A/A	G/G											
100 (father of 8)	No	No		G/A	G/G											
10	Yes	Yes	Paternal	A/Del	G/Del	M	28	40	2,900	Bleeding	8	120	Absence of radius with hypoplasia of humerus and ulna	No	No	Yes
11 (father of 10)	No	Yes		G/G	G/G											
12 (mother of 10)	No	No		G/A	G/G											
13	Yes	Yes	Maternal	A/Del	G/Del	F	5	40	2,670	Phototherapy	37	43	Radii absent or hypoplastic	Yes	No	No
14 (father of 13)	No	No		A/A	G/G											
15 (mother of 13)	No	Yes		G/G	G/G											
16	Yes	Yes	Paternal	A/Del	G/Del	F	15	40	3,150	Bleeding	29	64	Absence of radius, ulna and humerus; hypoplasia of scapula	Yes	No	Yes
17 (father of 16)	No	Yes		G/G	G/G											
18 (mother of 16)	No	No		G/A	G/C											
19	Yes	Yes	Unkn.	A/Del	G/Del	Unkn.	23	40	Unkn.	None	26	133	Radii absent or hypoplastic	Yes	No	No
20	Yes	Yes	Unkn.	A/Del	G/Del	Unkn.	23	40	2,750	Petechiae	10	110	Radii absent or hypoplastic	No	No	No
21	Yes	Yes	Unkn.	A/Del	G/Del	Unkn.	26	40	Unkn.	None	20	53	Radii absent or hypoplastic	Yes	No	No
22	Yes	Yes	Unkn.	A/Del	G/Del	Unkn.	27	40	Unkn.	None	10	40	Radii absent or hypoplastic	No	No	Yes
23	Yes	Yes	*De novo*	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
24 (parent of 23)	No	No		G/G	G/G											
25 (parent of 23)	No	No		G/A	G/G											
33	Yes	Het frameshift insertion chr1:145,508,476 (T/TAGCG)	n/a	G/A	G/G	M	29	41	3,110	Petechiae	13	Unkn.	Unkn.	Unkn.	Unkn.	Yes
31 (parent of 33)	No	Het frameshift insertion chr1:145,508,476 (T/TAGCG)		G/G	G/G											
32 (parent of 33)	No	No		G/A	G/G											
40	Yes	Yes	Parent	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
41	Yes	Yes	Parent	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
38 (parent 1 of 40 and 41)	No	Yes		G/Del	G/Del											
39 (parent 2 of 40 and 41)	No	No		G/A	G/G											
42	Yes	Yes	Maternal	G/Del	C/Del	F	17	40	2,750	Unkn.	9	58	Radii absent or hypoplastic	No	No	Yes
43 (mother of 42)	No	Yes		G/G	G/G											
44 (father of 42)	No	No		G/G	G/C											

47	Yes	Yes	Maternal	A/Del	G/Del	M	4 months	40	3,450	Unkn.	30	233	Radii absent or hypoplastic	Yes	No	No
48 (mother of 47)	No	Yes		G/Del	G/Del											
49 (father of 47)	No	No		G/A	G/Del											
50	Yes	Yes	Maternal	A/Del	G/Del	M	26	Unkn.	Unkn.	Unkn.	Unkn.	163	Radii absent or hypoplastic	Yes	No	No
51 (mother of 50)	No	Yes		G/Del	G/Del											
52 (father of 50)	No	No		G/A	G/G											
53	Yes	Yes	Paternal	A/Del	G/Del	F	1	Unkn.	2,610	None	10	28	Radii absent or hypoplastic	Yes	No	No
54 (mother of 53)	No	No		G/A	G/G											
55 (father of 53)	No	Yes		G/Del	G/Del											
56	Yes	Yes	*De novo*	A/Del	G/Del	F	34	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Radii absent or hypoplastic	Yes	No	No
57 (mother of 56)	No	No		G/G	G/G											
58 (father of 56)	No	No		G/A	G/G											
59	Yes	Yes	*De novo*	A/Del	G/Del	M	26	Unkn.	2,600	Unkn.	10	200	Radii absent or hypoplastic	Yes	Unkn.	Unkn.
60 (mother of 59)	No	No		G/G	G/G											
61 (father of 59)	No	No		G/A	G/G											
64	Yes	Yes	Unkn.	G/Del	C/Del	M	23	Unkn.	Unkn.	Unkn.	94	155	Radii absent or hypoplastic	Yes	Unkn.	Unkn.
65	Yes	Yes	Maternal	A/Del	G/Del	F	34	Unkn.	Unkn.	Unkn.	79	142	Radii absent or hypoplastic	Unkn.	Unkn.	Unkn.
66 (mother of 65)	No	Yes		G/Del	G/Del											
67 (father of 65)	No	No		G/A	G/G											
68	Yes	Yes	Maternal	G/Del	C/Del	M	1.5	Unkn.	Unkn.	Unkn.	79	169	Radii absent or hypoplastic	Unkn.	Unkn.	Unkn.
69 (mother of 68)	No	Yes		G/Del	G/Del											
70	Yes	Yes	Maternal	G/Del	C/Del	F	18	38	2,600	None	34	154	Radii absent or hypoplastic	Yes	No	Unkn.
71	Yes	Yes	Maternal	G/Del	C/Del	F	6 months	39	3,510	Unkn.	30	200	Radii absent or hypoplastic	Yes	Unkn.	Unkn.
72 (mother of 70 and 71)	No	Yes		G/Del	G/Del											
73 (father of 70 and 71)	No	No		G/G	G/C											
74	Yes	Yes	Unkn.	G/Del	C/Del	M	39	Unkn.	Unkn.	Unkn.	79	169	Radii absent or hypoplastic	Yes	No	No
75 (mother of 74)	No	Yes		G/Del	G/Del											
76	Yes	Yes	Maternal	G/Del	C/Del	M	2	40	3,220	Bleeding	8	130	Radii absent or hypoplastic	Yes	No	Unkn.
77 (mother of 76)	No	Yes		G/Del	G/Del											
78 (father of 76)	No	No		G/G	G/C											
83	Yes	Yes	Non-maternal	A/Del	G/Del	F	37	Unkn.	Unkn.	None	74	136	Radii absent or hypoplastic	Yes	Unkn.	Unkn.
84 (mother 83)	No	No		G/G	G/G											

85	Yes	Yes	*De novo*	A/Del	G/Del	F	17 days	37	2,800	Unkn.	Unkn.	167	Radii absent or hypoplastic	No	Yes	No
87 (father of 85)	No	No		G/A	G/G											
90 (mother of 85)	No	No		G/A	G/G											
88	Yes	Yes	Unkn.	A/Del	G/Del	F	4	Unkn.	2,720	Unkn.	Unkn.	34	Radii absent or hypoplastic	No	Unkn.	Unkn.
89	Yes	Yes	Unkn.	G/Del	C/Del	M	8	Unkn.	Unkn.	Unkn.	Unkn.	88	Radii absent or hypoplastic	Yes	Unkn.	Unkn.
92	Yes	Yes	*De novo*	A/Del	G/Del	M	1.5	Unkn.	Unkn.	Unkn.	7	65	Radii absent or hypoplastic	Unkn.	Unkn.	Unkn.
93 (mother of 92)	No	No		G/G	G/G											
94 (father of 92)	No	No		G/A	G/G											
95	Yes	Yes	Maternal	A/Del	G/Del	M	6	39	2,900	Unkn.	18	180	Radii absent or hypoplastic	Yes	Yes	Unkn.
96 (mother of 95)	No	Yes		G/Del	G/Del											
97 (father of 95)	No	No		G/A	G/G											
98	Yes	Yes	Unkn.	A/Del	G/Del	F	4	40	2,910	None	7	295	Radii absent or hypoplastic	No	No	Yes
101	Yes	Yes	Unkn.	A/Del	G/Del	F	17	Unkn.	Unkn.	Unkn.	31	91	Radii absent or hypoplastic	Yes	Unkn.	Unkn.
102 (mother of 101)	No	Yes		G/Del	G/Del											
113	Yes	Yes	Unkn.	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
114	Yes	Yes	*De novo*	G/Del	C/Del	Unkn.	8	42	2,960	Bleeding	Unkn.	43	Radii absent or hypoplastic	Yes	No	No
115 (parent of 114)	No	No		G/G	G/G											
131 (parent of 114)	No	No		G/G	G/C											
116	Yes	Yes	Maternal	A/Del	G/Del	F	8	40	3,062	Bleeding	12	91	Radii absent or hypoplastic	Yes	No	No
117 (father of 116)	No	No		G/A	G/G											
118 (mother of 116)	No	Yes		G/Del	G/Del											
121	Yes	Yes	Maternal	G/Del	G/Del	F	36	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Radii absent or hypoplastic	No	No	Yes
145 (mother of 121)	Yes	Yes	Unkn.	G/Del	G/Del	M	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
120 (father of 121)	No	No		G/G	G/G											
122	Yes	Yes	Unkn.	A/Del	G/Del	M	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
123	Yes	Yes	Unkn.	G/Del	C/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
124	Yes	Yes	Unkn.	A/Del	G/Del	F	22	Unkn.	Unkn.	Bruising	Unkn.	Unkn.	Radii absent or hypoplastic	Yes	No	Yes
125	Yes	Yes	Paternal	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
126 (father of 125)	No	Yes		G/Del	G/Del											
127 (mother of 125)	No	No		A/G	G/G											
128	Yes	Yes	Unkn.	G/Del	C/Del	F	8	41	3,544	Bleeding	11	178	Radii absent or hypoplastic	No	No	Yes
129 (parent of 128)	No	No		G/G	G/C											

134	Yes	Yes	Maternal	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
132 (mother of 134)	No	Yes		G/Del	G/Del											
133 (father of 132)	No	No		G/A	G/G											
136	Yes	Yes	Non-paternal	A/Del	G/Del	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.	Unkn.
135 (father of 136)	No	No		G/G	G/G											
139	Yes	Yes	Unkn.	A/Del	G/Del	F	17	42	3,459	Bleeding	20	55	Radii absent or hypoplastic	No	No	Unkn.
138 (parent of 138)	No	No		G/A	G/G											
140	Yes	Het gain of stop codon chr1:145,509,173 (C/T)	n/a	G/A	G/C	F	9	Unkn.	Unkn.	Bleeding	Unkn.	Unkn.	Unkn.	Unkn.	Yes	Yes
142	Yes	Yes	Paternal	A/Del	G/Del	M	16	40	Unkn.	Bleeding	Unkn.	78	Radii absent or hypoplastic	Yes	No	No
143 (mother of 142)	No	No		G/A	G/G											
144 (father of 142)	No	Yes		G/Del	G/Del											