

**Relapse of acute myeloid leukemia after allogeneic stem cell transplantation is associated with gain of *WT1* alterations and high mutation load**

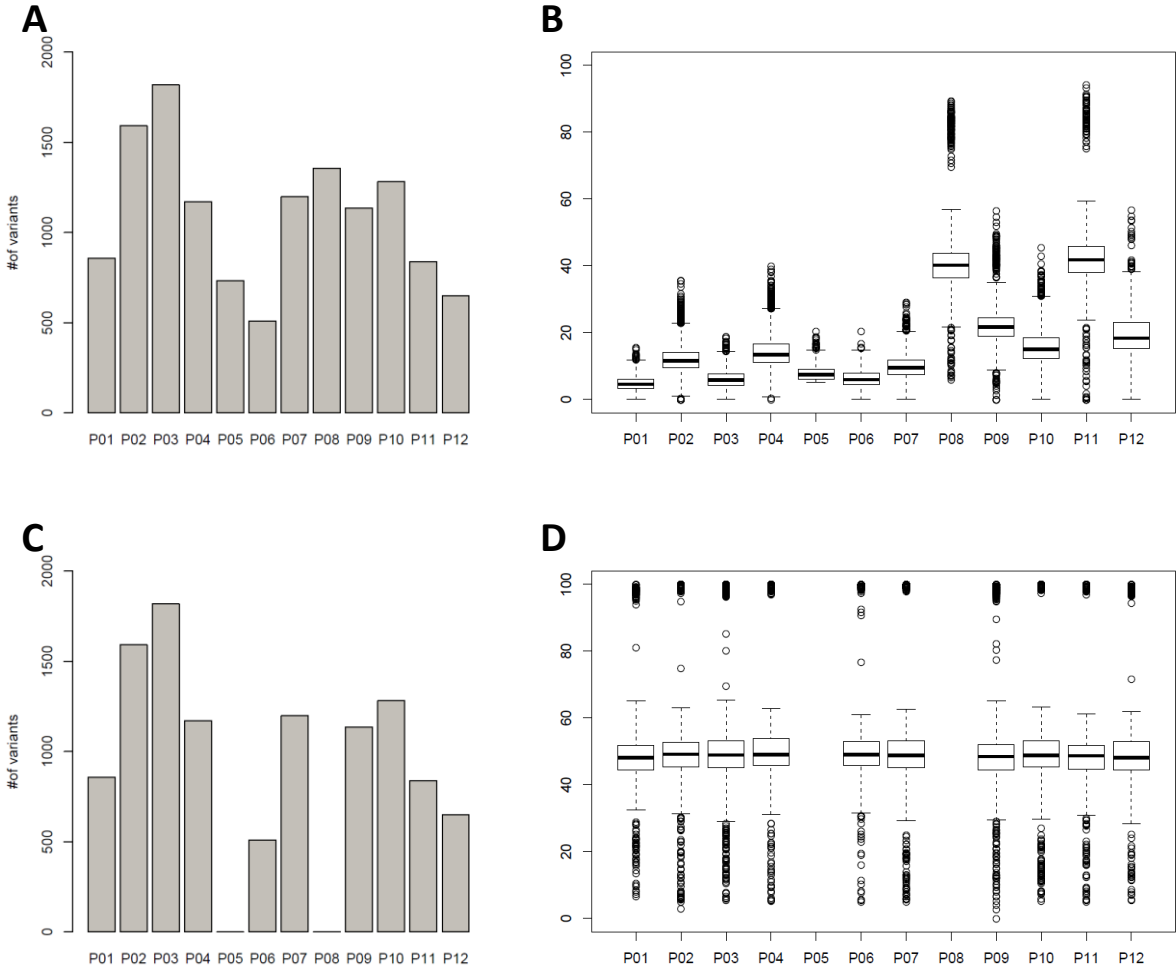
Sebastian Vosberg,<sup>1,2,3,4</sup> Luise Hartmann,<sup>1,2,3,4</sup> Klaus H. Metzeler,<sup>1,2,3,4</sup> Nikola P. Konstandin,<sup>4</sup> Stephanie Schneider,<sup>4,5</sup> Ashok Varadharajan,<sup>6</sup> Andreas Hauser,<sup>6</sup> Stefan Krebs,<sup>6</sup> Helmut Blum,<sup>6</sup> Stefan K. Bohlander,<sup>7</sup> Wolfgang Hiddemann,<sup>1,2,3,4</sup> Johanna Tischer,<sup>8</sup> Karsten Speikermann<sup>1,2,3,4</sup> and Philipp A. Greif<sup>1,2,3,4</sup>

<sup>1</sup>Experimental Leukemia and Lymphoma Research, Department of Medicine III, University Hospital, LMU Munich, Germany; <sup>2</sup>German Cancer Consortium (DKTK), partner site Munich, Germany; <sup>3</sup>German Cancer Research Center (DKFZ), Heidelberg, Germany; <sup>4</sup>Laboratory for Leukemia Diagnostics, Department of Medicine III, University Hospital, LMU Munich, Germany; <sup>5</sup>Institute of Human Genetics, University Hospital, LMU Munich, Germany; <sup>6</sup>Laboratory for Functional Genome Analysis, Gene Center, LMU Munich, Germany; <sup>7</sup>Leukaemia and Blood Cancer Research Unit, Department of Molecular Medicine and Pathology, The University of Auckland, New Zealand and <sup>8</sup>Hematopoietic Stem Cell Transplantation, Department of Medicine III, University Hospital, LMU Munich, Germany

Correspondence: [pgreif@med.uni-muenchen.de](mailto:pgreif@med.uni-muenchen.de)  
doi:10.3324/haematol.2018.193102

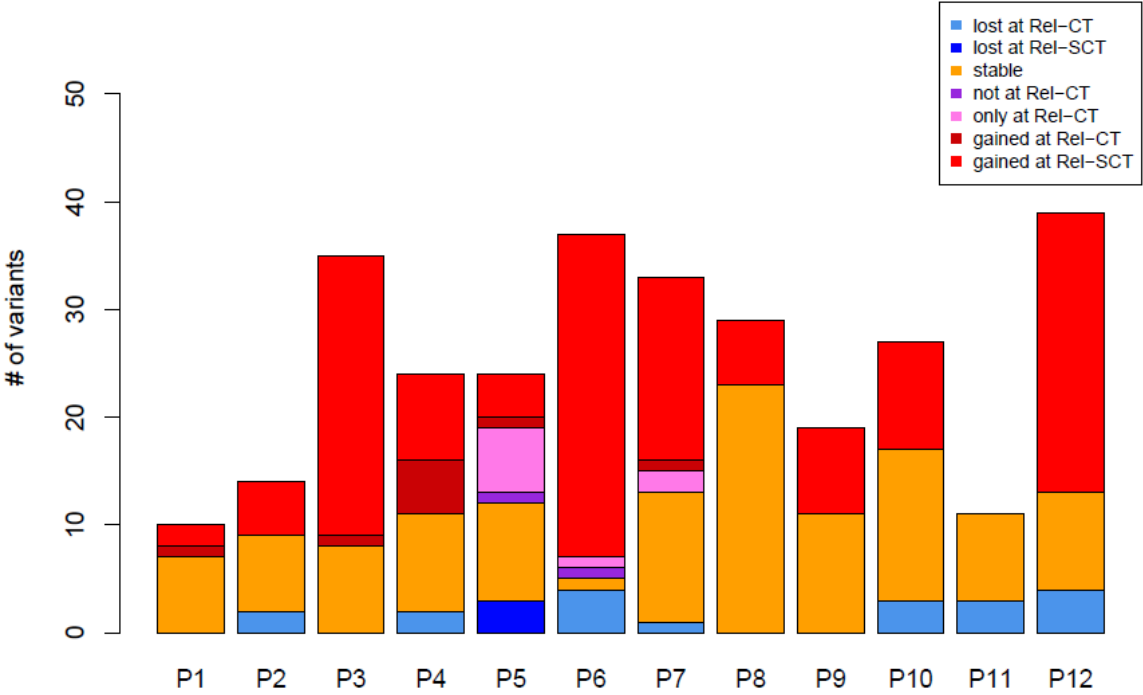
Supplementary Figure 1:

Variant allele frequencies of variants in samples after SCT likely representing donor germline SNPs. Variants were selected if present in samples after SCT but not prior to SCT (VAF=0%) at a minimum coverage of 100x in all samples, limited to common SNP positions (dbSNP138, MAF  $\geq 1\%$ ). (A) Number of variants detected in Rel-SCT samples (n=12). (B) VAFs detected in Rel-SCT samples. (C) Number of variants detected in CR-SCT samples (n=10). (D) VAFs detected in CR-SCT samples. SCT: stem cell transplantation; SNP: single nucleotide polymorphism; VAF: variant allele frequency; MAF: minor allele frequency; CR-SCT: complete remission after stem cell transplantation; Rel-SCT: relapse after stem cell transplantation.



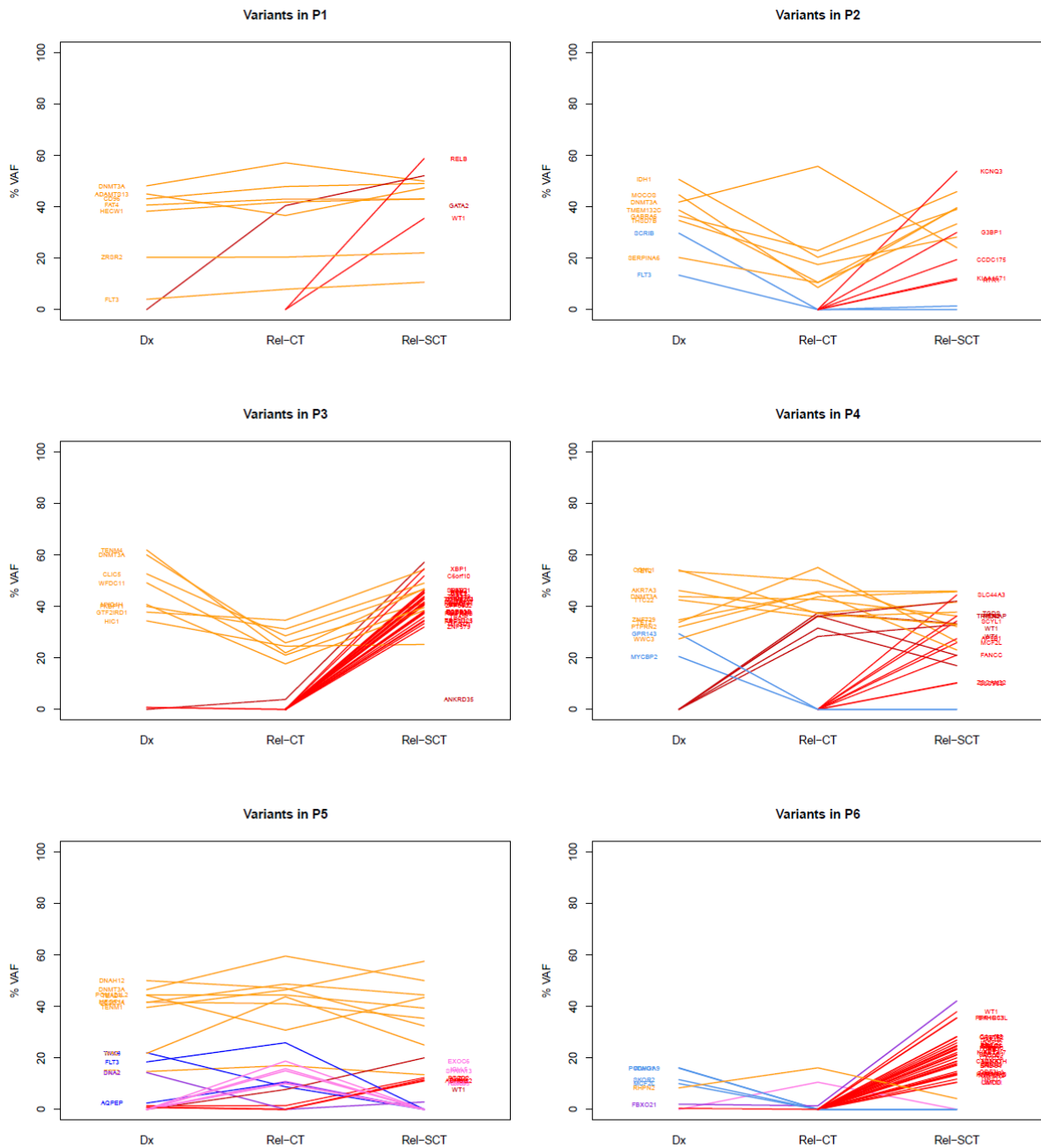
Supplementary Figure 2:

Number of variants detected in patients at individual time points.

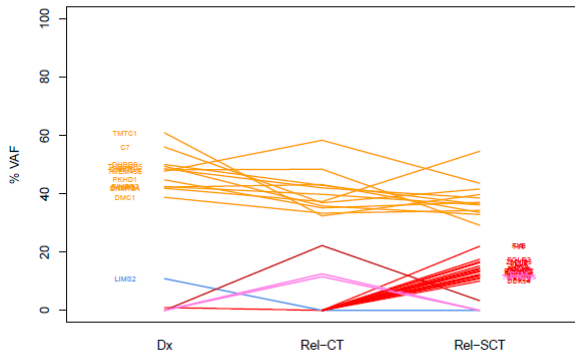


Supplementary Figure 3:

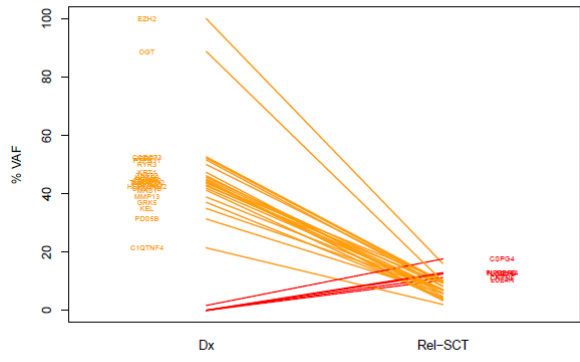
Clonal evolution of individual patients. Each mutation is displayed by a single line, colored according to its detection at individual time points. Orange: stable mutation; light blue: mutation lost after Dx; dark blue: mutation lost after Rel-CT; dark red: mutation gained at Rel-CT and stable at Rel-SCT; light red: mutation gained at Rel-SCT; light purple: mutation detected at Rel-CT only; dark purple: mutation detected at Dx and Rel-SCT, not at Rel-CT. Dx: diagnosis; Rel-CT: relapse after chemotherapy, before SCT; Rel-SCT: relapse after stem cell transplantation.



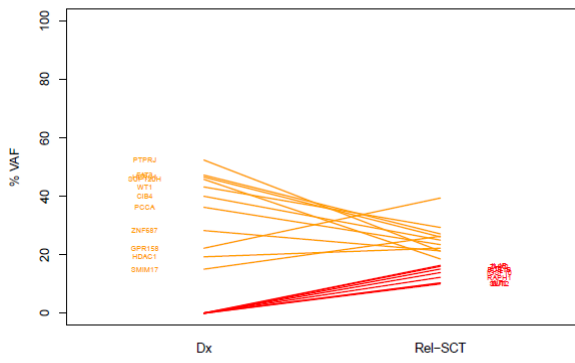
Variants in P7



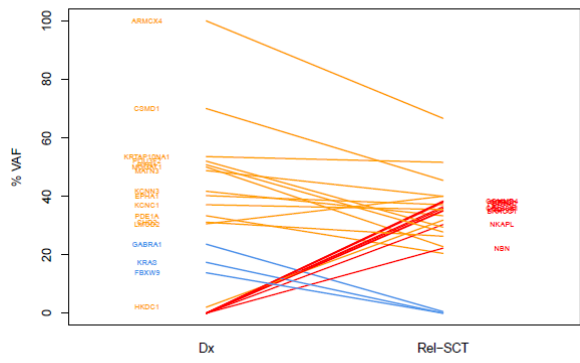
Variants in P8



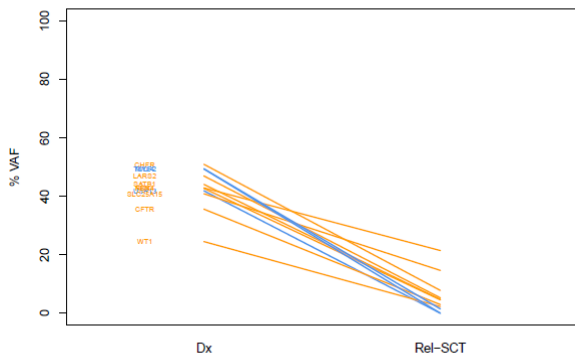
Variants in P9



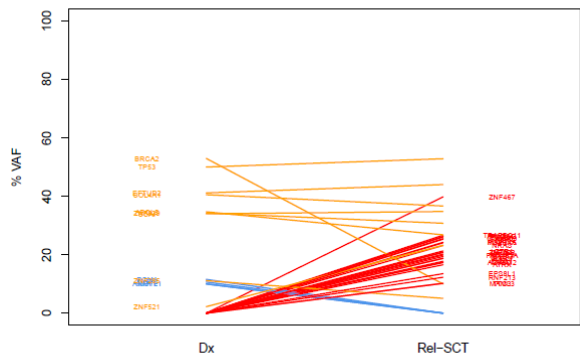
Variants in P10



Variants in P11



Variants in P12



Supplementary Table 1: Sequencing metrics.

UPN	Metrics	Dx	CR-CT	Rel-CT	CR-SCT	Rel-SCT
P1	# of total reads:	49,679,754	50,894,617	53,502,514	41,471,412	49,615,172
	mean target coverage:	104.58	106.82	112.14	85.14	103.93
	% of target sequenced, min 10x coverage:	96.14	96.09	96.14	95.96	95.99
	% of target sequenced, min 20x coverage:	93.47	93.54	93.76	93.43	93.31
	% of target sequenced, min 30x coverage:	89.30	89.55	90.16	88.66	89.14
P2	# of total reads:	52,125,219	53,503,834	58,461,678	49,445,123	53,911,707
	mean target coverage:	109.4	112.43	122.72	101.19	113.17
	% of target sequenced, min 10x coverage:	96.2	96.25	96.33	96.38	96.12
	% of target sequenced, min 20x coverage:	93.8	93.94	94.3	94.44	93.59
	% of target sequenced, min 30x coverage:	90.09	90.33	91.26	91.01	89.88
P3	# of total reads:	51,731,979	53,163,757	48,202,631	44,019,522	55,541,907
	mean target coverage:	108.71	111.57	101.4	90.19	116.69
	% of target sequenced, min 10x coverage:	96.07	96.18	95.97	96.21	96.34
	% of target sequenced, min 20x coverage:	93.53	93.78	93.07	93.79	94.1
	% of target sequenced, min 30x coverage:	89.62	90.14	88.56	89.47	90.7
P4	# of total reads:	44,377,025	48,026,764	37,743,180	45,009,741	47,944,302
	mean target coverage:	93.43	100.85	79.47	92.09	100.44
	% of target sequenced, min 10x coverage:	95.76	95.97	95.24	96.18	96.11
	% of target sequenced, min 20x coverage:	92.36	93.09	90.63	93.71	93.22
	% of target sequenced, min 30x coverage:	87.17	88.62	83.71	89.25	88.73
P5	# of total reads:	45,017,694	49,125,597	39,707,479	-	41,272,379
	mean target coverage:	94.56	102.94	83.29	-	86.47
	% of target sequenced, min 10x coverage:	95.05	95.55	94.74	-	94.87
	% of target sequenced, min 20x coverage:	91.4	92.53	90.3	-	90.69
	% of target sequenced, min 30x coverage:	86.2	88.18	83.95	-	84.74
P6	# of total reads:	46,055,892	44,931,714	37,969,394	46,409,837	39,498,278
	mean target coverage:	96.41	94.16	79.59	95.13	82.82
	% of target sequenced, min 10x coverage:	95.41	95.24	94.38	96.42	94.81
	% of target sequenced, min 20x coverage:	92.08	91.77	89.46	94.29	90.39
	% of target sequenced, min 30x coverage:	87.21	86.69	82.44	90.34	83.99
P7	# of total reads:	47,386,836	38,720,124	46,504,876	42,064,576	53,096,598
	mean target coverage:	99.49	81.43	97.63	86.06	111.18
	% of target sequenced, min 10x coverage:	95.95	95.27	95.78	95.87	96.23
	% of target sequenced, min 20x coverage:	93.01	90.88	92.62	92.96	93.78
	% of target sequenced, min 30x coverage:	88.45	84.24	87.84	87.78	90.09
P8	# of total reads:	53,309,663	50,191,545	-	-	49,257,003
	mean target coverage:	111.92	105.51	-	-	103.16
	% of target sequenced, min 10x coverage:	96.2	96.15	-	-	96.1
	% of target sequenced, min 20x coverage:	93.56	93.35	-	-	93.33
	% of target sequenced, min 30x coverage:	89.57	89.08	-	-	89.09
P9	# of total reads:	51,233,864	52,010,693	-	44,026,676	59,927,805
	mean target coverage:	107.66	109.47	-	90.18	125.68
	% of target sequenced, min 10x coverage:	95.99	96.16	-	96.29	96.56
	% of target sequenced, min 20x coverage:	93.34	93.64	-	93.88	94.65
	% of target sequenced, min 30x coverage:	89.32	89.78	-	89.43	91.72
P10	# of total reads:	37,705,107	51,512,450	-	45,442,141	39,938,239
	mean target coverage:	78.9	108.13	-	92.98	83.97
	% of target sequenced, min 10x coverage:	95.29	96.18	-	96.3	95.49
	% of target sequenced, min 20x coverage:	90.54	93.5	-	94.01	91.12
	% of target sequenced, min 30x coverage:	83.42	89.44	-	89.84	84.59
P11	# of total reads:	56,070,483	60,362,007	-	43,660,554	44,520,071
	mean target coverage:	117.43	126.49	-	89.37	93.4
	% of target sequenced, min 10x coverage:	96.09	96.45	-	96.21	95.72
	% of target sequenced, min 20x coverage:	93.6	94.44	-	93.75	92.23
	% of target sequenced, min 30x coverage:	89.96	91.42	-	89.2	86.94
P12	# of total reads:	46,344,724	31,587,528	-	40,279,295	62,086,705
	mean target coverage:	97.31	66.48	-	82.6	130.13

	% of target sequenced, min 10x coverage:	95.9	94.59	-	95.95	96.47
	% of target sequenced, min 20x coverage:	92.7	88.22	-	93.1	94.58
	% of target sequenced, min 30x coverage:	87.77	78.96	-	87.83	91.73

Supplementary Table 2: Patient characteristics.

UPN	Age	Gender	Donor	HLA setting	Rel-CT available	CR-SCT available	Karyotype at Diagnosis	Karyotype at Rel-CT	Karyotype at Rel-SCT
P01	66	female	related	identical	1	1	46,XX[25]	NA	46,XX[25]
P02	37	female	related	haploidentical	1	1	46,XX[25]	NA	NA
P03	58	female	related	identical	1	1	46,XX[20]	NA	46,XY[2]/ 46,XX,del(9)(q21q22) [23]
P04	61	female	related	identical	1	1	46,XX[20]	NA	NA
P05	65	female	unrelated	HLA-A mismatch	1	0	46,XX[20]	46,XX[20]	NA
P06	22	female	unrelated	identical	1	1	46,XX[20]	NA	46,XX,-6,+8[3]/46,XX[8]/ 46,XY[8]
P07	73	female	unrelated	identical	1	1	46,XX[20]	46,XX[25]	46,X,?del(X)(q11),der(3)[4]/ 46,XX[13]/46,XY[8]
P08	26	male	unrelated	identical	0	0	46,XY[16]	-	NA
P09	44	female	related	HLA-A mismatch	0	1	46,XX[25]	-	NA
P10	21	male	unrelated	identical	0	1	46,XY,t(9;11)(p11;q23)[17]	-	46,XY,t(9;11)(p22;q23)[4]/ 47,sl,+21[17]/46,XY[4]
P11	26	male	related	identical	0	1	47,XY,+8[18]/46,XY[7]	-	NA
P12	51	female	double cord blood	HLA-B/HLA-C mismatch	0	1	40-44,XX,der(4)t(4;7)-7, der(16)t(16;17), t(17;21),t(7;16),t(7;20),-7, der(20)t(20;21), t(21,21)(q10;q10),+21,der(22)t(16;22),+mar[cp4]/79-83,XXXX,der(4),t(4;7)x2, del(5)(q12),-7,-7,-8,-8, der(12)t(8;12),der(16)t(16;17), t(17;21),t(7;16), t(7;20)x2,-17,-17,der(20)t(20;21)x2, t(21,21)(q10;q10),-22,-22,der(22)t(16;22)[cp4]	-	43,X,der(X;14)(q10;q10), der(2)t(2;8),der(4;7)(p10;p10), del(5)(q1?5q3?3), der(16;17)(q10;p10), t(16;20)(q?23;?), dic(20;21)(p1?1;q2?2), +21,der(21)t(2;21), der(22)t(16;22)(?;q?)[cp5]



Supplementary Table 3: Variants detected by exome sequencing.

UPN	Chr	Position	Gene	Reference allele	Variant allele	VAF Dx	VAF CR-CT	VAF Rel-CT	VAF Rel-SCT	Transcript ID	Protein change
P1	chr2	25457242	DNMT3A	C	G	48.15	8.93	57.14	50	NM_022552.4	p.R882P
P1	chr3	111264108	CD96	G	A	43.08	-	47.89	49.09	NM_198196.2	p.E93K
P1	chr3	128200665	GATA2	G	C	0	-	40.45	52.11	NM_032638.4	p.H380Q
P1	chr4	126336423	FAT4	T	C	40.67	-	42.99	42.99	NM_001291303.1	p.I2102T
P1	chr7	43400530	HECW1	C	T	38.24	-	41.84	43.06	NM_015052.4	p.A169V
P1	chr9	136298496	ADAMTS13	A	G	45	-	36.59	47.37	NM_139025.4	splice_site
P1	chr11	32417910	WT1	G	T	0	-	0	35.48	NM_000378.4	p.S364*
P1	chr13	28608287	FLT3	AA	TT	4	1.54	7.89	10.64	NM_004119.2	p.F590Y
P1	chr19	45515534	RELB	G	A	0	-	0	58.82	NM_006509.3	p.S167N
P1	chrX	15841230	ZRSR2	C	CAGCCGG	20.29	4.76	20.41	22.06	NM_005089.3	p.G438GAG
P2	chr2	25457242	DNMT3A	C	T	41.82	42.62	55.77	24.07	NM_022552.4	p.R882H
P2	chr2	137814382	THSD7B	C	T	34.67	0.67	17.52	28.18	NM_001080427.1	p.R147W
P2	chr2	209113112	IDH1	C	T	50.68	-	20.34	38.98	NM_005896.3	p.R132H
P2	chr5	151170514	G3BP1	A	G	0	-	0	30	NM_005754.2	p.D81G
P2	chr5	161115980	GABRA6	G	A	36.47	-	22.92	45.88	NM_000811.2	p.R84H
P2	chr8	133152333	KCNQ3	G	A	0	-	0	53.85	NM_004519.3	p.R520*
P2	chr8	144873606	SCRIB	G	A	29.73	-	0	1.41	NM_182706.4	p.P1592L
P2	chr12	129180607	TMEM132C	C	T	38.71	-	10.59	33.33	NM_001136103.2	p.R630W
P2	chr13	28592642	FLT3	C	A	13.39	-	0	0	NM_004119.2	p.D835Y
P2	chr14	60027870	CCDC175	A	C	0	-	0	19.44	NM_001164399.1	p.L307R
P2	chr14	94776185	SERPINA6	C	T	20.25	0.55	10.48	39.44	NM_001756.3	p.V258M
P2	chr18	33795497	MOCOS	G	T	44.68	-	8.57	39.62	NM_017947.2	p.D452Y
P2	chr19	39058540	RYR1	C	A	0	-	0	11.54	NM_000540.2	p.Q4548K
P2	chr22	25424543	KIAA1671	C	A	0	-	0	12.07	NM_001145206.1	p.L193I
P3	chr1	145567026	ANKRD35	C	G	0	-	3.85	57.14	NM_144698.4	splice_site
P3	chr2	25457242	DNMT3A	C	T	60	8.77	25.86	41.27	NM_022552.4	p.R882H
P3	chr2	43452252	ZFP36L2	CG	C	0	-	0	40.43	NM_006887.4	p.G231fs
P3	chr3	28454565	ZCWPW2	T	G	0	-	0	42.86	NM_001040432.1	p.D2E
P3	chr3	49877233	TRAIIP	C	G	0	-	0	35.80	NM_005879.2	p.E286Q
P3	chr4	85674945	WDFY3	C	A	0	-	0	41.28	NM_014991.4	p.V1882L
P3	chr5	140188709	KCDHA4	G	A	0	-	0	37.80	NM_018907.3	p.R646H
P3	chr5	141309800	KIAA0141	C	T	0	-	0	41.25	NM_014773.4	p.L239F
P3	chr5	179020562	RUFY1	C	G	0.74	-	0	40.89	NM_025158.4	p.H443Q
P3	chr6	32306780	C6orf10	A	C	0	-	0	51.83	NM_006781.4	splice_site
P3	chr6	45881972	CLIC5	A	G	52.63	-	28.57	46.67	NM_001114086.1	p.V353A
P3	chr7	14027725	ETV1	G	A	0	-	0	45.11	NM_004956.4	p.A40V
P3	chr7	18067240	PRPS1L1	C	T	0	-	0	34.25	NM_175886.2	p.V56I
P3	chr7	73922436	GTF2IRD1	A	A	37.78	-	34.62	54.26	NM_001199207.1	p.D9G
P3	chr8	139263186	FAM135B	C	T	0	-	0	37.21	NM_015912.3	p.G147D
P3	chr8	145095366	SPATC1	T	A	0.76	-	0	46.20	NM_198572.2	splice_site
P3	chr10	64574229	EGR2	C	T	0	-	0	46.27	NM_000399.3	splice_site
P3	chr11	78516436	TENM4	C	T	61.90	-	21.95	46.88	NM_001098816.2	p.A694T
P3	chr12	49317616	FKBP11	T	C	40	-	31.21	49.02	NM_016594.2	p.I113V
P3	chr12	109880060	MYO1H	A	G	40.74	-	17.65	38.46	NM_001101421.3	p.S875G
P3	chr16	1584561	TMEM204	G	T	0	-	0	42.98	NM_001256541.1	splice_site
P3	chr16	58030857	ZNF319	T	G	0	-	0	43.90	NM_020807.1	p.K438T
P3	chr17	1961810	HIC1	T	TG	34.38	-	24.55	25.23	NM_001098202.1	p.L628fs
P3	chr17	27185807	ERAL1	T	G	0	-	0	40.80	NM_005702.2	p.S309A
P3	chr17	28656416	TMIGD1	C	A	0	-	0	33.33	NM_206832.1	p.G72W
P3	chr18	44560618	TCEB3B	G	A	0	-	0	38.18	NM_016427.2	p.L340F
P3	chr19	4703991	DPP9	C	T	0	-	0	45.22	NM_139159.4	p.D226N
P3	chr19	56089575	ZNF579	C	G	0	-	0	32	NM_152600.2	p.Q477H
P3	chr20	33450627	GGT7	C	G	0	-	0	43.48	NM_178026.2	p.G183R
P3	chr20	44277898	WFDC11	C	T	49.15	-	21.05	39.57	NM_147197.2	p.V81I
P3	chr21	43414135	ZBTB21	G	A	0	0.83	0	37.59	NM_020727.4	p.R24C
P3	chr22	29195143	XBP1	T	G	0	-	0	54.64	NM_001079539.1	splice_site
P3	chr22	37271990	NCF4	G	A	0	-	0	45.58	NM_013416.3	p.G308D
P3	chrX	48399764	TBC1D25	CG	T	0	-	0	34.62	NM_002536.2	p.V56fs
P3	chrX	144905448	SLITRK2	G	A	0	-	0	41.72	NM_032539.4	p.R502K
P4	chr1	19610518	AKR7A3	C	T	46.15	-	37.50	41.67	NM_012067.2	p.R269Q
P4	chr1	55266728	TTC22	A	G	42.52	-	35.92	37.78	NM_001114108.1	p.S37P
P4	chr1	95310984	SLC44A3	C	A	0	-	0	44.44	NM_001114106.2	p.L346I
P4	chr1	173552686	SLC9C2	C	CT	0	-	0	10.17	NM_178527.3	p.G200fs
P4	chr2	25468120	DNMT3A	AC	A	43.80	32.38	42.70	36.28	NM_022552.4	splice_site
P4	chr4	106197285	TET2	T	C	33.80	24.21	55.10	26.32	NM_001127208.2	p.I1873T
P4	chr4	106197387	TET2	T	A	53.70	32.50	50	32.14	NM_001127208.2	p.M1907K
P4	chr7	157959914	PTPRN2	C	T	32	-	45.74	45.87	NM_002847.3	p.G207R
P4	chr9	97912227	FANCC	C	T	0	-	0	21.05	NM_000136.2	p.A222T
P4	chr11	32417913	WT1	CG	CTC	0	-	31.58	16.96	NM_000378.4	p.R363fs
P4	chr11	32450115	WT1	AG	A	0	-	28.33	32.91	NM_000378.4	p.S233*
P4	chr11	65299069	SCYL1	A	C	0	-	0	34.33	NM_020680.3	p.E344A
P4	chr11	65546787	AP5B1	G	C	0	-	0	27.50	NM_138368.4	p.L393V
P4	chr13	77640211	MYCBP2	T	TCGGG	20.55	-	0	0	NM_015057.4	p.R4156fs
P4	chr13	95230288	TGDS	C	A	0	-	37.50	33.33	NM_014305.2	p.V266F
P4	chr13	113718729	MCF2L	C	T	0	-	0	25.93	NM_001112732.2	p.R201C
P4	chr15	57731260	CGNL1	C	G	54.14	-	37.23	32.98	NM_032866.4	p.Q355E
P4	chr16	3434487	ZSCAN32	G	T	0	-	0	10.34	NM_001284527.1	p.D402E

P4	chr17	8053390	PER1	C	T	0	-	0	36.26	NM_002616.2	p.R143Q
P4	chr19	22498208	ZNF729	A	T	34.78	-	45.24	23.08	NM_001242680.1	p.K663N
P4	chr19	45997994	RTN2	G	T	0	-	36.21	42.03	NM_005619.4	p.Q117K
P4	chr20	33623033	TRPC4AP	G	T	0	-	36.36	20.97	NM_015638.2	p.T315K
P4	chrX	9709460	GPR143	A	G	29.41	-	0	0	NM_000273.2	p.F268S
P4	chrX	10102553	WWC3	C	T	27.38	0.50	43.75	45.71	NM_015691.3	p.R894*
P5	chr1	42671456	FOXJ3	C	T	0	-	10.87	0	NM_014947.4	p.A203T
P5	chr2	25457242	DNMT3A	C	T	46.51	9.80	59.52	50	NM_022552.4	p.R882H
P5	chr3	57489762	DNAH12	C	T	50	-	47.06	32.43	NM_178504.4	p.R356Q
P5	chr3	186338565	AHSG	G	A	0.88	-	0	11.70	NM_001622.2	p.R317H
P5	chr4	15569411	CC2D2A	T	C	41.55	-	48.68	44.44	NM_001080522.2	splice_site
P5	chr4	106194014	TET2	AAAGTCAG CCCCATC	A	14.69	3.23	17	13.45	NM_001127208.2	p.E1492fs
P5	chr5	32091218	PDZD2	G	C	1.39	-	1.43	12.31	NM_178140.2	p.S2555T
P5	chr5	87502320	TMEM161B	TG	T	20.83	-	0	4.76	NM_001289007.1	splice_site
P5	chr5	115346513	AQPEP	G	A	2.44	-	10.53	0	NM_173800.4	p.W723*
P5	chr6	27279949	POM121L2	T	C	44.44	1.28	44.44	39.34	NM_033482.3	start_lost
P5	chr6	32038098	TNXB	G	A	22	-	8.77	0	NM_019105.6.7	p.T1695M
P5	chr10	28906712	WAC	AG	A	21.74	-	43.75	25	NM_016628.4	p.R625fs
P5	chr10	70204683	DNA2	A	T	14.29	-	0	2.86	NM_001080449.2	p.Y405*
P5	chr10	94679782	EXOC6	G	A	0	-	18.75	0	NM_019053.4	p.R288Q
P5	chr10	116085782	AFAP1L2	G	GCCC	0.81	0.63	0	11.02	NM_001287824.1	p.G111GG
P5	chr11	32417923	WT1	T	TCGAGTAA	0	-	7.69	20	NM_000378.4	p.T360fs
P5	chr11	66333175	CTSF	G	A	0	-	10	0	NM_003793.3	p.P338S
P5	chr12	3129938	TEAD4	C	G	44.29	1.37	30.77	43.51	NM_003213.3	p.T241R
P5	chr12	54427103	HOXC5	A	C	0	1.72	10.17	0	NM_018953.3	p.N66T
P5	chr13	24858341	SPATA13	T	C	0	-	15	0	NM_001286792.1	p.Y807H
P5	chr13	28592642	FLT3	C	A	18.42	-	25.88	0	NM_004119.2	p.D835Y
P5	chr15	66420719	MEGF11	A	T	41.67	-	41.07	35.37	NM_032445.2	p.L8H
P5	chr20	5903479	CHGB	A	G	0.77	-	0	11.19	NM_001819.2	p.H230R
P5	chr22	23237641	IGLL5	A	T	0	-	15.79	0	NM_001178126.1	p.I138F
P5	chrX	123615583	TENM1	T	C	39.58	-	46.43	57.50	NM_001163278.1	splice_site
P6	chr1	120478178	NOTCH2	C	A	0	-	0	13.33	NM_024408.3	p.C1191F
P6	chr1	152692739	C1orf68	T	A	0	-	0	28.30	NM_001024679.2	p.C248S
P6	chr1	197411322	CRB1	CCT	GC	0	-	0	23.47	NM_201253.2	p.A1302fs
P6	chr2	46602855	EPAS1	G	T	0	-	0	24.44	NM_001430.4	p.G305C
P6	chr4	94376849	GRID2	C	A	0	-	0	11.76	NM_001510.3	p.P528T
P6	chr5	137088914	HNRNPA0	C	A	0	-	0	13.46	NM_006805.3	p.W281L
P6	chr5	140783150	PCDHGA9	G	A	16.13	1.82	0	0	NM_018921.2	p.V211I
P6	chr5	149459869	CSF1R	T	A	0	-	10.53	0	NM_005211.3	p.Q113L
P6	chr5	167841567	WWC1	G	A	16	-	0	0	NM_001161661.1	p.D386N
P6	chr7	82580030	PCLO	G	T	0	-	0	21.05	NM_033026.5	p.Q3292K
P6	chr8	145057386	PARP10	C	A	0	1.10	0	23.71	NM_032789.3	p.G791C
P6	chr9	35714657	TLN1	C	A	0	-	0	14.06	NM_006289.3	p.V967L
P6	chr10	24813436	KIAA1217	G	A	0	0.82	0	22.09	NM_019590.4	p.G881S
P6	chr11	4843555	ORS1F2	C	G	0	-	0	28.17	NM_001004753.1	p.Q314E
P6	chr11	18732304	IGSF22	C	A	0	-	0	25.84	NM_173588.3	p.A824S
P6	chr11	32414242	WT1	G	T	0	-	0	10.48	NM_000378.4	p.Q420K
P6	chr11	32417910	WT1	G	T	0	-	0	37.93	NM_000378.4	p.S364*
P6	chr12	21628681	RECQL	C	A	0	-	0	24.85	NM_002907.3	p.A343S
P6	chr12	117627032	FBXO21	G	A	2.02	-	1.37	42.11	NM_033624.2	splice_site
P6	chr13	113736760	MCF2L	A	G	10	-	0	0	NM_001112732.2	p.D632G
P6	chr15	76019719	ODF3L1	G	T	0	-	0	14.73	NM_175881.3	p.M221I
P6	chr16	1268628	CACNA1H	C	T	0	-	0	18.75	NM_021098.2	p.T1955I
P6	chr16	16218648	ABCC1	G	T	0	-	0	17.86	NM_004996.3	p.W1198L
P6	chr16	20360284	UMOD	A	AGC	0	-	0	10.53	NM_001278614.1	p.A146fs
P6	chr16	20360294	UMOD	TC	T	0	-	0	17.50	NM_001278614.1	p.D143fs
P6	chr16	20360298	UMOD	C	G	0	-	0	18.60	NM_001278614.1	p.V142L
P6	chr17	47797201	FAM117A	AGGCAGG	A	0	-	0	13.85	NM_030802.3	p.PCL208P
P6	chr17	56404987	BZRAP1	G	T	0	-	0	21.43	NM_004758.3	p.Q99K
P6	chr17	78403686	ENDOV	C	A	0	-	0	17.11	NM_001164638.1	p.P253Q
P6	chr17	79411725	BAHCC1	G	GCCC	0	-	0	35.56	NM_001291324.1	p.L793fs
P6	chr18	23845205	TAF4B	G	A	0	-	0	26.90	NM_005640.1	p.E139K
P6	chr18	44772977	SKOR2	G	A	11.54	-	0	0	NM_001278063.1	p.H250P
P6	chr18	56401603	MALT1	G	T	0.40	0.43	0	24.63	NM_006785.3	p.G489*
P6	chr19	33490487	RHPN2	C	T	8.33	-	16.13	4.17	NM_033103.4	splice_site
P6	chr19	45895600	PPP1R13L	T	G	0	-	0	35.51	NM_006663.3	splice_site
P6	chrX	45060035	CXorf36	G	A	0	-	0	20	NM_176819.3	p.R13C
P7	chr1	26795536	DHDDS	C	T	50	-	42.86	36.36	NM_024887.3	p.R307W
P7	chr1	42048001	HIVEP3	T	G	0	-	0	12	NM_024503.4	p.K823T
P7	chr1	169696612	SELE	C	A	0.97	-	0	16.35	NM_000450.2	p.G508V
P7	chr2	25457242	DNMT3A	C	T	41.86	13.95	37.29	54.55	NM_022552.4	p.R882H
P7	chr2	25469982	DNMT3A	A	ACG	48.17	-	48.40	29.19	NM_022552.4	p.F354fs
P7	chr2	128397648	LIMS2	A	G	10.87	-	0	0	NM_017980.4	p.L316P
P7	chr2	135711035	CNT2	G	A	0	-	0	11.14	NM_058241.2	p.G337E
P7	chr4	47033959	GABRB1	G	C	42.25	0.52	43.13	33.57	NM_000812.3	p.V37L
P7	chr5	39119057	FYB	G	A	0	-	22.22	3.33	NM_001243093.1	p.Q784*
P7	chr5	40928700	C7	T	A	56	-	36.89	41.61	NM_000587.2	p.L9M
P7	chr5	140475570	PCDHB2	T	C	0	-	0	13.58	NM_018936.3	p.V399A
P7	chr6	51695697	PKHD1	G	A	44.74	-	35.14	37.04	NM_138694.3	p.T2755I
P7	chr7	2279310	FTSJ2	C	A	0	-	0	10.84	NM_013393.1	p.R14L
P7	chr8	21977683	HR	C	T	0	-	0	21.98	NM_005144.4	p.R927H
P7	chr8	41798487	KAT6A	C	A	0	-	0	14.50	NM_006766.3	p.R971L

P7	chr10	5436040	TUBAL3	C	A	0	-	0	13.39	NM_024803.2	p.E261*
P7	chr11	64375311	NRXN2	T	G	0	-	0	13.27	NM_015080.3	p.E1499A
P7	chr11	71932329	FOLR2	G	A	0	-	0	17.50	NM_000803.4	p.W150*
P7	chr11	129722508	TMEM45B	G	A	47.73	-	58.33	43.59	NM_138788.3	p.R44H
P7	chr12	2061664	DCP1B	G	C	0	-	0	11.83	NM_152640.3	p.A481G
P7	chr12	29669344	TMTC1	C	T	60.87	-	32.39	39.71	NM_001193451.1	p.G749R
P7	chr12	113614891	DDX54	C	G	0	-	0	10	NM_001111322.1	p.D208H
P7	chr12	130892338	RIMBP2	G	A	42.44	-	39.78	36.16	NM_015347.4	p.S953L
P7	chr15	22841116	TUBGCP5	C	T	49.40	-	35.80	32.86	NM_052903.4	p.P162S
P7	chr16	29675548	SPN	C	G	48.56	-	41.99	38.55	NM_003123.4	p.P167A
P7	chr17	7416520	POLR2A	C	T	0	-	0	12.39	NM_000937.4	p.T1646M
P7	chr19	17424902	DDA1	G	A	0	-	12.50	0	NM_024050.5	p.C25Y
P7	chr19	17954221	JAK3	C	A	0	-	0	15.22	NM_000215.3	p.D130Y
P7	chr19	41206229	ADCK4	C	T	0	-	0	14.10	NM_024876.3	p.D341N
P7	chr20	33337458	NCOA6	G	C	0	-	0	11.76	NM_014071.3	p.S847*
P7	chr22	37485713	TMPRSS6	C	T	0	-	11.54	0	NM_001289000.1	p.M247I
P7	chr22	38917700	DMC1	C	A	38.75	-	33.33	34.26	NM_007068.3	p.G289V
P7	chrX	70462897	ZMYM3	G	A	0	-	0	16.67	NM_005096.3	p.T1157M
P8	chr1	152681680	LCE4A	C	CAGTCTGGGGG TCGCTGT	0	-	-	12.44	NM_178356.3	p.S43SALGAAV
P8	chr1	158582642	SPTA1	C	T	42.97	-	-	7.44	NM_003126.2	p.G2367S
P8	chr2	68621240	PLEK	C	T	44.12	-	-	4.00	NM_002664.2	p.A283V
P8	chr2	219825563	CDK5R2	G	A	44.64	-	-	3.37	NM_003936.4	p.G341R
P8	chr6	31599583	PRRC2A	C	T	43.56	-	-	10.34	NM_004638.3	p.R1045*
P8	chr6	160328084	MAS1	C	A	41.18	-	-	3.57	NM_002377.2	p.P33T
P8	chr7	92848687	HEPACAM2	C	T	42.62	-	-	9.33	NM_001288804.1	p.V76I
P8	chr7	142641804	KEL	G	A	34.94	-	-	6.96	NM_000420.2	p.R447W
P8	chr7	148512096	EZH2	A	G	100	-	-	16	NM_004456.4	p.C528R
P8	chr10	45499398	ZNF22	T	TCCC	46.15	-	-	5.69	NM_006963.4	p.P194PP
P8	chr10	121086055	GRK5	G	C	37.04	-	-	4.35	NM_005308.2	p.S27T
P8	chr11	32636263	CCDC73	G	T	52.63	-	-	9.80	NM_001008391.3	p.P534Q
P8	chr11	47612306	C1QTNF4	C	CGGGCCAGGGCC CAGCAGGCCGCT	21.43	-	-	1.54	NM_031909.2	p.A12*
P8	chr11	99690461	CNTN5	A	G	0	-	-	11.28	NM_014361.3	p.N81S
P8	chr11	102826185	MMP13	G	A	38.86	-	-	6.83	NM_002427.3	p.A53V
P8	chr12	53073937	KRT1	T	TC	47.31	-	-	4.71	NM_006121.3	p.S66fs
P8	chr12	112888199	PTPN11	C	T	51.59	-	-	8.25	NM_002834.3	p.A72V
P8	chr13	33306277	PDS5B	A	AC	31.33	0.70	-	5.81	NM_015032.3	p.G721fs
P8	chr15	34127230	RYR3	G	T	50	-	-	9.52	NM_001036.3	p.G3842V
P8	chr15	43891129	CKMT1B	G	T	41.86	-	-	6.94	NM_020990.3	p.L372F
P8	chr15	75981944	CSPG4	C	T	1.69	-	-	17.65	NM_001897.4	p.A488T
P8	chr17	45669359	NPEPPS	T	G	0	-	-	12.92	NM_006310.3	p.F433C
P8	chr17	46703288	HOXB9	G	A	45.59	-	-	8.33	NM_024017.4	p.A115V
P8	chr19	2255228	JSRP1	T	A	52.24	-	-	9.43	NM_144616.3	p.E29V
P8	chr19	7935863	FLJ22184	G	T	0	-	-	12.77	NM_001190467.1	p.L756I
P8	chr19	47193921	PRKD2	A	C	45	-	-	7.14	NM_016457.4	p.I582S
P8	chr21	42852430	TMPRSS2	C	T	44.02	-	-	5.88	NM_001135099.1	p.R219Q
P8	chrX	70757751	OGT	T	A	88.76	-	-	9.87	NM_181672.2	p.N97K
P9	chr1	32797159	HDAC1	C	T	19.28	-	-	22.22	NM_004964.2	p.T355M
P9	chr1	151262327	ZNF687	G	GC	28.26	-	-	21.31	NM_020832.1	p.E936fs
P9	chr2	26818155	CIB4	C	T	40	-	-	25	NM_001029881.1	p.V73M
P9	chr2	204360027	RAPH1	C	T	0	-	-	12.26	NM_213589.2	p.S17N
P9	chr3	151155763	IGSF10	C	G	0	-	-	13.88	NM_178822.4	p.G2196R
P9	chr4	20525493	SLIT2	G	A	0	-	-	10.29	NM_004787.2	p.G414E
P9	chr8	123964807	ZHX2	G	T	0	-	-	16.01	NM_014943.3	p.A353S
P9	chr10	25886810	GPR158	G	A	22.22	-	-	39.39	NM_020752.2	p.R752H
P9	chr10	29759214	SVIL	A	ATTT	0	-	-	10	NM_021738.2	p.I1945IN
P9	chr11	32413581	WT1	G	A	43.18	-	-	29.33	NM_000378.4	p.Q440*
P9	chr11	32417943	WT1	C	CCA	0	-	-	10.34	NM_000378.4	p.R353fs
P9	chr11	48152200	PTPRJ	G	A	52.38	-	-	21.21	NM_002843.3	p.C516Y
P9	chr11	92616454	FAT3	C	T	47.22	1.82	-	27.12	NM_001008781.2	p.R4278W
P9	chr13	37605917	SUPT20H	C	T	45.78	-	-	18.55	NM_001278480.1	p.A275T
P9	chr13	100953752	PCCA	C	A	36.23	-	-	23.46	NM_000282.3	p.D368E
P9	chr15	40477801	BUB1B	G	T	0	-	-	15.09	NM_001211.5	p.S339I
P9	chr15	63986743	HERC1	G	A	46.58	-	-	26.09	NM_003922.3	p.Q1750*
P9	chr16	27374263	IL4R	G	T	0	-	-	16.35	NM_000418.3	p.M530I
P9	chr19	57166553	SMIM17	C	T	15.04	-	-	26.32	NM_001193628.1	p.T110M
P10	chr1	115256530	NRAS	G	T	0	-	-	38.16	NM_002524.4	p.Q61K
P10	chr1	154841527	KCNN3	G	A	41.71	-	-	33.33	NM_001204087.1	p.S305F
P10	chr2	20205592	MATN3	T	C	48.78	-	-	40	NM_002381.4	p.S235G
P10	chr2	183070674	PDE1A	C	T	33.33	-	-	20.45	NM_001258312.1	p.D319N
P10	chr2	223553059	MOGAT1	A	T	50	-	-	22.78	NM_058165.2	splice_site
P10	chr3	26751294	LRR3B	G	T	0	-	-	36.41	NM_052953.2	p.G44V
P10	chr4	134073627	PCDH10	C	G	0	-	-	37.84	NM_032961.1	p.R778G
P10	chr5	161324204	GABRA1	G	A	23.60	-	-	0.53	NM_000806.5	p.D383N
P10	chr6	28227707	NKAPL	GA	G	0	-	-	30.30	NM_001007531.2	p.K186fs
P10	chr6	99282912	POU3F2	C	T	52	-	-	29.41	NM_005604.3	p.L55F
P10	chr7	7571316	COL28A1	T	C	0	-	-	35.88	NM_001037763.2	p.Q115R
P10	chr7	123302500	LMOD2	C	T	30.56	-	-	40	NM_207163.1	p.A287V
P10	chr7	142572321	TRPV6	C	T	0	-	-	35.16	NM_018646.4	p.V459M
P10	chr7	143088836	EPHA1	G	T	40.22	-	-	37.11	NM_005232.4	p.S910*
P10	chr8	3889424	CSMD1	T	A	70	-	-	45.45	NM_033225.5	splice_site
P10	chr8	90993666	NBN	C	T	0	-	-	22.22	NM_002485.4	p.G86D

P10	chr10	71000489	HKDC1	G	A	2.04	-	-	31.82	NM_025130.3	p.E225K
P10	chr11	17793428	KCNC1	C	T	37.11	-	-	35.47	NM_001112741.1	p.R263C
P10	chr11	121384879	SORL1	G	T	0	-	-	38.10	NM_003105.5	p.A354S
P10	chr12	25398281	KRAS	C	T	17.39	-	-	0	NM_033360.3	p.G13D
P10	chr15	75630409	COMMD4	C	T	0	-	-	38.33	NM_017828.4	p.R4W
P10	chr15	93485160	CHD2	A	C	31.11	-	-	26.32	NM_001271.3	p.L267F
P10	chr17	79412683	BAHCC1	G	A	0	-	-	34.92	NM_001291324.1	p.A992T
P10	chr19	12800628	FBXW9	G	A	13.82	-	-	0	NM_032301.2	p.Q365*
P10	chr19	38987553	RYR1	A	C	50.77	-	-	27.78	NM_000540.2	p.N2284H
P10	chr21	45960023	KRTAP10-1	G	A	53.57	-	-	51.61	NM_198691.2	p.S4F
P10	chrX	100745806	ARMCX4	A	T	100	-	-	66.67	NM_001256155.2	p.T744S
P11	chr3	18390686	SATB1	G	GT	44.05	-	-	4.71	NM_001195470.1	p.D788fs
P11	chr3	45518000	LARS2	C	T	46.94	-	-	5.26	NM_015340.3	p.T300M
P11	chr3	179371153	USP13	A	T	41.86	-	-	0	NM_003940.2	p.N47I
P11	chr7	117174349	CFTR	G	A	35.59	-	-	2.94	NM_000492.3	p.R170H
P11	chr11	32417911	WT1	A	ACCGTACAAGAG	24.49	-	-	2.27	NM_000378.4	p.S364fs
P11	chr11	89133421	NOX4	G	A	42.70	0.38	-	4.57	NM_016931.3	p.R325*
P11	chr12	133434117	CHFR	C	T	50.93	0.89	-	7.79	NM_001161344.1	p.A326T
P11	chr13	41373252	SLC25A15	A	G	40.86	-	-	14.63	NM_014252.3	p.T39A
P11	chr15	48808490	FBN1	AG	A	42.86	-	-	21.43	NM_000138.4	p.L406fs
P11	chr17	45286901	MYL4	C	G	49.37	-	-	1.47	NM_002476.2	p.A38G
P11	chr19	49391385	TULP2	G	A	49.35	-	-	0	NM_003323.2	p.A257V
P12	chr1	24175149	FUCA1	T	C	0	-	-	26.15	NM_000147.4	p.T384A
P12	chr1	27684982	MAP3K6	C	T	0	-	-	24.05	NM_004672.3	p.G902R
P12	chr1	38155449	C1orf109	C	A	0	-	-	25.49	NM_017850.1	p.C35F
P12	chr1	114128074	MAGI3	C	G	0	-	-	19.39	NM_001142782.1	p.Q207E
P12	chr1	115258747	NRAS	C	A	0	-	-	23.15	NM_002524.4	p.G12V
P12	chr1	231830117	DISC1	G	T	0	0.81	-	17.74	NM_001164537.1	p.A205S
P12	chr2	62065845	FAM161A	G	C	0	-	-	19.86	NM_001201543.1	splice_site
P12	chr2	86327135	POLR1A	G	T	0	-	-	24.24	NM_015425.3	p.H80N
P12	chr3	12650286	RAF1	C	A	0	1.30	-	18.75	NM_002880.3	p.W187L
P12	chr3	132400855	NPHP3	A	C	0	-	-	20.20	NM_153240.4	p.L1298V
P12	chr4	141074093	MAML3	C	T	0	-	-	10.20	NM_018717.4	p.G130D
P12	chr4	184628067	TRAPP1	G	T	0	-	-	26.67	NM_021942.5	p.A1055S
P12	chr5	75587642	SV2C	C	A	0	-	-	17.28	NM_014979.1	p.T433K
P12	chr6	33995910	GRM4	G	T	0	-	-	20.91	NM_000841.3	p.N892K
P12	chr7	149463130	ZNF467	G	A	0	-	-	39.77	NM_207336.1	p.P154L
P12	chr8	17814172	PCM1	A	G	11.54	-	-	0	NM_006197.3	p.E511G
P12	chr9	127781402	SCAI	A	G	34	-	-	34.78	NM_173690.4	p.V254A
P12	chr10	127734635	ADAM12	G	T	0	-	-	17.42	NM_003474.5	p.H665N
P12	chr11	94587243	AMOTL1	A	G	10	-	-	0	NM_130847.2	p.Q647R
P12	chr12	53069000	KRT1	T	A	10	-	-	0	NM_006121.3	p.T638S
P12	chr12	130840202	PIWIL1	G	A	0	-	-	26.09	NM_004764.4	p.G465D
P12	chr13	32912331	BRCA2	A	T	52.94	42.11	-	10	NM_000059.3	p.V959I
P12	chr13	110833655	COL4A1	C	G	40.54	-	-	36.67	NM_001845.4	p.G726A
P12	chr14	102800984	ZNF839	G	A	34.25	-	-	30.77	NM_018335.4	splice_site
P12	chr17	7578550	TP53	G	A	50	-	-	52.83	NM_000546.5	p.S127F
P12	chr17	18144040	LLGL1	G	T	0	-	-	18.75	NM_004140.3	p.M785I
P12	chr17	18881328	FAM83G	G	T	0	1.12	-	26.20	NM_001039999.2	p.H551N
P12	chr17	42959054	EFTUD2	C	T	41.14	-	-	44	NM_004247.3	splice_site
P12	chr17	48431099	XYLT2	C	T	10.64	-	-	0	NM_022167.3	p.R82W
P12	chr17	78322037	RNF213	A	G	0	-	-	12.28	NM_001256071.2	p.Q3301R
P12	chr18	22806720	ZNF521	G	T	2.22	-	-	23.23	NM_015461.2	p.Q388K
P12	chr19	44564937	ZNF223	C	A	0	-	-	20.75	NM_013361.5	p.L60I
P12	chr19	44892153	ZNF285	G	C	11.11	1.15	-	5.06	NM_152354.4	p.T85S
P12	chr19	55597540	EPS8L1	C	A	0	-	-	13.51	NM_133180.2	p.Q544K
P12	chr21	38520922	TTC3	C	A	0	-	-	10.20	NM_003316.3	p.T698K
P12	chr22	21348030	LZTR1	T	G	0	-	-	21.28	NM_006767.3	p.F447C
P12	chr22	36122618	APOL5	T	C	34.68	-	-	26.80	NM_030642.1	p.M168T
P12	chrX	76777818	ATRX	C	A	0	-	-	16.55	NM_000489.4	p.V2300F
P12	chrX	151815559	GABRQ	G	C	0	-	-	25.29	NM_018558.3	p.V153L