

Plasmacytoid dendritic cells proliferation associated with acute myeloid leukemia: phenotype profile and mutation landscape

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Supplemental methods

PATIENT SAMPLES: Normal BM aspiration performed for peripheral thrombopenia/research of metastatic infiltration were performed on sternal puncture. After BM examination, no qualitative or quantitative abnormalities were noticed, and cytologists concluded to physiological hematopoiesis.

CELL LABELLING: After 20 minutes of incubation, cells were washed twice with PBS buffer (Gibco Life technologies, Paisley, UK). The percentage of positive expression of markers was determined by a threshold obtained using matched isotypic control mAbs.

CELL SORTING: 30,000 to one million cells were obtained per fraction and their purity was controlled by flow cytometry. The sorted cells were frozen in dry pellets at -20°C.

MOLECULAR BIOLOGY

After thawing the cell pellets at room temperature, the genomic DNA of each sorted fraction was extracted manually according to the manufacturer's instructions using the QIAamp® DNA Mini kit (Qiagen, Hilden, Germany) or the QIAamp® DNA Micro kit (Qiagen) for fractions below 150,000 cells. The DNA was eluted in 20-30 µL of elution buffer. Concentrations ranged from 0.58-33.86 ng/µL on a Nanodrop™ND-2000 spectrophotometer (Thermo Fisher Scientific, Wilmington, USA). After Whole Genome Amplification, the final DNA quantities obtained were about 40 µg on Nanodrop™ND-2000.

The HaloPlex^{HS} Target Enrichment System design included 5392 amplicons, a total sequenceable design size of 244.235 kbp and a total coverage of 98.56%. In addition to genes recurrently mutated in onco-hematology, this panel included mutated genes in BPDCN (*IKZF2*, *IKZF3*, *ZEB2*, *HOXB9*, *UBE2G2*, *APC*, *MET*, *RB1*, *VHL*, *MLH1*, *RET*, *SHOC2*, *NEFH*, *HNF1A*, *SSC5D*, *SF3A2*, *SF3BA*, *PAX3*, *GNB1*, *CRIPAK*, *MAP2K1*). *NPM1* and *CEBPA* variants were screened by Sanger sequencing with a BigDye®Terminator V3.1 Cycle Sequencing kit (Thermo Fisher Scientific) and *FLT3-ITD* mutations were confirmed by fragment analysis on an ABI3500 device (Applied Biosystems, Thermo Fisher Scientific). Some other variants (*ASXL1*, *RUNX1*, *PFH6*) detected by NGS were confirmed by Sanger sequencing on a capillary sequencer SeqStudio Genetic Analyser (Applied Biosystems, Thermo Fisher Scientific).

BIOINFORMATICS ANALYSIS

The reads were aligned with the reference genome sequence GHCh37 (hg19) using the BWA (Burrows-Wheeler Alignment) aligner. The calibration of the base scores (BSQR) and the realignment of the Indels were performed according to GATK Best Practices recommendations. Variant Calling was based on the VarScan algorithm. The

annotation of the variants was done via Generate Reports™ yielding VCF files (Variant Call Files). Finally, several filters were applied to eliminate intronic regions, synonymous mutations and polymorphisms with VAF>0.01%.

Supplemental tables

Table S1: Antibodies used for immunophenotype or cell sorting. Expression was evaluated on the selected populations for immunophenotyping. Intracellular detection of TCL1 was performed after permeabilization with PFA 8% (Sigma-Aldrich, Saint-Louis, Missouri, USA) and lysis perm buffer : Triton X100 10% (Euromedex, Souffelweyersheim, France). After 20 minutes of incubation, the cells were washed twice with PBS buffer (Gibco Life technologies, Paisley, UK). Conjugated fluorochromes: phycoerythrin (PE), allophycocyanin (APC), allophycocyanin-H7 (APC-H7), phycoerythrin-cyanin-7 (PE-Cy7)

Antigen	Conjugated fluorochrome	Clone	Firm	Purpose
CD45	Horizon V500	HI30	BD Biosciences	immunophenotype
CD34	Horizon V450	8G12	BD Biosciences	immunophenotype
CD117	APC	104D2	BD Pharmingen	immunophenotype
HLA-DR	FITC	G46-6	BD Pharmingen	immunophenotype
TdT	FITC	HT-6	Dako	immunophenotype
ILT7	APC	17G10.2	Affymetrix eBiosciences	immunophenotype
CD11c	PerCP-Cy5.5	B-ly6	BD Pharmingen	immunophenotype
CD1c	PE	L161	Biolegend, Ozyme	immunophenotype
CD141	APC	AD5-14H12	Miltenyi Biotec	immunophenotype
cMPO	FITC	MPO-7	Dako	immunophenotype
CD13	PE	L138	BD Pharmingen	immunophenotype
CD33	PerCP-Cy5.5	P67.6	BD Pharmingen	immunophenotype
CD14	APC-H7	MφP9	BD Pharmingen	immunophenotype
CD64	FITC	10.1	BD Pharmingen	immunophenotype
CD15	FITC	HI98	BD Pharmingen	immunophenotype
CD22	PerCP-Cy5.5	HIB22	BD Pharmingen	immunophenotype
CD7	Horizon V450	M-T701	BD Pharmingen	immunophenotype
CD5	FITC	BL1a	Beckman Coulter	immunophenotype
CD56	Horizon V450	B156	BD Biosciences	immunophenotype
CD4	APC-H7	SK3	BD Biosciences	immunophenotype
CD123	PE-Cy7	6H6	Biolegend, Ozyme	immunophenotype
CD303	APC	AC144	Miltenyi Biotec	immunophenotype
CD304	PE	AD5-17F6	Miltenyi Biotec	immunophenotype
cTCL1	PE	eBio1-21	Affymetrix eBiosciences	immunophenotype
CD45	BV421	HI30	BD Pharmingen	cell sorting
CD45	Horizon V500	HI30	BD Pharmingen	cell sorting
CD3	Horizon V500	SK7	BD Pharmingen	cell sorting
CD34	APC	8G12	BD Pharmingen	cell sorting
CD123	PE-Cy7	6H6	Biolegend, Ozyme	cell sorting
CD303	FITC	201A	Biolegend, Ozyme	cell sorting
CD304	FITC	12C2	Biolegend, Ozyme	cell sorting
CD1c	PE	L161	Biolegend, Ozyme	cell sorting
CD14	APC-H7	MφP9	BD Pharmingen	cell sorting
CD64	FITC	10.1	BD Pharmingen	cell sorting

SUPPLEMENTAL DOCUMENTS

Table S2: Amplicons included in the HaloPlex^{HS} Target Enrichment System design for Illumina Sequencing

Target ID (gene name_exon or region)	Chromosomal coordinates	Size (bp)	Coverage (%)
APC_P_ALA1582PRO	chr5:112175996-112176075	80	100
ASXL1_EX12	chr20:31022235-31027122	4888	99.71
ATM_EX1	chr11:108093559-108093913	355	100
ATM_EX10	chr11:108121428-108121799	372	100
ATM_EX11	chr11:108122564-108122758	195	100
ATM_EX12	chr11:108123544-108123639	96	90.62
ATM_EX13	chr11:108124541-108124766	226	100
ATM_EX14	chr11:108126942-108127067	126	100
ATM_EX15	chr11:108128208-108128333	126	100
ATM_EX16	chr11:108129713-108129802	90	100
ATM_EX17	chr11:108137898-108138069	172	100
ATM_EX18	chr11:108139137-108139336	200	100
ATM_EX19	chr11:108141791-108141873	83	100
ATM_EX2	chr11:108098322-108098423	102	100
ATM_EX20	chr11:108141978-108142133	156	100
ATM_EX21	chr11:108143259-108143334	76	100
ATM_EX22	chr11:108143449-108143579	131	100
ATM_EX23	chr11:108150218-108150335	118	100
ATM_EX24	chr11:108151722-108151895	174	100
ATM_EX25	chr11:108153437-108153606	170	100
ATM_EX26	chr11:108154954-108155200	247	100
ATM_EX27	chr11:108158327-108158442	116	100
ATM_EX28	chr11:108159704-108159830	127	56.69
ATM_EX29	chr11:108160329-108160528	200	100
ATM_EX3	chr11:108098503-108098615	113	100
ATM_EX30	chr11:108163346-108163520	175	100
ATM_EX31	chr11:108164040-108164204	165	100
ATM_EX32	chr11:108165654-108165786	133	100
ATM_EX33	chr11:108168014-108168109	96	89.58
ATM_EX34	chr11:108170441-108170612	172	100
ATM_EX35	chr11:108172375-108172516	142	100
ATM_EX36	chr11:108173580-108173756	177	100
ATM_EX37	chr11:108175402-108175579	178	100
ATM_EX38	chr11:108178624-108178711	88	100
ATM_EX39	chr11:108180887-108181042	156	100
ATM_EX4	chr11:108099905-108100050	146	100
ATM_EX40	chr11:108183138-108183225	88	100
ATM_EX41	chr11:108186550-108186638	89	100
ATM_EX42	chr11:108186738-108186840	103	100
ATM_EX43	chr11:108188100-108188248	149	100
ATM_EX44	chr11:108190681-108190785	105	93.33
ATM_EX45	chr11:108192028-108192147	120	100
ATM_EX46	chr11:108196037-108196271	235	100
ATM_EX47	chr11:108196785-108196952	168	100
ATM_EX48	chr11:108198372-108198485	114	100
ATM_EX49	chr11:108199748-108199965	218	100
ATM_EX5	chr11:108106397-108106561	165	100
ATM_EX50	chr11:108200941-108201148	208	100
ATM_EX51	chr11:108202171-108202284	114	100
ATM_EX52	chr11:108202606-108202764	159	100
ATM_EX53	chr11:108203489-108203627	139	100
ATM_EX54	chr11:108204613-108204695	83	100
ATM_EX55	chr11:108205696-108205836	141	100
ATM_EX56	chr11:108206572-108206688	117	100
ATM_EX57	chr11:108213949-108214098	150	100
ATM_EX58	chr11:108216470-108216635	166	100
ATM_EX59	chr11:108218006-108218092	87	0
ATM_EX6	chr11:108114680-108114845	166	100
ATM_EX60	chr11:108224493-108224607	115	100
ATM_EX61	chr11:108225538-108225601	64	100

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ATM_EX62	chr11:108235809-108235945	137	100
ATM_EX63	chr11:108236052-108239826	3775	90.07
ATM_EX7	chr11:108115515-108115753	239	100
ATM_EX8	chr11:108117691-108117854	164	100
ATM_EX9	chr11:108119660-108119829	170	100
BCL6_AMPL1	chr3:187440236-187440399	164	100
BCL6_AMPL2	chr3:187442719-187442876	158	100
BCL6_AMPL3	chr3:187443277-187443427	151	100
BCL6_AMPL4	chr3:187444509-187444696	188	100
BCL6_AMPL5	chr3:187446138-187446342	205	100
BCL6_AMPL6	chr3:187446828-187447819	992	100
BCL6_AMPL7	chr3:187449487-187449728	242	100
BCL6_AMPL8	chr3:187451311-187451491	181	100
BCOR_P_G1529EFSX4	chrX:39914632-39914711	80	100
BIRC3_EX6	chr11:102201730-102201972	243	100
BIRC3_EX7	chr11:102206697-102206951	255	100
BIRC3_EX8	chr11:102207491-102207532	42	100
BIRC3_EX9	chr11:102207640-102210135	2496	99.6
BRAF_EX15	chr7:140453075-140453193	119	100
BTK_AMPL1	chrX:100604863-100604954	92	100
BTK_AMPL10	chrX:100613595-100613694	100	100
BTK_AMPL11	chrX:100614271-100614345	75	100
BTK_AMPL12	chrX:100615066-100615148	83	100
BTK_AMPL13	chrX:100615546-100615753	208	100
BTK_AMPL14	chrX:100617151-100617238	88	100
BTK_AMPL15	chrX:100617539-100617687	149	100
BTK_AMPL16	chrX:100624976-100625077	102	100
BTK_AMPL17	chrX:100626611-100626699	89	100
BTK_AMPL18	chrX:100629514-100629632	119	100
BTK_AMPL19	chrX:100630122-100630282	161	100
BTK_AMPL2	chrX:100608172-100608349	178	100
BTK_AMPL3	chrX:100608848-100608986	139	100
BTK_AMPL4	chrX:100609608-100609692	85	100
BTK_AMPL5	chrX:100611030-100611266	237	100
BTK_AMPL6	chrX:100611762-100611953	192	100
BTK_AMPL7	chrX:100612487-100612581	95	100
BTK_AMPL8	chrX:100613137-100613158	22	100
BTK_AMPL9	chrX:100613284-100613435	152	100
CALR_AMPL1	chr19:13049484-13049594	111	100
CALR_AMPL2	chr19:13049938-13050059	122	100
CALR_AMPL3	chr19:13050232-13050455	224	100
CALR_AMPL4	chr19:13050857-13050971	115	100
CALR_AMPL5	chr19:13051047-13051276	230	100
CALR_AMPL6	chr19:13051345-13051478	134	100
CALR_AMPL7	chr19:13051548-13051711	164	100
CALR_AMPL8	chr19:13054341-13054453	113	100
CALR_AMPL9	chr19:13054517-13054737	221	100
CARD11_EX4	chr7:2985453-2985590	138	100
CARD11_EX5	chr7:2983846-2984171	326	100
CARD11_EX6	chr7:2979383-2979562	180	100
CARD11_EX7	chr7:2978313-2978465	153	100
CARD11_EX8	chr7:2977541-2977666	126	100
CARD11_EX9	chr7:2976671-2976868	198	100
CBL_EX8	chr11:119148876-119149007	132	100
CBL_EX9	chr11:119149220-119149423	204	100
CD79A_EX5	chr19:42384934-42385439	506	100
CD79B_EX5	chr17:62006794-62006835	42	100
CDKN2A_EXON1	chr9:21974647-21975127	481	100
CDKN2A_EXON2	chr9:21970871-21971237	367	100
CDKN2A_EXON3	chr9:21967722-21968271	550	100
CEBPA_EX1	chr19:33790840-33793470	2631	100
CLUU1_EX1	chr12:92817735-92819710	1976	99.65
CLUU1_EX2	chr12:92822810-92824778	1969	100
CRIPAK_NM_175918_C_294	chr4:1388554-1388680	127	0
CRIPAK_NM_175918_C_445_-CA	chr4:1388705-1388784	80	0

SUPPLEMENTAL DOCUMENTS

CRIPAK_NM_175918_P_X22	chr4:1388325-1388444	120	0
CSF3R_AMPL1	chr1:36931687-36931811	125	100
CSF3R_AMPL10	chr1:36937657-36937750	94	100
CSF3R_AMPL11	chr1:36937829-36938002	174	100
CSF3R_AMPL12	chr1:36938108-36938297	190	100
CSF3R_AMPL13	chr1:36939026-36939233	208	100
CSF3R_AMPL14	chr1:36939355-36939498	144	100
CSF3R_AMPL15	chr1:36940968-36941284	317	100
CSF3R_AMPL16	chr1:36945024-36945107	84	100
CSF3R_AMPL2	chr1:36931948-36932519	572	100
CSF3R_AMPL3	chr1:36932821-36932922	102	100
CSF3R_AMPL4	chr1:36933149-36933262	114	100
CSF3R_AMPL5	chr1:36933413-36933573	161	100
CSF3R_AMPL6	chr1:36933666-36933832	167	100
CSF3R_AMPL7	chr1:36934747-36934868	122	100
CSF3R_AMPL8	chr1:36935243-36935451	209	100
CSF3R_AMPL9	chr1:36937024-36937257	234	100
CXCR4_EX2	chr2:136871919-136873482	1564	100
DNMT3A_EX1	chr2:25565299-25565459	161	100
DNMT3A_EX10	chr2:25469489-25469645	157	100
DNMT3A_EX11	chr2:25469029-25469178	150	100
DNMT3A_EX12	chr2:25468889-25468933	45	100
DNMT3A_EX13	chr2:25468122-25468201	80	100
DNMT3A_EX14	chr2:25467409-25467521	113	100
DNMT3A_EX15	chr2:25467024-25467207	184	100
DNMT3A_EX16	chr2:25466767-25466851	85	100
DNMT3A_EX17	chr2:25464431-25464576	146	100
DNMT3A_EX18	chr2:25463509-25463599	91	100
DNMT3A_EX19	chr2:25463171-25463319	149	100
DNMT3A_EX2	chr2:25536782-25537030	249	100
DNMT3A_EX20	chr2:25461999-25462084	86	100
DNMT3A_EX21	chr2:25459805-25459874	70	100
DNMT3A_EX22	chr2:25458576-25458694	119	84.87
DNMT3A_EX23	chr2:25455830-25457289	1460	88.36
DNMT3A_EX3	chr2:25523008-25523112	105	100
DNMT3A_EX4	chr2:25505310-25505580	271	100
DNMT3A_EX5	chr2:25498369-25498412	44	100
DNMT3A_EX6	chr2:25497810-25497956	147	100
DNMT3A_EX7	chr2:25470906-25471121	216	100
DNMT3A_EX8	chr2:25470460-25470618	159	100
DNMT3A_EX9	chr2:25469920-25470027	108	100
ETV6_EX1	chr12:11802788-11803094	307	100
ETV6_EX2	chr12:11905384-11905513	130	100
ETV6_EX3	chr12:11992074-11992238	165	100
ETV6_EX4	chr12:12006361-12006495	135	100
ETV6_EX5	chr12:12022358-12022903	546	100
ETV6_EX6	chr12:12037379-12037521	143	100
ETV6_EX7	chr12:12038860-12038960	101	100
ETV6_EX8	chr12:12043875-12048325	4451	100
EZH2_EX1	chr7:148581256-148581441	186	100
EZH2_EX10	chr7:148514969-148515209	241	100
EZH2_EX11	chr7:148514314-148514483	170	100
EZH2_EX12	chr7:148513776-148513870	95	100
EZH2_EX13	chr7:148512598-148512638	41	63.41
EZH2_EX14	chr7:148512006-148512131	126	100
EZH2_EX15	chr7:148511051-148511229	179	100
EZH2_EX16	chr7:148508717-148508812	96	100
EZH2_EX17	chr7:148507425-148507506	82	100
EZH2_EX18	chr7:148506402-148506482	81	100
EZH2_EX19	chr7:148506163-148506247	85	100
EZH2_EX2	chr7:148544274-148544397	124	100
EZH2_EX20	chr7:148504464-148504798	335	100
EZH2_EX3	chr7:148543562-148543690	129	100
EZH2_EX4	chr7:148529726-148529842	117	100
EZH2_EX5	chr7:148526820-148526940	121	92.56

SUPPLEMENTAL DOCUMENTS

EZH2_EX6	chr7:148525832-148525972	141	100
EZH2_EX7	chr7:148524256-148524358	103	22.33
EZH2_EX8	chr7:148523546-148523724	179	97.21
EZH2_EX9	chr7:148516688-148516779	92	100
FBXW7_EX10	chr4:153247158-153247383	226	100
FBXW7_EX11	chr4:153245336-153245546	211	100
FBXW7_EX9	chr4:153249360-153249541	182	100
FLT3_AMPL1	chr13:28578179-28578321	143	100
FLT3_AMPL10	chr13:28602305-28602435	131	100
FLT3_AMPL11	chr13:28608014-28608138	125	100
FLT3_AMPL12	chr13:28608209-28608361	153	100
FLT3_AMPL13	chr13:28608428-28608554	127	100
FLT3_AMPL14	chr13:28609622-28609820	199	100
FLT3_AMPL15	chr13:28610062-28610190	129	100
FLT3_AMPL16	chr13:28611312-28611435	124	100
FLT3_AMPL17	chr13:28622402-28622590	189	100
FLT3_AMPL18	chr13:28623511-28623684	174	100
FLT3_AMPL19	chr13:28623762-28623921	160	100
FLT3_AMPL2	chr13:28588579-28588704	126	100
FLT3_AMPL20	chr13:28624222-28624369	148	100
FLT3_AMPL21	chr13:28626672-28626821	150	100
FLT3_AMPL22	chr13:28631474-28631609	136	100
FLT3_AMPL23	chr13:28635994-28636216	223	100
FLT3_AMPL24	chr13:28644618-28644759	142	100
FLT3_AMPL25	chr13:28674595-28674657	63	100
FLT3_AMPL3	chr13:28589284-28589403	120	100
FLT3_AMPL4	chr13:28589717-28589848	132	100
FLT3_AMPL5	chr13:28592585-28592736	152	100
FLT3_AMPL6	chr13:28597477-28597624	148	100
FLT3_AMPL7	chr13:28598988-28599090	103	100
FLT3_AMPL8	chr13:28599530-28599562	33	100
FLT3_AMPL9	chr13:28601215-28601388	174	100
GNB1_NM_002074_P_LYS57GLU	chr1:1747190-1747269	80	100
GNB1_NM_002074_P_LYS89GLU	chr1:1737877-1737956	80	98.75
HNF1A_NM_000545_	chr12:121432078-121432157	80	100
HNF1A_NM_000545_C_861_-G_P_X289X	chr12:121432075-121432157	83	100
HOXB9_P_ARG183HIS	chr17:46700428-46700507	80	100
IDH1_EX4	chr2:209113093-209113384	292	100
IDH2_EX4	chr15:90631819-90631979	161	100
IKZF1_AMPL1	chr7:50358648-50358707	60	100
IKZF1_AMPL2	chr7:50367224-50367363	140	100
IKZF1_AMPL3	chr7:50435694-50436078	385	100
IKZF1_AMPL4	chr7:50444221-50444501	281	100
IKZF1_AMPL5_P_H163Y_P_ILE164SERFS29	chr7:50450228-50450415	188	100
IKZF1_AMPL6_P_E221SFSX36	chr7:50455033-50455178	146	100
IKZF1_AMPL7	chr7:50459417-50459571	155	100
IKZF1_AMPL8_P_R502W	chr7:50467606-50468335	730	100
IKZF2_P_ALA91VAL	chr2:213921652-213921731	80	100
IKZF3_P_GLU318LYS	chr17:37922582-37922661	80	100
IKZF3_P_THR326HISFS25	chr17:37922559-37922638	80	100
IRF8_EX1	chr16:85932774-85932829	56	100
IRF8_EX2	chr16:85936621-85936795	175	100
IRF8_EX3	chr16:85942596-85942779	184	100
IRF8_EX4	chr16:85945176-85945264	89	100
IRF8_EX5	chr16:85946737-85946842	106	100
IRF8_EX6	chr16:85948079-85948126	48	100
IRF8_EX7	chr16:85952023-85952409	387	100
IRF8_EX8	chr16:85953715-85953830	116	100
IRF8_EX9	chr16:85954712-85956211	1500	94.73
JAK2_EX12	chr9:5069925-5070052	128	100
JAK2_EX14	chr9:5073698-5073785	88	100
KIT_EX10	chr4:55593384-55593490	107	100
KIT_EX11	chr4:55593582-55593708	127	100
KIT_EX17	chr4:55599236-55599358	123	91.87
KIT_EX9	chr4:55592023-55592216	194	100

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KRAS_EX2	chr12:25398208-25398329	122	100
KRAS_EX3	chr12:25380168-25380346	179	100
MAP2K1_EXON2	chr15:66727335-66727605	271	96.31
MAP2K1_EXON3	chr15:66729054-66729260	207	100
MET_P_ASN375SER	chr7:116340223-116340302	80	100
MET_P_GLU168ASP	chr7:116339603-116339682	80	100
MLH1_P_ARG385CYS	chr3:37067203-37067282	80	100
MPL_EX10	chr1:43814934-43815030	97	100
MYD88_EX3	chr3:38181879-38182083	205	100
MYD88_EX4	chr3:38182248-38182339	92	100
MYD88_EX5	chr3:38182623-38184512	1890	100
NEFH_NM_021076_P_X657LYS	chr22:29885559-29885638	80	65
NOTCH1_EX26	chr9:139399125-139399556	432	100
NOTCH1_EX27	chr9:139397634-139397782	149	100
NOTCH1_EX28	chr9:139396724-139396940	217	100
NOTCH1_EX34	chr9:139388885-139392010	3126	97.7
NOTCH2_EX26	chr1:120466260-120466607	348	100
NOTCH2_EX27	chr1:120465259-120465401	143	100
NOTCH2_EX28	chr1:120464859-120465069	211	100
NOTCH2_EX34	chr1:120454176-120459317	5142	99.75
NRAS_EX2	chr1:115258671-115258798	128	100
NRAS_EX3	chr1:115256421-115256599	179	100
PAX3_NM_181459_P_GLU28	chr2:223163214-223163293	80	100
PAX3_NM_181459_P_THR227ILE	chr2:223096870-223096949	80	100
PHF6_P_A140T	chrX:133527943-133528022	80	100
PTEN_EX5	chr10:89692770-89693008	239	100
PTEN_EX7	chr10:89717610-89717776	167	100
PTPN11_EX13	chr12:112926828-112926979	152	100
PTPN11_EX3	chr12:112888122-112888316	195	100
RB1_P_ARG358	chr13:48942646-48942725	80	100
RB1_P_ASP332GLY	chr13:48941646-48941725	80	100
RET_P_SER649LEU	chr10:43609955-43610034	80	100
RUNX1_EX3	chr21:36265222-36265260	39	100
RUNX1_EX4	chr21:36259140-36259393	254	100
RUNX1_EX5	chr21:36252854-36253010	157	100
RUNX1_EX6	chr21:36231771-36231875	105	100
RUNX1_EX7	chr21:36206707-36206898	192	100
RUNX1_EX8	chr21:36171598-36171759	162	100
RUNX1_EX9	chr21:36160098-36164907	4810	98.59
SETBP1_AMPL1	chr18:42281302-42281807	506	100
SETBP1_AMPL2	chr18:42449185-42449258	74	100
SETBP1_AMPL3	chr18:42456520-42456728	209	100
SETBP1_AMPL4	chr18:42529836-42533315	3480	100
SETBP1_AMPL5	chr18:42618440-42618630	191	100
SETBP1_AMPL6	chr18:42643034-42643673	640	100
SF3A2_NM_007165_P_X329X	chr19:2248095-2248174	80	100
SF3B1_EX13	chr2:198267673-198267759	87	100
SF3B1_EX14	chr2:198267280-198267550	271	100
SF3B1_EX15	chr2:198266709-198266854	146	100
SF3B1_EX16	chr2:198266466-198266612	147	100
SF3B1_EX17	chr2:198266124-198266249	126	100
SF3B1_EX18	chr2:198265439-198265660	222	100
SF3B4_NM_005850_P_X383X	chr1:149895522-149895601	80	100
SHOC2_P_S2G	chr10:112724081-112724160	80	100
SRSF2_EX1	chr17:74732881-74733493	613	100
SSC5D_NM_001144950_P_PRO1247X	chr19:56029344-56029423	80	100
SSC5D_NM_001144950_P_X1146X	chr19:56029041-56029120	80	100
STAT3_EX19	chr17:40475278-40475372	95	100
STAT3_EX20	chr17:40475022-40475161	140	100
STAT3_EX21	chr17:40474303-40474512	210	100
STAT3_EX22	chr17:40469200-40469242	43	100
STAT3_EX23	chr17:40468807-40468919	113	100
STAT3_EX24	chr17:40465343-40467818	2476	95.84
TCL1A_EX1	chr14:96180284-96180533	250	100
TCL1A_EX2	chr14:96178557-96178733	177	100

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TCL1A_EX3	chr14:96178086-96178139	54	100
TCL1A_EX4	chr14:96176304-96177218	915	100
TET2_EX1	chr4:106067842-106068136	295	100
TET2_EX10	chr4:106193721-106194075	355	100
TET2_EX11	chr4:106196205-106200960	4756	99.81
TET2_EX2	chr4:106111517-106111662	146	100
TET2_EX3	chr4:106155054-106158508	3455	98.9
TET2_EX4	chr4:106162496-106162586	91	100
TET2_EX5	chr4:106163991-106164084	94	100
TET2_EX6	chr4:106164727-106164935	209	100
TET2_EX7	chr4:106180776-106180926	151	100
TET2_EX8	chr4:106182916-106183005	90	100
TET2_EX9	chr4:106190767-106190904	138	100
TNFAIP3_EX1	chr6:138188325-138188631	307	100
TNFAIP3_EX2	chr6:138192350-138192659	310	100
TNFAIP3_EX3	chr6:138195982-138196172	191	100
TNFAIP3_EX4	chr6:138196825-138196972	148	100
TNFAIP3_EX5	chr6:138197133-138197303	171	100
TNFAIP3_EX6	chr6:138198213-138198393	181	100
TNFAIP3_EX7	chr6:138199569-138200488	920	100
TNFAIP3_EX8	chr6:138201208-138201389	182	100
TNFAIP3_EX9	chr6:138202172-138204451	2280	98.16
TP53_EX10	chr17:7573927-7574033	107	100
TP53_EX11	chr17:7571720-7573008	1289	98.37
TP53_EX2	chr17:7579839-7579940	102	100
TP53_EX3	chr17:7579700-7579721	22	100
TP53_EX4	chr17:7579312-7579590	279	100
TP53_EX5	chr17:7578371-7578554	184	100
TP53_EX6	chr17:7578177-7578289	113	100
TP53_EX7	chr17:7577499-7577608	110	100
TP53_EX8	chr17:7577019-7577155	137	100
TP53_EX9	chr17:7576853-7576926	74	100
U2AF1_AMPL1	chr21:44513202-44513369	168	100
U2AF1_AMPL10	chr21:44524425-44524512	88	100
U2AF1_AMPL11	chr21:44527551-44527614	64	100
U2AF1_AMPL2	chr21:44514571-44514683	113	100
U2AF1_AMPL3	chr21:44514755-44514908	154	100
U2AF1_AMPL4	chr21:44514765-44514898	134	100
U2AF1_AMPL5	chr21:44515538-44515656	119	100
U2AF1_AMPL6	chr21:44515794-44515863	70	100
U2AF1_AMPL7	chr21:44520553-44520639	87	100
U2AF1_AMPL8	chr21:44521466-44521552	87	100
U2AF1_AMPL9	chr21:44524415-44524522	108	100
UBE2G2_P_ASP63VAL	chr21:46197231-46197310	80	100
VHL_P_PRO81SER	chr3:10183733-10183812	80	100
WT1_AMPL1	chr11:32410594-32410735	142	100
WT1_AMPL10	chr11:32452066-32452095	30	100
WT1_AMPL11	chr11:32456236-32456901	666	100
WT1_AMPL2	chr11:32413508-32413620	113	100
WT1_AMPL3	chr11:32414202-32414311	110	100
WT1_AMPL4	chr11:32417793-32417963	171	100
WT1_AMPL5	chr11:32421484-32421600	117	100
WT1_AMPL6	chr11:32438026-32438096	71	100
WT1_AMPL7	chr11:32439113-32439210	98	100
WT1_AMPL8	chr11:32449492-32449614	123	100
WT1_AMPL9	chr11:32450033-32450175	143	100
ZEB2_P_ASP268ARGFS12	chr2:145161450-145161529	80	100
ZEB2_P_GLY626ARG	chr2:145156839-145156918	80	100
ZEB2_P_LEU420ARG	chr2:145157456-145157535	80	100
ZRSR2_AMPL1	chrX:15808609-15808669	61	100
ZRSR2_AMPL10	chrX:15838320-15838449	130	100
ZRSR2_AMPL11	chrX:15840844-15841375	532	94.92
ZRSR2_AMPL2	chrX:15809047-15809146	100	100
ZRSR2_AMPL3	chrX:15817985-15818086	102	100
ZRSR2_AMPL4	chrX:15821801-15821929	129	100

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ZRSR2_AMPL5	chr:15822224-15822330	107	100
ZRSR2_AMPL6	chr:15826346-15826404	59	100
ZRSR2_AMPL7	chr:15827313-15827451	139	100
ZRSR2_AMPL8	chr:15833790-15834023	234	100
ZRSR2_AMPL9	chr:15836700-15836775	76	100

Table S3: Purity of each fraction. ND: not done because small amounts of cells, stored for NGS analysis only.

Patient	fraction	purity
N1	Blasts	98.5
	pDCs	96.8
	Monocytes	98.9
	T-cells	99.0
N2	Blasts	99.1
	pDCs	98.9
	Monocytes	99.8
	T-cells	99.6
N7	Blasts	97.8
	pDCs	98.0
	Monocytes	98.3
	T-cells	99.0
N8	Blasts	98.6
	pDCs	99.1
	Monocytes	98.9
	T-cells	98.4
	cDCs	100.0
N9	Blasts	ND
	pDCs	ND
	Monocytes	ND
	T-cells	ND
N11	Blasts	99.3
	pDCs	94.9
	Monocytes	99.4
	T-cells	99.3
N12	Blasts	99.7
	pDCs	93.7
	Monocytes	99.5
	T-cells	99.3
N13	Blasts	98.2
	pDCs	84.1
	Monocytes	96.0
	T-cells	99.2
N14	Blasts	96.3
	pDCs	93.3
	Monocytes	97.9
	T-cells	97.3
N16	Blasts	97.0
	pDCs	90.3
	Monocytes	87.0
	T-cells	99.0
N19	Blasts	98.0
	pDCs	94.5
	Monocytes	94.6
	T-cells	98.9

patient	fraction	purity
N20	Blasts	98.6
	pDCs	96.1
	Monocytes	92.7
	T-cells	96.2
N34	Blasts	99.8
	pDCs	99.0
	Monocytes	99.4
	T-cells	99.0
N35	Blasts	95.8
	pDCs	ND
	Monocytes	99.0
	T-cells	96.8
N36	Blasts	98.8
	pDCs	96.2
	T-cells	99.4
	T-cells	99.4

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Table S4: Mutations detected by NGS.

Gene	Transcripts	Exon	cDNA coordinates	protein coordinates	Genomic coordinates	Type	Sample	Quality Score	Frequency	Number of mutated reads	Number of total reads	GenerateReports Class	InterVar	dbSNP	COSMIC ID	COSMIC tissue	ClinVar	PopFreqMax	EMAC_ALL	SIFT	polyphen2_HDIV	polyphen2_HVAR	LRT	MutationTaster	MutationAssessor	FATHMM	PROVEAN	VEST3	MetaSVM	MetaLR	MCAP	CADD	DANN	fathmm_MKL_coding	GERP				
ASXL1	NM_015338	12	c.A2123G	p.Q708R	chr20:g.3102263 8A>G	homozygous SNV	N11-BI	1061	38.3	203	530	Uncertain significance	Uncertain significance																										
ASXL1	NM_015338	12	c.A2123G	p.Q708R	chr20:g.3102263 8A>G	homozygous SNV	N11-mono	1044.43	58.25	113	194	Uncertain significance	Uncertain significance																										
ASXL1	NM_015338	12	c.A2123G	p.Q708R	chr20:g.3102263 8A>G	homozygous SNV	N11-pDC	2223.51	54.88	658	1199	Uncertain significance	Uncertain significance																										
ASXL1	NM_015338	12	c.G2362T	p.E788X	chr20:g.3102287 7G>T	stopgain	N12-BL	2122.04	48.27	474	982	Likely pathogenic	Uncertain significance																										
ASXL1	NM_015338	12	c.G2362T	p.E788X	chr20:g.3102287 7G>T	stopgain	N12-MONO	1351.34	46.92	183	390	Likely pathogenic	Uncertain significance																										
ASXL1	NM_015338	12	c.G2362T	p.E788X	chr20:g.3102287 7G>T	stopgain	N12-PDC	2567.24	49.28	785	1593	Likely pathogenic	Uncertain significance																										
ASXL1	NM_015338	12	c.C2757dupA	p.P920Tfs*1	chr20:g.3102327 1->A	Frameshift insertion	N9-BI	762.45	46.36	121	261	Likely pathogenic	Uncertain significance																										
ASXL1	NM_015338	12	c.C2757dupA	p.P920Tfs*1	chr20:g.3102327 1->A	Frameshift insertion	N9-Mono	299.57	26.32	34	93	Likely pathogenic	Uncertain significance																										
ASXL1	NM_015338	12	c.C2757dupA	p.P920Tfs*1	chr20:g.3102327 1->A	Frameshift insertion	N9-pDC	299.57	26.32	40	152	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.G1211A	p.C404Y	chr11:g.1191489 91G>A	homozygous SNV	N13-BI	214.63	48.3	398	824	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.G1211A	p.C404Y	chr11:g.1191489 91G>A	homozygous SNV	N13-Mono	192.75	44.09	496	1125	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.G1211A	p.C404Y	chr11:g.1191489 91G>A	homozygous SNV	N13-pDC	220.87	49.19	427	868	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.T1253G	p.F418C	chr11:g.1191492 45T>G	homozygous SNV	N8-BI	1030.8	62.69	84	134	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.T1253G	p.F418C	chr11:g.1191492 45T>G	homozygous SNV	N8-pDC	500	94.85	387	408	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.T1253G	p.F418C	chr11:g.1191492 45T>G	homozygous SNV	N8-mono	2654.79	90.11	246	273	Likely pathogenic	Uncertain significance																										
CBL	NM_005188	8	c.T1253G	p.F418C	chr11:g.1191492 45T>G	homozygous SNV	N8-pDC	500	98.37	301	306	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_175629	23	c.C2644T	p.R882C	chr2:g.25457243 G>A	homozygous SNV	N19-BL	630.03	42.26	251	594	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_175629	23	c.C2644T	p.R882C	chr2:g.25457243 G>A	homozygous SNV	N19-mono	640.96	48.69	167	343	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_022552	23	c.C2644T	p.R882C	chr2:g.25457243 G>A	homozygous SNV	N19-pDC	765.05	54.92	201	366	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_022552	23	c.C2644T	p.R882C	chr2:g.25457243 G>A	homozygous SNV	N2-BI	747.75	48.77	278	570	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_175629	23	c.C2644T	p.R882C	chr2:g.25457243 G>A	homozygous SNV	N2-Mono	851.01	48.67	514	1056	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_175629	23	c.C2644T	p.R882C	chr2:g.25457243 G>A	homozygous SNV	N2-pDC	775.42	45.41	475	1046	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_022552	23	c.G2645A	p.R882H	chr2:g.25457242 C>T	homozygous SNV	N36-BI	204.63	57.14	16	28	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_022552	23	c.G2645A	p.R882H	chr2:g.25457242 C>T	homozygous SNV	N36-Mono	9	22	248	248	Likely pathogenic	Uncertain significance																										
DNMT3A	NM_022552	23	c.G2645A	p.R882H	chr2:g.25457242 C>T	homozygous SNV	N36-pDC	577.67	53.3	105	197	Likely pathogenic	Uncertain significance																										
EZH2	NM_00120328	4	c.C889T	p.H297Y	chr7:g.14852356 4G>A	homozygous SNV	N13-BI	562.09	36.56	215	588	Likely pathogenic	Uncertain significance																										
EZH2	NM_00120328	4	c.C889T	p.H297Y	chr7:g.14852356 4G>A	homozygous SNV	N13-Mono	1967.64	55.65	754	1355	Likely pathogenic	Likely pathogenic																										
EZH2	NM_00120328	4	c.C889T	p.H297Y	chr7:g.14852356 4G>A	homozygous SNV	N13-Mono	11.69	70	599	599	Likely pathogenic	Uncertain significance																										

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ASXL1	NM_015338: exon12:c.193dupG(p.G646WfsX12)	E.1934dupG	p.G646WfsX12	p.1934dupG	chr20:g.3102244T>G	frameshift insertion	P89	P15.85	47.13	B2	L74	Benign	Uncertain significance	s756958159	CSOM1411076	I[large_intestine]	D.031	D.0016	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D							
ATM	NM_000051	E.74324C	p.Y1442H	chr11:10816041G>C	homozygous SNV	P5	142.59	21.92	32	146	Likely pathogenic	Uncertain significance	s20166889	CSOM41590	I[large_intestine]	D.0007	D.0004	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D						
CXCR4	NM_0010085140	E.C452T	p.P151L	chr2:136873058G>A	homozygous SNV	P2	142.59	21.92	32	146	Likely pathogenic	Uncertain significance											N	F	N	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D					
ETV6	NM_001987	E.C799T	p.Q267X	chr12:12022693C>T	stopgain	P4	1338.24	59.95	259	432	Likely pathogenic	Likely pathogenic																																	
DH2	NM_002168	E.G419A	p.R140Q	chr15:90631934C>T	homozygous SNV	P2		42.27	52	123	Likely pathogenic	Uncertain significance	s121913502	CSOM41590	I1[haematopoietic_and_lymphoid_tissue]1[NS]	D.0003	D.0001	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D					
DH2	NM_002168	E.G419A	p.R140Q	chr15:90631934C>T	homozygous SNV	P38	858.62	87.1	770	884	Likely pathogenic	Uncertain significance	s121913502	CSOM41590	I1[haematopoietic_and_lymphoid_tissue]1[NS]	D.0003	D.0001	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D					
KZF1	NM_001291847	E.G85T	p.E29X	chr7:503672786G>T	stopgain	P9	1156.66	46.12	892	850	Likely pathogenic	Likely pathogenic																																	
KZF1	NM_006060	E.C45T	p.Q149X	chr7:50450261C>T	stopgain	P89	335.98	33.43	228	582	Likely pathogenic	Likely pathogenic																																	
KZF3	NM_183229	E.C565A	p.L189I	chr17:37947696G>T	homozygous SNV	P5	478.39	33.92	58	171	Likely pathogenic	Uncertain significance																																	
IAK2	NM_004972	E.G1849T	p.V617F	chr9:5073770G>T	homozygous SNV	P9	2124.45	56.07	2706	4826	Likely pathogenic	Likely pathogenic	s77375493	CSOM12600	I[central_nervous_system]3[lung]3[haematopoietic_and_lymphoid_tissue]	D.0008	D.0007	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D				
KRAS	NM_004985	E.A183C	p.Q61H	chr12:25380275T>G	homozygous SNV	P1	1274.84	38.59	818	824	Likely pathogenic	Uncertain significance	s17851045	CSOM554 CSOM1135364	I[ovary]2[endometrium]1[breast]1[skin]2[small_intestine]4[liver]1[kidney]35[pancreas]1[urinary_tract]1[biliary_tract]5[large_intestine]13[lung]1[thyroid]1[haematopoietic_and_lymphoid_tissue]	D	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D		
MET	NM_000245	E.A1127G	p.K376R	chr7:116340265G>T	homozygous SNV	P11	2228.39	48.98	410	837	Uncertain significance	Uncertain significance																																	
NOTCH2	NM_00244084	E.A6295G	p.M2099V	chr11:120459050T>C	homozygous SNV	P11	84.73	26.37	807	1164	Likely benign	Likely benign	s201584590																																
NRAS	NM_002524	E.G35A	p.G12D	chr11:115258747G>T	homozygous SNV	P8	717.61	35.67	173	485	Likely pathogenic	Likely pathogenic	s121913237	CSOM564	I[skin]3[biliary_tract]4[large_intestine]1[genital_tract]1[kidney]1[pancreas]1[ovary]2[soft_tissue]4[endometrium]2[thyroid]1[lung]3[testis]3[haematopoietic_and_lymphoid_tissue]1[central_nervous_system]1[upper_aerodigestive_tract]2[NS]	D.0001	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D
NRAS	NM_002524	E.G34C	p.G12R	chr11:115258748C>G	homozygous SNV	P37	678.51	29.14	250	858	Likely pathogenic	Likely pathogenic	s121913250	CSOM561	I[haematopoietic_and_lymphoid_tissue]1[lung]1[NS]1[large_intestine]1[urinary_tract]5[skin]1[salivary_gland]	D	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D		
SRSF2	NM_003016	E.C284T	p.P95L	chr17:74732959G>A	homozygous SNV	P71	324.5	36.54	98	104	Likely pathogenic	Uncertain significance	s751713049	CSOM146288 CSOM211028	I[haematopoietic_and_lymphoid_tissue]	D.0001	D.0001	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D				
SRSF2	NM_003016	E.C284A	p.P95H	chr17:74732959G>T	homozygous SNV	P37	159.79	41.73	53	127	Likely pathogenic	Uncertain significance	s751713049	CSOM211029 CSOM211505 CSOM211504	I[haematopoietic_and_lymphoid_tissue]	D.0002	D.0001	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D				
TET2	NM_001127218	E.A285dupT	p.E1428fsX1	chr4:106193822G>T	frameshift insertion	P31	1737.4	96.88	2476	5714	Likely pathogenic																																		
TET2	NM_001127218	E.C64A	p.P22T	chr4:106155163C>A	homozygous SNV	P5	202.65	33	23	100	Likely pathogenic	Uncertain significance																																	
TET2	NM_001127218	E.G50delC	p.V218W	chr4:106155749C>	homozygous SNV	P9	9000	85.9	2279	2653	Likely pathogenic	Uncertain significance																																	
TET2	NM_001127218	E.G2392T	p.E798X	chr4:106157491G>T	stopgain	P31	881.34	27.36	776	2836	Likely pathogenic	Uncertain significance																																	
TET2	NM_001127218	E.A2698T	p.K900X	chr4:106157797A>T	stopgain	P3	603.39	48.78	801	517	Likely pathogenic	Uncertain significance																																	
TET2	NM_001127218	E.C2746T	p.Q916X	chr4:106157845C>T	stopgain	P71	1406.47	91.47	543	703	Likely pathogenic	Uncertain significance	s780710758	CSOM43417	I2[haematopoietic_and_lymphoid_tissue]	D.0001	D	D	D	D	D	M	F	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D	D					
TET2	NM_001127218	E.3140_3141delCT	L1048delXCT	chr4:106158239G>T	frameshift deletion	P36	38.38	147	883		Likely pathogenic																																		
TET2	NM_001127218	E.G3404A	p.C1135Y	chr4:106158503G>A	homozygous SNV	P11	623.5	41.53	98	236	Likely pathogenic	Uncertain significance	s769422572																																

SUPPLEMENTAL DOCUMENTS

TET2	NM_00112728	A3881G	p.Y1294C	chr4:106180853 A>G	homoynomous SNV	P6	1657.4	84.25	958	2797	Likely pathogenic	Uncertain significance					D s=0;rank=0.912	D s=1;rank=0.899	D s=0.999;rank=0.916	D s=1;rank=0.873	M s=2.51;rank=0.734	T s=2.43;rank=0.153	D s=8.03;rank=0.966	s=0.815;rank=0.953	T s=0.819;rank=0.54	T s=0.141;rank=0.461	s=0.466;rank=0.946	s=5.855;rank=0.802	s=0.998;rank=0.902	D s=0.986;rank=0.842	s=5;rank=0.651		
TET2	NM_00112728	T3986C	p.L1329P	chr4:106182947 T>C	homoynomous SNV	P37	1689.42	92.74	281	803	Likely pathogenic	Uncertain significance					D s=0;rank=0.912	D s=1;rank=0.899	D s=1;rank=0.9	D s=1;rank=0.873	M s=2.51;rank=0.734	T s=2.43;rank=0.153	D s=8.03;rank=0.966	s=0.815;rank=0.953	T s=0.819;rank=0.54	T s=0.141;rank=0.461	s=0.466;rank=0.946	s=5.855;rank=0.802	s=0.998;rank=0.902	D s=0.986;rank=0.842	s=5;rank=0.651		
TP53	NM_00112614	E367_368delTG	p.V122DfX	chr17:7579321C A>	frameshift substitution	P38	422.82	40.52	124	806	Likely pathogenic						D s=0;rank=0.912	D s=1;rank=0.899	D s=1;rank=0.9	D s=1;rank=0.873	M s=2.51;rank=0.734	T s=2.43;rank=0.153	D s=8.03;rank=0.966	s=0.815;rank=0.953	T s=0.819;rank=0.54	T s=0.141;rank=0.461	s=0.466;rank=0.946	s=5.855;rank=0.802	s=0.998;rank=0.902	D s=0.986;rank=0.842	s=5;rank=0.651		
TP53	NM_00127669	A503G	p.H168R	chr17:7578427T>C	homoynomous SNV	P5	679.95	98.25	112	114	Likely pathogenic	Uncertain significance	s867114783	COSM3388210 COSM3388206 COSM3388207 COSM3388208 COSM3388211 COSM43545 COSM3388209	1 breast 1 pancreas 2 large_intestine 1 soft_tissue 3 ovary 1 endometrium 1 lung 4 haematopoietic_and_lymphoid_tissue 1 central_nervous_system 5 stomach 1 oesophagus			D s=0.964;rank=0.622	D s=0.781;rank=0.644	N s=0.025;rank=0.262	D s=0.979;rank=0.395	M s=3.105;rank=0.879	D s=6.8;rank=0.998	D s=7.5;rank=0.952	D s=0.216;rank=0.649	D s=1.055;rank=0.982	D s=0.984;rank=0.995	s=0.479;rank=0.948	s=2.461;rank=0.357	s=0.975;rank=0.34	D s=0.968;rank=0.71	s=0.619;rank=0.167	
TP53	NM_00112614	T808A	p.F270I	chr17:757130A>T	homoynomous SNV	P8	168.47	22.22	96	162	Likely pathogenic	Uncertain significance		COSM43809 COSM1546805 COSM3937598 COSM43784	1 breast 1 large_intestine 2 oesophagus 2 lung			D s=0.002;rank=0.784	D s=1;rank=0.899	D s=0.994;rank=0.807	D s=0;rank=0.843	M s=1;rank=0.873	T s=2.225;rank=0.632	D s=6.8;rank=0.998	D s=5.2;rank=0.837	s=0.874;rank=0.862	T s=1.015;rank=0.974	D s=0.985;rank=0.996	s=0.693;rank=0.975	s=6.646;rank=0.911	s=0.992;rank=0.535	D s=0.998;rank=0.992	s=5.13;rank=0.696
ZEB2	NM_00117163	C801A	p.F267L	chr2:145158809 G>T	homoynomous SNV	P5	289.43	84.78	82	82	Likely pathogenic	Uncertain significance					D s=0.998;rank=0.715	D s=0.998;rank=0.875	D s=0;rank=0.843	D s=1;rank=0.588	M s=2.65;rank=0.778	T s=0.82;rank=0.741	D s=5.65;rank=0.87	T s=0.798;rank=0.911	T s=0.203;rank=0.776	T s=0.4;rank=0.752	s=0.099;rank=0.772	s=5.688;rank=0.776	s=0.998;rank=0.918	D s=0.977;rank=0.763	s=4.47;rank=0.536		
ZRSR2	NM_005089	E263delA	p.K88RfsX	chrX:15821871A>	frameshift deletion	P6	322.22	75.53	142	188	Uncertain significance	Uncertain significance					T s=0.13;rank=0.267	B s=0.047;rank=0.206	N s=0.024;rank=0.19	D s=0.043;rank=0.238	D s=0.726;rank=0.337	N s=1.795;rank=0.474	N s=2.03;rank=0.21	N s=1.78;rank=0.42	s=0.31;rank=0.371	T s=1.064;rank=0.107	s=0.036;rank=0.155	s=0.006;rank=0.162	s=2.645;rank=0.379	s=0.933;rank=0.224	D s=0.792;rank=0.389	s=1.22;rank=0.202	
ZRSR2	NM_005089	C505T	p.R169X	chrX:15827389C>T	stopgain	P2	93.07	121	130	Likely pathogenic	Likely pathogenic		COSM1716885	1 haematopoietic_and_lymphoid_tissue					N s=0.02;rank=0.272	A s=1;rank=0.873								s=12.662;rank=0.985	s=0.998;rank=0.919	D s=0.899;rank=0.503	s=5.46;rank=0.799		
ZRSR2	NM_005089	C700T	p.Q234X	chrX:15833942C>T	stopgain	P36	119.99	52.17	12	23	Likely pathogenic	Likely pathogenic							D s=0;rank=0.843	A s=1;rank=0.873								s=12.985;rank=0.988	s=0.998;rank=0.916	D s=0.906;rank=0.517	s=5.41;rank=0.782		
ZRSR2	NM_005089	G751A	p.G251R	chrX:15833993G>A	homoynomous SNV	P3	553.87	94.17	97	103	Likely pathogenic	Uncertain significance					D s=0;rank=0.912	D s=1;rank=0.899	D s=1;rank=0.971	D s=0;rank=0.843	H s=4.1;rank=0.974		D s=7.82;rank=0.96	s=0.938;rank=0.932	D s=0.449;rank=0.898	D s=0.605;rank=0.86	s=0.709;rank=0.976	s=6.676;rank=0.914	s=0.999;rank=0.996	D s=0.98;rank=0.788	s=5.41;rank=0.782		