Supplemental Table S1A: Details of amplicons used in Fluidigm Access Array

Chr	start	end	Gene	Amplicon ID	average read depth
chr20	31022180	31022428	ASXL1	ASXL1_13_1	1505
chr20	31023509	31023755	ASXL1	ASXL1_13_10	1826
chr20	31023671	31023920	ASXL1	ASXL1_13_11	1833
chr20	31023829	31024078	ASXL1	ASXL1_13_12	1782
chr20	31023981	31024218	ASXL1	ASXL1_13_13	1191
chr20	31024123	31024371	ASXL1	ASXL1_13_14	2066
chr20	31024278	31024518	ASXL1	ASXL1_13_15	1482
chr20	31024421	31024652	ASXL1	ASXL1_13_16	1880
chr20	31024557	31024806	ASXL1	ASXL1_13_17	1466
chr20	31024724	31024969	ASXL1	ASXL1_13_18	1370
chr20	31022350	31022586	ASXL1	ASXL1_13_2	1869
chr20	31022504	31022740	ASXL1	ASXL1_13_3	2497
chr20	31022651	31022894	ASXL1	ASXL1_13_4	2068
chr20	31022810	31023027	ASXL1	ASXL1_13_5	1958
chr20	31022949	31023198	ASXL1	ASXL1_13_6	1517
chr20	31023122	31023363	ASXL1	ASXL1_13_7	1463
chr20	31023306	31023533	ASXL1	ASXL1_13_8	1735
chr20	31023375	31023618	ASXL1	ASXL1_13_9	2860
chrX	39914487	39914706	BCOR	BCOR_11_1	2750
chrX	39914597	39914812	BCOR	BCOR_11_2	1932
chrX	39911465	39911709	BCOR	BCOR_14_1	1870
chrX	39933465	39933692	BCOR	BCOR_4a_1	1405
chrX	39932442	39932678	BCOR	BCOR_4b_1	1561
chrX	39932121	39932326	BCOR	BCOR_4c_1	2059
chrX	39922952	39923198	BCOR	BCOR_8_1	978
chr11	119155583	119155811	CBL	CBL_10_1	1287
chr11	119158381	119158620	CBL	CBL_12_2	1625
chr11	119158505	119158734	CBL	CBL_12_3	1007
chr11	119158610	119158859	CBL	CBL_12_5	736
chr11	119145434	119145649	CBL	CBL_5_1	1147
chr11	119145545	119145774	CBL	CBL_5_2	2148
chr11	119148351	119148598	CBL	CBL_8_1	1242
chr11	119148547	119148768	CBL	CBL_8_2	832
chr11	119148694	119148925	CBL	CBL_8_4	663
chr11	119148883	119149098	CBL	CBL_8_5	1429
chr11	119149095	119149343	CBL	CBL_9_1	837
chr11	119149237	119149461	CBL	CBL_9_2	1816
chr9	21974474	21974716	CDKN2A	CDKN2A_1_1	1084
chr9	21974588	21974803	CDKN2A	CDKN2A_1_2	1825
chr9	21970857	21971087	CDKN2A	CDKN2A_2_1	1593
chr9	21970995	21971236	CDKN2A	CDKN2A_2_2	2752
chr5	149441227	149441443	CSF1R	CSF1R_12_1	3087
chr5	149436727	149436974	CSF1R	CSF1R_17_1	930
chr5	149433804	149434052	CSF1R	CSF1R_21_1	1322
chr5	149433545	149433794	CSF1R	CSF1R_22_1	1418
chr5	149433611	149433828	CSF1R	CSF1R_22_2	2101
chr5	149459745	149459988	CSF1R	CSF1R_4_1	1533
chr5	149452803	149453037	CSF1R	CSF1R_7_1	2006
chr5	149452857	149453105	CSF1R	CSF1R_7_2	1803
chr5	149449646	149449894	CSF1R	CSF1R_9_1	1730
chr7	101877263	101877503	CUX1	CUX1_22_1	1937
chr7	101877337	101877579	CUX1	CUX1_22_2	1344
chr7	101747543	101747783	CUX1	CUX1_5_1	1426
chr5	149826388	149826610	del5q	del5q_SNP_1_1	1291

chr5	151169807	151170039	del5q	del5q_SNP_10_1	814
chr5	149460403	149460632	del5q	del5q_SNP_3_1	1595
chr5	149776061	149776288	del5q	del5q_SNP_4_1	1605
chr5	150489262	150489493	del5q	del5q_SNP_5_1	1899
chr5	150518846	150519090	del5q	del5q_SNP_6_1	1545
chr5	150639317	150639565	del5q	del5q_SNP_7_1	2357
chr5	150639317	150639565	del5q	del5q_SNP_8_1	2357
chr5	151043635	151043850	del5q	del5q_SNP_9_1	1439
chr2	25464383	25464623	DNMT3a	DNMT3a_17_1	1949
chr2	25463424	25463664	DNMT3a	DNMT3a_18_1	1978
chr2	25463106	25463352	DNMT3a	DNMT3a_19_1	1823
chr2	25461870	25462113	DNMT3A	DNMT3a_20_1	899
chr2	25459675	25459922	DNMT3a	DNMT3a_21_1	1682
chr2	25458516	25458751	DNMT3a	DNMT3a_22_1	719
chr2	25456997	25457198	DNMT3a	DNMT3a_23_1	2832
chr2	25457112	25457316	DNMT3a	DNMT3a_23_2	2003
chr2	25457153	25457372	DNMT3A	DNMT3a_23_3	2546
chr2	25470820	25471059	DNMT3a	DNMT3a_7_1	2667
chr2	25470931	25471160	DNMT3a	DNMT3a_7_2	1684
chr2	25470436	25470654	DNMT3a	DNMT3a_8_1	1515
chr2	25469856	25470097	DNMT3a	DNMT3a_9_1	2494
chr12	11992053	11992283	ETV6	ETV6_3_1	2090
chr12	12022589	12022825	ETV6	ETV6_5_1	1225
chr12	12022687	12022910	ETV6	ETV6_5_2	2041
chr12	12037320	12037562	ETV6	ETV6_6_1	1250
chr12	12038752	12038998	ETV6	ETV6_7_1	998
chr7	148508620	148508866	EZH2	EZH2_16_1	1582
chr13	28609548	28609764	FLT3	FLT3_ex12_1	2487
chr13	28609656	28609886	FLT3	FLT3_ex12_2	2276
chr13	28608365	28608586	FLT3	FLT3_ex13_1	1998
chr13	28608147	28608396	FLT3	FLT3_ex14_1	1356
chr13	28607941	28608157	FLT3	FLT3_ex15_1	2043
chr13	28602257	28602472	FLT3	FLT3_ex16_1	1792
chr13	28592520	28592767	FLT3	FLT3_ex20_1	1489
chr13	28635978	28636209	FLT3	FLT3_ex3_1	1750
chr13	28631412	28631660	FLT3	FLT3_ex4_1	610
chr13	28626652	28626877	FLT3	FLT3_ex5_1	867
chr13	28624182	28624429	FLT3	FLT3_ex6_1	526
chr13	28623460	28623681	FLT3	FLT3_ex8_1	2358
chr13	28623545	28623785	FLT3	FLT3_ex8_2	1310
chrX	48649448	48649664	GATA1	GATA1_2_1	1355
chrX	48649584	48649824	GATA1	GATA1_2_2	1054
chrX	48650115	48650359	GATA1	GATA1_3_1	1433
chrX	48650265	48650511	GATA1	GATA1_3_2	3308
chr2	209112978	209113226	IDH1	IDH1_ex4a_1	373
chr2	209112978	209113191	IDH1	IDH1_ex4a_2	1088
chr2	209113128	209113351	IDH1	IDH1_ex4a_3	1137
chr2	209113129	209113337	IDH1	IDH1_ex4a_4	1357
chr2	209113207	209113428	IDH1	IDH1_ex4a_5	2138
chr2	209113251	209113468	IDH1	IDH1_ex4a_6	2592
chr2	209109916	209110160	IDH1	IDH1_ex5_1	413
chr2	209110062	209110311	IDH1	IDH1_ex5_2	29
chr2	209104528	209104774	IDH1	IDH1_ex8_1	2189
chr2	209103763	209103997	IDH1	IDH1_ex9_1	1333
chr15	90631779	90631987	IDH2	IDH2_ex4_2	2007
chr15	90631793	90632008	IDH2	IDH2_ex4_3	1859
chr15	90631883	90632110	IDH2	IDH2_ex4_4	904
chr15	90628455	90628704	IDH2	IDH2_ex8_1	531

chr9	5069854	5070092	JAK2	JAK2_12_1	272
chr9	5072434	5072683	JAK2	JAK2_13_1	655
chr9	5073591	5073834	JAK2	JAK2_14_1	566
chr4	55593304	55593524	KIT	KIT_10_1	1881
chr4	55593504	55593748	KIT	KIT_11_1	904
chr4	55593920	55594154	KIT	KIT_12_1	1116
chr4	55594069	55594317	KIT	KIT_13_1	2207
chr4	55595383	55595606	KIT	KIT_14_1	964
chr4	55595485	55595718	KIT	KIT_14_2	1384
chr4	55597398	55597614	KIT	KIT_15_1	1100
chr4	55597967	55598196	KIT	KIT_16_1	1782
chr4	55599207	55599433	KIT	KIT_17_1	899
chr4	55602571	55602819	KIT	KIT_18_1	1484
chr4	55602826	55603070	KIT	KIT_19_1	1354
chr4	55561618	55561833	KIT	KIT_2_1	1790
chr4	55561741	55561967	KIT	KIT_2_2	2773
chr4	55603266	55603499	KIT	KIT_20_1	2039
chr4	55604524	55604773	KIT	KIT_21_1	1106
chr4	55564409	55564648	KIT	KIT_3_1	2027
chr4	55564525	55564769	KIT	KIT_3_2	1306
chr4	55565657	55565872	KIT	KIT_4_1	1681
chr4	55565765	55565981	KIT	KIT_4_2	1606
chr4	55569762	55570011	KIT	KIT_5_1	205
chr4	55569881	55570096	KIT	KIT_5_2	2300
chr4	55573148	55573392	KIT	KIT_6_1	1100
chr4	55573282	55573516	KIT	KIT_6_2	1870
chr4	55575514	55575763	KIT	KIT_7_1	619
chr4	55589685	55589925	KIT	KIT_8_1	1804
chr4	55591945	55592186	KIT	KIT_9_1	1305
chr4	55592015	55592263	KIT	KIT_9_2	1154
chr12	25398166	25398409	KRAS	KRAS_2_1	412
chr12	25380030	25380274	KRAS	KRAS_3_1	104
chr12	25380158	25380388	KRAS	KRAS_3_2	1038
chr12	25378404	25378624	KRAS	KRAS_4_1	1035
chr12	25378534	25378777	KRAS	KRAS_4_2	858
chr5	170834641	170834865	NPM1	NPM1_ex10_1	1297
chr5	170837475	170837718	NPM1	NPM1_ex11_1	664
chr1	115258607	115258834	NRAS	NRAS_2_4	1688
chr1	115256345	115256569	NRAS	NRAS_3_1	2585
chr1	115256425	115256648	NRAS	NRAS_3_2	246
chr4	55140956	55141194	PDFGRA	PDFGRA_12_1	1444
chr4	55144006	55144255	PDFGRA	PDFGRA_14_1	1378
chr4	55151953	55152176	PDFGRA	PDFGRA_18_1	1754
chrX	133559180	133559401	PHF6	PHF6_10_1	1652
chrX	133511574	133511822	PHF6	PHF6_2_1	985
chrX	133527442	133527657	PHF6	PHF6_4_1	1209
chrX	133527523	133527761	PHF6	PHF6_4_2	794
chrX	133527852	133528096	PHF6	PHF6_5_1	61
chrX	133547703	133547939	PHF6	PHF6_7_3	652
chrX	133547826	133548072	PHF6	PHF6_7_4	517
chrX	133548947	133549195	PHF6	PHF6_8_1	225
chrX	133551166	133551396	PHF6	PHF6_9_1	1318
chr10	89624181	89624400	PTEN	PTEN_1_1	1903
chr10	89692646	89692875	PTEN	PTEN_5_1	934
chr10	89692798	89693047	PTEN	PTEN_5_2	1434
chr10	89717566	89717800	PTEN	PTEN_7_1	1384
chr10	89717699	89717915	PTEN	PTEN_7_2	1161
chr10	89720550	89720789	PTEN	PTEN_8_1	139

chr10	89720693	89720932	PTEN	PTEN_8_2	871
chr12	112926779	112927028	PTPN11	PTPN11_ex13_1	1054
chr12	112888067	112888316	PTPN11	PTPN11_ex3_1	1382
chr12	112888130	112888375	PTPN11	PTPN11_ex3_2	2012
chr12	112915373	112915619	PTPN11	PTPN11_ex8_1	741
chr12	112915597	112915845	PTPN11	PTPN11_ex9_1	989
chr12	112915677	112915894	PTPN11	PTPN11_ex9_2	2378
chr21	36259031	36259263	RUNX1	RUNX1_1_6	1632
chr21	36259188	36259429	RUNX1	RUNX1_1_7	1995
chr21	36252795	36253038	RUNX1	RUNX1_2_1	1462
chr21	36231678	36231919	RUNX1	RUNX1_3_1	2134
chr21	36206617	36206863	RUNX1	RUNX1_4_1	617
chr21	36206757	36206993	RUNX1	RUNX1_4_2	619
chr21	36171562	36171810	RUNX1	RUNX1_5_1	1314
chr21	36160020	36160264	RUNX1	RUNX1_6_1	1444
chr21	36161185	36161410	RUNX1	RUNX1_6_10	1572
chr21	36161248	36161463	RUNX1	RUNX1_6_11	2230
chr21	36161393	36161639	RUNX1	RUNX1_6_12	1042
chr21	36161513	36161737	RUNX1	RUNX1_6_13	1252
chr21	36161665	36161914	RUNX1	RUNX1_6_14	326
chr21	36161829	36162078	RUNX1	RUNX1_6_15	479
chr21	36161993	36162220	RUNX1	RUNX1_6_16	1333
chr21	36162106	36162329	RUNX1	RUNX1_6_17	1127
chr21	36162260	36162502	RUNX1	RUNX1_6_18	847
chr21	36162443	36162675	RUNX1	RUNX1_6_19	30
chr21	36160176	36160425	RUNX1	RUNX1_6_2	1610
chr21	36162590	36162826	RUNX1	RUNX1_6_20	550
chr21	36162727	36162974	RUNX1	RUNX1_6_21	1627
chr21	36162885	36163134	RUNX1	RUNX1_6_22	770
chr21	36163042	36163285	RUNX1	RUNX1_6_23	1106
chr21	36163236	36163462	RUNX1	RUNX1_6_24	1972
chr21	36163331	36163570	RUNX1	RUNX1_6_25	804
chr21	36163482	36163726	RUNX1	RUNX1_6_26	42
chr21	36163666	36163915	RUNX1	RUNX1_6_27	1
chr21	36163758	36163975	RUNX1	RUNX1_6_28	3
chr21	36163891	36164119	RUNX1	RUNX1_6_29	641
chr21	36160333	36160566	RUNX1	RUNX1_6_3	1252
chr21	36164011	36164258	RUNX1	RUNX1_6_30	146
chr21	36164079	36164294	RUNX1	RUNX1_6_32	753
chr21	36164236	36164451	RUNX1	RUNX1_6_33	3
chr21	36164236	36164482	RUNX1	RUNX1_6_35	0
chr21	36164342	36164580	RUNX1	RUNX1_6_36	7
chr21	36164424	36164642	RUNX1	RUNX1_6_38	719
chr21	36164467	36164699	RUNX1	RUNX1_6_39	1106
chr21	36160483	36160732	RUNX1	RUNX1_6_4	857
chr21	36164617	36164843	RUNX1	RUNX1_6_41	1617
chr21	36164728	36164953	RUNX1	RUNX1_6_43	2580
chr21	36160624	36160851	RUNX1	RUNX1_6_5	580
chr21	36160716	36160962	RUNX1	RUNX1_6_6	318
chr21	36160821	36161020	RUNX1	RUNX1_6_7	90
chr21	36160940	36161158	RUNX1	RUNX1_6_8	1587
chr21	36161054	36161270	RUNX1	RUNX1_6_9	1400
chr2	198267134	198267383	SF3b1	SF3b1_14_1	539
chr2	198267302	198267540	SF3b1	SF3b1_14_2	2395
chr2	198267361	198267588	SF3b1	SF3b1_14_3	2019
chr2	198266592	198266833	SF3b1	SF3b1_15_1	525
chr2	198266721	198266957	SF3b1	SF3b1_15_2	988
chr2	198266399	198266616	SF3b1	SF3b1_16_1	457

chr2	198266496	198266743	SF3b1	SF3b1_16_2	189
chrX	53432360	53432608	SMC1A	SMC1A_ex11_1	1360
chrX	53432408	53432651	SMC1A	SMC1A_ex11_2	1607
chrX	53431881	53432117	SMC1A	SMC1A_ex13_1	2045
chrX	53430422	53430657	SMC1A	SMC1A_ex15_1	1096
chrX	53426455	53426681	SMC1A	SMC1A_ex16_1	2657
chrX	53423341	53423572	SMC1A	SMC1A_ex17_1	1758
chrX	53423089	53423338	SMC1A	SMC1A_ex18_1	1319
chrX	53421613	53421844	SMC1A	SMC1A_ex19_1	42
chrX	53409097	53409342	SMC1A	SMC1A_ex22_1	1722
chrX	53407479	53407716	SMC1A	SMC1A_ex24_1	1850
chrX	53440148	53440382	SMC1A	SMC1A_ex4_1	1354
chrX	53438830	53439075	SMC1A	SMC1A_ex6_1	1273
chrX	53438637	53438879	SMC1A	SMC1A_ex7_1	892
chr10	112343004	112343250	SMC3	SMC3_ex11_1	1185
chr10	112343146	112343381	SMC3	SMC3_ex11_2	2558
chr10	112343523	112343759	SMC3	SMC3_ex12_1	1098
chr10	112343847	112344094	SMC3	SMC3_ex13_1	1206
chr10	112356060	112356282	SMC3	SMC3_ex19_1	892
chr10	112356153	112356369	SMC3	SMC3_ex19_2	1814
chr10	112361612	112361861	SMC3	SMC3_ex25_1	486
chr10	112361779	112361994	SMC3	SMC3_ex25_2	1291
chr9	72933602	72933822	SMC5	SMC5_ex15_1	101
chr9	72933743	72933981	SMC5	SMC5_ex15_2	1083
chr9	72938809	72939058	SMC5	SMC5_ex17_2	206
chr9	72938912	72939149	SMC5	SMC5_ex17_3	892
chr9	72939017	72939246	SMC5	SMC5_ex17_4	334
chr9	72961917	72962164	SMC5	SMC5_ex20_1	466
chr9	72962725	72962940	SMC5	SMC5_ex22_1	601
chr9	72962856	72963105	SMC5	SMC5_ex22_2	14
chr9	72964940	72965168	SMC5	SMC5_ex23_1	1174
chr9	72965071	72965298	SMC5	SMC5_ex23_2	1797
chr17	74732803	74733040	SRSF2	SRSF2_1_1_1	483
chr17	74732965	74733210	SRSF2	SRSF2_1_1_2	1607
chr17	74733077	74733308	SRSF2	SRSF2_1_1_3	2067
chr17	74733227	74733459	SRSF2	SRSF2_1_1_4	2049
chrX	123197579	123197828	STAG2	STAG2_20_1	413
chrX	123197701	123197944	STAG2	STAG2_20_2	431
chrX	123217168	123217399	STAG2	STAG2_29_1	492
chrX	123217261	123217490	STAG2	STAG2_29_2	906
chrX	123220280	123220502	STAG2	STAG2_30_1	1989
chrX	123220430	123220677	STAG2	STAG2_30_2	315
chr4	106193655	106193871	TET2	TET2_10_1	2610
chr4	106193777	106194026	TET2	TET2_10_2	1751
chr4	106193887	106194135	TET2	TET2_10_3	868
chr4	106196139	106196382	TET2	TET2_11_1	793
chr4	106197178	106197419	TET2	TET2_11_10	1695
chr4	106197259	106197504	TET2	TET2_11_11	1615
chr4	106197335	106197574	TET2	TET2_11_12	1419
chr4	106197475	106197721	TET2	TET2_11_13	1428
chr4	106197624	106197862	TET2	TET2_11_14	1395
chr4	106197780	106198029	TET2	TET2_11_15	315
chr4	106197840	106198070	TET2	TET2_11_16	476
chr4	106198003	106198238	TET2	TET2_11_17	1365
chr4	106198117	106198356	TET2	TET2_11_18	783
chr4	106198234	106198470	TET2	TET2_11_19	848
chr4	106196304	106196541	TET2	TET2_11_2	2235
chr4	106198365	106198609	TET2	TET2_11_20	180

chr4	106198432	106198652	TET2	TET2_11_21	258
chr4	106198561	106198776	TET2	TET2_11_22	1302
chr4	106198728	106198976	TET2	TET2_11_23	1248
chr4	106198792	106199034	TET2	TET2_11_24	1129
chr4	106198952	106199198	TET2	TET2_11_25	2016
chr4	106199141	106199387	TET2	TET2_11_26	0
chr4	106199223	106199469	TET2	TET2_11_27	0
chr4	106199390	106199605	TET2	TET2_11_28	1036
chr4	106199507	106199747	TET2	TET2_11_29	147
chr4	106196432	106196680	TET2	TET2_11_3	1259
chr4	106199652	106199885	TET2	TET2_11_30	214
chr4	106199726	106199956	TET2	TET2_11_31	380
chr4	106199852	106200095	TET2	TET2_11_32	1159
chr4	106200045	106200286	TET2	TET2_11_33	674
chr4	106200156	106200404	TET2	TET2_11_34	788
chr4	106200320	106200558	TET2	TET2_11_35	569
chr4	106200470	106200706	TET2	TET2_11_36	1161
chr4	106200617	106200833	TET2	TET2_11_37	1267
chr4	106200746	106200983	TET2	TET2_11_38	66
chr4	106200866	106201113	TET2	TET2_11_39	25
chr4	106196565	106196788	TET2	TET2_11_4	2022
chr4	106196691	106196912	TET2	TET2_11_5	2056
chr4	106196754	106196976	TET2	TET2_11_6	2368
chr4	106196885	106197110	TET2	TET2_11_7	2479
chr4	106197045	106197294	TET2	TET2_11_8	1312
chr4	106197099	106197348	TET2	TET2_11_9	1236
chr4	106154923	106155165	TET2	TET2_3_1	287
chr4	106156339	106156580	TET2	TET2_3_10	1968
chr4	106156496	106156731	TET2	TET2_3_11	3287
chr4	106156624	106156873	TET2	TET2_3_12	1568
chr4	106156795	106157031	TET2	TET2_3_13	2224
chr4	106156947	106157174	TET2	TET2_3_14	2935
chr4	106157101	106157342	TET2	TET2_3_15	1473
chr4	106157245	106157488	TET2	TET2_3_16	1101
chr4	106157388	106157627	TET2	TET2_3_17	1556
chr4	106157509	106157758	TET2	TET2_3_18	1811
chr4	106157670	106157919	TET2	TET2_3_19	2741
chr4	106155100	106155324	TET2	TET2_3_2	2217
chr4	106157841	106158080	TET2	TET2_3_20	1455
chr4	106157921	106158168	TET2	TET2_3_21	1577
chr4	106158087	106158319	TET2	TET2_3_22	2927
chr4	106158142	106158391	TET2	TET2_3_23	2165
chr4	106158304	106158548	TET2	TET2_3_24	1218
chr4	106155248	106155477	TET2	TET2_3_3	2736
chr4	106155401	106155633	TET2	TET2_3_4	1749
chr4	106155553	106155789	TET2	TET2_3_5	1189
chr4	106155711	106155942	TET2	TET2_3_6	2483
chr4	106155864	106156112	TET2	TET2_3_7	1961
chr4	106156042	106156290	TET2	TET2_3_8	1110
chr4	106156190	106156425	TET2	TET2_3_9	1582
chr4	106162419	106162667	TET2	TET2_4_1	518
chr4	106163873	106164112	TET2	TET2_5_1	990
chr4	106164658	106164880	TET2	TET2_6_1	1963
chr4	106164737	106164979	TET2	TET2_6_2	1857
chr4	106180721	106180965	TET2	TET2_7_1	1566
chr4	106182802	106183001	TET2	TET2_8_2	206
chr4	106182855	106183070	TET2	TET2_8_4	385
chr4	106190625	106190860	TET2	TET2_9_1	943

chr4	106190769	106190984	TET2	TET2_9_2	1790
chr17	7573848	7574065	TP53	TP53_10_1	2354
chr17	7572225	7572469	TP53	TP53_11_10	822
chr17	7572369	7572607	TP53	TP53_11_11	1694
chr17	7572530	7572763	TP53	TP53_11_12	1957
chr17	7572671	7572920	TP53	TP53_11_13	778
chr17	7572812	7573049	TP53	TP53_11_14	648
chr17	7571643	7571869	TP53	TP53_11_2	1017
chr17	7571767	7572013	TP53	TP53_11_3	693
chr17	7571817	7572033	TP53	TP53_11_5	1852
chr17	7572006	7572211	TP53	TP53_11_6	76
chr17	7572009	7572258	TP53	TP53_11_7	53
chr17	7572174	7572392	TP53	TP53_11_8	2102
chr17	7579755	7579972	TP53	TP53_2_1	1144
chr17	7579547	7579777	TP53	TP53_3_1	926
chr17	7579259	7579508	TP53	TP53_4_1	924
chr17	7579378	7579618	TP53	TP53_4_2	623
chr17	7578253	7578502	TP53	TP53_5_1	1381
chr17	7578360	7578608	TP53	TP53_5_2	1639
chr17	7578089	7578320	TP53	TP53_6_1	1943
chr17	7577428	7577671	TP53	TP53_7_1	2954
chr17	7576957	7577194	TP53	TP53_8_1	1995
chr17	7576753	7576978	TP53	TP53_9_1	2588
chr21	44524292	44524535	U2AF1	U2AF1_1_1	895
chr21	44514679	44514928	U2AF1	U2AF1_5_1	1656
chr11	32417742	32417986	WT1	WT1_7_1	1138
chr11	32413390	32413637	WT1	WT1_9_1	1234
chrX	15821728	15821950	ZRSR2	ZRSR2_ex4_1	1333
chrX	15822159	15822392	ZRSR2	ZRSR2_ex5_1	1874

Supplemental Table S1B: Exon coverage of amplicon panel

Gene	Total Exon Count	No. of exons in gene	No. of exons covered by our panel	Exon(s) covered
ASXL1	12	12	1	12
BCOR	15	14	4	4 (partial), 8, 12, 15
CBL	16	16	6	5, 7-10, 12
CDKN2A	3	2	2	1, 2
CSF1R	22	21	8	3,4,7,9,12,17,21,22
CUX1	24	24	2	6, 22
DNMT3A	23	22	10	7-9, 17-23
ETV6	8	8	4	3, 5, 6, 7
EZH2	20	19	1	16
FLT3	24	24	10	3-6, 8, 12-16
GATA1	6	5	2	2, 3
IDH1	10	8	4	4, 5, 8, 9
IDH2	11	11	2	4, 8
JAK2	25	23	3	12-14
KIT	21	21	20	2-21
KRAS	6	4	3	2, 3, 4
NPM1	11	11	2	10, 11
NRAS	7	4	2	2, 3
PDGFRA	23	22	3	12, 14, 18
PHF6	10	9	5	4, 5, 7, 8, 9
PTEN	9	9	4	1, 5, 7, 8
PTPN11	16	15	4	3, 8, 9, 13
RUNX1	9	8	6	3-9
SF3B1	25	25	3	14-16

SMC1A	25	25	12	4, 6, 7, 11, 13, 15-19, 22, 24
SMC3	29	29	5	11-13, 19, 25
SMC5	25	25	5	15, 17, 20, 22, 23
SRSF2	3	2	1	1
STAG2	35	33	3	20, 29, 30
TET2	11	9	9	3-11
TP53	11	8	10	2-11
U2AF1	8	8	2	2, 6
WT1	10	10	2	7, 9
ZRSR2	11	11	2	4, 5

Supplemental Table S1C: FLT3 ITD detection by capillary electrophoresis and NGS

Patient ID		Genescan Capillary Electrophoresis	Detection by NGS and VAF%
1	FLT3 ITD	detected	15.22%
2	FLT3 ITD	detected	not detected
3	FLT3 ITD	detected	8.92%
4	FLT3 ITD	detected	not detected
6	FLT3 ITD	detected	not detected
7	FLT3 ITD	detected	not detected
10	FLT3 ITD	detected	42.32%
14	FLT3 ITD	detected	14.20%
15	FLT3 ITD	detected	not detected
17	FLT3 ITD	detected	1.12%
21	FLT3 ITD	detected	50.14%
23	FLT3 ITD	detected	not detected
25	FLT3 ITD	detected	not detected
27	FLT3 ITD	detected	not detected
29	FLT3 ITD	detected	not detected
31	FLT3 ITD	detected	5.27%
33	FLT3 ITD	detected	16.24%
37	FLT3 ITD	detected	40.18%
40	FLT3 ITD	detected	29.62%
41	FLT3 ITD	detected	40.22%
45	FLT3 ITD	detected	59.27%
49	FLT3 ITD	detected	not detected
51	FLT3 ITD	detected	not detected
53	FLT3 ITD	detected	22.63%
54	FLT3 ITD	detected	not detected
56	FLT3 ITD	detected	23.02%
60	FLT3 ITD	detected	not detected
61	FLT3 ITD	detected	33.50%
68	FLT3 ITD	detected	not detected
71	FLT3 ITD	detected	18.04%
76	FLT3 ITD	detected	not detected
77	FLT3 ITD	detected	36.47%
79	FLT3 ITD	detected	not detected
81	FLT3 ITD	detected	not detected
82	FLT3 ITD	detected	33.41%
85	FLT3 ITD	detected	16.85%
87	FLT3 ITD	detected	49.64%
93	FLT3 ITD	detected	17.07%
95	FLT3 ITD	detected	10.61%
104	FLT3 ITD	detected	39.60%
106	FLT3 ITD	detected	8.74%
111	FLT3 ITD	detected	6.94%
113	FLT3 ITD	detected	5.70%

Supplemental Table S1D: ddPCR primers and probes

Variant	ddPCR forward primer 5' - 3'	ddPCR reverse primer 5' - 3'	ddPCR wild type probe 5' HEX - *sequence* - 3' BHQ-1	ddPCR mutant probe 5' 6-FAM - *sequence* - 3' BHQ-1
NRAS Q61R	CAA ATA CAC AGA GGA AGC C	GGT GAA ACC TGT TTG TTG GAC	*/5'HEX/ CTG TAC TCT TCT T GT CCA GCT G /3'BHQ-1/*	*/5'6-FAM/ TG TAC TCT TCT C GT CCA GCT G /3'BHQ-1/*
NRAS Q61K	CAA ATA CAC AGA GGA AGC C	GGT GAA ACC TGT TTG TTG GAC	*/5'HEX/ CTG TAC TCT TCT T G T CCA GCT G /3'BHQ-1/*	*/5'6-FAM/ A CTG TAC TCT TCT T T T CCA GCT G /3'BHQ-1/*
IDH2 R140Q	TTG CAG ATG ATG GGC TC	CTC ACA GAG TTC AAG CTG	*/5'HEX/ TGG AAC TAT CC G GAA CAT CCT G /3'BHQ-1/*	*/5'6-FAM/ TGG AAC TAT CC A GAA CAT CCT GG /3'BHQ-1/*
SRSF2 P95R	CGT TTA CCT GCG GCT C	CTG AGG ACG CTA TGG ATG	*/5'HEX/ CGC C C C CCG GAC TCA CA /3'BHQ-1/*	*/5'6-FAM/ CGC C G C CCG GAC TCA C /3'BHQ-1/*
DNMT3A G706W	GTT GAC GAT GGA GAG GTC	CCT TTA TCC TCC CAG ATC C	*/5'HEX/ CTG CCC C C A ATC ACC AGA TC /3'BHQ-1/*	*/5'6-FAM/ CTG CCC C A A ATC ACC AGA TC /3'BHQ-1/*
DNMT3A K826R	CCA GCA GAG GTT CTA GAC	GTC TCT CTT CTG CCT CC	*/5'HEX/ CTG ACC T TG GCT ATC CTG C /3'BHQ-1/*	*/5'6-FAM/ CTG ACC C TG GCT ATC CTG C /3'BHQ-1/*
TET2 Y867H	GCA GGA AAC AAG ACC CAA AAC	ACC TGT GAA GAA GAT CTT GC	*/5'HEX/ CAT GCA A T A TTT TCC AAA TAA TGT GAT CC /3'BHQ-1/*	*/5'6-FAM/ CAT GCA A C A TTT TCC AAA TAA TGT GAT C /3'BHQ-1/*
TET2 P1723S	CAG CAG TTG TAC CAT TAG ACC	GAG GTG GCT CCC ATG AAG	*/5'HEX/ AAT TGC CT C CTT ATC CCA CTC ATG /3'BHQ-1/*	*/5'6-FAM/ A AAT TGC CT T CTT ATC CCA CTC ATG /3'BHQ-1/*

Supplemental Table S2: Mutations detected in patient cohort at diagnosis.

Patient ID	ASXL1	BCOR	CBL	CSF1R	DNMT3A R882C/H	DNMT3A other	ETV6	EZH2	FLT3 D835Y	FLT3 other point mutation	FLT3 ITD	IDH1 R132C/G/H/S	IDH1 E62K	IDH2 R140Q	IDH2 R172K	JAK2 V617F	KIT other poINT mutation	KIT D812V/ D816V	KRAS G12C/D/V	NPM1 W288fs	NPM1 S525C	NRAS G13D	NRAS G12C/D/S	NRAS Q61K/R	PHF6	PTPN11 other mutations	PTPN11 D61Y	RUNX1	RUNX1 splicing	SF3B1 K700E	SMC3	SMC5	SRSF2 P95A/H/L/R	TET2	TP53	U2AF1 Q157R	WT1	ZRSR2 R99W	total no. of mutations	cytogenetics	clinical outcome
1											0.15									0.39																			2	INT	R
2				0.51							det	0.21								0.48																			4	INT	
3											0.09			0.41						0.36																			4	INT	R
4											0.00	0.38																											2	INT	
5	0.39																																0.57						2	INT	
6	det		0.40								0.00	0.19																0.25	0.32				0.58						6	INT	
7									0.23		det	0.42								0.35		0.11																	6	INT	
8			0.28											0.42																			0.11						3	INT	
9														0.38																									1	INT	
10											0.42																												1	INT	R
11								0.36																		0.45		0.35											3	INT	R
12												0.24														0.19	0.11												3	INT	
13	0.20																																		0.09				2	INT	
14					0.45						0.14						0.32			0.39																			4	INT	
15	det										0.46	0.18																0.35, 0.36, 0.08											6	INT	
16							0.54																0.12			0.44													3	ADV	
17											0.01													0.05															2	INT	R
18					0.49				0.45					0.45																									3	INT	R
19																																			0.05				1	ADV	R
20																						0.29																	1	FAV	
21											0.50																												2	ADV	R
22																	0.45							0.33													0.41		3	INT	
23											0.01									0.38														0.15, 0.38					4	INT	
24																	0.36									0.06													2	FAV	
25											0.10																												1	ADV	R
26																												0.85										1	INT	
27											0.28																												1	INT	R
28																															0.51							1	INT	
29											0.02									0.30																			2	INT	
30																												0.25						0.20				2	INT	
31											0.05									0.46																			2	INT	
32												0.19																								0.40			2	INT	
33											0.16									0.43														0.45			0.31, 0.06		5	INT	
34	0.43																											0.11										2	INT	
35														0.46																			0.26						2	INT	R
36									0.49											0.39																			2	INT	
37											0.40	0.41																0.20					0.62	0.23					5	INT	
38							0.38		0.49	0.15										0.43														0.25					5	INT	R
39														0.46						0.34																			2	INT	R
40											0.30	0.44																											2	INT	R
41	0.51										0.40			0.52						0.44																			5	INT	R
42				0.84																								0.26				0.39			0.09				4	ADV	R
43					0.41															0.33																			2	INT	
44																																							0	INT	
45					0.41						0.59									0.32																	0.12		4	INT	
46																	0.35																				0.65		2	ADV	R
47																	0.46																	0.55					2	INT	
48																																							0	ADV	
49											det																	0.42						0.47					3	INT	R
50																								0.26										0.06					2	INT	R
51											0.68																												1	ADV	
52												0.44								0.43			0.23			0.14													4	INT	
53					0.22						0.23																												2	INT	
54											0.71																												1	FAV	R
55																																							0	INT	R
56					0.42						0.23									0.33																			3	INT	R

57																								0.62																1	INT	
58																				0.31																			0.63	2	INT	
59																																									0	INT	
60						0.44					0.02																													2	INT	R
61											0.33										0.39																		0.42		3	INT	R
62	0.51																								0.26			0.41, 0.41							0.86				5	ADV	R
63																																								0	INT	
64																														0.11										1	ADV	R
65																												0.56, 0.07					0.48	0.52					4	INT	R
66																								0.49																1	ADV	R
67												0.38													0.63			0.17												3	INT	
68											0.82										0.36																			2	INT	R
69																									0.41			0.30					0.42	0.58					4	INT	
70	0.44		0.95																															0.97					3	INT	
71							0.68				0.18																								0.43				3	INT	R
72																			0.32																				1	ADV	
73																																		0.49, 0.49					2	INT	
74																																		0.49					1	ADV	R
75														0.47																			0.07						2	INT	
76	0.45				0.38						0.17										0.29			0.28															5	INT	R
77											0.36	0.46									0.43																			3	INT	
78																																								0	INT	R
79											0.02								0.08	0.40																			3	ADV	
80																																		0.47					1	INT	
81											0.31														0.88								0.72	0.52					4	INT	
82											0.33																							0.46					2	INT	
83						0.09									0.15																								2	INT	R
84																												0.15					0.55						2	INT	R
85											0.17																								0.14				2	ADV	R
86																																		0.54	0.45				2	INT	
87											0.50										0.37																		2	INT	
88																												0.06					0.12						2	INT	R
89																	0.44	0.38																0.48					3	ADV	R
90						0.21									0.32																								2	INT	R
91		1.00				0.29																0.09																	3	ADV	R
92				0.47								0.41																					0.72	0.46					4	INT	R
93						0.25					0.17										0.31														0.13				4	INT	
94																																								0	INT	
95					0.49						0.11										0.41																			3	INT	R
96																				0.26																				1	ADV	R
97																												0.25											1	INT	
98										0.45						0.44																								2	INT	
99						0.90									0.47																									2	INT	
100										0.49				0.19		0.14								0.06					0.26					0.45, 0.40					7	INT	
101																																							0	FAV	R
102																																							0	ADV	R
103										0.50									0.37		0.43																			3	INT	
104											0.40																								0.36				2	INT	R
105																	0.58							0.37															2	UNK	
106											0.09										0.34													0.80					3	INT	
107													0.47	0.46							0.34																			3	INT	
108																																							0	FAV	
109																																							0	ADV	
110															0.15																								1	INT	
111											0.07																								0.07				2	ADV	R
112																																							0	ADV	R
113					0.46						0.06																							0.23					3	INT	R

Numbers beneath genes/ mutations refer to variant allele frequency as ratio of 1.0 (multiple by 100% for %VAF)

det= FLT3 ITD detected by non-NGS methods

cytogenetic risk groups: FAV= favourable, INT=intermediate, ADV= adverse Clinical outcome: R= relapse post-allogeneic SCT

Supplemental Table S3A: called nucleotide variants detected by NGS at diagnosis (excluding *FLT3* ITD)

Patient ID	Chr	Start	End	reference	alternative	Exon/ Intron	Gene	Amino acid change	VAF%	Exonic Function	dbSNP identifier	Cosmic identifier (v.68)	Notes for inclusion
1	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	38.95%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
2	chr5	170837547	170837547	-	CTTG	exonic	NPM1	W288fs	48.46%	frameshift insertion		COSM20811	Previously reported in haematopoietic samples in COSMIC database
2	chr2	209113113	209113113	G	C	exonic	IDH1	R132G	21.01%	nonsynonymous SNV		COSM28749	Previously reported in haematopoietic samples in COSMIC database
2	chr5	149433888	149433888	C	G	exonic	CSF1R	E920D	50.77%	nonsynonymous SNV	rs34030164		Previously reported in human variation database frequency less than 0.14%
3	chr2	25469933	25469933	T	-	exonic	DNMT3A	Y370fs	46.11%	frameshift deletion			frameshift indel, nonsense or splicing variant
3	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	36.11%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
3	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	41.31%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
4	chr2	209113112	209113112	C	T	exonic	IDH1	R132H	37.64%	nonsynonymous SNV	rs121913500	COSM28746	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
5	chr20	31022712	31022712	C	T	exonic	ASXL1	Q733X	38.96%	stopgain		COSM1318847	Previously reported in haematopoietic samples in COSMIC database
5	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	56.87%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
6	chr21	36171599	36171599	-	GA	exonic	RUNX1	S322fs	24.74%	frameshift insertion			Frameshift indel detected in Papaemmanuil NEJM 2016 in same codon
6	chr21	36206899	36206899	C	T	splicing	RUNX1	splicing	31.76%	Splicing			frameshift indel, nonsense or splicing variant
6	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	58.35%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
6	chr11	119148875	119148875	G	C	splicing	CBL	splicing	40.02%	Splicing		COSM34063	Previously reported in haematopoietic samples in COSMIC database
6	chr2	209113113	209113113	G	A	exonic	IDH1	R132C	19.33%	nonsynonymous SNV	rs121913499	COSM28747	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
7	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	34.58%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
7	chr2	209113112	209113112	C	T	exonic	IDH1	R132H	41.72%	nonsynonymous SNV	rs121913500	COSM28746	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
7	chr1	115258744	115258744	C	T	exonic	NRAS	G13D	10.84%	nonsynonymous SNV	rs121434596	COSM573	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
7	chr13	28592642	28592642	C	A	exonic	FLT3	D835Y	22.88%	nonsynonymous SNV	rs121913488	COSM783	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
7	chr2	25470590	25470590	A	T	exonic	DNMT3A	L295Q	38.75%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 1 codon COSM4383605 V296L
8	chr17	74732959	74732959	G	A	exonic	SRSF2	P95L	11.35%	nonsynonymous SNV		COSM146288,COSM211506,COSM211028	Previously reported in haematopoietic samples in COSMIC database
8	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	41.76%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
8	chr11	119145633	119145633	G	A	exonic	CBL	R280Q	27.72%	nonsynonymous SNV	rs145155035		Previously reported in human variation database frequency less than 0.14%
9	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	37.94%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
11	chr21	36252922	36252922	-	TGGGAAAA	exonic	RUNX1	A147fs	35.43%	frameshift insertion		COSM1717871	Previously reported in non-haematopoietic sample in COSMIC database, insertion detected within 1 codon COSM1717871 T148fs
11	chr7	148508788	148508788	C	T	exonic	EZH2	V626M	36.22%	nonsynonymous SNV		COSM1000723	Previously reported in haematopoietic samples in COSMIC database
11	chr12	112926887	112926887	G	A	exonic	PTPN11	G503R	44.67%	nonsynonymous SNV		COSM14259	Previously reported in haematopoietic samples in COSMIC database
12	chr12	112888165	112888165	G	T	exonic	PTPN11	D61Y	11.37%	nonsynonymous SNV		COSM13011	Previously reported in haematopoietic samples in COSMIC database
12	chr12	112926890	112926890	A	G	exonic	PTPN11	M504V	19.48%	nonsynonymous SNV		COSM4603683	Previously reported in haematopoietic samples in COSMIC database
12	chr2	209113112	209113112	C	T	exonic	IDH1	R132H	23.58%	nonsynonymous SNV	rs121913500	COSM28746	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
13	chr20	31022592	31022592	C	T	exonic	ASXL1	R693X	19.87%	stopgain		COSM51388	Previously reported in haematopoietic samples in COSMIC database
13	chr17	7577539	7577539	G	A	exonic	TP53	R116W,TP53	9.48%	nonsynonymous SNV	rs121912651	COSM120006,COSM120005,COSM1640831,COSM10656,COSM120007	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
14	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	39.27%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
14	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	45.43%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
14	chr4	55598108	55598108	T	A	exonic	KIT	L765M	31.79%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 3 codons COSM5496106 D768N
15	chr21	36252945	36252949	GTTTT	-	exonic	RUNX1	E138fs	35.89%	frameshift deletion			Frameshift indel detected in Papaemmanuil NEJM 2016 in same codon
15	chrX	36259234	36259234	G	-	exonic	RUNX1	P86fs	7.63%	frameshift deletion			frameshift indel, nonsense or splicing variant
15	chr20	31022403	31022425	CACCACTGCCATAGAGAGGCGGC	-	exonic	ASXL1	H630fs	58.04%	frameshift deletion		COSM51200	Previously reported in haematopoietic samples in COSMIC database
15	chr2	209113113	209113113	G	A	exonic	IDH1	R132C	17.78%	nonsynonymous SNV	rs121913499	COSM28747	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
15	chr21	36252943	36252943	T	A	exonic	RUNX1	Y140F	34.79%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 1 codon COSM1690275 S141L
16	chr12	11992215	11992220	TTCGCT	-	exonic	ETV6	102_104del	53.55%	nonframeshift deletion			frameshift indel, nonsense or splicing variant
16	chr12	112888210	112888210	G	C	exonic	PTPN11	E76Q	43.93%	nonsynonymous SNV		COSM13016	Previously reported in haematopoietic samples in COSMIC database
16	chr1	115258747	115258747	C	T	exonic	NRAS	G12D	11.73%	nonsynonymous SNV	rs121913237	COSM564	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
17	chr1	115256529	115256529	T	C	exonic	NRAS	Q61R	5%	nonsynonymous SNV	rs11554290	COSM584	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
18	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	45.28%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
18	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	49.13%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
18	chr13	28592642	28592642	C	A	exonic	FLT3	D835Y	45.06%	nonsynonymous SNV	rs121913488	COSM783	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%

19	chr17	7578203	7578203	C	T	exonic	TP53	V84M	5.34%	nonsynonymous SNV		COSM120097,COSM120095,COSM10667,COSM120096,COSM1644280,COSM120098	Previously reported in haematopoietic samples in COSMIC database
20	chr1	115258744	115258744	C	T	exonic	NRAS	G13D	28.51%	nonsynonymous SNV	rs121434596	COSM573	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
21	chr2	25464463	25464463	C	A	exonic	DNMT3A	V684F	47.94%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 1 codon COSM1690275 G685E
22	chr11	32413565	32413565	C	T	exonic	WT1	R462Q	41.16%	nonsynonymous SNV		COSM21434	Previously reported in haematopoietic samples in COSMIC database
22	chr1	115256530	115256530	G	T	exonic	NRAS	Q61K	33.08%	nonsynonymous SNV	rs121913254	COSM580	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
22	chr4	55593437	55593437	G	A	exonic	KIT	V528I	44.71%	nonsynonymous SNV	rs55792975		Previously reported in human variation database frequency less than 0.14%
23	chr4	106158253	106158253	A	-	exonic	TET2	K1052fs	14.83%	frameshift deletion			frameshift indel, nonsense or splicing variant
23	chr4	106180785	106180785	C	G	exonic	TET2	C1271W	38.37%	nonsynonymous SNV		COSM120176	Previously reported in haematopoietic samples in COSMIC database
23	chr5	170837547	170837547	-	CCGG	exonic	NPM1	W288fs	37.65%	frameshift insertion		COSM20806	Previously reported in haematopoietic samples in COSMIC database
24	chr4	55589771	55589776	ACGACA	-	exonic	KIT	418_420del	35.82%	nonframeshift deletion			frameshift indel, nonsense or splicing variant
24	chr12	112888198	112888198	G	A	exonic	PTPN11	A72T	6.21%	nonsynonymous SNV		COSM13014	Previously reported in haematopoietic samples in COSMIC database
26	chr21	36252866	36252866	G	A	exonic	RUNX1	R166X	84.85%	stopgain		COSM24769	Previously reported in haematopoietic samples in COSMIC database
28	chr10	110584233	110584233	G	A	exonic	SMC3	R381Q	50.53%	nonsynonymous SNV		COSM1317491	Previously reported in haematopoietic samples in COSMIC database
29	chr10	112343991	112343991	G	A	exonic	SMC3	R381Q	50.53%	nonsynonymous SNV		COSM1317491	Previously reported in haematopoietic samples in COSMIC database
29	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	29.90%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
30	chr21	36164830	36164830	A	-	exonic	RUNX1	Y349fs	25.46%	frameshift deletion			frameshift indel, nonsense or splicing variant
30	chr4	106196770	106196770	G	A	exonic	TET2	M1701I	20.45%	nonsynonymous SNV	rs62623390		Previously reported in human variation database frequency less than 0.14%
31	chr5	170837547	170837547	-	CCAG	exonic	NPM1	W288fs	46.44%	frameshift insertion		COSM20809	Previously reported in haematopoietic samples in COSMIC database
32	chrX	44514777	44514777	T	C	exonic	U2AF1	Q84R	40.02%	nonsynonymous SNV		COSM211532,COSM1724986	Previously reported in haematopoietic samples in COSMIC database
32	chr2	209113113	209113113	G	A	exonic	IDH1	R132C	18.69%	nonsynonymous SNV	rs121913499	COSM28747	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
33	chr11	32417908	32417908	C	-	exonic	WT1	S381fs	30.89%	frameshift deletion			Frameshift indel detected in Papaemmanuil NEJM 2016 in same codon
33	chr4	106157638	106157638	C	T	exonic	TET2	Q847X	45.35%	stopgain			frameshift indel, nonsense or splicing variant
33	chr5	170837547	170837547	-	TCAG	exonic	NPM1	W288fs	42.53%	frameshift insertion		COSM20856	Previously reported in haematopoietic samples in COSMIC database
33	chr11	32417914	32417914	G	C	exonic	WT1	R380G	5.16%	nonsynonymous SNV		COSM28984	Previously reported in haematopoietic samples in COSMIC database
34	chr21	36252936	36252936	-	GC	exonic	RUNX1	A142fs	11.02%	frameshift insertion			frameshift indel, nonsense or splicing variant
34	chr20	31022592	31022592	C	T	exonic	ASXL1	R693X	42.72%	stopgain		COSM51388	Previously reported in haematopoietic samples in COSMIC database
35	chr17	74732959	74732959	G	C	exonic	SRSF2	P95R	25.95%	nonsynonymous SNV		COSM211661	Previously reported in haematopoietic samples in COSMIC database
35	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	46.40%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
36	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	38.71%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
36	chr13	28592642	28592642	C	A	exonic	FLT3	D835Y	49.38%	nonsynonymous SNV	rs121913488	COSM783	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
37	chr4	106156959	106156959	C	-	exonic	TET2	Y620X	23.36%	stopgain			frameshift indel, nonsense or splicing variant
37	chr17	74732959	74732959	G	A	exonic	SRSF2	P95L	61.63%	nonsynonymous SNV		COSM146288,COSM211506,COSM211028	Previously reported in haematopoietic samples in COSMIC database
37	chr21	36164838	36164838	-	G	exonic	RUNX1	R346fs	20.35%	frameshift insertion		COSM36063	Previously reported in haematopoietic samples in COSMIC database
37	chr2	209113113	209113113	G	A	exonic	IDH1	R132C	40.99%	nonsynonymous SNV	rs121913499	COSM28747	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
38	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	42.76%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
38	chr4	106158237	106158237	-	A	exonic	TET2	T1047fs	25.00%	frameshift insertion		COSM211700	Previously reported in haematopoietic samples in COSMIC database
38	chr13	28608281	28608281	A	C	exonic	FLT3	V592G	15.41%	nonsynonymous SNV		COSM28045	Previously reported in haematopoietic samples in COSMIC database
38	chr13	28592642	28592642	C	A	exonic	FLT3	D835Y	18.17%	nonsynonymous SNV	rs121913488	COSM783	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
38	chr12	12037440	12037440	C	A	exonic	ETV6	F357L	38.06%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 2 codons COSM5683034 F359Q
39	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	34.04%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
39	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	45.97%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
40	chr2	209113113	209113113	G	T	exonic	IDH1	R132S	43.98%	nonsynonymous SNV	rs121913499	COSM28748	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
41	chr2	25462024	25462024	A	G	exonic	DNMT3A	W795R	46.15%	nonsynonymous SNV			ND, but variant 2bp downstream in same aa: W795C=ONCOGENIC
41	chr5	170837547	170837547	-	CCTG	exonic	NPM1	W288fs	43.73%	frameshift insertion		COSM17573	Previously reported in haematopoietic samples in COSMIC database
41	chr20	31023821	31023821	G	T	exonic	ASXL1	E1102D	50.98%	nonsynonymous SNV	rs139115934	COSM36205	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
41	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	51.89%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
42	chr5	149441413	149441413	C	T	splicing	CSF1R		84.05%	Splicing			frameshift indel, nonsense or splicing variant
42	chr21	36164601	36164601	G	A	exonic	RUNX1	P425L	26.05%	nonsynonymous SNV		COSM87293	Previously reported in haematopoietic samples in COSMIC database
42	chr17	7577539	7577539	G	A	exonic	TP53	R116W	9.48%	nonsynonymous SNV	rs121912651	COSM120006,COSM120005,COSM1640831,COSM10656,COSM120007	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
42	chr9	72965035	72965035	T	G	exonic	SMC5	D965E	38.78%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 2 codons COSM3716331 D967H
43	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	33.05%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
43	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	40.62%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
45	chr11	32417915	32417915	-	ACAA	exonic	WT1	L378fs	12.43%	frameshift insertion			frameshift indel, nonsense or splicing variant
45	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	31.94%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database

45	chr2	25457243	25457243	G	A	exonic	DNMT3A	R882C	40.96%	nonsynonymous SNV		COSM53042,COSM1166704	Previously reported in haematopoietic samples in COSMIC database
46	chr11	32413539	32413548	TGGTGTGGGT	-	exonic	WT1	T468fs	64.68%	frameshift deletion			frameshift indel, nonsense or splicing variant
46	chr4	55593437	55593437	G	A	exonic	KIT	V528I	34.96%	nonsynonymous SNV	rs55792975		Previously reported in human variation database frequency less than 0.14%
47	chr4	55589773	55589775	GAC	-	exonic	KIT	419_419del	46.11%	nonframeshift deletion		COSM29014	Previously reported in haematopoietic samples in COSMIC database
47	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	54.53%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
49	chr22	36259163	36259163	T	G	exonic	RUNX1	K110Q	42.07%	nonsynonymous SNV		COSM26691	Previously reported in haematopoietic samples in COSMIC database
49	chr4	106157374	106157374	A	C	exonic	TET2	T759P	47.09%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 1 codon COSM4383839 Q758X
50	chr4	106164914	106164914	G	A	exonic	TET2	R1261H	5.66%	nonsynonymous SNV		COSM211643	Previously reported in haematopoietic samples in COSMIC database
50	chr1	115256529	115256529	T	C	exonic	NRAS	Q61R	25.54%	nonsynonymous SNV	rs11554290	COSM584	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
52	chr12	112926884	112926884	T	C	exonic	PTPN11	S502P	14.46%	nonsynonymous SNV		COSM13020	Previously reported in haematopoietic samples in COSMIC database
52	chr5	170837547	170837547	-	CATG	exonic	NPM1	W288fs	42.57%	frameshift insertion		COSM17571	Previously reported in haematopoietic samples in COSMIC database
52	chr2	209113112	209113112	C	T	exonic	IDH1	R132H	44.18%	nonsynonymous SNV	rs121913500	COSM28746	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
52	chr1	115258747	115258747	C	T	exonic	NRAS	G12D	23.02%	nonsynonymous SNV	rs121913237	COSM564	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
53	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	21.83%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
56	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	33.33%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
56	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	41.89%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
57	chr1	115258747	115258747	C	T	exonic	NRAS	G12D	61.98%	nonsynonymous SNV	rs121913237	COSM564	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
58	chr12	25398285	25398285	C	A	exonic	KRAS	G12C	30.91%	nonsynonymous SNV	rs121913530	COSM1140136,COSM516	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
58	chrX	15821902	15821902	C	T	exonic	ZRSR2	R99W	63.29%	nonsynonymous SNV	rs181618355		Previously reported in human variation database frequency less than 0.14%
60	chr2	25459806	25459806	T	C	exonic	DNMT3A	K826R	43.60%	nonsynonymous SNV		COSM1583124	Previously reported in haematopoietic samples in COSMIC database
61	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	39.34%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
61	chr11	32413565	32413565	C	T	exonic	WT1	R462Q	42.26%	nonsynonymous SNV		COSM21434	Previously reported in haematopoietic samples in COSMIC database
62	chr21	36252938	36252938	-	CGGCCTGAGCG	exonic	RUNX1	S330delinsTS	41.06%	frameshift insertion			Frameshift indel detected in Papaemmanuil NEJM 2016 in same codon
62	chrX	133559250	133559250	-	CAA	exonic	PHF6	H329_S330delinsPTX	26.45%	nonframeshift insertion			frameshift indel, nonsense or splicing variant
62	chr21	36252940	36252940	-	GCCTGAGCGCG	exonic	RUNX1	S141fs	40.86%	frameshift insertion			frameshift indel, nonsense or splicing variant
62	chr17	7578394	7578394	T	G	exonic	TP53	H47P	85.78%	nonsynonymous SNV		COSM44218	Previously reported in haematopoietic samples in COSMIC database
62	chr20	31023821	31023821	G	T	exonic	ASXL1	E1102D	50.69%	nonsynonymous SNV	rs139115934	COSM36205	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
64	chr2	198266834	198266834	T	C	exonic	SF3B1	K700E	11.45%	nonsynonymous SNV		COSM84677	Previously reported in haematopoietic samples in COSMIC database
65	chr21	36231791	36231791	T	C	exonic	RUNX1	D198G	55.84%	nonsynonymous SNV		COSM24799	Previously reported in haematopoietic samples in COSMIC database
65	chr17	74732960	74732960	G	C	exonic	SRSF2	P95A	47.58%	nonsynonymous SNV		COSM307352	Previously reported in haematopoietic samples in COSMIC database
65	chr21	36231860	36231860	A	G	exonic	RUNX1	L175P	7.48%	nonsynonymous SNV		COSM96548	Previously reported in haematopoietic samples in COSMIC database
65	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	51.79%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
66	chr1	115258748	115258748	C	T	exonic	NRAS	G12S	48.80%	nonsynonymous SNV	rs121913250	COSM563	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
67	chr21	36252865	36252865	C	T	exonic	RUNX1	R166Q	16.87%	nonsynonymous SNV		COSM36055	Previously reported in haematopoietic samples in COSMIC database
67	chr2	209113113	209113113	G	A	exonic	IDH1	R132C	38.10%	nonsynonymous SNV	rs121913499	COSM28747	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
67	chrX	133551305	133551305	T	C	exonic	PHF6	I314T	62.92%	nonsynonymous SNV			Previously reported in Papaemmanuil NEJM 2016
68	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	35.71%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
69	chr4	106190802	106190816	GGGTCTGAAGGAAGG	-	exonic	TET2	1360_1365del	58.05%	nonframeshift deletion			frameshift indel, nonsense or splicing variant
69	chrX	36259253	36259253	C	A	exonic	RUNX1	E80X	29.67%	stopgain			frameshift indel, nonsense or splicing variant
69	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	41.73%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
69	chrX	133549137	133549137	G	A	exonic	PHF6	R274Q	40.80%	nonsynonymous SNV		COSM306061	Previously reported in haematopoietic samples in COSMIC database
70	chr20	31022245	31022245	C	G	exonic	ASXL1	S577X	43.89%	stopgain			frameshift indel, nonsense or splicing variant
70	chr11	119148991	119148991	G	A	exonic	CBL	C404Y	95.06%	nonsynonymous SNV	rs192712314	COSM34068	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
70	chr4	106180897	106180897	T	C	exonic	TET2	F1309L	96.79%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 1 codon COSM4383839 K1310T
71	chr13	12038956	12038956	T	C	exonic	ETV6	F417L	68.01%	nonsynonymous SNV			Different SNV in same codon in haematopoietic sample in COSMIC confirmed somatic COSM5879372 F417C
71	chr17	7578548	7578548	G	T	exonic	TP53	P89T	43.33%	nonsynonymous SNV			Mutations in TP53 are widely distributed across the gene. FATHMM algorithm predicts a strong association with cancer.
72	chr4	55599321	55599321	A	T	exonic	KIT	D812V	31.69%	nonsynonymous SNV	rs121913507	COSM1314	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
73	chr4	106196834	106196834	C	T	exonic	TET2	P1723S	48.74%	nonsynonymous SNV		COSM1235472	Previously reported in haematopoietic samples in COSMIC database
73	chr4	106157698	106157698	T	C	exonic	TET2	Y867H	48.75%	nonsynonymous SNV	rs146348065	COSM1235472	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
74	chr4	106157710	106157710	A	T	exonic	TET2	N871Y	48.56%	nonsynonymous SNV			Somatic mutation documented in haematopoietic sample in COSMIC within 3 codons COSM220395 P874A
75	chr17	74732959	74732959	G	C	exonic	SRSF2	P95R	7.17%	nonsynonymous SNV		COSM211661	Previously reported in haematopoietic samples in COSMIC database

75	chr17	90631934	90631934	C	T	exonic	IDH2	R140Q	46.87%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
76	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	29.14%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
76	chr20	31023821	31023821	G	T	exonic	ASXL1	E1102D	45.47%	nonsynonymous SNV	rs139115934	COSM36205	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
76	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	38.33%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
76	chr1	115258747	115258747	C	T	exonic	NRAS	G12D	27.57%	nonsynonymous SNV	rs121913237	COSM564	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
77	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	43.08%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
77	chr2	209113113	209113113	G	C	exonic	IDH1	R132G	46.02%	nonsynonymous SNV		COSM28749	Previously reported in haematopoietic samples in COSMIC database
79	chr12	25398284	25398284	C	T	exonic	KRAS	G12D	40.43%	nonsynonymous SNV	rs121913529	COSM1135366,COSM521	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
79	chr4	55599321	55599321	A	T	exonic	KIT	D812V	8.38%	nonsynonymous SNV	rs121913507	COSM1314	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
80	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	46.92%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
81	chrX	133559234	133559234	-	CC	exonic	PHF6	L324fs	88%	frameshift insertion			frameshift indel, nonsense or splicing variant
81	chr17	74732959	74732959	G	A	exonic	SRSF2	P95L	72.37%	nonsynonymous SNV		COSM146288,COSM211506,COSM211028	Previously reported in haematopoietic samples in COSMIC database
81	chr4	106156384	106156384	G	A	exonic	TET2	G429R	52.08%	nonsynonymous SNV		COSM219042	Previously reported in haematopoietic samples in COSMIC database
82	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	45.61%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
83	chr2	25459807	25459807	T	C	exonic	DNMT3A	K826E	8.79%	nonsynonymous SNV			Different SNV in same codon in haematopoietic sample in COSMIC confirmed somatic COSM5708156 K826N
83	chr15	90631838	90631838	C	T	exonic	IDH2	R172K	14.93%	nonsynonymous SNV	rs121913503	COSM33733	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
84	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	55.32%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
84	chr21	36252940	36252940	G	A	exonic	RUNX1	S141L	15.28%	nonsynonymous SNV		COSM24737	Previously reported in haematopoietic samples in COSMIC database
85	chr11	32413557	32413557	G	A	exonic	WT1	H465Y	14.30%	nonsynonymous SNV		COSM1317334,COSM1317335	Previously reported in haematopoietic samples in COSMIC database
86	chr4	106156915	106156925	ACTGGAAATTC	-	exonic	TET2	T606fs	53.60%	frameshift deletion			Frameshift indel detected in Papaemmanuil NEJM 2016 in same codon
86	chr17	7577093	7577093	C	T	exonic	TP53	R282Q	45.38%	nonsynonymous SNV		COSM99936,COSM44338,COSM1646815	Previously reported in haematopoietic samples in COSMIC database
87	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	37.30%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
88	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	12.27%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
88	chrX	36259172	36259172	G	A	exonic	RUNX1	R107C	6.46%	nonsynonymous SNV		COSM24736	Previously reported in haematopoietic samples in COSMIC database
89	chr4	55561860	55561860	A	C	exonic	KIT	T84P	43.55%	nonsynonymous SNV			Different SNV in same codon in haematopoietic sample in COSMIC confirmed somatic COSM3380948 T84M
89	chr4	55599321	55599321	A	T	exonic	KIT	D812V	37.58%	nonsynonymous SNV	rs121913507	COSM1314	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
89	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	48.44%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
90	chr2	25458602	25458602	G	-	exonic	DNMT3A	D857fs	20.92%	frameshift deletion		COSM1683137,COSM1683138	Previously reported in haematopoietic samples in COSMIC database
90	chr15	90631838	90631838	C	T	exonic	IDH2	R172K	32.11%	nonsynonymous SNV	rs121913503	COSM33733	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
91	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	24.66%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
91	chr2	25463541	25463541	G	C	exonic	DNMT3A	S714C	47.92%	nonsynonymous SNV		COSM442677,COSM87011	Previously reported in haematopoietic samples in COSMIC database
91	chrX	39932564	39932564	C	T	exonic	BCOR	V679I	99.73%	nonsynonymous SNV	rs144722432		Previously reported in human variation database frequency less than 0.14%
92	chr4	106157945	106157945	A	G	exonic	TET2	H949R	45.92%	nonsynonymous SNV			Previously reported in Papaemmanuil NEJM 2016
92	chr2	209113113	209113113	G	A	exonic	IDH1	R132C	41.34%	nonsynonymous SNV	rs121913499	COSM28747	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
92	chr5	149436930	149436930	C	T	exonic	CSF1R	G747R	46.85%	nonsynonymous SNV	rs41355444		Previously reported in human variation database frequency less than 0.14%
92	chr17	74732959	74732959	G	A	exonic	SRSF2	P95L	71.82%	nonsynonymous SNV		COSM146288,COSM211506,COSM211028	Previously reported in haematopoietic samples in COSMIC database
93	chr2	25457281	25457281	C	-	exonic	DNMT3A	G869fs	14.10%	frameshift deletion		COSM133716	Previously reported in haematopoietic samples in COSMIC database
93	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	31.08%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
93	chr11	32417907	32417907	-	CCGA	exonic	WT1	R380fs	12.71%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
95	G	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	40.90%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
95	C	25457242	25457242	C	T	exonic	DNMT3A	R882H	48.95%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
96	C	25398284	25398284	C	A	exonic	KRAS	G12V	25.97%	nonsynonymous SNV		COSM1140133,COSM520	Previously reported in haematopoietic samples in COSMIC database
97	chr21	36231773	36231773	C	T	exonic	RUNX1	R204Q	25.12%	nonsynonymous SNV		COSM24731	Previously reported in haematopoietic samples in COSMIC database
98	chr13	28602329	28602329	G	A	exonic	FLT3	A680V	44.69%	nonsynonymous SNV		COSM786	Previously reported in haematopoietic samples in COSMIC database
98	chr9	5073770	5073770	G	T	exonic	JAK2	V617F	44.24%	nonsynonymous SNV	rs77375493	COSM12600	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
99	chr2	25463286	25463286	C	T	exonic	DNMT3A	R736H	90.37%	nonsynonymous SNV	rs139293773	COSM133737,COSM1318940	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
99	chr17	90631838	90631838	C	T	exonic	IDH2	R172K	47.39%	nonsynonymous SNV	rs121913503	COSM33733	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
100	chr13	28623585	28623585	G	T	exonic	FLT3	D324E	49.35%	nonsynonymous SNV			Different SNV in same codon in haematopoietic sample in COSMIC confirmed somatic COSM46508 D324N
100	chr4	106164914	106164914	G	A	exonic	TET2	R1261H	45.21%	nonsynonymous SNV		COSM211643	Previously reported in haematopoietic samples in COSMIC database
100	chr21	36253011	36253011	C	T	splicing	RUNX1	splicing	25.61%	Splicing		COSM26032	Previously reported in haematopoietic samples in COSMIC database
100	chr4	106156747	106156747	C	T	exonic	TET2	R550X	39.72%	stopgain		COSM26032	Previously reported in haematopoietic samples in COSMIC database

100	chr9	5073770	5073770	G	T	exonic	JAK2	V617F	13.82%	nonsynonymous SNV	rs77375493	COSM12600	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
100	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	19.22%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
100	chr1	115258747	115258747	C	T	exonic	NRAS	G12D	5.62%	nonsynonymous SNV	rs121913237	COSM564	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
103	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	43.04%	frameshift insertion		COSM1314	Previously reported in haematopoietic samples in COSMIC database
103	chr13	28626716	28626716	C	T	exonic	FLT3	V194M	50.05%	nonsynonymous SNV		COSM28039	Previously reported in haematopoietic samples in COSMIC database
103	chr4	55599321	55599321	A	T	exonic	KIT	D812V	36.64%	nonsynonymous SNV	rs121913507	COSM1314	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
104	chr11	32417943	32417943	-	GTCG	exonic	WT1	R369fs	36.13%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
105	chr1	115258748	115258748	C	A	exonic	NRAS	G12C	37.39%	nonsynonymous SNV		COSM1155	Previously reported in haematopoietic samples in COSMIC database
105	chr4	55593431	55593431	G	A	exonic	KIT	V526I	57.50%	nonsynonymous SNV		COSM27295	Previously reported in haematopoietic samples in COSMIC database
106	chr5	170837547	170837547	-	CCTG	exonic	NPM1	W288fs	33.59%	frameshift insertion		COSM17573	Previously reported in haematopoietic samples in COSMIC database
106	chr4	106197377	106197377	C	G	exonic	TET2	H1904D	80.05%	nonsynonymous SNV		COSM17573	Previously reported in haematopoietic samples in COSMIC database
107	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	39.02%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
107	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	45.57%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
110	chr17	90631838	90631838	C	T	exonic	IDH2	R172K	15.11%	nonsynonymous SNV	rs121913503	COSM33733	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
113	chr4	106194004	106194010	ATGAAAA	-	exonic	TET2	N1489fs	22.76%	frameshift deletion		COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database
113	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	45.82%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%

Supplemental Table S3B: called nucleotide variants detected by NGS at relapse (excluding *FLT3* ITD)

Patient ID	Chr	Start	End	reference	alternative	Exon/ Intron	Gene	Amino acid change	VAF%	Exonic Function	dbSNP identifier	Cosmic identifier (v.68)	Notes for inclusion
1	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	34.27%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
11	chr12	12037403	12037403	T	A	exonic	ETV6	V345D	27.47%	nonsynonymous SNV			not detected in patient at diagnosis
11	chr12	11992216	11992216	-	GGGGGCC	exonic	ETV6	F102fs	11.12%	frameshift insertion			frameshift indel, nonsense or splicing variant
11	chr21	36252922	36252922	-	TGGGAAAA	exonic	RUNX1	A147fs	37.42%	frameshift insertion			Previously reported in non-haematopoietic sample in COSMIC database, insertion detected within 1 codon COSM1717871 T148fs
11	chr7	148508788	148508788	C	T	exonic	EZH2	V626M	42.88%	nonsynonymous SNV		COSM1000723	Previously reported in haematopoietic samples in COSMIC database
11	chr12	112926887	112926887	G	A	exonic	PTPN11	G503R	46.44%	nonsynonymous SNV		COSM14259	Previously reported in haematopoietic samples in COSMIC database
20	chr4	106158350	106158350	A	C	exonic	TET2	Q1084P	52.88%	nonsynonymous SNV	rs75056899		Somatic mutation documented in haematopoietic sample in COSMIC within same codon, not detected in patient at diagnosis
23	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	45.75%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
25	chr1	115256530	115256530	G	T	exonic	NRAS	Q61K	13.10%	nonsynonymous SNV	rs121913254	COSM580	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
29	chrX	39914710	39914710	G	C	exonic	BCOR	S1517C	51.71%	nonsynonymous SNV			not detected in patient at diagnosis
35	chr2	25463566	25463566	C	A	exonic	DNMT3A	G706W	17.21%	nonsynonymous SNV			not detected in patient at diagnosis
35	chr1	115256530	115256530	G	T	exonic	NRAS	Q61K	23.68%	nonsynonymous SNV	rs121913254	COSM580	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
35	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	18.28%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
56	chr2	209113112	209113112	C	T	exonic	IDH1	R132H	41.31%	nonsynonymous SNV	rs121913500	COSM28746	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
56	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	44.22%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
61	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	20.87%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
61	chr11	32413565	32413565	C	T	exonic	WT1	R462Q	18.06%	nonsynonymous SNV		COSM21434	Previously reported in haematopoietic samples in COSMIC database
62	chr21	36252940	36252940	-	GCCTGAGCGCG	exonic	RUNX1	S141fs	32.44%	frameshift insertion			frameshift indel, nonsense or splicing variant
62	chrX	133559247	133559247	-	CCACCTAA	exonic	PHF6	H329_S330delinsPTX	30.88%	stopgain			frameshift indel, nonsense or splicing variant
62	chr20	31023821	31023821	G	T	exonic	ASXL1	E1102D	36.32%	nonsynonymous SNV	rs139115934	COSM36205	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
62	chr17	7578394	7578394	T	G	exonic	TP53	H140P	54.92%	nonsynonymous SNV		COSM44218	Previously reported in haematopoietic samples in COSMIC database
65	chrX	133527545	133527545	C	A	exonic	PHF6	C85X	33.13%	stopgain			frameshift indel, nonsense or splicing variant
65	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	13.28%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
65	chr4	106158350	106158350	A	C	exonic	TET2	Q1084P	26.96%	nonsynonymous SNV	rs75056899		Somatic mutation documented in haematopoietic sample in COSMIC within same codon, not detected in patient at diagnosis
65	chr21	36231791	36231791	T	C	exonic	RUNX1	D198G	29.85%	nonsynonymous SNV		COSM24799	Previously reported in haematopoietic samples in COSMIC database
65	chr17	74732960	74732960	G	C	exonic	SRSF2	P95A	20.30%	nonsynonymous SNV		COSM307352	Previously reported in haematopoietic samples in COSMIC database
66	chr1	115258748	115258748	C	T	exonic	NRAS	G12S	21.84%	nonsynonymous SNV	rs121913250	COSM563	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
68	chr4	106157698	106157698	T	C	exonic	TET2	Y867H	10.89%	nonsynonymous SNV	rs144386291	COSM327337	Previously reported in haematopoietic samples in COSMIC database
68	chr4	106196834	106196834	C	T	exonic	TET2	P1723S	11.06%	nonsynonymous SNV	rs146348065	COSM1235472	Previously reported in haematopoietic samples in COSMIC database
71	chr12	12038956	12038956	T	C	exonic	ETV6	F417L	72.97%	nonsynonymous SNV			Different SNV in same codon in haematopoietic sample in COSMIC confirmed somatic COSM5879372 F417C

71	chr17	7578548	7578548	G	T	exonic	TP53	P89T	32.18%	nonsynonymous SNV			Mutations in TP53 are widely distributed across the gene. FATHMM algorithm predicts a strong association with cancer.
78	chr11	32417914	32417917	GTAC	-	exonic	WT1	V379fs	54.80%	frameshift deletion			frameshift indel, nonsense or splicing variant
81	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	47.11%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
84	chr21	36164627	36164627	-	A	exonic	RUNX1	F416fs	28.13%	frameshift insertion			frameshift indel, nonsense or splicing variant
84	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	17.14%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
88	chr21	36259172	36259172	G	A	exonic	RUNX1	R107C	41.99%	nonsynonymous SNV		COSM24736	Previously reported in haematopoietic samples in COSMIC database
88	chr17	74732959	74732959	G	T	exonic	SRSF2	P95H	66.09%	nonsynonymous SNV		COSM211505,COSM211029,COSM211504	Previously reported in haematopoietic samples in COSMIC database
89	chr4	55561860	55561860	A	C	exonic	KIT	T84P	57.11%	nonsynonymous SNV			Different SNV in same codon in haematopoietic sample in COSMIC confirmed somatic COSM3380948 T84M
89	chr4	55599321	55599321	A	T	exonic	KIT	D812V	46.68%	nonsynonymous SNV	rs121913507	COSM1314	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
89	chr4	106196819	106196819	G	T	exonic	TET2	V1718L	50.76%	nonsynonymous SNV	rs142312318	COSM41742	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
91	chrX	39932564	39932564	C	T	exonic	BCOR	V679I	28.54%	nonsynonymous SNV	rs144722432		Previously reported in human variation database frequency less than 0.14%
91	chr2	25463541	25463541	G	C	exonic	DNMT3A	S714C	9.25%	nonsynonymous SNV		COSM442677,COSM87011	Previously reported in haematopoietic samples in COSMIC database
92	chr17	7579419	7579428	AGGGGGCTGG	-	exonic	TP53	P48fs	50.44%	frameshift deletion			frameshift indel, nonsense or splicing variant
92	chr4	106157945	106157945	A	G	exonic	TET2	H949R	50.71%	nonsynonymous SNV			Previously reported in Papaemmanuil NEJM 2016
92	chr17	7578406	7578406	C	T	exonic	TP53	R136H	48.82%	nonsynonymous SNV	rs28934578	COSM10648,COSM99022,COSM1640851,COSM99024,COSM99023,COSM99914	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
92	chr5	149436930	149436930	C	T	exonic	CSF1R	G747R	52.90%	nonsynonymous SNV	rs41355444		Previously reported in human variation database frequency less than 0.14%
92	chr17	74732959	74732959	G	A	exonic	SRSF2	P95L	43.59%	nonsynonymous SNV		COSM146288,COSM211506,COSM211028	Previously reported in haematopoietic samples in COSMIC database
95	chr2	25457242	25457242	C	T	exonic	DNMT3A	R882H	11.24%	nonsynonymous SNV	rs147001633	COSM52944,COSM442676	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
95	chr5	170837547	170837547	-	TCTG	exonic	NPM1	W288fs	10.87%	frameshift insertion		COSM17559	Previously reported in haematopoietic samples in COSMIC database
96	chr12	25398284	25398284	C	A	exonic	KRAS	G12V	46.39%	nonsynonymous SNV	rs121913529	COSM1140133,COSM520	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%
111	chr11	32417911	32417911	-	C	exonic	WT1	S381fs	6.85%	frameshift insertion		COSM41864,COSM1166635	Previously reported in haematopoietic samples in COSMIC database
112	chr15	90631934	90631934	C	T	exonic	IDH2	R140Q	20.83%	nonsynonymous SNV	rs121913502	COSM41590	Previously reported in haematopoietic samples in COSMIC database, human variation database frequency less than 0.14%

Supplemental Table S4. Patients with changes in genetic mutation profile only between diagnosis and relapse.

Patient ID	Genetic aberrations at diagnosis	genetic aberrations pre-allo	genetic aberrations at relapse
20	FLT3 ITD 2.04	N/A	FLT3 ITD 0.00
	NRAS G13D 28.51		NRAS G13D 0.00
	TET2 Q1084P 0.00		TET2 Q1084P 52.88
	46,XY,inv(16)(p13q22)[8]/ 47,idem,+22[2] CBFB gene rearrangement by FISH [47/91] CBFB/MYH11 to ABL1 ratio of 1.82	cDNA molecular analysis: CBFB/MYH11 positive; CBFB/MYH11 to ABL1 ratio of 0.0136.	46,XY,inv(16)(p13q22)[2]/46,XY[28] CBFB gene rearrangement positive by metaphase FISH [3/21] CBFB gene rearrangement positive by interphase FISH [3/121] CBFB/MYH11 to ABL1 ratio of 0.169
56	DNMT3A R882H 41.89	N/A	DNMT3A R882H 44.22
	FLT3 ITD 28.05		FLT3 ITD 52.83
	NPM1 insTCTG 33.33		NPM1 insTCTG 60.00
	IDH1 R132H 1.15		IDH1 R132H 41.31
	46,XY[20]		46, XY[20]
112	IDH2 R140Q 0.38	N/A	IDH2 R140Q 20.83
	t(10;11) in 20/20 cells KMT2A rearranged 94/100 by interphase FISH		t(10;11) in 16/20 cells [KMT2A 55/100 by FISH] KMT2A rearranged 55/100 by interphase FISH
60	DNMT3A K826R 43.6	DNMT3A K826R 2.92	DNMT3A K826R 0.45
	46,XX[20]	N/A	46,XX[20]
65*	PHF6 C85X 0.09	PHF6 C85X 25.46	PHF6 C85X 33.13
	RUNX1 D198G 55.84	RUNX1 D198G 34.43	RUNX1 D198G 29.85
	RUNX1 L175P 7.48	RUNX1 L175P 0.94	RUNX1 L175P 0.51
	SRSF2 P95A 47.58	SRSF2 P95A 38.54	SRSF2 P95A 20.3
	TET2 V1718L 51.79	TET2 V1718L 50.54	TET2 V1718L 13.28
	TET2 Q1084P 0.02	TET2 Q1084P 0.03	TET2 Q1084P 26.96
	47,XY,+13[10]	46,XY[60]	47, XY, +13[5]/46,XY[5]
68	NPM1 insTCTG 35.71%	NPM1 insTCTG 0.00	NPM1 insTCTG 35.84
	TET2 Y867H 0.27	TET2 Y867H 0.15	TET2 Y867H 10.89
	TET2 P1723S 0.00	TET2 P1723S 0.11	TET2 P1723S 11.06
	FLT3 ITD 84bp 32.4%	FLT3 ITD 0.0	FLT3 ITD 84bp 27.8%
	46,XX[20]	N/A	46,XX[19]//46,XY[1] XX[94]//XY[17] by interphase FISH (85% host)
78	WT1 V379fs 0.00	WT1 V379fs 0.00	WT1 V379fs 54.8
	46,XX,t(9;11)(p22;q23)[10] KMT2A gene rearrangement positive by metaphase FISH [5]	KMT2A gene rearrangement negative by metaphase FISH [65] KMT2A gene rearrangement negative by interphase FISH [100]	46,XX,t(9;11)(p22;q23)[10] XX[93]//XY[7] by interphase FISH (93% host) KMT2A gene rearrangement positive by metaphase FISH [5] KMT2A gene rearrangement positive by interphase FISH [98/100]

N/A= sample not available

***pre-allo-SCT sample was taken prior to patient achieving CR.**

Supplemental Table S5. Patients with changes in genetic mutation and karyotypic profile between diagnosis and relapse.

Patient ID	genetic aberrations at diagnosis	genetic aberrations at pre-allo	genetic aberrations at relapse
11	EZH2 V626M 36.22	N/A	EZH2 V626M 42.88
	PTPN11 G503R 44.67		PTPN11 G503R 46.44
	ETV6 F102fs 0.00		ETV6 F102fs 11.12
	ETV6 V345D 0.00		ETV6 V345D 27.47
	RUNX1 A147fs 35.43		RUNX1 A147fs 37.42
	46,XY[20]	N/A	46,XY[53]/47,X,idic(Y)(q11.2q11.2),+idic(Y)[12]/ 48,X,idic(Y),+idic(Y)x2[11]/49,X,idic(Y),+idic(Y)x3[2]/46,XX[6]
17	6bp FLT3 ITD 1.12	FLT3 ITD 0.00	FLT3 ITD 0.00
	NRAS Q61R 5.00	NRAS Q61R 0.01 (ddPCR-ND)	NRAS Q61R 0.00 ddPCR ND
	46,XX,t(12;17)(p11;q11)[3]/ 46,XX[18]	46,XX[60]	46,XX,-2, add(7)(q32),+8,add(8)(q24),add(16)(q1)[3]/ 46,idem,add(X)(q2),add(3)(p21),add(3)(p25),add(10)(p1),add(18)(q2),add(21)(q22)[3]/ 46,idem,add(4)(q3),add(5)(q3),add(9)(q2),del(13)(q1q2)[3]/ 46,XX,-2,der(7)add(7)(p2)add(7)(q32),add(10)(p1),add(16)(q1),add(19)(q1),+mar[5]//46,XY[2]
25	NRAS Q61K 0.00 ddPCR	NRAS Q61K 0.00 (ddPCR-ND)	NRAS Q61K 13.10 ddPCR
	46,XX,t(6;11)(q27;q23)[9]/46,XX[1] KMT2A rearrangement confirmed by FISH	No evidence of a KMT2A gene translocation by FISH [100]	47,X,add(X)(p1),t(1;2)(q25;q31),der(3)add(3)(p2)add(3)(q2),t(6;8)(p21;q24),t(6;11)(q27;q23),add(9)(p1), add(11)(p15),add(15)(q1),add(17)(q2),+mar[10] KMT2A rearrangement confirmed by FISH
35	DNMT3A G706W 0.00	DNMT3A G706W 0.00	DNMT3A G706W 17.82
	IDH2 R140Q 43.84	IDH2 R140Q 44.61	IDH2 R140Q 21.23
	NRAS Q61K 0.00	NRAS Q61K 0.81	NRAS Q61K 21.24
	SRSF2 P95R 40.86	SRSF2 P95R 46.23	SRSF2 P95R 21.25
	46,XY,t(12;22)(p13;q12)[10] ETV6 gene rearrangement positive [5/5]	46,XY,t(12;22)(p13;q12)[2]/47,XY,+8,t(12;22)[3]/ 48,XY,+8,+8,t(12;22)[2]/46,XY[23]	46,X,-Y,+8,t(12;22)(p13;q12)[10]/46,idem,add(20)(q13)[2]
84*	RUNX1 S141L 15.28	RUNX1 S141L 0.49	RUNX1 S141L 0.00
	RUNX1 F416fs 10.57	RUNX1 F416fs 85.13	RUNX1 F416fs 28.13
	SRSF2 55.32	SRSF2 36.92	SRSF2 17.14
	46,XY[20]	46,XY[26]	46,XY,inv(3)(q21q26)[9]/46,XY[12]
92	CSF1R G747R 46.85	CSF1R G747R 51.12	CSF1R G747R 52.9
	IDH1 R132C 41.34	IDH1 R132C 0.30	IDH1 R132C 0.00
	SRSF2 P95L 71.82	SRSF2 P95L 36.1	SRSF2 P95L 43.59
	TET2 H949R 45.93	TET2 H949R 46.54	TET2 H949R 50.71
	TP53 R136H 0.08	TP53 R136H 3.74	TP53 R136H 48.82
	TP53 P48fs 0.00	TP53 P48fs 0.00	TP53 P48fs 50.44
	46,XY[20]	N/A	47,XY,+8[4]/46,XY,add(3)(p2)[3]/46,XY,t(2;15)(q37;q2)[2]/46,XY,dup(18)(q11.2q21)[1]/46,XY[3]//46XX[1]

***patient not in CR pre-allo-SCT**

N/A= sample not available

Supplemental Table S6. Patients with changes in karyotypic profile only between diagnosis and relapse.

Patient ID	genetic aberrations at diagnosis	genetic aberrations at CR pre-allo	genetic aberrations at relapse
10	46,XY,t(5;11)(q35;p15) [20] NSD1 (5q35) gene rearrangement by FISH [93/100]	No evidence of an NSD1 (5q35) gene rearrangement by FISH [34]	45,X,-Y, add(1)(P3),add(1)(q2),add(2)(q3),der(3)t(3;11)(p1;p1),add(4)(q2),der(5)t(5;11)(q35.3;p15.4),add(6)(p2),der(11)t(3;11)(p1;p1)add(11)(q2),der(11)t(5;11)add(11)(q1),-13,-16,add(17)(p1),add(18)(q2),add(19)(q13),+2mar[8]/46,XY[2]
54	45,X,X,t(8;21)(q22;q22),del(9)(q1q22)[10]	46,XX[60]	45,X,-X,add(1)(q32),add(7)(q22),-8,t(8;21)(q22;q22),add(8)(q1), del(9)(q1q22),add(10)(q22),add(11)(p15),add(12)(q24),add(13)(q3),-16, -17,add(17)(q2),add(18)(q22),+3mar[cp7]/46,XX[3]
66	46,XY,del(7)(q22q36), der(10)t(10;11)(p12~13;q13~21),der(11) add(11)(p1)t(10;11),del(12)(q2?2q2?4), del(13)(q1?4q2?2)[12]/46,XY[3]	N/A	45~46,XY,del(6)(q1q2),del(7)(q2q3),der(10)t(10;11)(p12~13;q13~21),der(11)add(11)(p1)t(10;12),del(13)(q1q2), add(17)(p1),+1~3mar[cp7]//46,XY[5]
71	46,XX[20]	N/A	45~46,XX,+1,add(1)(p1),der(1;10)(q10;p10),add(2)(p2),t(3;7)(p1;q3),add(4)(q3),-5, add(5)(q2),+6,der(6)t(6;18)(q1;q1),add(13)(q2),-18,-20,add(20)(p1),add(21)(q2),+3mar[cp14]/46,XX[1]/ /46, XY[3]
88	46,XY[20]	N/A	51,XY,der(4)t(1;4)(q21;q3),+9,+12,+13,+15,+21[7]/46,XY,t(7;16)(q2;q2)[2],46XY[1]
89	46,XY,t(3;6)(p25;q25)?c,del(5)(q1q3)[4]/ 47,XY,t(3;6)(p25;q25)?c,del(5)(q1q3),+9[6]	N/A	46,XY,add(1)(p36),t(3;6)(p25;q25)?c,del(5)(q1q3)[5]/83,XXYY,-1,-2,der(3)t(3;6)?c,der(3)t(3;6)?ct(3;22)(q2;q13), -4,del(5)(q1q3)x2,der(6)t(3;6)?cx2,-7,add(9)(q2),add(11)(p1),-13,-15,-16,der(16)t(7;16)(q1;p1),-17, -18,der(22)t(3;22)[6]
90	46,XX[30]	46,XX[20]	46,XX,add(5)(q13)[9]/46,XX[1]//46,XY[3]
91	45,XY,-7[10]	45,XY,-7[2]/46,XY[2] Monosomy 7 detected by interphase FISH [71/101]	45,XY,-7,inv(12)(q13q2)x2,del(15)(q?1q2)[5]/46,idem,+del(15)(q?1q2)[3]/46,XY[2]
96	46,XX,der(6)t(6;11)(q21;q23),der(11)t(11;16)(q2;q2),der(16)t(11;16)(q2;q2)t(6;11)(q2;q23)[10] KMT2A gene rearrangement confirmed by metaphase FISH [4/5]	46,XX[30]	50, XX, der(6)t(6;11)(q21;q23),+8,+8,der(11)t(11;16)(q2;q2),+16,der(16)t(11;16)q2;q2)t(6;11)(q2;q23)x2,+19[2]/50,idem, t(X;15)(p22;q1)[8] KMT2A gene rearrangement confirmed by interphase FISH [94/100]
102	46, XX, del 11q, add 21q [20]. Undefined abnormal 5p, 6q detected. MLL 11q23 rearrangement detected by FISH [99/100]. 46,XX,add(5)(p?),add(6)(q?),del(11)(q?), add(21)(q?)[20] KMT2A rearrangement detected by FISH [99/100]	N/A	48,XX,t(1;14),+6 +8,add (9)(p?)[10/10]

N/A= sample not available

Supplemental Table S7. Patients with no changes in profile of genetic aberrations between diagnosis and relapse.

Patient ID	genetic aberrations at diagnosis	genetic aberrations pre-allo	genetic aberrations at relapse
1	FLT3 ITD 15.22	FLT3 ITD 0.00	FLT3 ITD 20.42
	NPM1 insTCTG 38.95	NPM1 insTCTG 0.06	NPM1 insTCTG 34.27
	46,XX[20]	N/A	XX[90]/XY[10]
61*	FLT3 ITD 33.50	FLT3 ITD 0.00	FLT3 ITD 42.26
	NPM1 insTCTG 39.34	NPM1 insTCTG 7.64	NPM1 insTCTG 10.58
	WT1 R462Q 42.26	WT1 R462Q 20.87	WT1 R462Q 18.06
	46,XX[20]	46,XX,t(3;5)(p2;q1)[7]/ 46,XX,t(5;16)(q2;q1)[3]/ 46,XX[15]	46,XX[20]
62*	ASXL1 E1102D 50.69	ASXL1 E1102D 49.39	ASXL1 E1102D 36.32
	PHF6 S330fs 26.56	PHF6 S330fs 30.22	PHF6 S330fs 30.88
	RUNX1 S141fs 40.86	RUNX1 S141fs 32.12	RUNX1 S141fs 32.44
	TP53 H179P 85.78	TP53 H179P 62.69	TP53 H179P 54.92
	47~49,XX,+X,del(6)(q2),+10,+12,del(12)(p1), del(17)(p1),+21,inc[cp8]	48~49,XX,+X,del(6)(q13q21),+12,add(12)(p1), del(12)(p11p13),del(17)(p1),+21[cp11]/ 46,XX[4]	48~49,X,+X,add(X)(q2),del(6)(q1q2),+12,add(12)(p1),del(12)(p1), del(17)(p1),+21,+2~3mar[cp6]/46,XY[4]
64	SF3B1 K700E 11.45	SF3B1 K700E 1.13	SF3B1 K700E 3.01
	46,XY,inv(3)(q21q26)[10]/ 46,XY[5])	46,XY,inv(3)(q21q26)[8]/ 46,XY[2]	46,XY,inv(3)(q21q26)[9]/46,XY[1]

***patient not in CR pre-allo-SCT**

N/A= sample not available

patient #102 had no mutations detected by NGS at diagnosis or at relapse and is not shown in this table