

Utility of plasma cell-free DNA for *de novo* detection and quantification of clonal hematopoiesis

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SUPPLEMENTARY DATA

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Supplementary Methods

Cohort and samples

BLSA samples assayed in this study were collected from 2000-2011 during participants' clinical visits. The BLSA is a longitudinal observational study started in 1958. The BLSA continuously enrolls community dwelling volunteers who are 20 years and older, and extremely healthy at study entry-based on strict objective, clinical criteria. Enrolled participants have follow-up visits (either clinical or home) in an age-dependent frequency: if aged <60 every 4 years, 60–79 every 2 years and ≥ 80 every year.

In the AA group, serial plasma samples were collected from nine patients enrolled on the protocol at baseline and at 6 months post-treatment, and an additional 17 samples were collected at 6 months or 2 years post-treatment with no baseline samples. In the MDS group, serial plasma samples were collected from all patients both post-treatment and at progression as defined in the protocol (*Online Supplementary Figure S1*).¹ All samples were sent to a CLIA-certified laboratory for DNA extraction and targeted massively parallel. For the healthy and MDS cohorts, stored plasma and blood cells that had been previously collected, isolated and stored in liquid nitrogen

were sent for screening, whereas for AA patients, whole peripheral blood and marrow samples were sent shortly after collection with both peripheral plasma cfDNA and DNA from BM cells extracted up to 48 h after collection.

Patients were either enrolled on clinical trials (NCT00932156 and NCT01623167) or were seen in consultation.

Commercial massively parallel sequencing of targeted genes

In healthy individuals and MDS patients, plasma and PB/BM cells were independently sent for CH screening and results were separately reported by the laboratory. In contrast, cfDNA and BM results from AA were reported simultaneously for each patient since their marrow and PB were sent for parallel screening of plasma and cells.

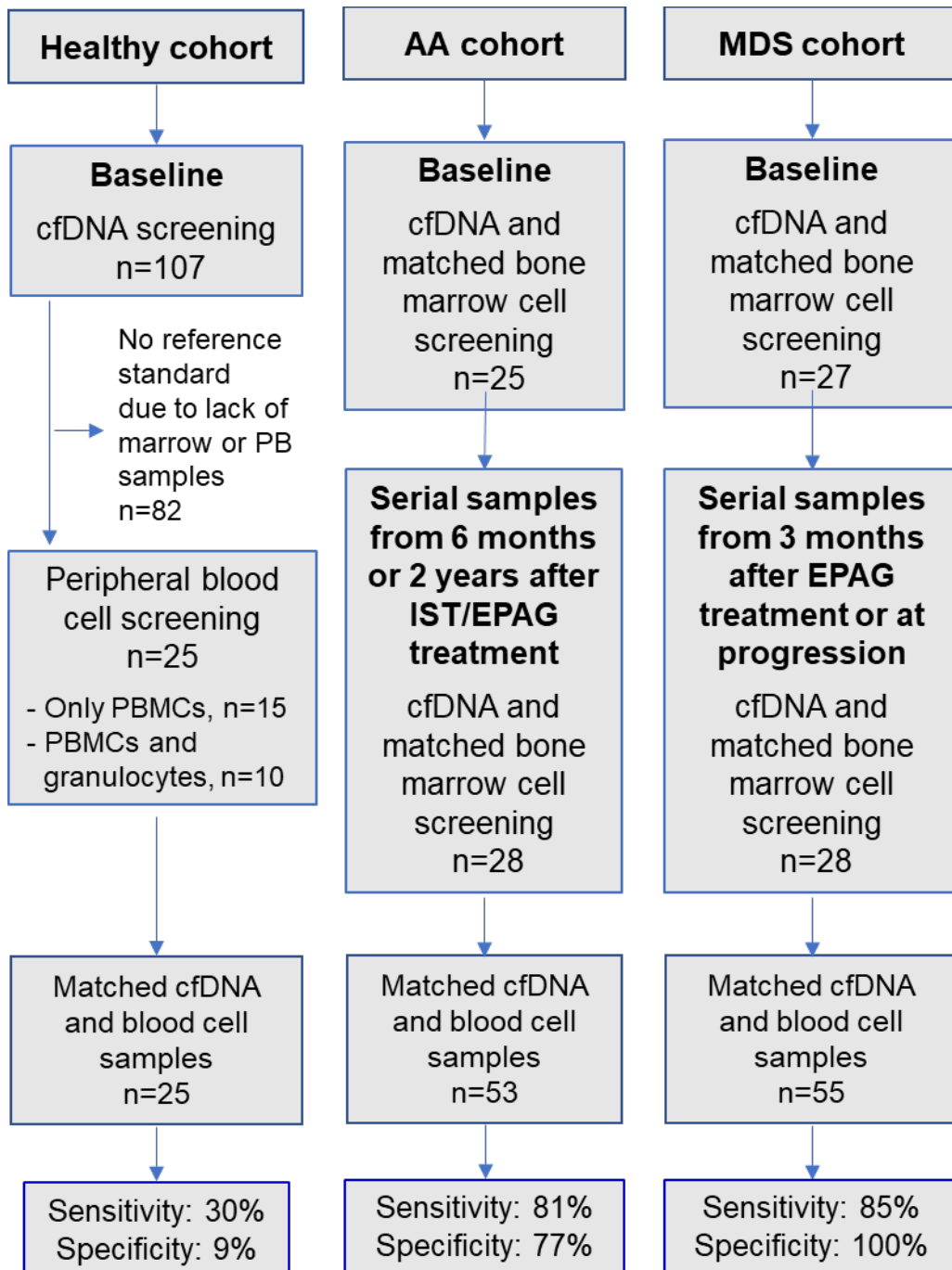
Per vendor, the library for targeted DNA sequencing is based on Single Primer Extension (SPE) chemistry and variant calling was performed using the DRAGEN somatic pipeline using tumor-only analysis against the GRCh37 reference genome. The assay has a typical sensitivity of 5% for overall variant detection and 3% for detecting specific somatic variants in hot-spots. However, somatic variations as low as 0.9% were reported back by the laboratory, which were all included in the analysis. In addition, the assay is specifically indicated for the molecular profiling of genomic abnormalities (SNV and indels) in DNA from patients with hematologic neoplasms using BM fresh cells, PB fresh cells, PB cfDNA and non-decalcified lymphoid tissue in formalin-fixed paraffin-embedded (FFPE). The cfDNA testing was validated to be used only for detecting abnormalities in myeloid neoplasms (AML, MDS, MPN and AA) but not for non-myeloid neoplasms; samples from healthy individuals were screened for research purposes only.

Stringent criteria for filtering variants

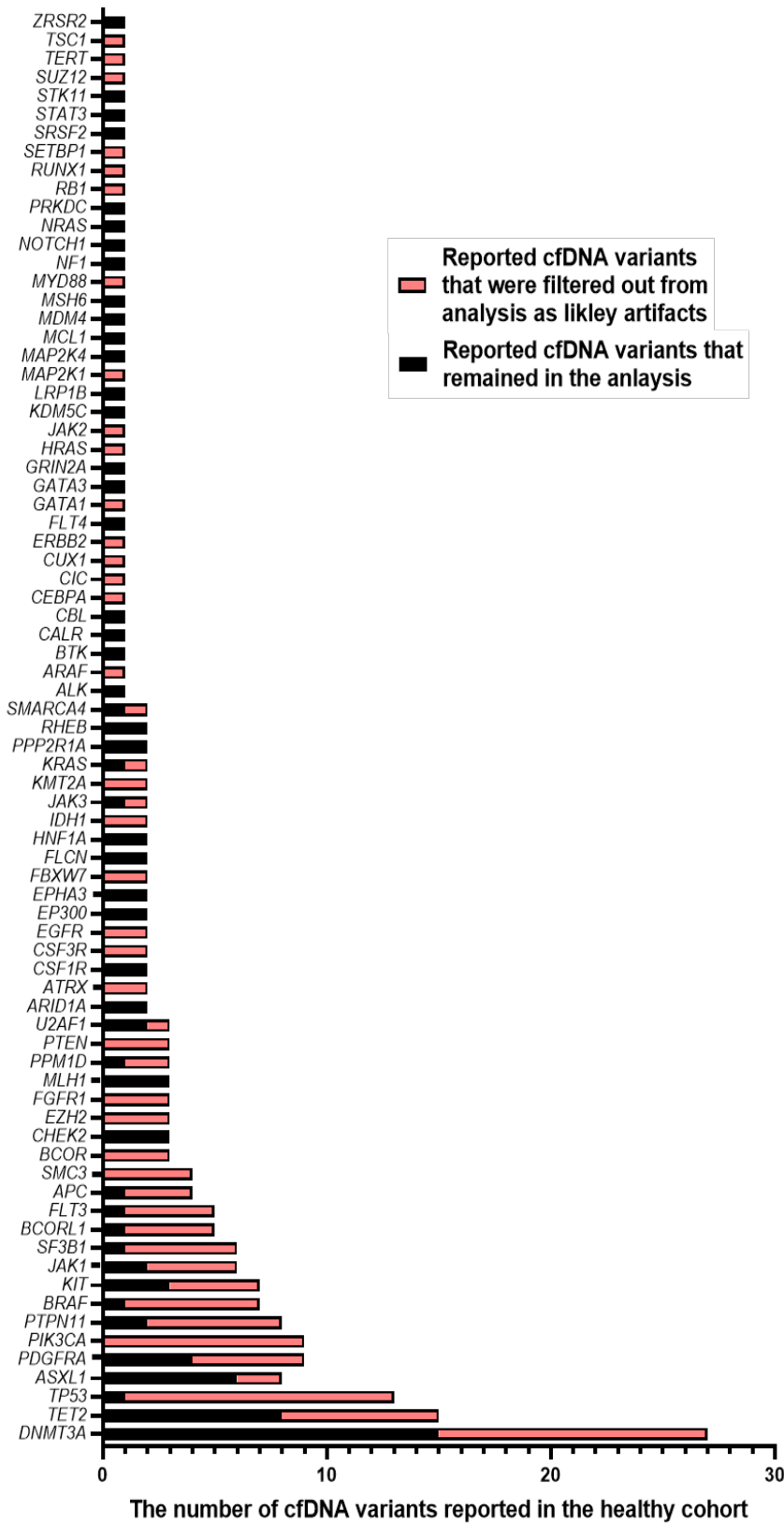
Results obtained from CLIA-reports were refined by stringent analysis that filtered out variants below the VAF and read depth thresholds of confirmed negative variants (limit of detection). We first discarded variants at VAF<1% and read depth<700X, and variants at VAF<3.5% and read depth<400X (more stringent than the minimal of 350X observed as the upper 95% CI in confirmed negative variants). cfDNA variants at VAF<10% were only included in the analysis if read depth was >100X (Figure 2D). Of note, the read depth of a variant at the lowest VAF (0.96%) found in both cfDNA and blood cells was 750X.

Targeted error-corrected DNA sequencing

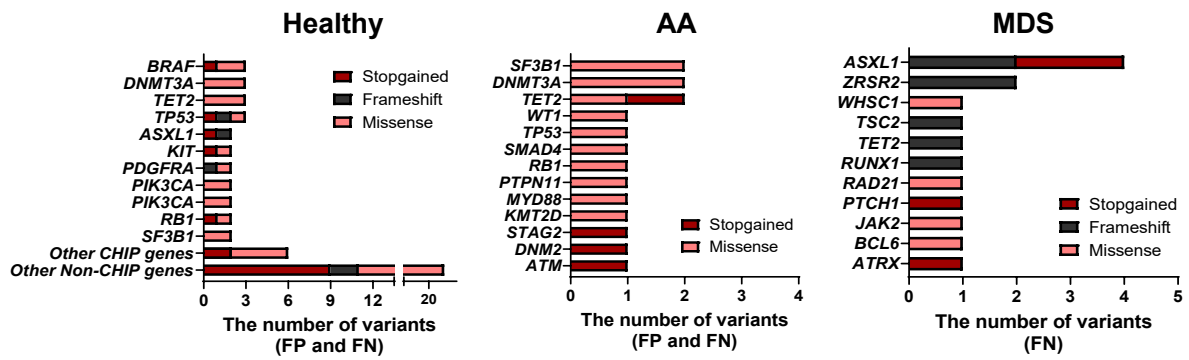
For selected HC samples with DNA available from PBMCs and grans, ultra-deep error correcting sequencing utilizing unique molecular indices was performed using a custom anchored multiplex PCR-based targeted DNA sequencing panel (VariantPlex, ArcherDx, Boulder, CO, USA) with coverage of 13 gene regions (*ASXL1*, *DNMT3A*, *FLT3*, *IDH1*, *IDH2*, *JAK2*, *KIT*, *NPM1*, *NRAS*, *RUNX1*, *SF3B1*, *TET2* and *TP53*) as previously described with slight modifications.⁴ Libraries were prepared with 100 ng of genomic DNA input utilizing pre- and post-PCR separation on liquid handling robots. The resulting libraries were subjected to paired-end 150-bp sequencing on either a HiSeq 2500 in Rapid Run Mode or a MiSeq (Illumina, San Diego, CA, USA). An average of 8,603,953 read pairs was collected per patient. FASTQ files are available in the NCBI Small Reads Archive (SRA). Raw FASTQ files were analyzed using Archer Analysis software version 6.0.2.3 using default settings. *De novo* variant calling was performed with minimum allele frequency for a variant calling set to 0.1%. An average de-duplicated depth per gene-specific primer per sample was 4,663X.



Supplementary Figure S1. A flow diagram of participants enrolled in the study. This study evaluated performance of cfDNA (index test) by detecting and quantifying clonal hematopoiesis in comparison to the screening of blood cells (reference test).



Supplementary Figure S2. The number of reported cfDNA variants found in 107 healthy subjects, according to individual genes. During analysis, the original data from genetic reports were filtered out to remove variants that were likely false positive and false negative, due to low VAF and low sequencing coverage. Variants originally reported by the CLIA assay were filtered out from analysis if were at VAF<10% and had been sequenced at read depth <100X, or were at VAF<3.5% and had been sequenced at read depth<400X.



Supplementary Figure S3. The number of discordant variants originally reported in the CLIA cfDNA assay in healthy individuals and patients with AA and MDS. Shown are mutated genes and types of FP and FN variants.

Supplementary Table S1. Lists of genes screened in this study by the commercial targeted massively parallel sequencing panel.

Genes commonly associated with clonal hematopoiesis (CHIP genes)*					
<i>ASXL1</i>	<i>CUX1</i>	<i>HRAS</i>	<i>KRAS</i>	<i>SETBP1</i>	<i>TP53</i>
<i>BCOR</i>	<i>DNMT3A</i>	<i>IDH1</i>	<i>NF1</i>	<i>SETD2</i>	<i>U2AF1</i>
<i>BCORL1</i>	<i>EP300</i>	<i>IDH2</i>	<i>NPM1</i>	<i>SF3B1</i>	<i>ZRSR2</i>
<i>BRAF</i>	<i>ETV6</i>	<i>IKZF1</i>	<i>NRAS</i>	<i>SMC1A</i>	<i>WT1</i>
<i>CBL</i>	<i>EZH2</i>	<i>IKZF3</i>	<i>PHF6</i>	<i>SMC3</i>	
<i>CBLB</i>	<i>FLT3</i>	<i>JAK1</i>	<i>PPM1D</i>	<i>SRSF2</i>	
<i>CEBPA</i>	<i>GATA1</i>	<i>JAK2</i>	<i>PTEN</i>	<i>STAG2</i>	
<i>CREBBP</i>	<i>GATA2</i>	<i>JAK3</i>	<i>PTPN11</i>	<i>SUZ12</i>	
<i>CSF1R</i>	<i>GATA3</i>	<i>KDM6A</i>	<i>RAD21</i>	<i>TERT</i>	
<i>CSF3R</i>	<i>GNAS</i>	<i>KIT</i>	<i>RUNX1</i>	<i>TET2</i>	
Cancer-related genes (non-CHIP genes)					
<i>ABL1</i>	<i>CALR</i>	<i>ERBB3</i>	<i>KAT6A</i>	<i>MYD88</i>	<i>RHOA</i>
<i>AKT1</i>	<i>CARD11</i>	<i>ESR1</i>	<i>KDM5C</i>	<i>NFKBIA</i>	<i>RHEB</i>
<i>AKT2</i>	<i>CBLC</i>	<i>ERBB2</i>	<i>KDR</i>	<i>NOTCH1</i>	<i>RNF43</i>
<i>AKT3</i>	<i>CCND1</i>	<i>ERG</i>	<i>KEAP1</i>	<i>NOTCH2</i>	<i>SDHB</i>
<i>ALK</i>	<i>CCND3</i>	<i>FAM175A</i>	<i>KMT2A</i>	<i>NOTCH3</i>	<i>SMAD2</i>
<i>AMER1</i>	<i>CD274</i>	<i>FAM46C</i>	<i>KMT2B</i>	<i>NSD1</i>	<i>SMAD4</i>
<i>APC</i>	<i>CD79A</i>	<i>FANCA</i>	<i>KMT2C</i>	<i>PALB2</i>	<i>SMARCA4</i>
<i>ARAF</i>	<i>CD79B</i>	<i>FANCC</i>	<i>KMT2D</i>	<i>PAX5</i>	<i>SMARCB1</i>
<i>ARID1A</i>	<i>CDH1</i>	<i>FANCD2</i>	<i>LRP1B</i>	<i>PBRM1</i>	<i>SMO</i>
<i>ARID1B</i>	<i>CDK12</i>	<i>FANCE</i>	<i>MAP2K1</i>	<i>PDGFRA</i>	<i>SOCS1</i>
<i>ARID2</i>	<i>CDK4</i>	<i>FANCF</i>	<i>MAP2K2</i>	<i>PDGFRB</i>	<i>SRC</i>
<i>ATM</i>	<i>CDK6</i>	<i>FANCG</i>	<i>MAP2K4</i>	<i>PIK3CA</i>	<i>STAT3</i>
<i>ATRX</i>	<i>CDKN2A</i>	<i>FAS</i>	<i>MAP3K1</i>	<i>PIK3R1</i>	<i>STK11</i>
<i>AXIN2</i>	<i>CDKN2B</i>	<i>FBXW7</i>	<i>MAP3K14</i>	<i>PIK3R2</i>	<i>TGFBR2</i>
<i>B2M</i>	<i>CDKN2C</i>	<i>FGFR1</i>	<i>MAPK1</i>	<i>PIM1</i>	<i>TSC1</i>
<i>BCL2</i>	<i>CHEK1</i>	<i>FLCN</i>	<i>MCL1</i>	<i>PLCG1</i>	<i>TSC2</i>
<i>BCL2L1</i>	<i>CHEK2</i>	<i>FLT4</i>	<i>MDM2</i>	<i>POLD1</i>	<i>TSHR</i>
<i>BCL6</i>	<i>CIC</i>	<i>GEN1</i>	<i>MDM4</i>	<i>POLE</i>	<i>ZNF217</i>
<i>BCR</i>	<i>CRLF2</i>	<i>GNAQ</i>	<i>MED12</i>	<i>PPP2R1A</i>	<i>MEF2B</i>
<i>BIRC3</i>	<i>CTNNA1</i>	<i>GRIN2A</i>	<i>MRE11A</i>	<i>PRKDC</i>	<i>WHSC1</i>
<i>BLM</i>	<i>CTNNB1</i>	<i>H3F3A</i>	<i>MLH1</i>	<i>PTCH1</i>	
<i>BRCA1</i>	<i>CXCR4</i>	<i>HNF1A</i>	<i>MTOR</i>	<i>RAD50</i>	
<i>BRCA2</i>	<i>DDR2</i>	<i>HOXB13</i>	<i>MSH6</i>	<i>RAD51</i>	
<i>BRIP1</i>	<i>DICER1</i>	<i>HSP90AA1</i>	<i>MUTYH</i>	<i>RB1</i>	
<i>BTK</i>	<i>DNM2</i>	<i>IGF1R</i>	<i>MYC</i>		
	<i>EPHA3</i>	<i>IRF4</i>			
*A gene list based on previously published data ^{2, 5-8}					

Supplementary Table S2. Sequencing results from healthy individuals reported by the CLIA-certified cfDNA test

Patient ID	Sex	Age	Status	Gene	HGVS _c	HGVS _p	Aminoacid change	Codons	cfDNA		PBMC		Peripheral granulocytes		Variant-level concordance	filtered data VAF <10% with read depth <100 and VAF<3.5% with read depth <400
									VAF (%)	Read depth	VAF (%)	Read depth	VAF (%)	Read depth		
HC-001	M	78	Somatic	ASXL1	NM_015338.5:c.4135A>T	NP_056153.2:p.Lys1379Ter	K/*	Aag/Tag	9.9	141	0	x	NA	NA	FP	FP
HC-001	M	78	Somatic	ARAF	NM_001256196.1:c.803C>A	NP_001243125.1:p.Ser268Ter	S/*	tCg/tAg	10.4	77	0	x	NA	NA	FP	FP
HC-001	M	78	Somatic	KIT	NM_000222.2:c.1757G>A	NP_000213.1:p.Arg586Lys	R/K	aGa/aAa	15.9	63	0	x	NA	NA	FP	FP
HC-002	M	71	Somatic	all NEG					0	x	0	x	NA	NA	TN	TN
HC-003	M	74	Somatic	BIRC3	NM_001165.4:c.1202G>A	NP_001156.1:p.Arg401Lys	R/K	aGa/aAa	0	x	23.5	617	NA	NA	FN	FN
HC-003	M	74	Somatic	MAP2K1	NM_002755.3:c.390T>A	NP_002746.1:p.Tyr130Ter	Y/*	taT/taA	8.7	69	0	x	NA	NA	FP	x
HC-003	M	74	Somatic	NOTCH1	NM_017617.3:c.6550C>T	NP_060087.3:p.Gln2184Ter	Q/*	Cag/Tag	13	115	0	x	NA	NA	FP	FP
HC-004	M	70	Somatic	IDH1	NM_005896.2:c.1169A>G	NP_005887.2:p.Asp390Gly?	D/G	gAc/gGc	11.7	60	0	x	NA	NA	FP	FP
HC-005	M	77	Somatic	GATA3	NM_001002295.1:c.881T>G	NP_001002295.1:p.Met294Arg	M/R	aTg/aGg	8	200	0	x	NA	NA	FP	FP
HC-006	M	44	Somatic	EZH2	NM_004456.4:c.1850delA	NP_004447.2:p.Lys617SerfsTer58	K/X	aAg/ag	1.23	325	NA	NA	NA	NA		
HC-006	M	44	Somatic	KIT	NM_000222.2:c.2447A>T	NP_000213.1:p.Asp816Val	D/V	gAc/gTc	1.74	345	NA	NA	NA	NA		
HC-006	M	44	Somatic	FLT3	NM_004119.2:c.2380C>T	NP_004110.2:p.Gln794Ter	Q/*	Caa/Taa	6.63	422	NA	NA	NA	NA		
HC-006	M	44	Somatic	PDGFRA	NM_006206.4:c.2428G>A	NP_006197.1:p.Ala810Thr(0.99)	A/T	Gct/Act	10.76	409	NA	NA	NA	NA		
HC-006	M	44	Somatic	LRP1B	NM_018557.2:c.11227G>A	NP_061027.2:p.Gly3743Ser	G/S	Ggt/Agt	35.88	301	NA	NA	NA	NA		
HC-007	M	84	Somatic	APC	NM_000038.5:c.2626C>T	NP_000029.2:p.Arg876Ter	R/*	Cga/Tga	1.03	390	NA	NA	NA	NA		
HC-007	M	84	Somatic	JAK1	NM_002227.2:c.848delC	NP_002218.2:p.Ala283ValfsTer10	A/X	gCt/gt	1.19	588	NA	NA	NA	NA		
HC-007	M	84	Somatic	BCORL1	NM_021946.4:c.3957_3959delGG	NP_068765.3:p.Glu1324del	E/-	GAG/-	2.31	216	NA	NA	NA	NA		
HC-007	M	84	Somatic	DNMT3A	NM_175629.2:c.2094G>A	NP_783328.1:p.Trp698Ter	W/*	tgG/tgA	6.06	297	NA	NA	NA	NA		
HC-008	M	75	Somatic	CSF1R	NM_005211.3:c.1596_1598delGCG	NP_005202.2:p.Leu537del	LL/L	cGCTc/cctc	1.87	481	NA	NA	NA	NA		
HC-009	F	51	Somatic	FLT3	NM_004119.2:c.2073delT	NP_004110.2:p.Phe691LeufsTer13	F/X	tTt/tt	1.39	432	NA	NA	NA	NA		
HC-009	F	51	Somatic	FGFR1	NM_001174067.1:c.854G>T	NP_001167538.1:p.Arg285Leu	R/L	cGg/cTg	1.86	377	NA	NA	NA	NA		
HC-009	F	51	Somatic	TET2	NM_001127208.2:c.3854T>C	NP_001120680.1:p.Phe1285Ser	F/S	tTc/Cc	1.89	317	NA	NA	NA	NA		
HC-009	F	51	Somatic	PPP2R1A	NM_014225.5:c.1015C>T	NP_055040.2:p.Gln339Ter	Q/*	Caa/Taa	6.52	537	NA	NA	NA	NA		
HC-009	F	51	Somatic	DNMT3A	NM_175629.2:c.2255T>G	NP_783328.1:p.Phe752Cys(0.96)	F/C	tTc/tGc	6.78	605	NA	NA	NA	NA		
HC-010	M	66	Somatic	KRAS	NM_033360.2:c.179G>A	NP_203524.1:p.Gly60Asp	G/D	gGt/gAt	1.09	549	NA	NA	NA	NA		
HC-010	M	66	Somatic	ARID1A	NM_006015.4:c.60_62delGCC	NP_006006.3:p.Pro21del	P/-	CCG/-	7.24	138	NA	NA	NA	NA		
HC-012	M	78	Somatic	IDH1	NM_005896.2:c.395G>A	NP_005887.2:p.Arg132His	R/H	cGt/cAt	0.91	328	NA	NA	NA	NA		
HC-012	M	78	Somatic	DNMT3A	NM_175629.2:c.2059G>A	NP_783328.1:p.Val687Ile(0.93)	V/I	Gtc/Atc	6.18	97	NA	NA	NA	NA		
HC-013	M	72	Somatic	DNMT3A	NM_175629.2:c.2645G>A	NP_783328.1:p.Arg882His	R/H	cGc/cAc	1.32	528	NA	NA	NA	NA		
HC-013	M	72	Somatic	PTPN11	NM_002834.3:c.181G>C	NP_002825.3:p.Asp61His(0.99)	D/H	Gat/Cat	1.56	320	NA	NA	NA	NA		
HC-014	M	85	Somatic	TET2	NM_001127208.2:c.2827C>T	NP_001120680.1:p.Gln943Ter	Q/*	Cag/Tag	5.06	474	NA	NA	NA	NA		
HC-014	M	85	Somatic	TET2	NM_001127208.2:c.1852C>T	NP_001120680.1:p.Gln618Ter	Q/*	Caa/Taa	5.94	640	NA	NA	NA	NA		
HC-014	M	85	Somatic	TET2	NM_001127208.2:c.4546C>T	NP_001120680.1:p.Arg1516Ter	R/*	Cga/Tga	13.58	405	NA	NA	NA	NA		
HC-015	M	76	Somatic	EGFR	NM_005228.3:c.2235_2249delGG	NP_005219.2:p.Glu746_Ala750del	KELREA/K	TAAGAGA	1.66	302	NA	NA	NA	NA		
HC-016	M	75	Somatic	DNMT3A	NM_175629.2:c.1669T>C	NP_783328.1:p.Cys557Arg	C/R	Tgc/Cgc	2.08	289	NA	NA	NA	NA		
HC-016	M	75	Somatic	PDGFRA	NM_006206.4:c.2525A>T	NP_006197.1:p.Asp842Val	D/V	gAc/gTc	2.21	453	NA	NA	NA	NA		
HC-016	M	75	Somatic	EPHA3	NM_005233.5:c.2369G>A	NP_005224.2:p.Trp790Ter	W/*	tGg/tAg	6.32	411	NA	NA	NA	NA		
HC-016	M	75	Somatic	CHEK2	NM_001005735.1:c.247C>T	NP_001005735.1:p.Gln83Ter	Q/*	Caa/Taa	7.28	302	NA	NA	NA	NA		
HC-016	M	75	Somatic	DNMT3A	NM_175629.2:c.1122+1G>A				12.96	324	NA	NA	NA	NA		
HC-017	F	96	Somatic	PPM1D	NM_003620.3:c.1280G>A	NP_003611.1:p.Trp427Ter	W/*	tGg/tAg	1.61	311	NA	NA	NA	NA		
HC-017	F	96	Somatic	PDGFRA	NM_006206.4:c.2525A>T	NP_006197.1:p.Asp842Val	D/V	gAc/gTc	1.71	468	NA	NA	NA	NA		
HC-017	F	96	Somatic	TET2	NM_001127208.2:c.4700delA	NP_001120680.1:p.Asn1567IlefsTer4	N/X	Aat/at	2.73	330	NA	NA	NA	NA		
HC-018	F	94	Somatic	SF3B1	NM_012433.2:c.1984C>G	NP_036565.2:p.His662Asp	H/D	Cac/Gac	1.75	228	NA	NA	NA	NA		
HC-018	F	94	Somatic	ZRSR2	NM_005089.3:c.376C>T	NP_005080.1:p.Arg126Ter	R/*	Cga/Tga	1.91	419	NA	NA	NA	NA		
HC-019	F	61	Somatic	RUNX1	NM_001754.4:c.484A>G	NP_001745.2:p.Arg162Gly	R/G	Agg/Ggg	1.46	412	NA	NA	NA	NA		
HC-019	F	61	Somatic	ARID1A	NM_006015.4:c.2296delC	NP_006006.3:p.Gln766SerfsTer67	S/X	tCc/tc	6.36	110	NA	NA	NA	NA		
HC-019	F	61	Somatic	DNMT3A	NM_175629.2:c.1579C>T	NP_783328.1:p.Gln527Ter(0.98)	Q/*	Cag/Tag	33.13	323	NA	NA	NA	NA		
HC-020	M	70	Somatic	BRAF	NM_004333.4:c.1802A>G	NP_004324.2:p.Lys601Arg	K/R	aAa/aGa	0.94	427	NA	NA	NA	NA		
HC-020	M	70	Somatic	FLCN	NM_144997.5:c.1285delC	NP_659434.2:p.His429ThrfsTer39	H/X	Cac/ac	4.17	384	NA	NA	NA	NA		
HC-021	M	72	Somatic	KIT	NM_000222.2:c.2447A>T	NP_000213.1:p.Asp816Val	D/V	gAc/gTc	1.24	403	NA	NA	NA	NA		
HC-021	M	72	Somatic	DNMT3A	NM_175629.2:c.2193_2195delCT	NP_783328.1:p.Phe732del	FF/F	ttCTT/ttt	6.65	481	NA	NA	NA	NA		
HC-021	M	72	Likely germline	CD79A	NM_001783.3:c.379+1G>A				47.13	314	NA	NA	NA	NA		
HC-022	M	67	Somatic	all NEG					0	x	NA	NA	NA	NA		

HC-023	M	91	Likely germline	CEBPA	NM_004364.3:c.584_589delACC	NP_004355.2:p.His195_Pro196del	HPP/P	CCCCGcg/c	40.62	64	NA	NA	NA	NA		
HC-024	M	55	Somatic	all NEG					0	x	NA	NA	NA	NA		
HC-025	M	76	Somatic	PIK3CA	NM_006218.2:c.1634A>C	NP_006209.2:p.Glu545Ala	E/A	gAg/gCg	1.18	424	NA	NA	NA	NA		
HC-025	M	76	Somatic	RHEB	NM_005614.3:c.73C>T	NP_005605.1:p.Gln25Ter	Q/*	Caa/Taa	4.06	197	NA	NA	NA	NA		
HC-025	M	76	Somatic	RHEB	NM_005614.3:c.68C>G	NP_005605.1:p.Thr23Arg(1.0)	T/R	aCg/aGc	4.06	197	NA	NA	NA	NA		
HC-026	M	83	Somatic	EGFR	NM_005228.3:c.2327G>T	NP_005219.2:p.Arg776Leu(0.98)	R/L	cCg/cTc	1.1	364	NA	NA	NA	NA		
HC-026	M	83	Somatic	EZH2	NM_004456.4:c.2068C>T	NP_004447.2:p.Arg690Cys	R/C	Cgt/Tgt	1.4	356	NA	NA	NA	NA		
HC-026	M	83	Somatic	PDGFRA	NM_006206.4:c.2525A>T	NP_006197.1:p.Asp842Val	D/V	gAc/gTc	1.45	552	NA	NA	NA	NA		
HC-026	M	83	Somatic	NRAS	NM_002524.4:c.35G>A	NP_002515.1:p.Gly12Asp	G/D	gGt/gAt	1.95	409	NA	NA	NA	NA		
HC-026	M	83	Somatic	KRAS	NM_033360.2:c.34G>C	NP_203524.1:p.Gly12Arg	G/R	Ggt/Cgt	30.96	449	NA	NA	NA	NA		
HC-026	M	83	Somatic	TET2	NM_001127208.2:c.2106_2110del	NP_001120680.1:p.His702GlnfsTer8	HL/X	cACTTg/c	40.76	314	NA	NA	NA	NA		
HC-026	M	83	Somatic	TET2	NM_001127208.2:c.4272_4278del	NP_001120680.1:p.Asp1425MetfsTer21	SDV/X	TTGACGTG/	42.46	544	NA	NA	NA	NA		
HC-026	M	83	Somatic	U2AF1	NM_006758.2:c.101C>T	NP_006749.1:p.Ser34Phe	S/F	tCt/tTt	51.05	333	NA	NA	NA	NA		
HC-026	M	83	Somatic	DNMT3A	NM_175629.2:c.2711C>T	NP_783328.1:p.Pro904Leu(0.99)	P/L	cCg/cTg	52.7	518	NA	NA	NA	NA		
HC-027	M	89	Somatic	PIK3CA	NM_006218.2:c.1634A>C	NP_006209.2:p.Glu545Ala	E/A	gAg/gCg	1.05	475	NA	NA	NA	NA		
HC-027	M	89	Somatic	KMT2A	NM_001197104.1:c.1142delA	NP_001184033.1:p.Lys381ArgfsTer19	A/X	gcA/gc	1.97	355	NA	NA	NA	NA		
HC-027	M	89	Somatic	ASXL1	NM_015338.5:c.1900_1922delAG	NP_056153.2:p.Glu635ArgfsTer15	HHCHREAA/X	CATAGAG/	4.07	344	NA	NA	NA	NA		
HC-027	M	89	Somatic	SRSF2	NM_003016.4:c.284C>A	NP_003007.2:p.Pro95His	P/H	cCg/cAc	5.18	309	NA	NA	NA	NA		
HC-028	F	82	Somatic	TP53	NM_000546.5:c.701A>G	NP_000537.3:p.Tyr234Cys	Y/C	tAc/tGc	0.94	533	NA	NA	NA	NA		
HC-028	F	82	Somatic	DNMT3A	NM_175629.2:c.1322_1323dupTC	NP_783328.1:p.Glu442TrpfsTer210	-X	-/TG	19.76	506	NA	NA	NA	NA		
HC-029	M	44	Somatic	CALR	NM_004343.3:c.1099_1150delCT	NP_004334.1:p.Leu367ThrfsTer46	EEEEEDKKRKK	GAAGCAA	1.85	541	NA	NA	NA	NA		
HC-030	M	53	Somatic	GFR1	NM_001174067.1:c.854G>T	NP_001167538.1:p.Arg285Leu	R/L	cGg/cTg	1.42	423	NA	NA	NA	NA		
HC-030	M	53	Somatic	DNMT3A	NM_175629.2:c.1238delG	NP_783328.1:p.Gly413AlafsTer238	G/X	gGc/gc	1.54	325	NA	NA	NA	NA		
HC-031	M	66	Somatic	PIK3CA	NM_006218.2:c.1634A>C	NP_006209.2:p.Glu545Ala	E/A	gAg/gCg	1.51	396	NA	NA	NA	NA		
HC-032	M	72	Somatic	U2AF1	NM_006758.2:c.467G>A	NP_006749.1:p.Arg156His(0.98)	R/H	cGt/cAt	1.06	473	NA	NA	NA	NA		
HC-032	M	72	Somatic	PIK3CA	NM_006218.2:c.3146G>A	NP_006209.2:p.Gly1049Asp(0.97)	G/D	gGt/gAt	1.11	270	NA	NA	NA	NA		
HC-032	M	72	Somatic	ASXL1	NM_015338.5:c.1534C>T	NP_056153.2:p.Gln512Ter(1.0)	Q/*	Cag/Tag	7.02	285	NA	NA	NA	NA		
HC-033	M	73	Somatic	PTEN	NM_000314.4:c.470A>G	NP_000305.3:p.Glu157Gly	E/G	gAa/gGa	1.23	406	NA	NA	NA	NA		
HC-033	M	73	Somatic	PTPN11	NM_002834.3:c.922A>G	NP_002825.3:p.Asn308Asp(1.0)	N/D	Aat/Gat	1.36	369	NA	NA	NA	NA		
HC-033	M	73	Somatic	TP53	NM_000546.5:c.736A>G	NP_000537.3:p.Met246Val	M/V	Atg/Gtg	1.67	240	NA	NA	NA	NA		
HC-033	M	73	Somatic	DNMT3A	NM_175629.2:c.2645G>A	NP_783328.1:p.Arg882His	R/H	cGc/cAc	2.35	468	NA	NA	NA	NA		
HC-033	M	73	Somatic	JAK3	NM_000215.3:c.115delC	NP_000206.2:p.Gln39SerfsTer108	Q/X	Cag/ag	4.33	231	NA	NA	NA	NA		
HC-034	F	63	Somatic	SF3B1	NM_012433.2:c.1873C>T	NP_036565.2:p.Arg625Cys	R/C	Cgt/Tgt	1.25	399	NA	NA	NA	NA		
HC-034	F	63	Somatic	EZH2	NM_004456.4:c.1850delA	NP_004447.2:p.Lys617SerfsTer58	K/X	aAg/ag	2.33	301	NA	NA	NA	NA		
HC-035	F	63	Somatic	PIK3CA	NM_006218.2:c.1634A>C	NP_006209.2:p.Glu545Ala	E/A	gAg/gCg	1.59	378	NA	NA	NA	NA		
HC-035	F	63	Somatic	TP53	NM_000546.5:c.725G>C	NP_000537.3:p.Cys242Ser	C/S	tCg/tCc	1.64	183	NA	NA	NA	NA		
HC-036	F	71	Somatic	PIK3CA	NM_006218.2:c.3073A>G	NP_006209.2:p.Thr1025Ala(1.0)	T/A	Acc/Gcc	1.51	396	NA	NA	NA	NA		
HC-037	F	60	Somatic	DNMT3A	NM_175629.2:c.2711C>T	NP_783328.1:p.Pro904Leu(0.99)	P/L	cCg/cTg	0.96	416	NA	NA	NA	NA		
HC-038	F	88	Somatic	ERBB2	NM_004448.2:c.2305G>A	NP_004439.2:p.Asp769Asn(0.99)	D/N	Gac/Aac	1.58	190	NA	NA	NA	NA		
HC-038	F	88	Somatic	SMC3	NM_005445.3:c.127delT	NP_005436.1:p.Tyr43MetfsTer69	F/X	Ttt/tt	2.3	304	NA	NA	NA	NA		
HC-038	F	88	Somatic	EP300	NM_001429.3:c.6970delC	NP_001420.2:p.His2324ThrfsTer29	P/X	Ccc/cc	4.37	183	NA	NA	NA	NA		
HC-038	F	88	Somatic	CBL	NM_005188.3:c.1211G>A	NP_005179.2:p.Cys404Tyr(0.99)	C/Y	tGt/tAt	8.21	280	NA	NA	NA	NA		
HC-039	F	67	Somatic	BRAF	NM_004333.4:c.722C>T	NP_004324.2:p.Thr241Met	T/M	aCg/aTg	1.57	318	NA	NA	NA	NA		
HC-040	M	82	Somatic	JAK1	NM_002227.2:c.848delC	NP_002218.2:p.Ala283ValfsTer10(0.99)	A/X	gCt/gt	1.05	380	NA	NA	NA	NA		
HC-040	M	82	Somatic	CSF3R	NM_156039.3:c.1404delC	NP_724781.1:p.Ser469AlafsTer22?	P/X	ccC/cc	2.71	258	NA	NA	NA	NA		
HC-041	F	50	Somatic	CUX1	NM_001202543.1:c.1867C>T	NP_001189472.1:p.Gln623Ter	Q/*	Cag/Tag	1.13	443	NA	NA	NA	NA		
HC-041	F	50	Somatic	ATRX	NM_000489.3:c.2518delA	NP_000480.2:p.Arg840GlnfsTer29	R/X	AgA/ga	2.72	368	NA	NA	NA	NA		
HC-041	F	50	Somatic	SMC3	NM_005445.3:c.127delT	NP_005436.1:p.Tyr43MetfsTer69	F/X	Ttt/tt	2.93	239	NA	NA	NA	NA		
HC-042	F	70	Somatic	PTPN11	NM_002834.3:c.226G>A	NP_002825.3:p.Glu76Lys	E/K	Gag/Aag	1.46	273	NA	NA	NA	NA		
HC-042	F	70	Somatic	PDGFRA	NM_006206.4:c.2525A>T	NP_006197.1:p.Asp842Val	D/V	gAc/gTc	1.59	378	NA	NA	NA	NA		
HC-042	F	70	Somatic	BCORL1	NM_021946.4:c.5042delC	NP_068765.3:p.Pro1681GlnfsTer20	S/X	tCc/tc	2.4	417	NA	NA	NA	NA		
HC-043	F	42	Somatic	DNMT3A	NM_175629.2:c.958C>T	NP_783328.1:p.Arg320Ter(0.96)	R/*	Cga/Tga	1.55	323	NA	NA	NA	NA		
HC-044	M	83	Somatic	PIK3CA	NM_006218.2:c.263G>A	NP_006209.2:p.Arg88Gln	R/Q	cGc/cAa	1.26	395	NA	NA	NA	NA		
HC-045	F	80	Somatic	FLCN	NM_144997.5:c.1285delC	NP_659434.2:p.His429ThrfsTer39	H/X	Cac/ac	5.26	399	NA	NA	NA	NA		
HC-045	F	80	Somatic	DNMT3A	NM_175629.2:c.1803G>A	NP_783328.1:p.Trp601Ter	W/*	tGt/gTgA	6.76	340	NA	NA	NA	NA		
HC-045	F	80	Somatic	DNMT3A	NM_175629.2:c.1320G>A	NP_783328.1:p.Trp440Ter	W/*	tgGt/gtA	8.27	399	NA	NA	NA	NA		
HC-046	M	72	Somatic	BCORL1	NM_021946.4:c.249delA	NP_068765.3:p.Lys83AsnfsTer33	E/X	gAa/ga	2.41	207	NA	NA	NA	NA		
HC-047	M	77	Somatic	TP53	NM_000546.5:c.712T>C	NP_000537.3:p.Cys238Arg	C/R	Tgt/Cgt	1.24	564	NA	NA	NA	NA		
HC-047	M	77	Somatic	JAK1	NM_002227.2:c.2795T>C	NP_002218.2:p.Leu932Pro	L/P	cTc/cCc	1.56	320	NA	NA	NA	NA		
HC-048	M	71	Somatic	DNMT3A	NM_175629.2:c.2245C>T	NP_783328.1:p.Arg749Cys	R/C	Cgc/Tgc	0.93	645	NA	NA	NA	NA		

HC-049	M	70	Somatic	<i>all NEG</i>					0	x	NA	NA	NA	NA		
HC-050	F	87	Somatic	<i>PDGFRA</i>	NM_006206.4:c.1906A>T	NP_006197.1:p.Ser636Cys	S/C	AgT/Tgt	8.4	143	0	x	NA	NA	FP	FP
HC-050	F	87	Somatic	<i>CHEK2</i>	NM_001005735.1:c.172G>T	NP_001005735.1:p.Gly58Trp	G/W	Ggg/Tgg	10.1	109	0	x	NA	NA	FP	FP
HC-051	M	37	Somatic	<i>TP53</i>	NM_000546.5:c.1146delA	NP_000537.3:p.Lys382AsnfsTer40	K/X	aaA/aa	1.14	438			NA	NA		
HC-051	M	37	Likely germline	<i>CHEK2</i>	NM_001005735.1:c.1229delC	NP_001005735.1:p.Thr410MetfsTer15	T/X	aCt/at	50	332	NA	NA	NA	NA		
HC-052	M	75	Somatic	<i>KIT</i>	NM_000222.2:c.2447A>T	NP_000213.1:p.Asp816Val	D/V	gAc/gTc	1.36	441	NA	NA	NA	NA		
HC-053	M	71	Somatic	<i>BRAF</i>	NM_004333.4:c.1802A>G	NP_004324.2:p.Lys601Arg	K/R	aAa/aGa	1.33	376	NA	NA	NA	NA		
HC-053	M	71	Somatic	<i>CHEK2</i>	NM_001005735.1:c.599T>C	NP_001005735.1:p.Ile200Thr	I/T	aTt/aCt	36.36	297	NA	NA	NA	NA		
HC-054	F	63	Somatic	<i>PTPN11</i>	NM_002834.3:c.173A>G	NP_002825.3:p.Asn58Ser	N/S	aAc/aGc	48.89	270	NA	NA	NA	NA		
HC-055	M	65	Somatic	<i>all NEG</i>					0	x	NA	NA	NA	NA		
HC-056	M	70	Somatic	<i>HRAS</i>	NM_005343.2:c.182A>G	NP_005334.1:p.Gln61Arg	Q/R	cAg/cGg	0.73	410	NA	NA	NA	NA		
HC-056	M	70	Somatic	<i>DNMT3A</i>	NM_175629.2:c.2246G>A	NP_783328.1:p.Arg749His	R/H	cGc/cAc	1.52	461	NA	NA	NA	NA		
HC-056	M	70	Somatic	<i>BRAF</i>	NM_004333.4:c.1742A>G	NP_004324.2:p.Asn581Ser	N/S	aAt/aGt	1.93	311	NA	NA	NA	NA		
HC-056	M	70	Somatic	<i>BCORL1</i>	NM_021946.4:c.1115_1118delTCC	NP_068765.3:p.Leu372ProfsTer45	L/X	cTCATc/cc	4.3	93	NA	NA	NA	NA		
HC-057	M	65	Somatic	<i>PIK3CA</i>	NM_006218.2:c.1634A>C	NP_006209.2:p.Glu545Ala	E/A	gAg/gCg	1.34	448	NA	NA	NA	NA		
HC-058	M	61	Somatic	<i>KMT2A</i>	NM_001197104.1:c.1142delA	NP_001184033.1:p.Lys381ArgfsTer19	A/X	gcA/gc	1.94	309	NA	NA	NA	NA		
HC-058	M	61	Somatic	<i>FLT4</i>	NM_182925.4:c.89delC	NP_891555.2:p.Pro30ArgfsTer3	P/X	cCg/cg	7.33	191	NA	NA	NA	NA		
HC-059	M	41	Somatic	<i>MYD88</i>	NM_001172567.1:c.818T>C	NP_001166038.1:p.Leu273Pro	L/P	cTg/cCg	0.57	528	NA	NA	NA	NA		
HC-059	M	41	Somatic	<i>DNMT3A</i>	NM_175629.2:c.2204A>G	NP_783328.1:p.Tyr735Cys	Y/C	tAc/tGc	0.6	663	NA	NA	NA	NA		
HC-059	M	41	Somatic	<i>SF3B1</i>	NM_012433.2:c.1997A>G	NP_036565.2:p.Lys666Arg	K/R	aAg/aGg	0.82	367	NA	NA	NA	NA		
HC-059	M	41	Somatic	<i>PPM1D</i>	NM_003620.3:c.1535delA	NP_003611.1:p.Asn512IlefsTer2	Q/X	cAa/ca	1.64	426	NA	NA	NA	NA		
HC-059	M	41	Somatic	<i>JAK1</i>	NM_002227.2:c.425delA	NP_002218.2:p.Lys142ArgfsTer26	K/X	aAg/ag	5.23	287	NA	NA	NA	NA		
HC-060	M	63	Somatic	<i>DNMT3A</i>	NM_175629.2:c.2185C>T	NP_783328.1:p.Arg729Trp	R/W	Cgg/Tgg	1.09	642	NA	NA	NA	NA		
HC-060	M	63	Somatic	<i>FLT3</i>	NM_004119.2:c.52delT	NP_004110.2:p.Ser18LeufsTer3	S/X	Tct/tc	1.38	506	NA	NA	NA	NA		
HC-060	M	63	Somatic	<i>JAK1</i>	NM_002227.2:c.425delA	NP_002218.2:p.Lys142ArgfsTer26	K/X	aAg/ag	4.78	272	NA	NA	NA	NA		
HC-061	M	32	Somatic	<i>MLH1</i>	NM_000249.3:c.2174G>A	NP_000240.1:p.Arg725His	R/H	cGc/cAc	24.63	540	NA	NA	NA	NA		
HC-062	F	87	Somatic	<i>CSF3R</i>	NM_156039.3:c.1404delC	NP_724781.1:p.Ser469AlafsTer22	P/X	ccC/cc	2.5	200	NA	NA	NA	NA		
HC-062	F	87	Somatic	<i>ASXL1</i>	NM_015338.5:c.619_621delAGC	NP_056153.2:p.Ser207del	SS/S	tcCAGc/tcc	2.95	237	NA	NA	NA	NA		
HC-062	F	87	Somatic	<i>BCORL1</i>	NM_021946.4:c.249delA	NP_068765.3:p.Lys83AsnfsTer33	E/X	gAa/ga	3.02	232	NA	NA	NA	NA		
HC-063	M	84	Somatic	<i>all NEG</i>					0	x	NA	NA	NA	NA		
HC-064	F	74	Somatic	<i>BRAF</i>	NM_004333.4:c.1742A>G	NP_004324.2:p.Asn581Ser	N/S	aAt/aGt	1.2	334	NA	NA	NA	NA		
HC-064	F	74	Somatic	<i>SMC3</i>	NM_005445.3:c.127delT	NP_005436.1:p.Tyr43MetfsTer69	F/X	Ttt/tt	2.67	262	NA	NA	NA	NA		
HC-064	F	74	Somatic	<i>TP53</i>	NM_000546.5:c.367A>G	NP_000537.3:p.Thr123Ala	T/A	Act/Gct	50	202	NA	NA	NA	NA		
HC-065	M	53	Somatic	<i>MLH1</i>	NM_000249.3:c.1852A>G	NP_000240.1:p.Lys618Glu	K/E	Aag/Gag	26.37	474	NA	NA	NA	NA		
HC-065	M	53	Somatic	<i>MLH1</i>	NM_000249.3:c.1853A>C	NP_000240.1:p.Lys618Thr	K/T	aAg/aGc	26.53	475	NA	NA	NA	NA		
HC-066	M	90	Somatic	<i>DNMT3A</i>	NM_175629.2:c.1906G>A	NP_783328.1:p.Val636Met	V/M	Gtg/Atg	0.9	443	NA	NA	NA	NA		
HC-066	M	90	Somatic	<i>TP53</i>	NM_000546.5:c.512A>G	NP_000537.3:p.Glu171Gly	E/G	gAg/gGg	0.96	311	NA	NA	NA	NA		
HC-066	M	90	Somatic	<i>SF3B1</i>	NM_012433.2:c.1873C>T	NP_036565.2:p.Arg625Cys	R/C	Cgt/Tgt	1.11	449	NA	NA	NA	NA		
HC-067	M	51	Somatic	<i>HNF1A</i>	NM_000545.5:c.864delG	NP_000536.5:p.Pro291GlnfsTer51	G/X	Ggg/gg	6.27	383	NA	NA	NA	NA		
HC-068	M	78	Somatic	<i>PDGFRA</i>	NM_006206.4:c.2525A>T	NP_006197.1:p.Asp842Val	D/V	gAc/gTc	1.16	516	NA	NA	NA	NA		
HC-068	M	78	Somatic	<i>KIT</i>	NM_000222.2:c.1674G>C	NP_000213.1:p.Lys558Asn(0.81)	K/N	aaG/aaC	7.24	456	NA	NA	NA	NA		
HC-068	M	78	Somatic	<i>TET2</i>	NM_001127208.2:c.1648C>T	NP_001120680.1:p.Arg550Ter	R/*	Cga/Tga	8.14	565	NA	NA	NA	NA		
HC-068	M	78	Somatic	<i>ASXL1</i>	NM_015338.5:c.2404G>T	NP_056153.2:p.Glu802Ter	E/*	Gag/Tag	51.71	321	NA	NA	NA	NA		
HC-069	M	82	Somatic	<i>APC</i>	NM_000038.5:c.2609dupC	NP_000029.2:p.Gly871ArgfsTer41	-/X	-/C	4.08	147	NA	NA	NA	NA		
HC-069	M	82	Somatic	<i>TET2</i>	NM_001127208.2:c.685delA	NP_001120680.1:p.Thr229HisfsTer21	E/X	gAa/ga	4.33	439	NA	NA	NA	NA		
HC-070	M	77	Somatic	<i>TET2</i>	NM_001127208.2:c.4160A>G	NP_001120680.1:p.Asn1387Ser	N/S	aAc/aGc	1.63	367	NA	NA	NA	NA		
HC-070	M	77	Somatic	<i>DNMT3A</i>	NM_175629.2:c.2645G>A	NP_783328.1:p.Arg882His	R/H	cGc/cAc	16.33	447	NA	NA	NA	NA		
HC-071	M	87	Somatic	<i>all NEG</i>					0	x	NA	NA	NA	NA		
HC-072	M	81	Somatic	<i>TET2</i>	NM_001127208.2:c.3893G>A	NP_001120680.1:p.Cys1298Tyr(0.99)	C/Y	tGt/At	1.34	373	NA	NA	NA	NA		
HC-072	M	81	Somatic	<i>JAK2</i>	NM_004972.3:c.1849G>T	NP_004963.1:p.Val617Phe	V/F	Gtc/Ttc	2.55	196	NA	NA	NA	NA		
HC-073	M	70	Somatic	<i>SETBP1</i>	NM_015559.2:c.2608G>A	NP_056374.2:p.Gly870Ser	G/S	Ggc/Agc	0.77	517	NA	NA	NA	NA		
HC-073	M	70	Somatic	<i>APC</i>	NM_000038.5:c.4393_4394delAG	NP_000029.2:p.Ser1465TrpfsTer3	K/X	aAG/a	1.06	473	NA	NA	NA	NA		
HC-073	M	70	Somatic	<i>JAK1</i>	NM_002227.2:c.2795T>C	NP_002218.2:p.Leu932Pro	L/P	cTc/cCc	2.25	267	NA	NA	NA	NA		
HC-073	M	70	Somatic	<i>DNMT3A</i>	NM_175629.2:c.2711C>T	NP_783328.1:p.Pro904Leu	P/L	cCg/cTg	2.38	461	NA	NA	NA	NA		
HC-073	M	70	Somatic	<i>JAK3</i>	NM_000215.3:c.115delC	NP_000206.2:p.Gln359SerfsTer108	Q/X	Cag/ag	2.64	265	NA	NA	NA	NA		
HC-074	M	68	Somatic	<i>TET2</i>	NM_001127208.2:c.4075C>T	NP_001120680.1:p.Arg1359Cys(0.98)	R/C	Cgt/Tgt	1.19	252	NA	NA	NA	NA		
HC-074	M	68	Somatic	<i>TP53</i>	NM_000546.5:c.725G>C	NP_000537.3:p.Cys242Ser	C/S	tGct/Cc	1.9	158	NA	NA	NA	NA		
HC-074	M	68	Somatic	<i>FLT3</i>	NM_004119.2:c.2027A>G	NP_004110.2:p.Asn676Ser	N/S	aAc/aGc	2.84	141	NA	NA	NA	NA		
HC-075	F	67	Somatic	<i>FBXW7</i>	NM_033632.3:c.1513C>T	NP_361014.1:p.Arg505Cys	R/C	Cgc/Tgc	0.81	490	NA	NA	NA	NA		
HC-075	F	67	Somatic	<i>PPM1D</i>	NM_003620.3:c.1535delA	NP_003611.1:p.Asn512IlefsTer2	Q/X	cAa/ca	2.42	289	NA	NA	NA	NA		

HC-076	F	84	Somatic	ASXL1	NM_015338.5:c.2893C>T	NP_056153.2:p.Arg965Ter	R/	Cga/Tga	1.23	244	NA	NA	NA	NA		
HC-076	F	84	Somatic	SF3B1	NM_012433.2:c.2098A>G	NP_036565.2:p.Lys700Glu(0.99)	K/E	Aaa/Gaa	12.97	239	NA	NA	NA	NA		
HC-077	F	83	Somatic	FBXW7	NM_033632.3:c.1513C>T	NP_361014.1:p.Arg505Cys(0.94)	R/C	Cgc/Tgc	1.15	435	NA	NA	NA	NA		
HC-077	F	83	Somatic	FGFR1	NM_001174067.1:c.854G>T	NP_001167538.1:p.Arg285Leu(0.96)	R/L	cGg/cTg	1.72	291	NA	NA	NA	NA		
HC-077	F	83	Somatic	DNMT3A	NM_175629.2:c.2255T>G	NP_783328.1:p.Phe752Cys(0.96)	F/C	tTc/tGc	4.08	417	NA	NA	NA	NA		
HC-078	F	45	Somatic	SF3B1	NM_012433.2:c.1868A>G	NP_036565.2:p.Tyr623Cys(0.99)	Y/C	tAt/tGt	0.95	421	NA	NA	NA	NA		
HC-078	F	45	Somatic	FLT3	NM_004119.2:c.2039C>T	NP_004110.2:p.Ala680Val(0.99)	A/V	gCg/gTg	1.12	267	NA	NA	NA	NA		
HC-078	F	45	Somatic	TP53	NM_000546.5:c.536A>G	NP_000537.3:p.His179Arg(0.99)	H/R	cAt/cGt	1.2	334	NA	NA	NA	NA		
HC-079	F	54	Somatic	PIK3CA	NM_006218.2:c.1637A>G	NP_006209.2:p.Gln546Arg	Q/R	cAg/cGg	0.99	404	NA	NA	NA	NA		
HC-079	F	54	Somatic	TP53	NM_000546.5:c.512A>G	NP_000537.3:p.Glu171Gly	E/G	gAg/gGg	1.52	329	NA	NA	NA	NA		
HC-079	F	54	Somatic	NF1	NM_001042492.2:c.7063-2A>T	pathogenic (0.99)			5.76	399	NA	NA	NA	NA		
HC-079	F	54	Somatic	PPP2R1A	NM_014225.5:c.1015C>T	NP_055040.2:p.Gln339Ter	Q/	Caa/Taa	6.84	468	NA	NA	NA	NA		
HC-079	F	54	Somatic	EPHA3	NM_005233.5:c.2369G>A	NP_005224.2:p.Trp790Ter	W/	tGg/tAg	8.82	374	NA	NA	NA	NA		
HC-080	M	92	Somatic	PTEN	NM_000314.4:c.407G>A	NP_000305.3:p.Cys136Tyr	C/Y	tGt/tAt	1.83	327	NA	NA	NA	NA		
HC-080	M	92	Somatic	CEBPA	NM_004364.3:c.68delC	NP_004355.2:p.Pro23ArgfsTer137	P/X	cCg/cg	2.55	196	NA	NA	NA	NA		
HC-080	M	92	Somatic	MAP2K4	NM_001281435.1:c.25_26insCC	NP_001268364.1:p.Gly9AlafsTer17	G/AX	ggc/gCCgc	5.97	134	NA	NA	NA	NA		
HC-081	F	86	Somatic	PTEN	NM_000314.4:c.470A>G	NP_000305.3:p.Glu157Gly	E/G	gAa/gGa	1.31	305	NA	NA	NA	NA		
HC-081	F	86	Somatic	BCOR	NM_001123385.1:c.3621delA	NP_001116857.1:p.Lys1207AsnfsTer31	K/X	aaA/aa	1.61	248	NA	NA	NA	NA		
HC-081	F	86	Somatic	GATA1	NM_002049.3:c.1203delC	NP_002040.1:p.Thr402ProfsTer30	P/X	Ccc/cc	2.69	223	NA	NA	NA	NA		
HC-081	F	86	Somatic	KIT	NM_000222.2:c.2133delG	NP_000213.1:p.Glu711AspfsTer14	E/X	gaG/ga	5.16	155	NA	NA	NA	NA		
HC-082	M	83	Somatic	all NEG					0	x	NA	NA	NA	NA		
HC-083	M	89	Somatic	ASXL1	NM_015338.5:c.2423delC	NP_056153.2:p.Pro808LeufsTer10	P/X	Cct/ct	21.88	329	NA	NA	NA	NA		
HC-084	M	84	Somatic	TET2	NM_001127208.2:c.3604A>G	NP_001120680.1:p.Arg1202Gly	R/G	Aga/Gga	0.96	312	NA	NA	NA	NA		
HC-084	M	84	Somatic	SMC3	NM_005445.3:c.127delT	NP_005436.1:p.Tyr43MetfsTer69	F/X	Ttt/tt	3.28	305	NA	NA	NA	NA		
HC-084	M	84	Somatic	SUZ12	NM_015355.2:c.80delG	NP_056170.2:p.Gly27ValfsTer45	G/X	Ggg/gg	4.41	68	NA	NA	NA	NA		
HC-084	M	84	Somatic	SMARCA4	NM_001128849.1:c.1663C>T	NP_001122321.1:p.Gln555Ter	Q/	Cag/Tag	8.31	445	NA	NA	NA	NA		
HC-085	F	74	Somatic	HNF1A	NM_000545.5:c.864delG	NP_000536.5:p.Pro291GlnfsTer51	G/X	Ggg/gg	6.69	299	NA	NA	NA	NA		
HC-086	M	55	Somatic	TP53	NM_000546.5:c.887A>G	NP_000537.3:p.His296Arg	H/R	cAc/cGc	1.22	411	NA	NA	NA	NA		
HC-086	M	55	Somatic	PTPN11	NM_002834.3:c.1504T>C	NP_002825.3:p.Ser502Pro	S/P	Tca/Cca	1.26	318	NA	NA	NA	NA		
HC-086	M	55	Somatic	PDGFRA	NM_006206.4:c.2525A>T	NP_006197.1:p.Asp842Val	D/V	gAc/gTc	1.36	440	NA	NA	NA	NA		
HC-086	M	55	Somatic	PTPN11	NM_002834.3:c.181G>C	NP_002825.3:p.Asp61His	D/H	Gat/Cat	1.62	308	NA	NA	NA	NA		
HC-086	M	55	Somatic	TET2	NM_001127208.2:c.3854T>C	NP_001120680.1:p.Phe1285Ser	F/S	tTc/tCc	1.89	265	NA	NA	NA	NA		
HC-087	M	65	Somatic	all NEG					0	x	NA	NA	NA	NA		
HC-088	M	24	Somatic	DNMT3A	NM_175629.2:c.2312G>A	NP_783328.1:p.Arg771Gln	R/Q	cGa/cAa	0	x	2.38	252	0	x	FN	TN
HC-089	F	27	Somatic	TP53	NM_000546.5:c.86delA	NP_000537.3:p.Asn29ThrfsTer15	N/X	aAc/ac	0	x	1.6	187	0	x	FN	TN
HC-089	F	27	Somatic	PIK3CA	NM_006218.2:c.263G>A	NP_006209.2:p.Arg88Gln	R/Q	cGa/cAa	0	x	1.78	225	0	x	FN	x
HC-089	F	27	Somatic	PHF6	NM_032458.2:c.767C>G	NP_115834.1:p.Ser256Ter	S/	tCa/tGa	0	x	1.95	205	0	x	FN	x
HC-089	F	27	Somatic	BRAF	NM_004333.4:c.1742A>G	NP_004324.2:p.Asn581Ser	N/S	aAt/aGt	0	x	0	x	1.83	327	FN	x
HC-089	F	27	Somatic	GATA1	NM_002049.3:c.49C>T	NP_002040.1:p.Gln17Ter	Q/	Cag/Tag	0	x	0	x	1.8	333	FN	x
HC-089	F	27	Somatic	SF3B1	NM_012433.2:c.1997A>G	NP_036565.2:p.Lys666Arg	K/R	aAg/aGg	0	x	0	x	1.67	300	FN	x
HC-090	M	38	Likely germline	NPM1	NM_002520.6:c.519_521delTGA	NP_002511.1:p.Asp173del	D/-	GAT/-	39.94	353	43.8	379	46.71	456	TP	TP
HC-091	M	47	Somatic	RB1	NM_000321.2:c.2663+2T>C				0	x	0	x	2.34	214	FN	x
HC-091	M	47	Somatic	TET2	NM_001127208.2:c.4346C>T	NP_001120680.1:p.Ser1449Phe	S/F	tCt/tTt	0	x	0	x	2.56	273	FN	x
HC-091	M	47	Somatic	GRIN2A	NM_000833.3:c.4021delA	NP_000824.1:p.Ser1341AlafsTer56	S/X	Agc/gc	4.65	172	0	x	0	x	FP	FP
HC-091	M	47	Likely germline	BRCA2	NM_000059.3:c.9976A>T	NP_000050.2:p.Lys3326Ter	K/	Aaa/Taa	58.98	256	45.78	308	57.84	306	TP	TP
HC-092	M	41	Somatic	all NEG					0	x	0	x	0	x	TN	TN
HC-093	F	53	Somatic	TP53	NM_000546.5:c.509C>T	NP_000537.3:p.Thr170Met	T/M	aCg/aTg	2.17	138	0	x	0	x	FP	x
HC-093	F	53	Likely germline	MSH6	NM_000179.2:c.431G>T	NP_000170.1:p.Ser144Ile	S/I	aGc/aTc	46.39	194	52.81	267	44.83	290	TP	TP
HC-093	F	53	Likely germline	PDGFRA	NM_006206.4:c.661C>T	NP_006197.1:p.Leu221Phe, snp	L/F	Ctt/tTt	52.55	274	53.51	342	54.98	442	TP	TP
HC-094	M	59	Somatic	TET2	NM_001127208.2:c.4346C>T	NP_001120680.1:p.Ser1449Phe	S/F	tCt/tTt	0	x	3.64	220	0	x	FN	FN
HC-094	M	59	Somatic	FGFR1	NM_001174067.1:c.2060A>G	NP_001167538.1:p.Lys687Arg	K/R	aAg/aGg	0	x	2.19	274	0	x	FN	x
HC-094	M	59	Somatic	EPHA3	NM_005233.5:c.1095delA	NP_005224.2:p.Lys365AsnfsTer6	K/X	Aaa/aa	0	x	0	x	2.29	305	FN	x
HC-094	M	59	Somatic	SF3B1	NM_012433.2:c.2219G>C	NP_036565.2:p.Gly740Ala	G/A	gGa/gCa	0	x	0	x	2.02	347	FN	x
HC-094	M	59	Somatic	BRAF	NM_004333.4:c.1447A>G	NP_004324.2:p.Lys483Glu	K/E	Aaa/Gaa	2.65	151	0	x	0	x	FP	x
HC-094	M	59	Somatic	PDGFRA	NM_006206.4:c.2426delT	NP_006197.1:p.Leu809TrpfsTer24	F/X	Ttt/tt	4.67	107	0	x	0	x	FP	FP
HC-094	M	59	Somatic	KIT	NM_000222.2:c.1671G>A	NP_000213.1:p.Trp557Ter	W/	tgG/tgA	4.81	104	0	x	0	x	FP	FP
HC-095	F	69	Somatic	PTEN	NM_000314.4:c.235G>A	NP_000305.3:p.Ala79Thr	A/T	Gcc/acc	0	x	2.55	235	0	x	FN	TN
HC-095	F	69	Somatic	BRCA2	NM_000059.3:c.5073delA	NP_000050.2:p.Lys1691AsnfsTer15	A/X	gcA/gc	0	x	0	x	3.39	354	FN	x

HC-096	F	57	Somatic	ASXL1	NM_015338.5:c.1934delG	NP_056153.2:p.Gly645ValfsTer58	G/X	Ggg/gg	5.61	196	0	x	0	x	FP	FP
HC-097	M	25	Somatic	PIK3CA	NM_006218.2:c.263G>A	NP_006209.2:p.Arg88Gln	R/Q	cGa/cAa	1.26	318	0	x	0	x	FP	TN
HC-097	M	25	Somatic	DNMT3A	NM_175629.2:c.2312G>A	NP_783328.1:p.Arg771Gln	R/Q	cGa/cAa	1.63	246	0	x	0	x	FP	x
HC-098	M	71	Somatic	TSC1	NM_000368.4:c.163C>T	NP_000359.1:p.Gln55Ter	Q/*	Cag/Tag	10.8	83	0	x	NA	NA	FP	FP
HC-098	M	71	Somatic	RB1	NM_000321.2:c.1399C>T	NP_000312.2:p.Arg467Ter	R/*	Cga/Tga	11.3	53	0	x	NA	NA	FP	FP
HC-098	M	71	Somatic	PRKDC	NM_006904.6:c.8306C>A	NP_008835.5:p.Ser2769Ter	S/*	tCg/tAg	13.9	110	0	x	NA	NA	FP	FP
HC-098	M	71	Likely germline	CHEK2	NM_001005735.1:c.1392delT	NP_001005735.1:p.Ser465ValfsTer15	L/X	ctT/ct	58.3	48	42.6	344	NA	NA	TP	TP
HC-099	M	84	Somatic	APC	NM_000038.5:c.2388T>G	NP_000029.2:p.Tyr796Ter	Y/*	taT/taG	9.9	911	0	x	NA	NA	FP	x
HC-099	M	84	Somatic	BCOR	NM_001123385.1:c.1160G>A	NP_001116857.1:p.Arg387Lys	R/K	aGg/aAg	11.8	68	0	x	NA	NA	FP	FP
HC-099	M	84	Somatic	DNMT3A	NM_175629.2:c.886G>C	NP_783328.1:p.Val296Leu	V/L	Gtg/Ctg	19.7	71	0	x	NA	NA	FP	FP
HC-099	M	84	Somatic	MSH6	NM_000179.2:c.2910G>A	NP_000170.1:p.Trp970Ter	W/*	tgG/tgA	28.7	87	0	x	NA	NA	FP	FP
HC-100	M	73	Somatic	SMARCA4	NM_001128849.1:c.2620C>T	NP_001122321.1:p.Arg874Cys	R/C	Cgt/Tgt	10.5	57	0	x	NA	NA	FP	FP
HC-100	M	73	Somatic	MDM4	NM_002393.4:c.1031C>A	NP_002384.2:p.Ser344Tyr	S/Y	tCt/tAt	11.7	145	0	x	NA	NA	FP	FP
HC-100	M	73	Somatic	MCL1	NM_021960.4:c.391C>T	NP_068779.1:p.Pro131Ser	P/S	Ccg/Tcg	20.2	84	0	x	NA	NA	FP	FP
HC-100	M	73	Somatic	DNMT3A	NM_175629.2:c.1903C>T	NP_783328.1:p.Arg635Trp	R/W	Cgg/Tgg	5	162	10.7	431	NA	NA	TP	TP
HC-101	M	84	Somatic	all NEG					0	x	0	x	NA	NA	TN	TN
HC-101	M	77	Somatic	PTPN11	NM_002834.3:c.181G>C	NP_002825.3:p.Asp61His	D/H	Gat/Cat	1.53	327	NA	NA	NA	NA		
HC-101	M	77	Somatic	BCOR	NM_001123385.1:c.1005delC	NP_001116857.1:p.Ser336ArgfsTer42	P/X	ccC/cc	2.55	235	NA	NA	NA	NA		
HC-102	M	86	Somatic	BRAF	NM_004333.4:c.1249C>T	NP_004324.2:p.Gln417Ter	Q/*	Cag/Tag	9.3	162	0	x	NA	NA	FP	FP
HC-102	M	86	Somatic	TET2	NM_001127208.2:c.5711A>G	NP_001120680.1:p.His1904Arg	H/R	cAt/cGt	10.4	278	0	x	NA	NA	FP	FP
HC-102	M	86	Somatic	EP300	NM_001429.3:c.6798_6800delGC	NP_001420.2:p.Gln2268del	Q/-	CAG/-	38.1	194	25.7	344	NA	NA	TP	TP
HC-103	M	38	Somatic	TP53	NM_000546.5:c.437G>A	NP_000537.3:p.Trp146Ter	W/*	tGg/tAg	1.25	321	0	x	NA	NA	FP	TN
HC-104	M	76	Somatic	CSF1R	NM_005211.3:c.149G>A	NP_005202.2:p.Trp50Ter	W/*	tGg/tAg	7.2	138	0	x	NA	NA	FP	FP
HC-104	M	76	Somatic	CIC	NM_015125.3:c.1361C>T	NP_055940.3:p.Ala454Val	A/V	gCt/gTt	12	92	0	x	NA	NA	FP	FP
HC-104	M	76	Somatic	ATRX	NM_000489.3:c.5288A>G	NP_000480.2:p.Asn1763Ser	N/S	aAt/aGt	15.3	59	0	x	NA	NA	FP	FP
HC-104	M	76	Somatic	STK11	NM_000455.4:c.200T>A	NP_000446.1:p.Leu67Gln	L/Q	cTg/cAg	15.8	77	0	x	NA	NA	FP	FP
HC-104	M	76	Somatic	U2AF1	NM_006758.2:c.43A>G	NP_006749.1:p.Lys15Glu	K/E	Aaa/Gaa	16.9	65	0	x	NA	NA	FP	FP
HC-105	M	77	Somatic	PTPN11	NM_002834.3:c.262T>A	NP_002825.3:p.Leu88Ile	L/I	Tta/Ata	9	133	0	x	NA	NA	FP	FP
HC-105	M	77	Somatic	STAT3	NM_139276.2:c.1810G>A	NP_644805.1:p.Gly604Ser	G/S	Ggc/Agc	9.9	112	0	x	NA	NA	FP	FP
HC-105	M	77	Somatic	TERT	NM_198253.2:c.16C>T	NP_937983.2:p.Arg6Cys	R/C	Cgc/Tgc	12	75	0	x	NA	NA	FP	FP
HC-105	M	77	Somatic	ALK	NM_004304.4:c.4530G>A	NP_004295.2:p.Trp1510Ter	W/*	tgG/tgA	15.9	82	0	x	NA	NA	FP	FP
HC-105	M	77	Somatic	BTK	NM_000061.2:c.1001A>T	NP_000052.1:p.Tyr334Phe	Y/F	tAt/tTt	20	70	0	x	NA	NA	FP	FP
HC-106	F	78	Somatic	all NEG					0	x	0	x	NA	NA	TN	TN
HC-107	F	72	Somatic	KDM5C	NM_004187.3:c.1231A>G	NP_004178.2:p.Met411Val	M/V	Atg/Gtg	13	100	0	x	NA	NA	FP	FP

Supplementary Table S3. Sequencing results from patients with aplastic anemia reported by the CLIA-certified cfDNA test

Patient ID	Time of collection in the protocol or at first consultation	Age	Gender	Status	Gene	Hgvsp	Hgvsc	Aminoacid change	Codon	Bone marrow		Peripheral blood cfDNA		Variant-level concordance	Variant-level concordance with filtered data (VAF <10% with read depth >100 and VAF<3.5% with read depth >400)
										VAF (%)	Read depth	VAF (%)	Read depth		
AA-01	2Y	59	M	Somatic	DNMT3A	NP_783328.1.p.Arg882His	NM_175629.2.c.2645G>A	R/H	cGc/cAc	6.12	294	11.93	N/A	TP	TP
AA-02	6M	16	M	Somatic	ASXL1	NP_056153.2.p.Lys618Ter	NM_015338.5.c.1852A>T	K/*	Aaa/Taa	15.64	179	8.58	233	TP	TP
AA-02	6M	16	M	Somatic	ASXL1	NP_056153.2.p.Thr754LeufsTer18	NM_015338.5.c.2256delT	A/X	gcT/gc	9.29	269	6.87	320	TP	TP
AA-02	6M	16	M	Somatic	CBLC	NP_036248.3.p.Thr223Ala	NM_012116.3.c.667A>G	T/A	Aca/Gca	26.49	117	34.21	114	TP	TP
AA-02	6M	16	M	Somatic	EGLN1	NP_071334.1.p.Asp211His	NM_022051.2.c.631G>C	D/H	Gac/Cac	37.78	270	29.14	278	TP	TP
AA-02	Baseline	15	M	Somatic	KMT2D	NP_003473.3.p.Glu363Lys	NM_003482.3.c.1087G>A	E/K	Gag/Aag	0	0	6.79	162	FP	FP
AA-03	Baseline	21	M	Somatic	BCOR	NP_001116857.1.p.Gly893GlufsTer21	NM_001123385.1.c.2676_2677dupAG	G/EX	ggg/gAGgg	3.43	175	3.14	223	TP	x
AA-03	Baseline	21	M	Somatic	BCOR	NP_001116857.1.p.Glu1337LysfsTer32	NM_001123385.1.c.4009delG	E/X	Gaa/aa	5.17	116	2.3	174	TP	FN
AA-03	Baseline	21	M	Somatic	KMT2C	NP_733751.2.p.Ser1614Tyr	NM_170606.2.c.4841C>A	S/Y	ICU/At	40.66	182	45.31	245	TP	TP
AA-03	Baseline	21	M	Somatic	WT1	NP_077744.3.p.Cys277Arg	NM_024426.4.c.829T>C		Tgc/Cgc	3.25	123	0	0	FN	x
AA-04	2Y	13	M	Somatic	All neg					0	0	0	0	TN	TN
AA-05	2Y	52	F	Somatic	All neg					0	0	0	0	TN	TN
AA-06	Baseline	23	F	Somatic	All neg					0	0	0	0	TN	TN
AA-07	Baseline	71	F	Somatic	All neg					0	0	0	0	TN	TN
AA-08	7Y	49	M	Somatic	ASXL1	NP_056153.2.p.Glu635ArgfsTer15	NM_015338.5.c.1900_1922delAG AGAGGC	HHCHREAA/X	G CCATAGAG AGC	23.3	103	35	N/A	TP	TP
AA-08	7Y	49	M	Somatic	PHF6	NP_115834.1.p.Arg274Gln	NM_032458.2.c.821G>A	R/Q	cGa/cAa	5.65	177	5.4	N/A	TP	TP
AA-08	7Y	49	M	Somatic	RUNX1	NP_001745.2.p.Arg107Cys	NM_001754.4.c.319C>T	R/C	Cgc/Tgc	51.61	186	48	N/A	TP	TP
AA-08	7Y	49	M	Somatic	SETBP1	NP_056374.2.p.Asp868Asn	NM_015559.2.c.2602G>A	D/N	Gac/Aac	53.12	128	59	N/A	TP	TP
AA-08	7Y	49	M	Somatic	SETBP1	NP_056374.2.p.Asp868Asn	NM_015559.2.c.2602G>A	D/N	Gac/Aac	53.12	128	59	N/A	TP	TP
AA-09	2Y	41	F	Somatic	All neg					0	0	0	0	TN	TN
AA-10	Baseline	36	M	Somatic	All neg					0	0	0	0	TN	TN
AA-11	6M	38	F	Somatic	All neg					0	0	0	0	TN	TN
AA-12	Baseline	25	M	Somatic	All neg					0	0	0	0	TN	TN
AA-12	6M	26	M	Somatic	RB1	SPLICE_SITE:c.265-2A>T	NM_000321.2.c.265-2A>T	0	0	6.36	173	7.89	76	TP	FN
AA-13	6M	63	M	Somatic	All neg					0	0	0	0	TN	TN
AA-14	Baseline	40	F	Somatic	DNMT3A	NP_783328.1.p.Asn802Ser	NM_175629.2.c.2405A>G	N/S	aAc/aGc	2.31	130	0	0	FN	x
AA-14	Baseline	40	F	Somatic	SF3B1	NP_036565.2.p.Asp621Asn	NM_012433.2.c.1861G>A	D/N	Gat/Aat	0	0	2.28	175	FP	TN
AA-14	Baseline	40	F	Somatic	SF3B1	NP_036565.2.p.Arg625Cys	NM_012433.2.c.1873C>T	R/C	Cgl/Tgt	0	0	1.79	168	FP	x
AA-15	Baseline	40	F	Somatic	ATM	NP_000442.3.p.Trp1710Ter	NM_000051.3.c.5129G>A	W/*	lGg/TAg	0	0	1.58	190	FP	TN
AA-15	Baseline	40	F	Somatic	SMAD4	NP_005350.1.p.Cys499Arg	NM_005359.5.c.1495T>C	C/R	Tgc/Cgc	1.65	182	0	0	FN	x
AA-15	Baseline	40	F	Somatic	TP53	NP_000537.3.p.Leu330Pro	NM_000546.5.c.989T>C	L/P	cTt/cCt	2.46	122	0	0	FN	x
AA-16	2Y	80	F	Somatic	All neg					0	0	0	0	TN	TN
AA-17	Baseline	32	M	Somatic	All neg					0	0	0	0	TN	TN
AA-18	2Y	29	F	Somatic	All neg					0	0	0	0	TN	TN
AA-19	Baseline	55	M	Somatic	All neg					0	0	0	0	TN	TN
AA-19	6M	56	M	Somatic	All neg					0	0	0	0	TN	TN
AA-20	2Y	12	M	Somatic	All neg					0	0	0	0	TN	TN
AA-21	Baseline	41	M	Somatic	PTPN11	NP_002825.3.p.Arg498Gln	NM_002834.3.c.1493G>A	R/Q	cGg/cAg	0	0	2.1	143	FP	TN
AA-21	Baseline	41	M	Somatic	TET2	NP_001120680.1.p.Lys1950Glu	NM_001127208.2.c.5848A>G	K/E	Aaa/Gaa	0	0	1.45	207	FP	x
AA-22	2Y	53	F	Somatic	All neg					0	0	0	0	TN	TN
AA-23	Baseline	71	M	Somatic	All neg					0	0	0	0	TN	TN
AA-24	Baseline	46	F	Somatic	BCOR	NP_001116857.1.p.Gln1423ArgfsTer61	NM_001123385.1.c.4268delA	Q/X	cAg/cg	13.69	168	29.17	N/A	TP	TP
AA-24	Baseline	46	F	Somatic	BCOR	NP_001116857.1.p.Gln231Ter	NM_001123385.1.c.691C>T	Q/*	Cag/Tag	10.27	146	2	N/A	TP	TP
AA-25	2Y	35	M	Somatic	All neg					0	0	0	0	TN	TN
AA-26	6M	24	M	Somatic	All neg					0	0	0	0	TN	TN
AA-27	Baseline	15	F	Somatic	All neg					0	0	0	0	TN	TN
AA-27	6M	15	M	Somatic	All neg					0	0	0	0	TN	TN
AA-28	6M	18	F	Somatic	PMS1	NP_000525.1.p.Arg630Ter	NM_000534.4.c.1888C>T	R/*	Cga/Tga	27.08	421	29.33	416	TP	TP
AA-29	Baseline	36	M	Somatic	PHF6	NP_115834.1.p.Arg225Gln	NM_032458.2.c.674G>A	R/Q	cGa/cAa	3.6	194	4.03	124	TP	TP
AA-30	2Y	28	M	Somatic	ARID1B	NP_065783.3.p.Gln284Ter	NM_020732.3.c.850C>T	Q/*	Cag/Tag	3.6	222	1.36	294	TP	x
AA-30	2Y	28	M	Somatic	BCOR	NP_001116857.1.p.Gln1626Ter	NM_001123385.1.c.4876C>T	Q/*	Cag/Tag	16.4	128	9.68	155	TP	TP
AA-30	2Y	28	M	Somatic	DNM2	NP_001005360.1.p.Gln648Ter	NM_001005360.2.c.1942C>T	Q/*	Caa/Taa	3.03	228	0	0	FN	x
AA-30	2Y	28	M	Somatic	DNMT3A	NP_783328.1.p.Leu648TrpfsTer3	NM_175629.2.c.1942delC	L/X	Ctg/tg	1.53	196	6.38	94	TP	x
AA-30	2Y	28	M	Somatic	DNMT3A	NP_783328.1.p.Asp765Gly	NM_175629.2.c.2294A>G	D/G	gAc/gGc	0	0	1.72	174	FP	x
AA-30	2Y	28	M	Somatic	PTEN	NP_000305.3.p.Arg173His	NM_000314.4.c.518G>A	R/H	cGc/cAc	0.34	294	1.49	201	TP	x
AA-31	Baseline	68	F	Somatic	All neg					0	0	0	0	TN	TN
AA-31	6M	68	F	Somatic	TET2	NP_001120680.1.p.Arg1516Ter	NM_001127208.2.c.4546C>T	R/*	Cga/Tga	0	0	3.37	89	FP	TN
AA-32	Baseline	24	F	Somatic	KMT2C	NP_733751.2.p.Ser3492Ter	NM_170606.2.c.10475delC	S/X	tCa/ta	6.73	297	2.83	283	TP	FN
AA-33	Baseline	39	F	Somatic	All neg					0	0	0	0	TN	TN

AA-33	6M	40	F	Somatic	All neg					0	0	0	0	TN	TN
AA-34	2Y	19	F	Somatic	All neg					0	0	0	0	TN	TN
AA-35	2Y	66	F	Somatic	MYD88	NP_001166038.1:p.Arg209Cys	NM_001172567.1:c.625C>T	R/C	Cgc/Tgc	2.29	131	0	0	FN	TN
AA-36	Baseline	13	F	Somatic	PTPN11	NP_002825.3:p.Tyr279Cys	NM_002834.3:c.836A>G	Y/C	tAt/tGt	48.8	129	54.54	121	TP	TP
AA-37	Baseline	43	F	Somatic	CXCR4	NP_001008540.1:p.Arg338Ter	NM_001008540.1:c.1012C>T	R/*	Cga/Tga	0.5	400	2.46	122	TP	x
AA-37	Baseline	43	F	Somatic	RB1	NP_000312.2:p.Arg579Ter	NM_000321.2:c.1735C>T	R/*	Cga/Tga	0	0	2.8	143	FP	x
AA-37	Baseline	43	F	Somatic	STAG2	NP_001036214.1:p.Gln615Ter	NM_001042749.1:c.1843C>T	Q/*	Cag/Tag	0	0	4.12	97	FP	TN
AA-38	Baseline	58	M	Somatic	All neg					0	0	0	0	TN	TN
AA-38	6M	58	M	Somatic	All neg					0	0	0	0	TN	TN
AA-39	Baseline	74	F	Somatic	All neg					0	0	0	0	TN	TN
AA-40	Baseline	15	M	Somatic	All neg					0	0	0	0	TN	TN
AA-40	6M	16	M	Somatic	All neg					0	0	0	0	TN	TN
AA-41	Baseline	64	M	Somatic	All neg					0	0	0	0	TN	TN
AA-42	Baseline	47	F	Somatic	All neg					0	0	0	0	TN	TN
AA-42	6M	47	F	Somatic	All neg					0	0	0	0	TN	TN

Supplementary Table S4. Sequencing results from patients with myelodysplastic syndromes reported by the CLIA-certified cfDNA test

Patient ID	Age	Timepoint	Status	Gene	Amino Acids	HGVS	HGVS	HGVS	Variant-level concordance	Variant-level concordance with filtered data (VAF <10% with read depth >100 AND VAF <3.5% with read depth >400)	Bone marrow		cfDNA		Peripheral blood CD3+		Peripheral blood granulocytes		Peripheral blood CD3-		
											VAF (%)	Read Depth	VAF (%)	Read Depth	VAF (%)	Read Depth	VAF (%)	Read Depth	VAF (%)	Read Depth	
UPN02	76	at progression	Somatic	NF1	R*	NM_001042492.2:c.4600C>T	NP_001035957.1:p.Arg1534Ter, in BAM	TP	FN		14.6	192	8	40							
UPN02	76	at progression	Somatic	EZH2	G/R	NM_004456.4:c.475G>C	NP_004447.2:p.Gly159Arg	TP	TP		16.5	242	22.7	22							
UPN03	74	Primary endpoint	Somatic	ATM	R/C	NM_000051.3:c.94C>T	NP_000042.3:p.Arg32Cys	TP	TP		20.2	485	29	665	0	0	qns	qns	8.1	101	
UPN03	74	Primary endpoint	Likely germline	JAK2	S/C	NM_004972.3:c.2498C>G	NP_004963.1:p.Ser833Cys	TP	TP		51.3	430	51.8	606	52.7	303	qns	qns	58.9	191	
UPN03	74	Primary endpoint	Somatic	TSC2	S/R/S/X	NM_000548.3:c.5184_5190delCCGCT	NP_000539.2:p.Ser1728ArgfsTer96	FN	FN		20.79	101	0	0							
UPN04	79	Baseline	Somatic	ATM	W/R	NM_000051.3:c.5572T>A	NP_000042.3:p.Trp1858Arg (0.94)	NA	NA		na	na	8	177							
UPN04	79	Baseline	Somatic	ATM	D/N	NM_000051.3:c.7342G>A	NP_000042.3:p.Asp2448Asn(1.0)	NA	NA		na	na	22.5	80							
UPN04	79	Baseline	Somatic	TERT	K/N	NM_198253.2:c.3150G>C	NP_937983.2:p.Lys1050Asn (0.74)	NA	NA		na	na	26.6	418							
UPN04	79	Baseline	Likely germline	MSH6	V/A	NM_000179.2:c.1526T>C	NP_000170.1:p.Val509Ala (0.99)	NA	NA		na	na	40.8	174							
UPN04	79	Primary endpoint	Somatic	SETBP1	D/N	NM_015559.2:c.2602G>A	NP_056374.2:p.Asp868Asn	TP	TP		19	344	0.96	728							
UPN04	79	Primary endpoint	Somatic	TERT	K/N	NM_198253.2:c.3150G>C	NP_937983.2:p.Lys1050Asn (0.74)	TP	TP		23.6	551	19.8	618							
UPN04	79	Primary endpoint	Likely germline	MSH6	V/A	NM_000179.2:c.1526T>C	NP_000170.1:p.Val509Ala (0.99)	TP	TP		30.8	494	31.3	520							
UPN04	79	Primary endpoint	Somatic	U2AF2	Q/E	NM_007279.2:c.439C>G	NP_009210.1:p.Gln147Glu	TP	TP		18.2	435	37.8	612							
UPN05	46	Baseline	Somatic	all NEG				TN	TN		0	0	0	0							
UPN05	46	Primary endpoint	Somatic	all NEG				TN	TN		0	0	0	0							
UPN06	53	Primary endpoint	Somatic	BIRC3	R/K	NM_001165.4:c.1202G>A	NP_001156.1:p.Arg401Lys (0.90)	TP	TP		24.1	547	24.7	747							
UPN06	53	Baseline	Somatic	ASXL1	G/X	NM_015338.5:c.1886_1887delGT	NP_056153.2:p.Gly629AlafsTer5	TP	TP		23.95	503	27.6	740							
UPN06	53	Primary endpoint	Somatic	ASXL1	G/X	NM_015338.5:c.1886_1887delGT	NP_056153.2:p.Gly629AlafsTer5	TP	TP		34.5	394	33.1	456							
UPN06	53	Progression	Somatic	ASXL1	G/X	NM_015338.5:c.1886_1887delGT	NP_056153.2:p.Gly629AlafsTer5	TP	TP		34.5	321	43.9	338							
UPN06	53	Progression	Somatic	RUNX1	D/N	NM_001754.4:c.592G>A	NP_001745.2:p.Asp198Asn (0.94)	TP	TP		54.5	382	55.7	291							
UPN07	73	Baseline	Likely germline	TE72	S/L	NM_001127208.2:c.3116C>T	NP_001120680.1:p.Ser1039Leu	TP	TP		54.4	601	47.9	585	52.7	408	50.7	367	58.3	156	
UPN07	73	Baseline	Likely germline	CDK12	L/Q	NM_016507.2:c.1697T>A	NP_057591.2:p.Leu566Gln	TP	TP		48.7	392	50.2	445	50.7	305	48.7	238	52	100	
UPN07	73	Baseline	Somatic	TE72	Y/I	NM_001127208.2:c.575_576insAAT	NP_001120680.1:p.Tyr192delinsTer	TP	TP		37	392	45	360	0	0	52.8	220	33.3	69	
UPN07	73	Baseline	Somatic	TE72	Q/N/X	NM_001127208.2:c.1118_1122delAAA	NP_001120680.1:p.Gln373ArgfsTer15	TP	TP		24.23	587	25.2	706	0	0	0	0	0	0	
UPN07	73	Primary endpoint	Somatic	TE72	Q/N/X	NM_001127208.2:c.1118_1122delAAA	NP_001120680.1:p.Gln373ArgfsTer15	FN	FN		29.2	662	0	0							
UPN07	73	Primary endpoint	Somatic	TE72	Y/I	NM_001127208.2:c.575_576insAAT	NP_001120680.1:p.Tyr192delinsTer	TP	TP		35.4	465	36.8	563							
UPN07	73	Primary endpoint	Likely germline	CDK12	L/Q	NM_016507.2:c.1697T>A	NP_057591.2:p.Leu566Gln	TP	TP		50.6	449	45.7	579							
UPN07	73	Primary endpoint	Likely germline	TE72	S/L	NM_001127208.2:c.3116C>T	NP_001120680.1:p.Ser1039Leu	TP	TP		49.6	740	48.3	706							
UPN08	47	Baseline	Somatic	CDH1	V/M	NM_004360.3:c.2494G>A	NP_004351.1:p.Val832Met	NA	NA		na	na	20.5	703							
UPN08	47	Baseline	Somatic	SETBP1	P/Q	NM_015559.2:c.1610C>A	NP_056374.2:p.Pro537Gln	NA	NA		na	na	28.6	729							
UPN08	47	Baseline	Somatic	KDR	R/Q	NM_002253.2:c.2360G>A	NP_002244.1:p.Arg787Gln (0.99)	NA	NA		na	na	51.5	390							
UPN08	47	Primary endpoint	Somatic	all NEG				TN	TN		0	0	0	0							
UPN09	67	Primary endpoint	Somatic	RUNX1	VGG/X	NM_001754.4:c.1256_1262delTGGCC	NP_001745.2:p.Val419AlafsTer173	FN	FN		5.17	58	0	0							
UPN09	67	Baseline	Likely germline	GATA2	G/R	NM_032638.4:c.1096G>A	NP_116027.2:p.Gly366Arg (0.97)	TP	TP		47.3	207	46.8	711							
UPN09	67	at progression	Likely germline	GATA2	G/R	NM_032638.4:c.1096G>A	NP_116027.2:p.Gly366Arg (0.97)	TP	TP		47.3	292	49.7	300							
UPN09	67	Primary endpoint	Likely germline	GATA2	G/R	NM_032638.4:c.1096G>A	NP_116027.2:p.Gly366Arg (0.97)	TP	TP		56.7	277	53.2	487							
UPN10	76	Baseline	Somatic	ASXL1	C/*	NM_015338.5:c.2616C>A	NP_056153.2:p.Cys872Ter	TP	TP		15.6	365	16.3	312	qns	0	9.8	287	9	162	
UPN10	76	Baseline	Somatic	ZFRS2	G/X	NM_005089.3:c.1074delG	NP_005080.1:p.Lys359ArgfsTer?	FN	FN		11.65	371	0	0							
UPN10	76	Primary endpoint	Somatic	ZFRS2	G/X	NM_005089.3:c.1074delG	NP_005080.1:p.Lys359ArgfsTer?	FN	FN		12.38	205	0	0							
UPN10	76	Primary endpoint	Somatic	ASXL1	C/*	NM_015338.5:c.2616C>A	NP_056153.2:p.Cys872Ter	TP	TP		22.3	332	15.6	355							
UPN10	76	Primary endpoint	Somatic	ZFRS2	S/S/X	NM_005089.3:c.120dupA	NP_005080.1:p.Gly41ArgfsTer14	TP	TP		32.7	339	32.7	370							
UPN10	76	Baseline	Somatic	ZFRS2	S/S/X	NM_005089.3:c.120dupA	NP_005080.1:p.Gly41ArgfsTer14	TP	TP		43.1	400	38.8	304							
UPN11	35	Baseline	Somatic	all NEG				TN	TN		0	0	0	0							
UPN11	35	Primary endpoint	Somatic	all NEG				TN	TN		0	0	0	0							
UPN12	76	Baseline	Likely germline	CREBBP	V/M	NM_004380.2:c.712G>A	NP_004371.2:p.Val238Met (0.99)	TP	TP		39.4	175	38.6	377	48.9	315	47.4	139	43.5	264	
UPN12	76	Baseline	Somatic	SF3B1	K/E	NM_012433.2:c.2098A>G	NP_036565.2:p.Lys700Glu (0.99)	TP	TP		28.5	158	43.6	182	0	0	30.1	123	100	0	
UPN12	76	Baseline	Somatic	ATRX	L/*	NM_000489.3:c.2801T>A	NP_000480.2:p.Leu634Ter	FN	FN		10.3	145	0	0							
UPN12	76	Baseline	Somatic	BCL6	R/C	NM_001706.4:c.1375C>T	NP_001697.2:p.Arg459Cys	FN	FN		64.14	198	0	0							
UPN12	76	Primary endpoint	Somatic	SF3B1	K/E	NM_012433.2:c.2098A>G	NP_036565.2:p.Lys700Glu (0.99)	TP	TP		44.1	288	34.4	250							
UPN12	76	Primary endpoint	Somatic	BCL6	R/C	NM_001706.4:c.1375C>T	NP_001697.2:p.Arg459Cys	TP	TP		61.2	408	52.8	743							
UPN12	76	Primary endpoint	Likely germline	CREBBP	V/M	NM_004380.2:c.712G>A	NP_004371.2:p.Val238Met (0.99)	TP	TP		45.8	273	54.5	479							
UPN13	54	Baseline	Likely germline	BRIP1	CCCAC/C	NM_032043.2:c.258_269delTTGTTGT	NP_114432.2:p.Cys87_Cys90del	TP	TP		27.23	235	27.9	594	29	231	41.7	400	43.9	244	
UPN13	54	Baseline	Somatic	WHSC1	Q/E	NM_001042424.2:c.3010C>G	NP_001035889.1:p.Gln1004Glu	FN	FN		14.67	228	0	0							
UPN13	54	Baseline	Somatic	LRP1B	L/F	NM_018557.2:c.1102C>T	NP_061027.2:p.Leu3368Phe (0.96)	TP	TP		48.4	308	43.9	578							
UPN14	85	Baseline	Somatic	SETBP1	G/S	NM_015559.2:c.2608G>A	NP_056374.2:p.Gly870Ser (0.99)	TP	TP		24.1	299	27.1	851							
UPN14	85	Primary endpoint	Somatic	SETBP1	G/S	NM_015559.2:c.2608G>A	NP_056374.2:p.Gly870Ser (0.99)	TP	TP		23.8	491	29.7	1112							
UPN14	85	Primary endpoint	Somatic	ASXL1	HHCHREAA/X	NM_015338.5:c.1900_1922delAAGAGAA	NP_056153.2:p.Glu635ArgfsTer15	TP	TP		34.18	163	31.5	727							
UPN15	64	Baseline	Somatic	all NEG				TN	TN		0	0	0	0							
UPN15	64	Primary endpoint	Somatic	BIRC3	D/Y	NM_001165.4:c.73G>T	NP_001156.1:p.Asp251Trp (0.99)	TP	TP		34.3	310	9.5	147							
UPN15	64	Primary endpoint	Somatic	LRP1B	G/S	NM_018557.2:c.1122T>G>A	NP_061027.2:p.Gly3743Ser (0.97)	TP	TP		38	309	20	70							
UPN15	64	Primary endpoint	Somatic	JAK2</																	

UPN20	63	Primary endpoint	Somatic	EZH2	C/R	NM_004456.4.c.1693T>C	NP_004447.2.p.Cys565Arg	TP	TP	96	327	90.1	533	0	0	95.4	151	26.1	116
UPN20	63	Primary endpoint	Somatic	RAD21	R/G	NM_006265.2.c.193C>G	NP_006256.1.p.Arg65Gly	FN	FN	10.3	408	0	0						
UPN20	63	Baseline	Somatic	RAD21	R/G	NM_006265.2.c.193C>G	NP_006256.1.p.Arg65Gly	NA	NA	na	na	14.7	170						
UPN20	63	Baseline	Somatic	ETV6	F/L	NM_001987.4.c.1251C>G	NP_001978.1.p.Phe417Leu	NA	NA	na	na	52.2	584						
UPN20	63	Baseline	Somatic	EZH2	C/R	NM_004456.4.c.1693T>C	NP_004447.2.p.Cys565Arg	NA	NA	na	na	92.3	416						
UPN20	63	Primary endpoint	Somatic	ASXL1	HHCHREAA/X	NM_015338.5.c.1900_1922delAGAG	NP_056153.2.p.Glu635ArgfsTer15	TP	TP	20.98	206	43.9	312						
UPN21	68	Baseline	Somatic	all NEG	0			TN	TN	0	0	0	0						
UPN21	68	Primary endpoint	Somatic	all NEG	0			TN	TN	0	0	0	0						
UPN22	72	Baseline	Somatic	TE72	A/V	NM_001127208.2.c.5645C>T	NP_001120680.1.p.Ala1882Val	TP	TP	28.1	472	14.8	551						
UPN22	72	Primary endpoint	Somatic	TE72	A/V	NM_001127208.2.c.5645C>T	NP_001120680.1.p.Ala1882Val	TP	TP	27.1	332	18.9	653						
UPN22	72	Baseline	Somatic	SF3B1	H/Q	NM_012433.2.c.1986C>A	NP_036565.2.p.His662Gln	TP	TP	44.4	374	24.2	372						
UPN22	72	Primary endpoint	Somatic	SF3B1	H/Q	NM_012433.2.c.1986C>A	NP_036565.2.p.His662Gln	TP	TP	47.2	254	27.5	241						
UPN23	76	Baseline	Likely germline	C1C	P/S	NM_015125.3.c.2224C>T	NP_055940.3.p.Pro742Ser (0.88)	TP	TP	41.7	207	45.4	579						
UPN23	76	Primary endpoint	Likely germline	C1C	P/S	NM_015125.3.c.2224C>T	NP_055940.3.p.Pro742Ser (0.88)	TP	TP	50	199	50.1	741						
UPN24	63	Baseline	Somatic	ASXL1	R/*	NM_015338.5.c.2077C>T	NP_056153.2.p.Arg693Ter	FN	FN	6.21	145	0	0						
UPN24	63	Baseline	Somatic	ASXL1	L/X	NM_015338.5.c.2468delT	NP_056153.2.p.Leu823Ter	FN	FN	7.14	183	0	0						
UPN24	63	Baseline	Somatic	ASXL1	CQ/X	NM_015338.5.c.2279_2283delAGGCC	NP_056153.2.p.Gln760LeufsTer12	FN	FN	20.17	233	0	0						
UPN24	63	Primary endpoint	Somatic	CUX1	S/C	NM_001202543.1.c.3401_3402delCC	NP_001189472.1.p.Ser1134Cys	TP	TP	31.51	344	24.4	582						
UPN24	63	Primary endpoint	Somatic	ASXL1	CQ/X	NM_015338.5.c.2279_2283delAGGCC	NP_056153.2.p.Gln760LeufsTer12	TP	TP	14.71	238	26.1	602						
UPN24	63	Baseline	Somatic	CUX1	S/C	NM_001202543.1.c.3401_3402delCC	NP_001189472.1.p.Ser1134Cys	TP	TP	30.53	269	28.1	563						
UPN24	63	Primary endpoint	Somatic	ATM,C11ORF65	G/R	NM_000051.3.c.6067G>A	NP_000042.3.p.Gly2023Arg (0.99)	TP	TP	43.3	225	37	185						
UPN24	63	Primary endpoint	Somatic	GRIN2A	D/G	NM_000833.3.c.197A>G	NP_000824.1.p.Asp66Gly	TP	TP	51.5	274	46	426						
UPN24	63	Baseline	Somatic	ATM,C11ORF65	G/R	NM_000051.3.c.6067G>A	NP_000042.3.p.Gly2023Arg (0.99)	TP	TP	22.2	54	47.2	248						
UPN25	72	Baseline	Somatic	TE72	E/V	NM_001127208.2.c.5636A>T	NP_001120680.1.p.Glu1879Val	TP	TP	64.98	219	41.5	648	0	0	67	284	38.6	298
UPN25	72	Baseline	Likely germline	AXIN2	A/V	NM_004655.3.c.2051C>T	NP_004646.3.p.Ala684Val (0.97)	TP	TP	47.1	238	42.5	368	38	252	50	215	41.9	217
UPN25	72	Baseline	Somatic	SF3B1	W/S	NM_012433.2.c.1973G>C	NP_036565.2.p.Trp658Ser (0.99)	TP	TP	40.5	331	38.6	339	0	0	43	142	26	237
UPN25	72	Primary endpoint	Somatic	SF3B1	W/S	NM_012433.2.c.1973G>C	NP_036565.2.p.Trp658Ser (0.99)	TP	TP	38.7	375	44	498						
UPN25	72	Primary endpoint	Somatic	TE72	E/V	NM_001127208.2.c.5636A>T	NP_001120680.1.p.Glu1879Val	TP	TP	52.5	446	50.1	636						
UPN25	72	Primary endpoint	Likely germline	AXIN2	A/V	NM_004655.3.c.2051C>T	NP_004646.3.p.Ala684Val (0.97)	TP	TP	53.3	311	55.8	353						
UPN26	36	Baseline	Likely germline	NPM1	D/-	NM_002501.6.c.495_497delCGA	NP_002511.1.p.Asp168del	TP	TP	14.54	110	28.4	850	36.7	98	36.7	98	46.1	219
UPN26	36	Primary endpoint	Somatic	ESR1	D/N	NM_001122742.1.c.652G>A	NP_001116214.1.p.Asp218Asn (0.98)	TP	TP	46.5	229	42.8	276						
UPN26	36	Baseline	Somatic	ESR1	D/N	NM_001122742.1.c.652G>A	NP_001116214.1.p.Asp218Asn (0.98)	TP	TP	55.9	170	56.3	206						
UPN27	47	Primary endpoint	Somatic	all NEG	0			TN	TN	0	0	0	0						
UPN27	47	Baseline	Somatic	FANCD2	N/S	NM_033084.3.c.2372A>G	NP_149075.2.p.Asn791Ser	TP	TP	31.91	257	22.7	458						
UPN27	47	Baseline	Somatic	ERBB4	P/R	NM_005235.2.c.1760C>G	NP_005226.1.p.Pro587Arg	TP	TP	23	452	25.8	469						
UPN28	79	Primary endpoint	Likely germline	AXIN2	A/V	NM_004655.3.c.2051C>T	NP_004646.3.p.Ala684Val (0.97)	TP	TP	43.8	385	50.3	485	48.5	231	49.3	148	41.4	158
UPN28	79	Baseline	Somatic	TE72	Q/*	NM_001127208.2.c.2218C>T	NP_001120680.1.p.Gln740Ter	TP	FP	2.36	297	11.4	316						
UPN28	79	Primary endpoint	Somatic	ASXL1	-JX	NM_015338.5.c.1934dupG	NP_056153.2.p.Gly646TrpfsTer12	TP	TP	7.14	285	5.8	312						
UPN28	79	Primary endpoint	Somatic	MED12	-/QQHQ	NM_005120.2.c.6348_6359dupCCAGC	NP_005111.2.p.His2116_Gln2119dup	TP	TP	34.2	121	30.1	196						
UPN28	79	Baseline	Somatic	ASXL1	-JX	NM_015338.5.c.1934dupG	NP_056153.2.p.Gly646TrpfsTer12	FN	x	5.68	95	0	0						
UPN29	76	Baseline	Somatic	TERT	R/W	NM_198253.2.c.1714A>T	NP_937983.2.p.Arg572Trp	TP	TP	36.3	335	30	777						
UPN29	76	Primary endpoint	Somatic	TERT	R/W	NM_198253.2.c.1714A>T	NP_937983.2.p.Arg572Trp	TP	TP	33.8	681	31.2	617						
UPN29	76	Primary endpoint	Somatic	GEN1	R/W	NM_182625.3.c.277C>T	NP_872431.3.p.Arg93Trp (0.92)	TP	TP	37.8	999	35.4	733						
UPN29	76	Primary endpoint	Somatic	SF3B1	K/E	NM_012433.2.c.2098A>G	NP_036565.2.p.Lys700Glu	TP	TP	39.9	349	41.4	256						
UPN29	76	Baseline	Somatic	SF3B1	K/E	NM_012433.2.c.2098A>G	NP_036565.2.p.Lys700Glu	TP	TP	37.4	190	42.5	208						
UPN30	47	Primary endpoint	Somatic	BRCA1	S/-	NM_007300.3.c.1846_1848delTCT	NP_009231.2.p.Ser616del (pathogenic)	TP	TP	30.9	554	27.5	728						
UPN30	47	Primary endpoint	Somatic	ERBB3	RSRS/-	NM_001982.3.c.3358_3369delAGGAG	NP_001973.2.p.Arg1120_Ser1123del	TP	TP	37.7	253	37.6	648						
UPN30	47	Baseline	Somatic	ERBB3	RSRS/-	NM_001982.3.c.3358_3369delAGGAG	NP_001973.2.p.Arg1120_Ser1123del	TP	TP	32.8	61	40.8	588						

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