

**RECURRENT MUTATIONS IN THE *U2AF1* SPLICING FACTOR
IN MYELODYSPLASTIC SYNDROMES**

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SUPPLEMENTARY NOTE

Clinical history of index case (UPN 266395). The patient was a 65 year old male of European ancestry who presented with anemia and thrombocytopenia (white blood cell count 5,900/mcl, hemoglobin 9.6 g/dl, platelet count 53,000/mcl). A bone marrow biopsy and aspirate showed 60% cellularity with trilineage hematopoiesis, without excess blasts or overt dysplasia. His marrow was felt to be abnormal, but not diagnostic of a myelodysplastic or myeloproliferative disorder. Cytogenetics showed a 46, XY karyotype. He was initially treated with recombinant erythropoietin without improvement, and two months later was referred for evaluation with the following blood counts: white blood cell count 40,300/mcl, hemoglobin 9.4 g/dl, platelet count 33,000/mcl. A bone marrow biopsy and aspirate were repeated (including specimens obtained for tissue banking after obtaining informed consent with specific language authorizing whole genome sequencing which was approved by the Washington University Human Research Protection Office), and showed a 100% cellular marrow, with trilineage dysplasia and 7% blasts, interpreted as MDS/RAEB-1. Cytogenetics again showed a normal male karyotype. His IPSS score was 1.5 (Intermediate-2). At that time, erythropoietin was continued and hydroxyurea was added (1500 mg daily).

One month later, the patient presented with increased fatigue, blurred vision, and dyspnea and was found to have a white blood cell count of 147,000/mcl, hemoglobin 8.9 g/dl, platelet count 36,000/mcl, with 27% circulating blasts. A bone marrow biopsy and aspirate were repeated (including specimens obtained for tissue banking after obtaining informed consent with specific language authorizing whole genome sequencing which was approved by the Washington University Human Research Protection Office). The bone marrow biopsy showed >90% cellularity, with 60% blasts with irregular nuclear contours (NBE positive, largely myeloperoxidase negative). Flow cytometry demonstrated coexpression of CD13 and CD33, with variable expression of CD64 and CD117, most consistent with AML M5b (AML with monocytic differentiation). He received emergency leukopheresis and began 5-azacytidine (160 mg subcutaneously, days 1-7 of 28), for a total of 6 cycles.

Seven months later, the patient developed mental status changes and was found to have leukemic relapse in the central nervous system. He received supportive care until his death 322 days after initial presentation, and 246 days after evolution to secondary AML (sAML).

The patient had a prior history of coronary artery disease, hyperlipidemia, and recurrent pneumonia. There was no significant family history of cancer.

RESULTS

Genome coverage. Individual reads were aligned to the reference genome (NCBI build 36) and only unique reads (deduplicated reads) were used to determine haploid coverage of the genome for the normal and sAML samples (38.23x and 39.12x, respectively, **Supplementary Table 1**). We obtained at least 32x haploid coverage for all autosomal chromosomes in the normal genome and ~half this coverage for chromosomes X and Y, as expected (**Supplementary Fig. 1**). At least 99.42% of the consensus coding DNA sequence (CCDS) was covered by at least 1 read in both genomes (**Supplementary Table 1**). We identified all the heterozygous and homozygous SNPs present in the normal and sAML genomes using the Affymetrix SNP 6.0 arrays and determined diploid coverage for each genome based on the number of these SNPs that were identified using the WGS data, as previously described¹. Using this approach, we achieved 98.91% and 99.31% diploid coverage of the normal and sAML genomes, respectively (**Supplementary Table 1**).

Detection and validation of somatic single nucleotide variants and insertions/deletions.

1281 potential somatic single nucleotide variants (SNVs) and 8801 insertions/deletions (indels) were identified in the sAML genome after filtering out low quality data and variants present in the matched normal genome and previously identified SNPs. These variant calls were prioritized into non-overlapping tiers for downstream validation, as previously described¹. 1233 SNV predictions were tested using the custom capture approach, of which 507 validated (**Supplementary Table 2**). This represents an overall validation rate of 507/1233 (41%) and 16/38 (42%) for tier 1 high confidence (HC). 16 indel predictions were validated (none in tier 1). 505 of the 507 SNVs (99.6 %) were present in the MDS genome, and 2 were new in the sAML genome (sAML-specific) (**Supplementary Table 2**).

Copy number alterations and loss of heterozygosity. Using WGS data, we identified 42 putative amplifications. None of these calls were supported by LOH analysis and there were no instances of copy number neutral LOH (**Supplementary Fig. 1**). 4 putative copy number alterations were detected using the Affymetrix SNP 6.0 array data (SNPa). Deep sequencing after solid phase capture of the called sites in the normal, MDS, and sAML samples did not support any of the copy number predictions. Therefore, no confirmed structural variants were detected in this genome.

Luciferase- β -galactosidase double-reporter assay. Transient co-expression of the pTN24 double-reporter plasmid and the wild-type or p.Ser34Phe mutant U2AF1 cDNA, with or without the Tra2 α splicing enhancer or hnRNPG splicing inhibitor, was performed in 293T cells in 3 independent experiments, with one representative example shown in **Fig. 3a**. Each experiment had 6 replicate data values for each combination of conditions. A significant increase in splicing (as detected by an increase in the luciferase/ β -galactosidase ratio) occurred with expression of the mutant U2AF1 compared to wild-type *U2AF1* when co-expressed with pTN24 in 3 experiments ($P \leq 0.004$). Co-expression of the Tra2 α splicing enhancer or hnRNPG splicing inhibitor resulted in increased and decreased splicing of the pTN24 reporter plasmid, as expected, that was independent of wild-type or mutant U2AF1 expression. Experiments were analyzed independently. Transient co-expression of a control plasmid (vector) or the mutant U2AF1 with or without a siRNA targeting the endogenous *U2AF1* was performed in 3 independent experiments, with one representative example shown in **Fig. 3b**. Each experiment had 6 replicate data values for each combination of conditions. A significant increase in splicing (as detected by an increase in the luciferase/ β -galactosidase ratio) occurred in all 3 experiments when comparing the vector and U2AF1 mutant cDNAs, independent of knockdown of *U2AF1* ($P \leq 0.04$).

Alternative splicing of *FMR1*. The *FMR1* gene, fragile X mental retardation 1, contains several alternative cryptic 3' splice acceptor sites in exon 15². We performed RT-PCR using primers that flanked the cryptic splice sites and measured the relative abundance of splice acceptor site usage, as previously described². We used RNA extracted from 7 unfractionated bone marrow samples harvested from MDS patients with a *U2AF1* mutation and 5 samples from MDS patients without a *U2AF1* mutation. Two independent experiments were performed using cDNA obtained from these samples and results are shown in **Supplementary Fig. 4a**. The splicing ratio is defined as the ratio of mRNAs resulting from the use of an alternative 3' cryptic splice acceptor site located within exon 15 (resulting in 124 or 85 base pair amplicons) compared to the normal splice acceptor site (160 base pair amplicon). There was an increased utilization of a cryptic 3' splice acceptor site in MDS samples harboring a *U2AF1* mutation compared to MDS samples without *U2AF1* mutations ($p < 0.03$ for both independent experiments, $p = 0.0001$ for pooled data).

Next, we validated this result by creating a *FMR1* minigene splicing reporter construct using the same genomic locus interrogated in the clinical samples. Each experiment was

repeated with four biologic replicates. The *FMR1* minigene was transiently transfected into 293T cells with a control plasmid (pcDNA3.1-YFP), wild-type U2AF1 cDNA, or mutant (p.Ser34Phe) U2AF1 cDNA in the presence of a control siRNA or siRNA targeting the endogenous *U2AF1*. RNA was harvested 48 hours later and a reverse transcriptase (RT) reaction was performed to create cDNA. Expression of the p.Ser34Phe mutant U2AF1 resulted in an increase in cryptic 3' splice acceptor usage compared to control or wild-type *U2AF1*, independent of endogenous *U2AF1* levels ($P < 0.04$) (**Supplementary Fig. 4b**).

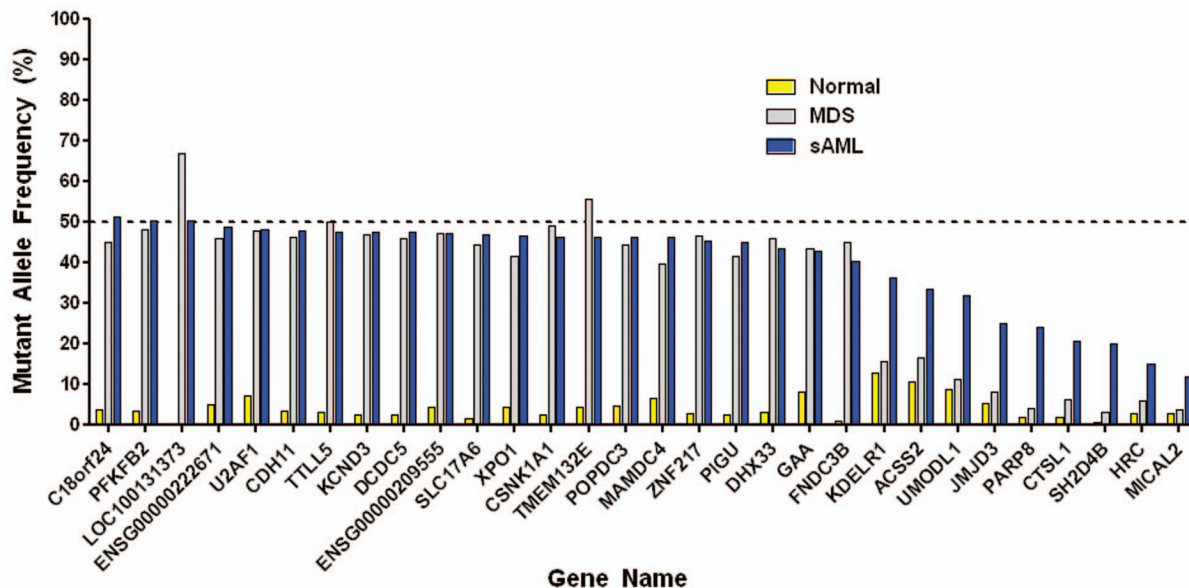
RT-PCR of U2AF1. Quantitative RT-PCR of *U2AF1* revealed no difference in the expression levels between mutant and wild-type MDS samples ($p = 0.73$, **Supplementary Fig. 2a, left panel**). A dominant RT-PCR product was identified for both wild-type and mutant U2AF1, consistent with isoform “a” expression (**Supplementary Fig. 2a, right panel**). Cloning and sequencing of RT-PCR products obtained from U2AF1 mutant MDS samples UPN 300813 and UPN 571656 revealed isoform “a” by Sanger sequencing (data not shown) and next-generation sequencing (**Fig. 1c**).

Gene expression profiling. We compared the expression level of the 30 genes containing tier 1 mutations in our index sAML sample (UPN 266395) with the expression in 43 *de novo* AML samples for which exon array data was available (T. Ley, unpublished data). 28 genes had at least 3 probesets on the array. For these 28 genes, at least one probeset was called present in 27 genes in the sAML case (**Supplementary Table 9**). All 27 genes were also expressed in >50% of the *de novo* AML cases and the one gene not expressed in the sAML case (*POPDC3*) had only 1/13 probesets expressed in ~50% of AML cases (**Supplementary Table 9**). 27 of the 30 mutated genes had probesets in the mutated exon. 14 of these were expressed in sAML (and in *de novo* AML) and 13 were not (vs. 11 absent in >50% of *de novo* AML cases). Overall, the expression pattern of the 28 genes with tier 1 mutations analyzed by microarray was highly similar in the sAML and *de novo* AML samples.

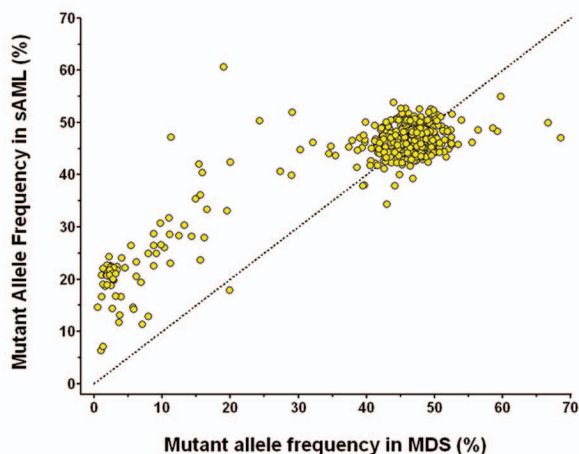
Affymetrix U133plus2 gene expression array data was available for 15 MDS and 4 control samples, as previously described³. RNA was extracted from CD34+ purified bone marrow cells for all samples. Array data was generated from the same batch analysis for the 19 samples and 6 of the 15 MDS samples had *U2AF1* mutations. First, we performed unsupervised hierarchical clustering using 19 arrays (4 control, 6 *U2AF1* mutant MDS samples, and 9 *U2AF1* wild-type MDS samples) and observed that clustering of samples was independent of disease or mutation status. When we restricted our clustering analysis to the 4 control and 6 *U2AF1* mutant samples, using 37,552 probesets that remained after removing probesets absent in all samples and using Ward's, UPGMA, or WPGMA clustering algorithms with Euclidean distance, we observed segregation of the mutant samples (**Supplementary Fig. 2b, left panel**). To identify the most significantly differentially expressed genes between control and *U2AF1* mutant MDS samples from the list of 37,552 probesets, we used the Significance Analysis of Microarray (SAM)⁴ and Gene Set Enrichment Analysis (GSEA)⁵ supervised algorithms. While we found no significant gene sets using GSEA, we did identify 50 probesets that were up-regulated and 351 probesets that were down-regulated in *U2AF1* mutant samples compared to controls with a false discovery rate (FDR) <0.005 (**Supplementary Table 6**). *U2AF1* mutant and control samples segregated by hierarchical clustering when we used 401 SAM defined probesets, as expected (**Supplementary Fig. 2b, right panel**). We analyzed up- and down-regulated genes to identify common pathways based on functional annotation categories (including gene ontology) using DAVID⁶. Nine annotation clusters were identified by DAVID using the 351 down-regulated probesets (300 with unique gene symbols) in *U2AF1* mutant samples that had an enrichment score >2. Three of the 9 clusters contained “mRNA splicing/processing” genes (enrichment score = 2.46) and two clusters with “RRM”(RNA recognition motif) genes (enrichment scores = 3-4.3) involved in splicing (**Supplementary Fig.**

2c, Supplementary Table 6). When genes that segregated control samples from *U2AF1* wild-type MDS samples were analyzed in DAVID, “splicing” and “RRM” categories were not enriched, suggesting that down-regulation of splicing and RRM genes is not common to all MDS samples, but instead is associated with *U2AF1* mutations. Collectively, these results suggest that a regulatory feedback loop may exist for mRNA splicing genes when *U2AF1* is mutated.

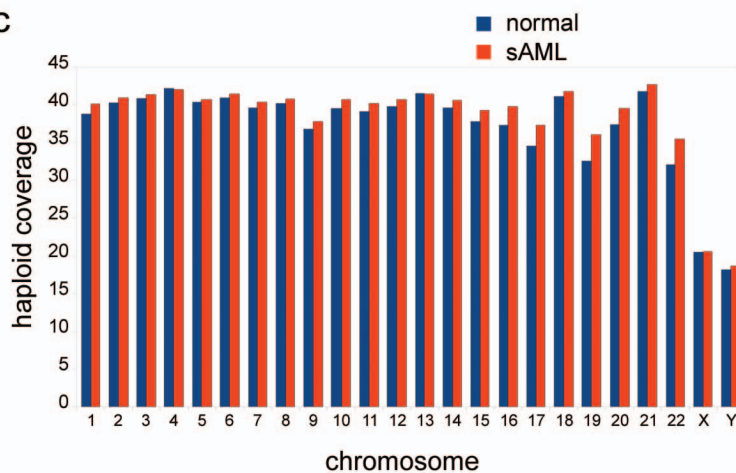
a



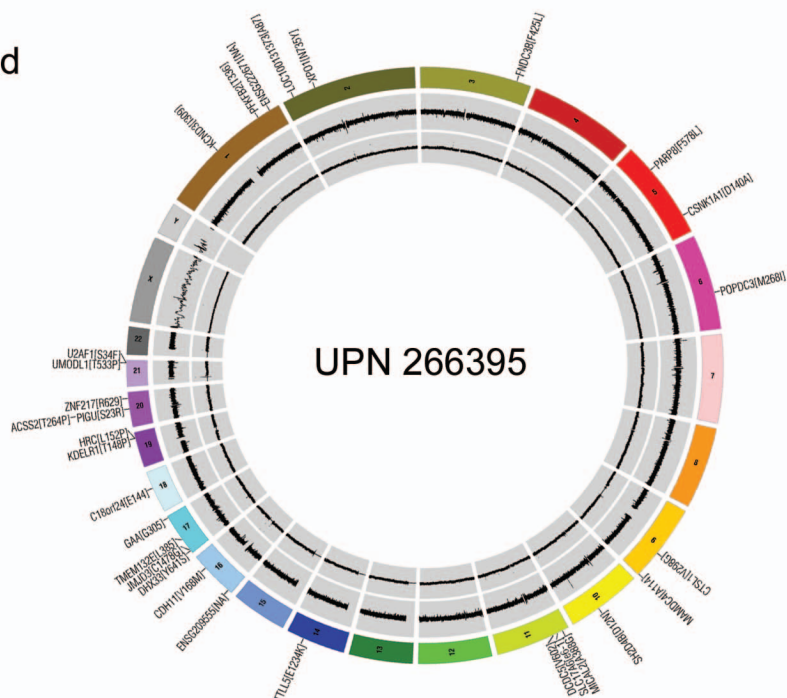
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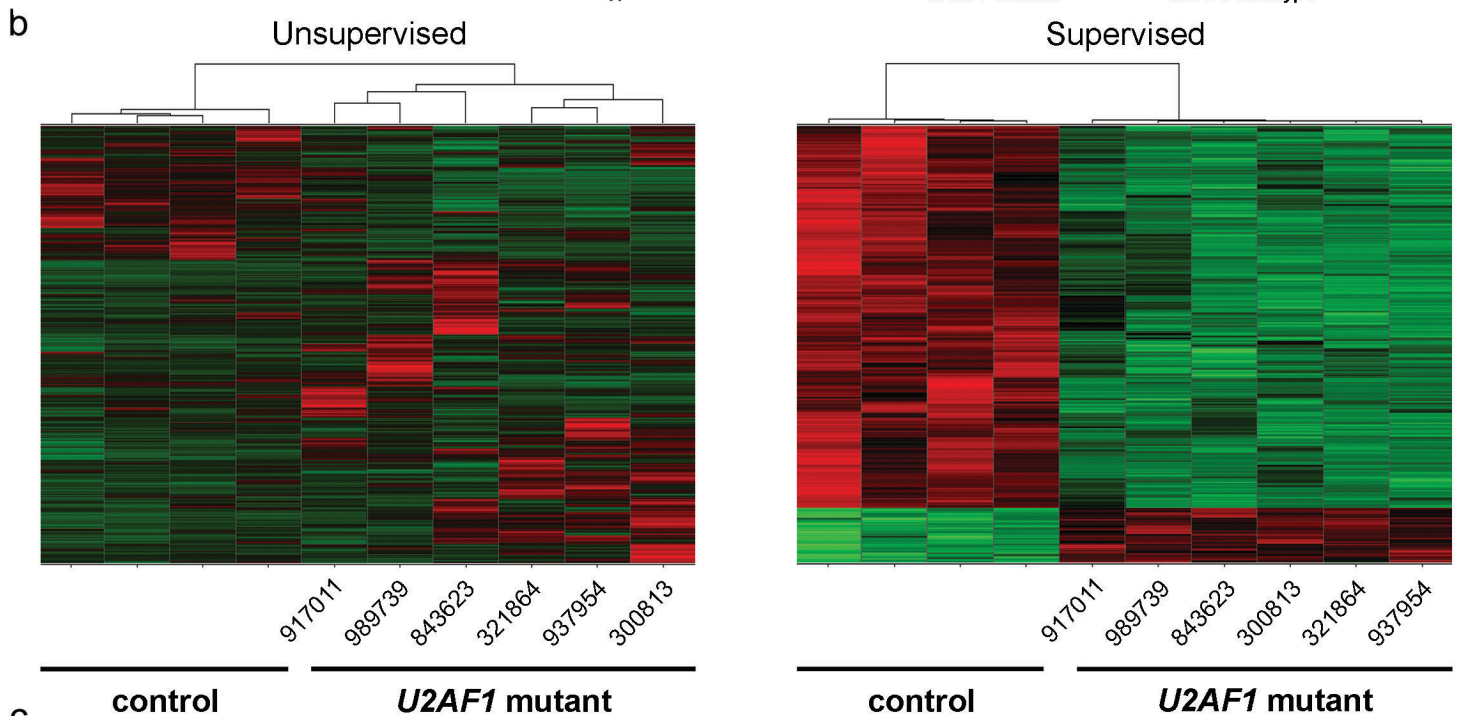
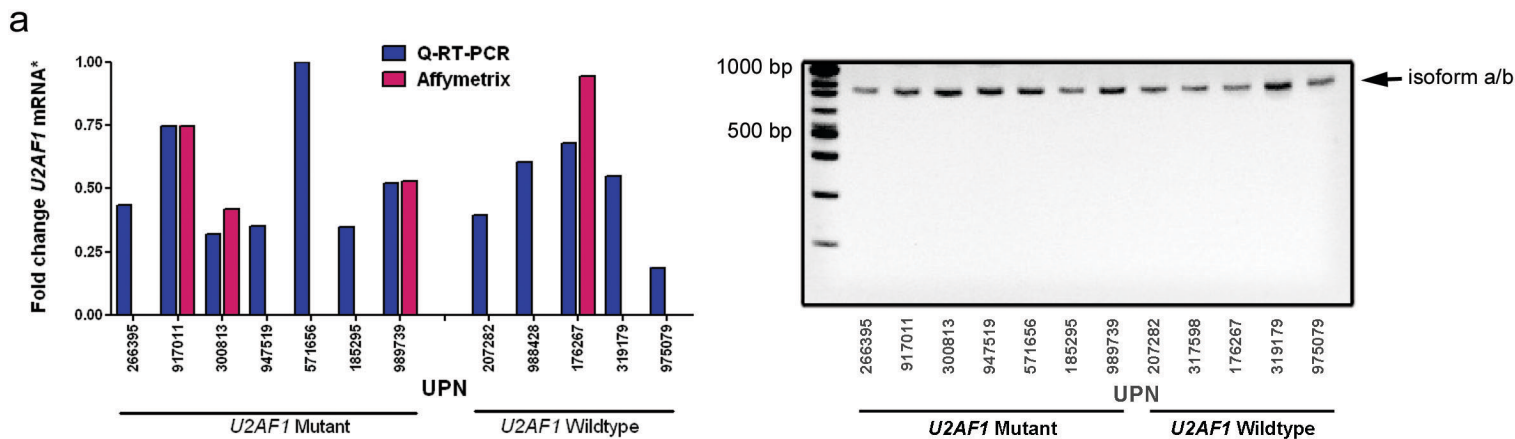
c



d



Supplementary Figure 1. Genomic architecture of UPN266395 index MDS patient. (a) Mutant allele frequencies of tier 1 SNVs from capture/deep sequencing of normal (skin), MDS, and sAML samples are shown. (b) Mutant allele frequencies of 507 validated somatic tier 1-3 SNVs from capture/deep sequencing are shown for the MDS vs. sAML samples. A single dominant clone was present in MDS and persisted at the sAML stage. Only 2 SNVs were acquired or enriched between MDS and sAML. (c) Sequence coverage from WGS was uniform across chromosomes and between normal (skin) and sAML samples. (d) Circos plot summarizing somatic changes identified by WGS in index case. Outer ring: validated tier 1 SNVs; middle ring: heterozygosity of sAML sample (no loss of heterozygosity detected); inner ring: copy number of sAML sample (no copy number alterations detected). SNV, single nucleotide variant. UPN, unique patient number.

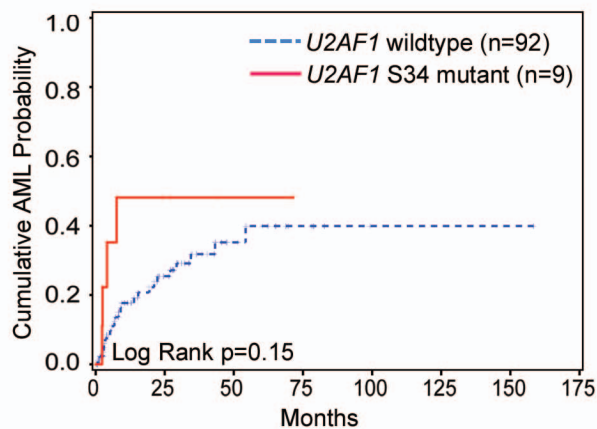
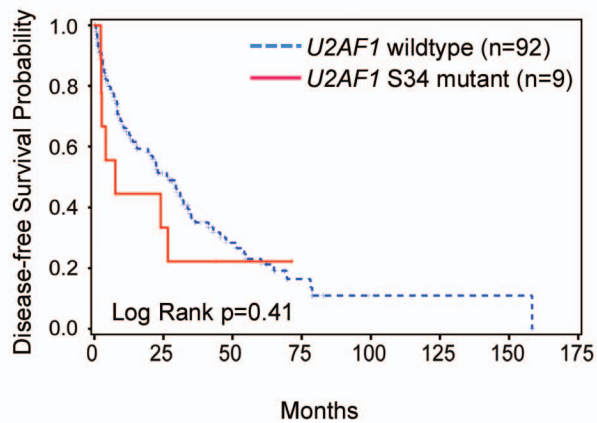
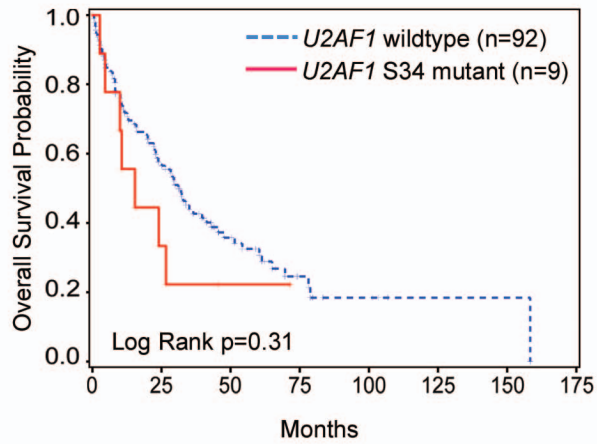


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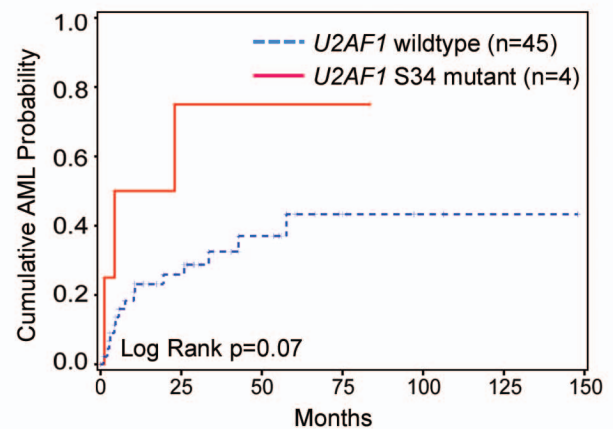
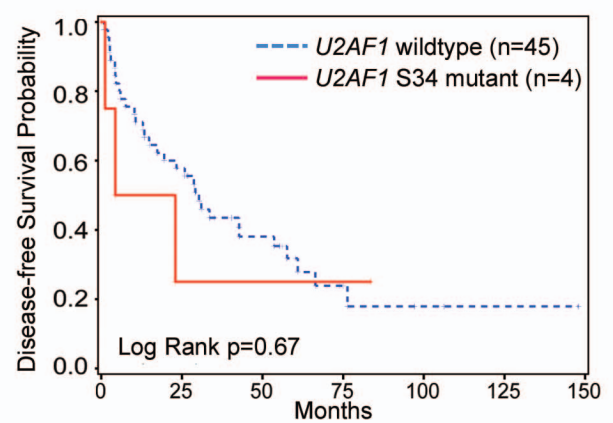
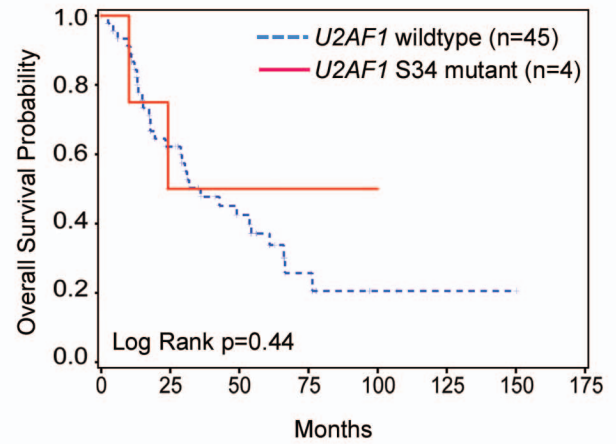
Annotation Cluster 1		Enrichment Score: 7.715			
Category	Term	# genes	% total genes	Fold Enrichment	Benjamini p-value
GOTERM_CC_FAT	GO:0031974:membrane-enclosed lumen	67	22.33	2.09	8.89E-07
GOTERM_CC_FAT	GO:0070013:intracellular organelle lumen	64	21.33	2.08	1.41E-06
GOTERM_CC_FAT	GO:0043233:organelle lumen	64	21.33	2.03	2.29E-06
GOTERM_CC_FAT	GO:0031981:nuclear lumen	52	17.33	2.07	3.73E-05
Annotation Cluster 2		Enrichment Score: 4.311			
Category	Term	# genes	% total genes	Fold Enrichment	Benjamini p-value
SMART	SM00360:RRM	14	4.67	4.18	3.33E-03
INTERPRO	IPR000504:RNA recognition motif, RNP-1	14	4.67	3.92	3.70E-02
INTERPRO	IPR012677:Nucleotide-binding, alpha-beta plait	14	4.67	3.88	2.05E-02

Supplementary Figure 2. U2AF1 quantitative RT-PCR and global expression profiling. (a, left panel) qRT-PCR of U2AF1 (exons 2-3 of isoform A) mRNA was performed using RNA extracted from unpurified bone marrow cells from patients with (n=7) and without (n=5) U2AF1 mutations. Affymetrix U133plus2 array data were available for 4 of the patients and are plotted for comparison. There was no significant difference in the expression of U2AF1 in mutant vs. wild-type MDS patient samples (p=0.73). (a, right panel) RT-PCR was performed using RNA extracted from unpurified bone marrow cells from patients with (n=7) and without (n=5) U2AF1 mutations. Primers were designed to detect all 3 isoforms of U2AF1. Full-length isoform a was detected in all samples, independent of mutation status. (b, left panel) Unsupervised hierarchical clustering results using Ward's method and 37,552 probesets from Affymetrix U133plus2 arrays (probesets were excluded only if absent in all samples being compared). RNA was obtained from CD34+ purified bone marrow cells from 4 normal healthy controls and 6 MDS samples with a U2AF1 mutation. (b, right panel) Supervised hierarchical clustering using Ward's method and 401 probesets identified by Significance Analysis of Microarrays (SAM), comparing control and U2AF1 mutant MDS samples with a false discovery rate (FDR) < 0.005. (c) Nine gene ontology annotation clusters were identified by DAVID using 351 probesets (300 gene symbols) that were down-regulated in U2AF1 mutant compared to control samples (identified by SAM) and had an enrichment score > 2 in DAVID. The top 2 clusters are shown. RNA recognition motif (RRM) gene categories were down-regulated in the U2AF1 samples compared to control samples. UPN, unique patient number. * relative to GAPDH mRNA expression.

No Transplant



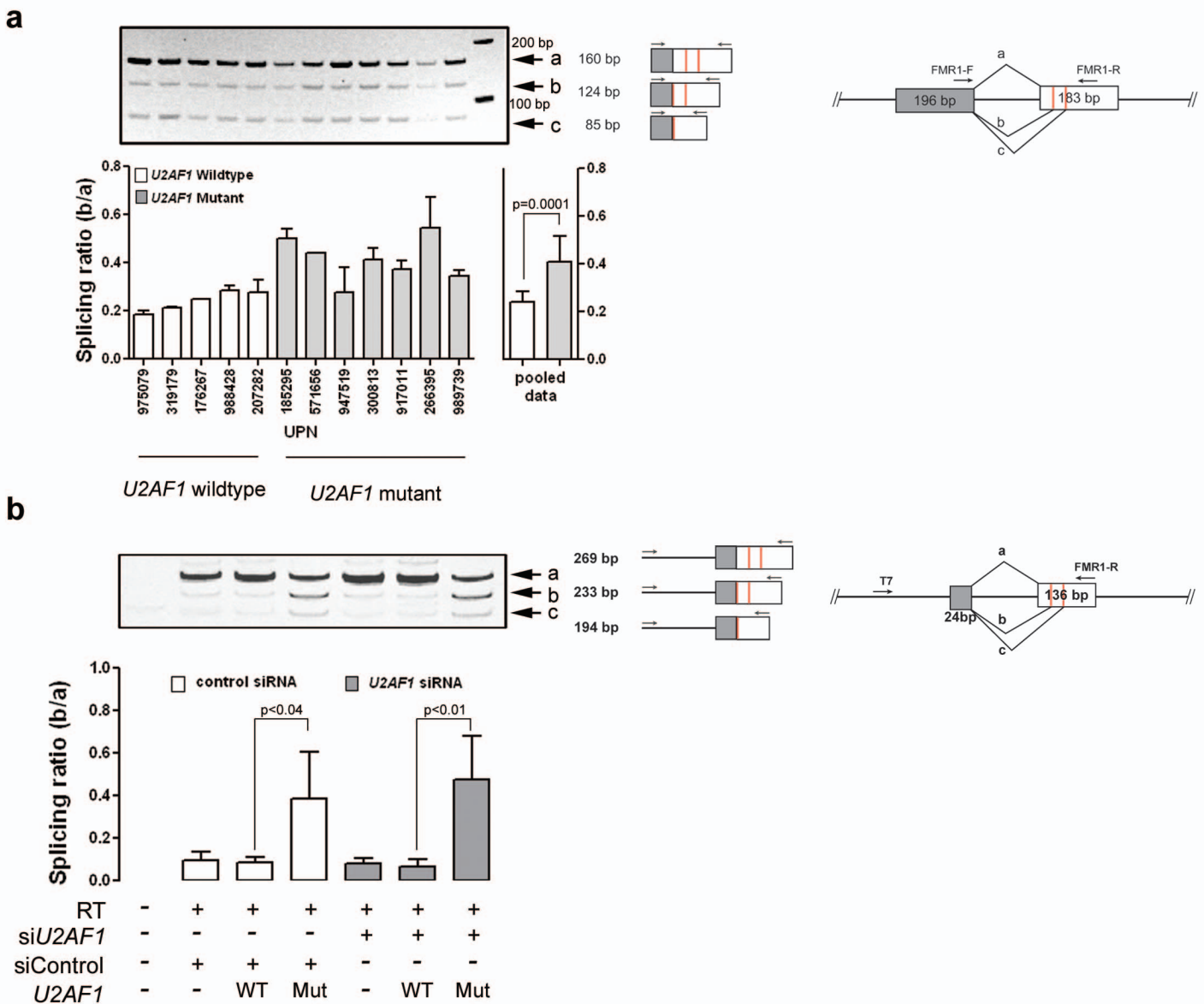
Transplant



Supplementary Figure 3. Survival analysis in transplanted vs. non-transplanted patients.

(left column) Overall, event-free survival, and probability of progression to sAML were not significantly different in U2AF1 wildtype vs. mutant MDS patients who *did not* undergo stem cell transplantation.

(right column) Overall, event-free survival, and probability of progression to sAML were not significantly different in U2AF1 wildtype vs. mutant MDS patients who *did* undergo stem cell transplantation.



Supplementary Figure 4. U2AF1 S34F mutation induces splicing alterations in FMR1. (a) The relative ratios of alternatively spliced products was measured following RT-PCR of RNA extracted from unpurified bone marrow cells from patients with (n=7) and without (n=5) U2AF1 mutations. A cartoon of FMR1 exons 14 and 15 that are interrogated by the assay and the possible splice products are indicated (labeled a, b, or c). A representative PCR gel image is shown and the ratio of the middle band (amplicon b, utilizing the 3' cryptic splice acceptor site) relative to the fully spliced upper band (amplicon a) is shown for each condition. The ratio of splice products was measured by densitometry and plotted in the bar graph with the p-value listed. Patient samples with U2AF1 mutations had increased usage of a cryptic 3' splice acceptor site in exon 15 of FMR1 relative to nonmutant patient samples (left panel, error bars represent two independent experiments) (wildtype vs. mutant splicing ratio, $p < 0.03$ for both independent experiments in the left panel, $p = 0.0001$ for pooled data in the right panel). (b) The FMR1 minigene (generated using primers shown in panel a) was transiently transfected into 293T cells with a control plasmid (pcDNA3.1-YFP), wild-type U2AF1 cDNA, or mutant (S34F) U2AF1 cDNA in the presence of a control siRNA or siRNA targeting endogenous U2AF1. RNA was harvested 48 hours later and a reverse transcriptase reaction was performed to create cDNA. PCR using the indicated primers resulted in a fully spliced 269 base pair amplicon or smaller amplicons (233 or 194 basepairs) that skip the first splice acceptor site. A representative PCR gel image is shown and the ratio of the middle band (cryptic 3' splice acceptor site = amplicon b) relative to the fully spliced upper band (amplicon a) is shown for each condition. Expression of S34F mutant U2AF1 resulted in an increase in cryptic 3' splice acceptor usage compared to control or wild-type U2AF1, independent of endogenous U2AF1 levels (n=4, $P < 0.04$).

Supplementary Table 1. Sequence metrics.

Parameter	Normal Genome	MDS Tumor Genome	sAML Tumor Genome
Whole Genome Sequence			
lanes	8	ND	12
sequence production (Gbp)	134.91	ND	132.24
aligned (Gbp)	122.80	ND	124.37
deduplicated (Gbp)	114.50	ND	116.32
haploid coverage	38.23	ND	39.12
bases with $\geq 1x$ coverage (%)	92.97	ND	93.00
bases with $>10x$ coverage (%)	91.87	ND	92.49
bases with $>20x$ coverage (%)	86.23	ND	89.12
CCDS bases with $\geq 1x$ coverage (%)	99.42	ND	99.67
CCDS bases with $\geq 10x$ coverage (%)	92.24	ND	97.21
CCDS bases with $\geq 20x$ coverage (%)	73.61	ND	88.52
Het SNPs (Affy)	226,603	ND	224,808
Hom SNPs (Affy)	166,483	ND	167,609
Diploid coverage Het SNPs (%)	98.91	ND	99.31
Diploid coverage Hom SNPs (%)	99.55	ND	99.17
Capture Validation			
target bases with $\geq 1x$ coverage (%)	93.06	93.78	94.86
target bases with $>10x$ coverage (%)	87.16	88.53	91.34
target bases with $>20x$ coverage (%)	84.22	86.15	89.84

CCDS, consensus coding DNA sequence; ND, not done.

Het SNPs, single nucleotide polymorphic sites genotyped as heterozygous using Affymetric 6.0 SNP array and both wildtype and variant alleles detected in whole genome sequence data

Hom SNPs, single nucleotide polymorphic sites genotyped as homozygous using Affymetric 6.0 SNP array and only variant allele detected in whole genome sequence data

Supplementary Table 2. Validated Tier 1-3 SNVs in the MDS and sAML genomes.

Chr	Start	Ref Allele	Var Allele	Gene Name	Tier	Mutation	Ref Reads in Normal	Var Reads in Normal	Var Freq in Normal	Ref Reads in MDS	Var Reads in MDS	Var Freq in MDS	Copy number adjusted Var Freq in MDS	Var Type MDS	Somatic P-value	Ref Reads in AML	Var Reads in AML	Var Freq in AML	Copy number adjusted Var Freq in AML	Var Type AML	Somatic P-value	Mutation Cluster	
1	19,065,087	G	A	TAS1R2	3	5_prime_flanking_region	559	16	2.78	82	79	49.07	49.07	Somatic	0.000	241	236	49.48	49.48	Somatic	0.000	1	
1	19,575,544	G	A	CAPZB	3	intrinsic	240	14	5.51	90	70	43.75	43.75	Somatic	0.000	220	186	45.81	45.81	Somatic	0.000	1	
1	35,025,125	A	G	GJA4	2	5_prime_flanking_region	891	11	1.22	383	318	45.36	45.36	Somatic	0.000	437	345	44.12	44.12	Somatic	0.000	1	
1	43,382,798	G	A	FAM183A	3	5_prime_untranslated_region	1361	56	3.95	559	480	46.20	46.20	Somatic	0.000	406	341	45.65	45.65	Somatic	0.000	1	
1	53,979,456	C	T	GLIS1	3	5_prime_flanking_region	219	28	11.34	231	203	46.77	46.77	Somatic	0.000	201	180	47.24	47.24	Somatic	0.000	1	
1	54,441,310	C	T	MRPL37	2	intrinsic	691	34	4.69	274	259	48.59	48.59	Somatic	0.000	286	254	47.04	47.04	Somatic	0.000	1	
1	60,329,124	A	G	C1orf87	2	5_prime_flanking_region	664	25	3.63	679	571	45.68	45.68	Somatic	0.000	394	355	47.40	47.40	Somatic	0.000	1	
1	61,483,596	C	T	NFIA	3	intrinsic	134	10	6.94	127	120	48.58	48.58	Somatic	0.000	203	171	45.72	45.72	Somatic	0.000	1	
1	63,400,074	G	T	ENSG00000210785	2	3_prime_flanking_region	792	27	3.30	942	938	49.89	49.89	Somatic	0.000	470	422	47.31	47.31	Somatic	0.000	1	
1	66,393,703	C	T	PDE4B	3	intrinsic	638	27	4.06	591	451	43.28	43.28	Somatic	0.000	306	273	47.15	47.15	Somatic	0.000	1	
1	84,880,057	A	C	C1orf180	3	5_prime_flanking_region	366	15	3.94	930	61	6.16	6.16	Somatic	0.201	599	183	23.40	23.40	Somatic	0.000	0	
1	91,923,288	C	T	TGFR3	3	intrinsic	603	27	4.29	488	507	50.95	50.95	Somatic	0.000	329	321	49.38	49.38	Somatic	0.000	1	
1	97,037,795	T	C	PTBP2	3	intrinsic	269	8	2.89	568	529	48.22	48.22	Somatic	0.000	296	259	46.67	46.67	Somatic	0.000	1	
1	106,376,327	C	G	LOC126987	3	3_prime_flanking_region	242	9	3.59	279	280	50.09	50.09	Somatic	0.000	325	293	47.41	47.41	Somatic	0.000	1	
1	111,623,853	C	T	CHIA	2	5_prime_flanking_region	1328	27	1.99	897	735	45.04	45.04	Somatic	0.000	567	416	42.32	42.32	Somatic	0.000	1	
1	112,325,945	G	T	KCND3	3	silent	708	17	2.34	278	245	46.85	46.85	Somatic	0.000	298	268	47.35	47.35	Somatic	0.000	1	
1	146,213,819	A	T	LOC10132219	3	5_prime_flanking_region	801	40	4.76	206	173	45.65	45.65	Somatic	0.000	332	286	46.28	46.28	Somatic	0.000	1	
1	157,603,769	G	T	OR10J1	3	intrinsic	616	31	4.79	462	372	44.60	44.60	Somatic	0.000	278	243	46.64	46.64	Somatic	0.000	1	
1	163,243,072	C	T	ENSG00000210270	3	5_prime_flanking_region	595	18	2.94	654	556	45.95	45.95	Somatic	0.000	310	249	44.54	44.54	Somatic	0.000	1	
1	165,197,653	G	A	ILDR2	3	intrinsic	1699	61	3.47	475	437	47.92	47.92	Somatic	0.000	469	423	47.42	47.42	Somatic	0.000	1	
1	165,827,772	C	T	CREG1	3	5_prime_flanking_region	238	13	5.18	218	165	43.08	43.08	Somatic	0.000	311	254	44.96	44.96	Somatic	0.000	1	
1	174,944,913	C	T	PAPPA2	3	intrinsic	946	1	0.11	1713	47	2.67	2.67	Somatic	0.000	514	136	20.92	20.92	Somatic	0.000	2	
1	175,516,166	C	A	FAM5B	3	splice_region	679	27	3.82	680	565	45.38	45.38	Somatic	0.000	329	311	48.59	48.59	Somatic	0.000	1	
1	205,309,412	C	G	PFKFB2	1	silent	593	21	3.42	447	414	48.08	48.08	Somatic	0.000	331	332	50.08	50.08	Somatic	0.000	1	
1	206,921,075	G	A	ENSG00000217293	3	3_prime_flanking_region	396	11	2.70	275	256	48.21	48.21	Somatic	0.000	389	293	42.96	42.96	Somatic	0.000	1	
1	215,059,819	C	T	ESRRG	3	intrinsic	321	8	2.43	306	271	46.97	46.97	Somatic	0.000	417	311	42.72	42.72	Somatic	0.000	1	
1	215,500,402	A	T	ENSG00000210753	2	3_prime_flanking_region	830	23	2.70	933	745	44.40	44.40	Somatic	0.000	428	371	46.43	46.43	Somatic	0.000	1	
1	225,042,030	C	T	ITPKB	3	5_prime_flanking_region	422	19	4.31	197	181	47.88	47.88	Somatic	0.000	215	193	47.30	47.30	Somatic	0.000	1	
1	229,096,537	T	C	ENSG00000222671	1	rna	406	20	4.69	172	146	45.91	45.91	Somatic	0.000	231	219	48.67	48.67	Somatic	0.000	1	
1	232,624,711	A	C	TARBP1	3	intrinsic	444	21	4.52	964	122	11.23	11.23	Somatic	0.048	287	257	47.24	47.24	Somatic	0.000	1	
1	242,682,289	T	G	ADSS	2	5_prime_flanking_region	110	0	0.00	372	7	1.85	1.85	Somatic	0.165	460	133	22.43	22.43	Somatic	0.000	2	
1	244,630,678	C	T	SMYD3	2	intrinsic	292	10	3.31	299	198	39.84	39.84	Somatic	0.000	302	303	50.08	50.08	Somatic	0.000	1	
1	246,007,658	G	A	OR1C1	3	5_prime_flanking_region	434	8	1.81	579	510	46.83	46.83	Somatic	0.000	369	286	43.66	43.66	Somatic	0.000	1	
2	863,187	C	T	LOC391343	3	3_prime_flanking_region	379	21	5.25	106	46	30.26	30.26	Somatic	0.000	310	252	44.84	44.84	Somatic	0.000	0	
2	12,407,391	G	A		0	3	0	1171	40	3.30	634	556	46.72	46.72	Somatic	0.000	403	394	49.44	49.44	Somatic	0.000	1
2	20,289,990	C	T	LOC100131373	1	silent	20	0	0.00	8	16	66.67	66.67	Somatic	0.000	92	92	50.00	50.00	Somatic	0.000	0	
2	35,703,943	G	A		0	3	0	682	24	3.40	511	518	50.34	50.34	Somatic	0.000	344	293	46.00	46.00	Somatic	0.000	1
2	35,720,616	T	C		0	3	0	501	24	4.57	608	526	46.38	46.38	Somatic	0.000	427	368	46.29	46.29	Somatic	0.000	1
2	39,902,719	T	C	THUMP2D	3	5_prime_flanking_region	340	7	2.02	538	477	47.00	47.00	Somatic	0.000	189	169	47.21	47.21	Somatic	0.000	1	
2	61,569,230	T	A	XPO1	1	missense	386	17	4.22	206	145	41.31	41.31	Somatic	0.000	373	322	46.33	46.33	Somatic	0.000	1	
2	69,027,275	A	G	GKN2	3	intrinsic	519	3	0.57	1093	51	4.46	4.46	Somatic	0.000	545	156	22.25	22.25	Somatic	0.000	2	
2	77,345,681	G	T	LRRTM4	3	intrinsic	540	25	4.42	1135	1029	47.55	47.55	Somatic	0.000	399	352	46.87	46.87	Somatic	0.000	1	
2	80,591,943	A	G	CTNNA2	3	intrinsic	548	18	3.18	547	453	45.30	45.30	Somatic	0.000	392	309	44.08	44.08	Somatic	0.000	1	
2	89,010,893	C	T	ENSG00000219044	3	5_prime_flanking_region	198	4	1.98	226	200	46.95	46.95	Somatic	0.000	274	228	45.42	45.42	Somatic	0.000	1	
2	98,221,020	T	A	VWA3B	3	intrinsic	736	52	6.60	442	407	47.94	47.94	Somatic	0.000	283	263	48.17	48.17	Somatic	0.000	1	
2	123,201,256	G	A		0	3	0	126	2	1.56	158	116	42.34	42.34	Somatic	0.000	194	167	46.26	46.26	Somatic	0.000	1
2	125,241,196	C	T	CNTNAP5	2	intrinsic	683	75	9.89	925	708	43.36	43.36	Somatic	0.000	303	289	48.82	48.82	Somatic	0.000	1	
2	126,115,254	G	T		0	3	0	537	11	2.01	690	544	44.08	44.08	Somatic	0.000	265	232	46.68	46.68	Somatic	0.000	1
2	129,312,063	T	C		0	3	0	222	3	1.33	165	124	42.91	42.91	Somatic	0.000	227	119	34.39	34.39	Somatic	0.000	0
2	132,910,452	A	C	GPR39	3	intrinsic	335	1	0.30	398	383	49.04	49.04	Somatic	0.000	361	266	42.42	42.42	Somatic	0.000	1	
2	138,312,675	C	T		0	3	0	917	14	1.50	557	438	44.02	44.02	Somatic	0.000	444	360	44.78	44.78	Somatic	0.000	1
2	148,169,122	C	G		0	3	0	776	39	4.79	384	367	48.87	48.87	Somatic	0.000	371	365	49.59	49.59	Somatic	0.000	1
2	148,826,888	C	G	MBD5	3	intrinsic	403	31	7.14	332	320	49.08	49.08	Somatic	0.000	233	239	50.64	50.64	Somatic	0.000	1	
2	151,024,156	A	T	RND3	3	3_prime_flanking_region	517	10	1.90	500	442	46.92	46.92	Somatic	0.000	252	254	50.20	50.20	Somatic	0.000	1	
2	163,476,519	G	T		0	3	0	609	14	2.25	474	360	43.17	43.17	Somatic	0.000	412	403	49.45	49.45	Somatic	0.000	1
2	164,335,975	T	C	FIGN	3	5_prime_flanking_region	598	22	3.55	555	470	45.85	45.85	Somatic	0.000	395	325	45.14	45.14	Somatic	0.000	1	
2	165,485,434	G	A	SLC38A11	3	intrinsic	876	27	2.99	965	915	48.67	48.67	Somatic	0.000	369	361	49.45	49.45	Somatic	0.000	1	
2	173,997,180	T	A	LOC100130171	3	3_prime_flanking_region	28	0	0.00	20	29	59.18	59.18	Somatic	0.000	78	73	48.34	48.34	Somatic	0.000	0	
2	176,980,601	G	A	ENSG00000221304	2	5_prime_flanking_region	537	23	4.11	416	316	43.17	43.17	Somatic	0.000	251	219	46.60	46.60	Somatic	0.000	1	
2	177,423,844	A	C	LOC100131991	3	5_prime_flanking_region	663	6	0.90	1615	18	1.10	1.10	Somatic	0.048	748	150	16.70	16.70	Somatic	0.000	1	
2	184,200,811	G	A	LOC644877	3	5_prime_flanking_region	393	10	2.48	542	476	46.76	46.76	Somatic	0.000	309	247	44.42	44.42	Somatic	0.000	1	
2	193,919,617	T	C		0	3	0	92	5	5.15	235	129	35.44	35.44	Somatic	0.000	228	177	43.70	43.70	Somatic	0.000	0

3	13,411,614	G	A	NUP210	3	intronic	193	6	3.02	65	61	48.41	48.41	Somatic	0.000	161	136	45.79	45.79	Somatic	0.000	1	
3	21,855,819	A	C	ENSG00000221384	3	3_prime_flanking_region	0	405	53	11.57	791	197	19.94	19.94	Somatic	0.000	303	223	42.40	42.40	Somatic	0.000	0
3	26,376,984	G	A		0	3	269	9	3.24	302	283	48.38	48.38	Somatic	0.000	163	132	44.75	44.75	Somatic	0.000	1	
3	26,998,497	G	A		0	3	692	37	5.08	983	698	41.52	41.52	Somatic	0.000	411	347	45.78	45.78	Somatic	0.000	1	
3	40,732,099	G	T	LOC729505	2	5_prime_flanking_region	683	26	3.67	626	616	49.60	49.60	Somatic	0.000	458	376	45.08	45.08	Somatic	0.000	1	
3	45,981,158	T	G	FYCO1	3	intronic	345	17	4.70	326	31	8.68	8.68	Somatic	0.324	377	136	26.51	26.51	Somatic	0.000	0	
3	47,603,402	G	T	SMARCC1	3	3_prime_untranslated_region	564	70	11.04	275	42	13.25	13.25	Somatic	0.533	404	176	30.34	30.34	Somatic	0.000	0	
3	53,603,765	T	G	CACNA1D	3	intronic	821	89	9.78	1229	297	19.46	19.46	Somatic	0.000	411	204	33.17	33.17	Somatic	0.000	0	
3	75,444,179	C	T	ENSG00000179799	3	3_prime_flanking_region	1238	2	0.16	1384	31	2.19	2.19	Somatic	0.000	598	163	21.42	21.42	Somatic	0.000	2	
3	75,472,731	A	C	LOC728135	3	3_prime_flanking_region	322	12	3.59	226	186	45.15	45.15	Somatic	0.000	187	164	46.72	46.72	Somatic	0.000	1	
3	76,099,526	G	A	ROBO2	3	intronic	277	5	1.77	476	225	32.10	32.10	Somatic	0.000	230	198	46.26	46.26	Somatic	0.000	0	
3	76,586,575	C	T	ROBO2	3	intronic	233	2	0.85	354	144	28.92	28.92	Somatic	0.000	293	195	39.96	39.96	Somatic	0.000	0	
3	80,602,658	G	A		0	3	126	3	2.33	171	147	46.23	46.23	Somatic	0.000	243	234	49.06	49.06	Somatic	0.000	1	
3	81,055,536	G	A		0	3	490	35	6.67	686	616	47.31	47.31	Somatic	0.000	429	366	46.04	46.04	Somatic	0.000	1	
3	85,219,286	T	G	CADM2	3	intronic	159	15	8.62	261	169	39.30	39.30	Somatic	0.000	286	236	45.21	45.21	Somatic	0.000	1	
3	86,451,526	T	C	ENSG00000222934	3	5_prime_flanking_region	25	3	10.71	39	55	58.51	58.51	Somatic	0.000	51	49	49.00	49.00	Somatic	0.000	0	
3	99,901,474	T	G	ST3GAL6	2	5_prime_flanking_region	36	5	12.20	81	19	19.00	19.00	Somatic	0.285	55	85	60.71	60.71	Somatic	0.001	0	
3	100,667,116	C	T		0	3	819	2	0.24	2861	55	1.89	1.89	Somatic	0.000	614	181	22.77	22.77	Somatic	0.000	2	
3	111,192,335	T	C		0	3	338	7	2.03	438	411	48.41	48.41	Somatic	0.000	302	247	44.99	44.99	Somatic	0.000	1	
3	111,431,333	C	T		0	3	153	1	0.65	287	213	42.60	42.60	Somatic	0.000	258	181	41.23	41.23	Somatic	0.000	1	
3	115,134,962	G	T	GRAMD1C	3	intronic	141	2	1.40	252	200	44.25	44.25	Somatic	0.000	236	188	44.34	44.34	Somatic	0.000	1	
3	115,150,252	C	G	ZDHHC23	2	intronic	296	7	2.31	480	393	45.02	45.02	Somatic	0.000	271	236	46.55	46.55	Somatic	0.000	1	
3	133,430,465	T	G	LOC729677	3	3_prime_flanking_region	1116	22	1.93	1852	136	6.84	6.84	Somatic	0.000	614	448	19.42	19.42	Somatic	0.000	0	
3	133,441,990	C	A	LOC729677	3	3_prime_flanking_region	542	37	6.39	721	670	48.17	48.17	Somatic	0.000	450	412	47.80	47.80	Somatic	0.000	1	
3	134,386,054	A	T	TMEM108	3	intronic	134	8	5.63	147	128	46.55	46.55	Somatic	0.000	233	185	44.26	44.26	Somatic	0.000	1	
3	136,219,503	A	G	EPHB1	3	intronic	1277	59	4.42	1050	100	8.70	8.70	Somatic	0.004	342	100	22.62	22.62	Somatic	0.000	0	
3	136,463,072	C	T	EPHB1	3	3_prime_flanking_region	332	8	2.35	245	187	43.29	43.29	Somatic	0.000	177	151	46.04	46.04	Somatic	0.000	1	
3	158,727,296	T	A	C3orf55	3	5_prime_flanking_region	545	25	4.39	313	272	46.50	46.50	Somatic	0.000	256	205	44.47	44.47	Somatic	0.000	1	
3	163,509,409	G	C		0	3	815	7	2.17	549	459	45.54	45.54	Somatic	0.000	381	349	47.81	47.81	Somatic	0.000	1	
3	166,201,621	C	T	SI	3	intronic	39	3	3.26	152	122	44.53	44.53	Somatic	0.000	109	83	43.23	43.23	Somatic	0.000	1	
3	170,819,805	C	T	MDS1	3	intronic	687	20	2.83	683	638	48.30	48.30	Somatic	0.000	312	250	44.48	44.48	Somatic	0.000	1	
3	173,529,456	C	A	FNDC3B	1	missense	262	2	0.76	439	356	44.78	44.78	Somatic	0.000	219	146	40.00	40.00	Somatic	0.000	0	
3	177,936,424	A	T	ENSG00000208490	3	5_prime_flanking_region	614	30	4.66	602	426	41.44	41.44	Somatic	0.000	460	380	45.24	45.24	Somatic	0.000	1	
3	188,400,850	C	A	RTP1	3	3_prime_untranslated_region	341	5	1.45	405	383	48.60	48.60	Somatic	0.000	278	215	43.61	43.61	Somatic	0.000	1	
4	2,261,310	G	A	ZFYVE28	2	intronic	131	11	7.75	37	26	41.27	41.27	Somatic	0.000	283	249	46.80	46.80	Somatic	0.000	1	
4	7,365,576	T	A	SORCS2	3	intronic	1048	8	0.76	270	3	1.10	1.10	Somatic	0.401	446	117	20.78	20.78	Somatic	0.000	2	
4	7,395,182	A	T	SORCS2	3	intronic	148	3	1.99	113	97	46.19	46.19	Somatic	0.000	121	130	51.79	51.79	Somatic	0.000	1	
4	16,550,472	C	T	LDB2	3	5_prime_flanking_region	241	14	5.49	252	214	45.92	45.92	Somatic	0.000	229	213	48.19	48.19	Somatic	0.000	1	
4	18,139,490	C	T	ENSG00000209956	3	3_prime_flanking_region	392	25	6.00	687	531	43.60	43.60	Somatic	0.000	305	224	42.34	42.34	Somatic	0.000	1	
4	22,362,173	A	G	GBA3	3	intronic	1156	44	3.67	896	834	48.21	48.21	Somatic	0.000	371	366	49.66	49.66	Somatic	0.000	1	
4	29,185,860	G	A		0	3	304	11	3.49	536	498	48.16	48.16	Somatic	0.000	344	313	47.64	47.64	Somatic	0.000	1	
4	29,629,786	G	A		0	3	397	3	0.75	378	266	41.30	41.30	Somatic	0.000	273	199	42.16	42.16	Somatic	0.000	1	
4	31,623,175	A	G	LOC100133713	3	5_prime_flanking_region	246	15	5.75	264	253	48.94	48.94	Somatic	0.000	316	315	49.92	49.92	Somatic	0.000	1	
4	32,768,631	G	A		0	3	398	15	3.63	653	505	43.61	43.61	Somatic	0.000	307	268	46.61	46.61	Somatic	0.000	1	
4	36,443,632	T	A		0	3	374	16	4.10	365	341	48.30	48.30	Somatic	0.000	382	293	43.41	43.41	Somatic	0.000	1	
4	43,458,657	C	T		0	3	104	3	2.80	138	88	38.94	38.94	Somatic	0.000	209	186	47.09	47.09	Somatic	0.000	1	
4	44,155,349	C	T	KCTD8	3	5_prime_flanking_region	1221	18	1.45	614	514	45.57	45.57	Somatic	0.000	437	365	45.51	45.51	Somatic	0.000	1	
4	63,541,472	C	A		0	3	268	1	0.37	410	315	43.45	43.45	Somatic	0.000	287	221	43.50	43.50	Somatic	0.000	1	
4	77,073,409	T	C	NAAA	3	intronic	79	2	2.47	57	55	49.11	49.11	Somatic	0.000	188	147	43.88	43.88	Somatic	0.000	1	
4	86,518,791	A	C		0	2	805	62	7.15	574	503	46.70	46.70	Somatic	0.000	410	346	45.77	45.77	Somatic	0.000	1	
4	110,783,030	T	G	CCDC109B	2	intronic	784	63	7.44	1497	250	14.31	14.31	Somatic	0.000	512	201	28.19	28.19	Somatic	0.000	0	
4	112,604,557	G	A		0	3	193	7	3.50	418	416	49.88	49.88	Somatic	0.000	263	226	46.22	46.22	Somatic	0.000	1	
4	118,188,338	A	G	TRAM1L1	3	3_prime_flanking_region	551	32	5.49	392	397	50.32	50.32	Somatic	0.000	297	263	46.96	46.96	Somatic	0.000	1	
4	125,746,802	G	A		0	3	372	9	2.36	677	499	42.43	42.43	Somatic	0.000	295	220	42.72	42.72	Somatic	0.000	1	
4	126,682,546	A	T	FAT4	3	3_prime_flanking_region	561	15	2.60	506	477	48.52	48.52	Somatic	0.000	424	431	50.41	50.41	Somatic	0.000	1	
4	128,277,343	C	T		0	3	637	24	3.63	843	699	45.33	45.33	Somatic	0.000	360	347	49.08	49.08	Somatic	0.000	1	
4	132,318,397	C	T		0	3	273	10	3.53	410	324	44.14	44.14	Somatic	0.000	336	205	37.89	37.89	Somatic	0.000	0	
4	136,046,582	T	A	LOC646272	3	5_prime_flanking_region	585	12	2.01	456	243	34.76	34.76	Somatic	0.000	281	234	45.44	45.44	Somatic	0.000	0	
4	136,492,542	G	A	ENSG00000207188	3	5_prime_flanking_region	62	2	3.13	182	130	41.67	41.67	Somatic	0.000	183	145	44.21	44.21	Somatic	0.000	1	
4	137,716,185	C	T		0	2	400	14	3.38	269	229	45.98	45.98	Somatic	0.000	293	273	48.23	48.23	Somatic	0.000	1	
4	139,423,585	T	A	SLC7A11	2	5_prime_flanking_region	46	1	2.13	57	61	51.69	51.69	Somatic	0.000	39	30	43.48	43.48	Somatic	0.000	1	
4	145,369,376	C	T		0	3	473	21	4.25	375	302	44.61	44.61	Somatic	0.000	252	213	45.81	45.81	Somatic	0.000	1	
4	146,313,010	T	C	OTUD4	3	intronic	335	15	4.29	387	429	52.57	52.57	Somatic	0.000	306	326	51.58	51.58	Somatic	0.000	1	
4	155,766,017	C	T	FGA	3	5_prime_flanking_region	693	13	1.84	661	581	46.78	46.78	Somatic	0.000	433	395	47.71	47.71	Somatic	0.000	1	
4	161,787,610	C	T		0	3	830	36	4.16	742	5												

5	51,503,256	G	A		0	2	0	571	25	4.19	523	486	48.17	48.17	Somatic	0.000	358	330	47.97	47.97	Somatic	0.000	1
5	68,314,881	T	A	ENSG00000222762	3	5_prime_flanking_region		328	16	4.65	679	591	46.54	46.54	Somatic	0.000	300	285	48.72	48.72	Somatic	0.000	1
5	71,888,102	G	A	ZNF366	2	5_prime_flanking_region		549	16	2.83	444	345	43.73	43.73	Somatic	0.000	301	254	45.77	45.77	Somatic	0.000	1
5	89,673,080	A	T		0	3	0	302	18	5.63	341	327	48.95	48.95	Somatic	0.000	258	234	47.56	47.56	Somatic	0.000	1
5	92,816,744	G	A		0	3	0	499	17	3.29	423	349	45.21	45.21	Somatic	0.000	382	338	46.94	46.94	Somatic	0.000	1
5	96,065,480	A	G	CAST	2	intronic		278	5	1.77	255	251	49.60	49.60	Somatic	0.000	320	325	50.39	50.39	Somatic	0.000	1
5	100,118,060	T	C	ENSG00000207269	3	3_prime_flanking_region		504	17	3.26	343	326	48.73	48.73	Somatic	0.000	346	311	47.34	47.34	Somatic	0.000	1
5	113,227,599	C	T		0	3	0	624	28	4.29	663	547	45.21	45.21	Somatic	0.000	322	306	48.73	48.73	Somatic	0.000	1
5	117,884,100	G	A		0	3	0	782	0	0.00	1820	51	2.73	2.73	Somatic	0.000	714	194	21.37	21.37	Somatic	0.000	2
5	123,015,301	C	T	ENSG00000208978	3	5_prime_flanking_region		815	26	3.09	821	687	45.56	45.56	Somatic	0.000	406	335	45.21	45.21	Somatic	0.000	1
5	123,877,689	C	T		0	2	0	313	12	3.69	172	122	41.50	41.50	Somatic	0.000	280	234	45.53	45.53	Somatic	0.000	1
5	132,897,276	C	T	FSTL4	3	intronic		595	16	2.62	922	804	46.58	46.58	Somatic	0.000	433	360	45.40	45.40	Somatic	0.000	1
5	140,439,825	T	C	PCDHB2	3	5_prime_flanking_region		349	13	3.59	250	186	42.66	42.66	Somatic	0.000	384	359	48.32	48.32	Somatic	0.000	1
5	140,469,201	G	T	PCDHB5	3	5_prime_flanking_region		1075	33	2.98	195	146	42.82	42.82	Somatic	0.000	383	345	47.39	47.39	Somatic	0.000	1
5	147,696,652	G	A	SPINK9	3	intronic		530	18	3.28	665	573	46.28	46.28	Somatic	0.000	342	353	50.79	50.79	Somatic	0.000	1
5	148,880,083	T	G	CSNK1A1	1	missense		531	13	2.39	473	450	48.75	48.75	Somatic	0.000	379	326	46.24	46.24	Somatic	0.000	1
5	149,525,443	T	C	PDGFRB	3	5_prime_flanking_region		262	4	1.50	172	7	3.91	3.91	Somatic	0.168	529	106	16.69	16.69	Somatic	0.000	0
5	160,513,086	G	A		0	3	0	327	17	4.94	238	183	43.47	43.47	Somatic	0.000	295	251	45.97	45.97	Somatic	0.000	1
5	162,193,041	C	A	ENSG00000208481	3	3_prime_flanking_region		452	5	1.09	433	333	43.47	43.47	Somatic	0.000	325	250	43.48	43.48	Somatic	0.000	1
5	164,071,958	G	A		0	3	0	576	18	3.03	581	494	45.95	45.95	Somatic	0.000	417	415	49.88	49.88	Somatic	0.000	1
5	165,820,268	C	A		0	3	0	790	37	4.47	768	743	49.17	49.17	Somatic	0.000	456	454	49.89	49.89	Somatic	0.000	1
5	166,002,175	C	A		0	2	0	561	17	2.94	586	460	43.98	43.98	Somatic	0.000	372	328	46.86	46.86	Somatic	0.000	1
6	900,039	T	C	LOC730397	3	5_prime_flanking_region		304	12	3.80	399	400	50.06	50.06	Somatic	0.000	347	276	44.30	44.30	Somatic	0.000	1
6	5,996,224	G	A	NRN1	3	5_prime_flanking_region		606	15	2.42	517	562	52.09	52.09	Somatic	0.000	360	294	44.95	44.95	Somatic	0.000	1
6	6,260,424	C	T	F13A1	3	intronic		483	12	2.42	404	344	45.99	45.99	Somatic	0.000	411	402	49.45	49.45	Somatic	0.000	1
6	9,584,137	C	T		0	2	0	1427	97	6.36	962	905	48.47	48.47	Somatic	0.000	503	470	48.30	48.30	Somatic	0.000	1
6	12,118,719	G	T	HIVEP1	2	5_prime_flanking_region		246	6	2.38	261	208	44.35	44.35	Somatic	0.000	307	287	48.32	48.32	Somatic	0.000	1
6	23,624,936	C	T		0	3	0	163	6	3.55	224	220	49.55	49.55	Somatic	0.000	260	216	45.38	45.38	Somatic	0.000	1
6	33,967,359	C	G	LOC100132252	3	3_prime_flanking_region		268	15	5.30	89	94	51.37	51.37	Somatic	0.000	336	269	44.46	44.46	Somatic	0.000	1
6	44,532,555	C	T	LOC100128935	2	5_prime_flanking_region		483	26	5.11	392	336	46.15	46.15	Somatic	0.000	224	227	50.33	50.33	Somatic	0.000	1
6	45,133,962	A	C	SUPT3H	3	intronic		492	21	4.09	783	649	45.32	45.32	Somatic	0.000	339	274	44.70	44.70	Somatic	0.000	1
6	51,440,235	T	C	ENSG00000212532	3	3_prime_flanking_region		396	14	3.41	349	277	44.25	44.25	Somatic	0.000	253	201	44.27	44.27	Somatic	0.000	1
6	54,067,267	T	G	C6orf142	3	intronic		287	19	6.21	253	215	45.94	45.94	Somatic	0.000	266	225	45.82	45.82	Somatic	0.000	1
6	77,277,763	C	T	LOC643281	3	3_prime_flanking_region		175	10	5.41	278	276	49.82	49.82	Somatic	0.000	290	230	44.23	44.23	Somatic	0.000	1
6	85,673,094	G	A		0	3	0	621	7	1.11	566	546	49.10	49.10	Somatic	0.000	226	210	48.17	48.17	Somatic	0.000	1
6	89,065,419	C	T	ENSG00000220267	3	5_prime_flanking_region		980	46	4.48	928	868	48.33	48.33	Somatic	0.000	448	432	49.09	49.09	Somatic	0.000	1
6	91,891,502	G	T		0	2	0	786	31	3.79	644	494	43.41	43.41	Somatic	0.000	367	338	47.94	47.94	Somatic	0.000	1
6	95,400,882	C	T		0	3	0	632	0	0.00	864	14	1.59	1.59	Somatic	0.000	499	115	18.73	18.73	Somatic	0.000	2
6	105,713,110	C	A	POPDC3	1	missense		174	8	4.40	175	138	44.09	44.09	Somatic	0.000	246	211	46.17	46.17	Somatic	0.000	1
6	113,767,508	C	T		0	3	0	821	5	0.61	745	615	45.22	45.22	Somatic	0.000	362	266	42.36	42.36	Somatic	0.000	1
6	125,900,869	G	A		0	3	0	400	3	0.74	341	254	42.69	42.69	Somatic	0.000	344	263	43.33	43.33	Somatic	0.000	1
6	133,329,889	T	C	ENSG00000213122	3	3_prime_flanking_region		244	8	3.17	447	370	45.29	45.29	Somatic	0.000	262	208	44.26	44.26	Somatic	0.000	1
6	134,376,650	C	G	SLC2A12	3	intronic		325	10	2.99	522	379	42.06	42.06	Somatic	0.000	264	250	48.64	48.64	Somatic	0.000	1
6	135,937,118	C	T	ENSG00000213112	3	3_prime_flanking_region		383	8	2.05	413	364	46.85	46.85	Somatic	0.000	346	292	45.77	45.77	Somatic	0.000	1
6	137,632,989	G	A		0	3	0	677	32	4.51	363	305	45.66	45.66	Somatic	0.000	220	246	52.79	52.79	Somatic	0.000	1
6	138,308,151	T	C	ENSG00000219463	3	5_prime_flanking_region		1695	93	5.20	1166	1159	49.85	49.85	Somatic	0.000	404	436	51.90	51.90	Somatic	0.000	1
6	146,176,900	G	T	FBXO30	2	intronic		415	8	1.89	492	391	44.28	44.28	Somatic	0.000	378	325	46.23	46.23	Somatic	0.000	1
6	150,184,625	C	T	LRP11	2	intronic		389	8	2.02	320	271	45.85	45.85	Somatic	0.000	316	309	49.44	49.44	Somatic	0.000	1
6	162,878,522	C	T	PARK2	3	intronic		690	17	2.40	561	488	46.52	46.52	Somatic	0.000	384	322	45.61	45.61	Somatic	0.000	1
6	166,045,320	A	G	PDE10A	3	5_prime_flanking_region		171	8	4.47	79	60	43.17	43.17	Somatic	0.000	344	334	49.26	49.26	Somatic	0.000	1
7	14,766,804	G	A	DGKB	3	intronic		311	3	0.96	435	376	46.36	46.36	Somatic	0.000	398	285	41.73	41.73	Somatic	0.000	1
7	19,322,718	C	T		0	3	0	325	16	4.69	236	195	45.24	45.24	Somatic	0.000	243	197	44.77	44.77	Somatic	0.000	1
7	23,245,489	G	A	GNPMB	2	5_prime_flanking_region		1053	56	5.05	409	367	47.29	47.29	Somatic	0.000	249	249	50.00	50.00	Somatic	0.000	1
7	40,891,373	G	A	C7orf10	3	3_prime_flanking_region		374	15	3.86	405	346	46.07	46.07	Somatic	0.000	342	327	48.88	48.88	Somatic	0.000	1
7	41,710,975	T	C	INHBA	2	5_prime_flanking_region		633	48	7.05	375	318	45.89	45.89	Somatic	0.000	287	309	51.85	51.85	Somatic	0.000	1
7	42,368,306	C	T		0	3	0	866	27	3.02	604	436	41.92	41.92	Somatic	0.000	385	303	44.04	44.04	Somatic	0.000	1
7	44,255,955	C	T	CAMK2B	2	intronic		305	0	0.00	200	5	2.44	2.44	Somatic	0.040	460	118	20.42	20.42	Somatic	0.000	2
7	49,636,687	C	T		0	3	0	830	30	3.49	1210	1124	48.16	48.16	Somatic	0.000	395	304	43.49	43.49	Somatic	0.000	1
7	50,762,444	A	G	GRB10	3	intronic		608	2	0.33	709	17	2.34	2.34	Somatic	0.001	482	124	20.46	20.46	Somatic	0.000	2
7	51,944,112	C	A		0	3	0	710	0	0.00	789	45	5.40	5.40	Somatic	0.000	433	156	26.49	26.49	Somatic	0.000	0
7	67,497,348	G	A		0	3	0	680	17	2.44	515	483	48.40	48.40	Somatic	0.000	380	354	48.23	48.23	Somatic	0.000	1
7	91,954,454	T	C	PEX1	3	3_prime_untranslated_region		116	2	1.69	293	322	52.36	52.36	Somatic	0.000	170	161	48.64	48.64	Somatic	0.000	1
7	104,632,275	C	A	SRPK2	3	intronic		949	50	5.01	723	643	47.07	47.07	Somatic	0.000	313						

8	36,228,367	G	A	ENSG00000210631	3	5_prime_flanking_region	451	20	4.25	704	599	45.97	45.97	Somatic	0.000	296	244	45.19	45.19	Somatic	0.000	1
8	39,742,880	T	A	ADAM2	3	intronic	210	11	4.98	404	287	41.53	41.53	Somatic	0.000	284	204	41.80	41.80	Somatic	0.000	1
8	52,586,888	C	T	PXDNL	3	intronic	1045	33	3.06	530	504	48.74	48.74	Somatic	0.000	400	377	48.52	48.52	Somatic	0.000	1
8	66,534,946	A	T		0	2	905	44	4.64	590	493	45.52	45.52	Somatic	0.000	461	403	46.64	46.64	Somatic	0.000	1
8	68,491,540	A	G	CPA6	3	3_prime_flanking_region	61	2	3.17	118	116	49.57	49.57	Somatic	0.000	225	215	48.86	48.86	Somatic	0.000	1
8	70,103,300	G	A	LOC100129096	2	intronic	143	1	0.69	254	203	44.42	44.42	Somatic	0.000	315	281	47.15	47.15	Somatic	0.000	1
8	75,925,059	T	C	P115	3	3_prime_untranslated_region	440	16	3.51	391	388	49.81	49.81	Somatic	0.000	321	344	51.73	51.73	Somatic	0.000	1
8	77,914,894	C	G	ZFH4	2	intronic	506	16	3.07	801	781	49.37	49.37	Somatic	0.000	258	286	52.57	52.57	Somatic	0.000	1
8	84,408,136	G	A		0	3	634	45	6.63	758	690	47.65	47.65	Somatic	0.000	459	428	48.25	48.25	Somatic	0.000	1
8	85,565,386	G	A	RALYL	3	intronic	618	34	5.21	414	382	47.99	47.99	Somatic	0.000	184	179	49.31	49.31	Somatic	0.000	1
8	86,568,817	T	G	CA2	3	intronic	202	18	8.18	192	196	50.52	50.52	Somatic	0.000	267	262	49.53	49.53	Somatic	0.000	1
8	87,836,982	T	A	CNG83	3	5_prime_flanking_region	424	13	2.97	319	190	37.33	37.33	Somatic	0.000	287	238	45.33	45.33	Somatic	0.000	1
8	88,641,249	C	T		0	3	641	13	1.99	753	711	48.57	48.57	Somatic	0.000	330	272	45.18	45.18	Somatic	0.000	1
8	90,190,284	C	T		0	3	308	6	1.91	326	252	43.60	43.60	Somatic	0.000	281	241	46.17	46.17	Somatic	0.000	1
8	93,512,629	G	A		0	3	537	11	2.01	231	218	48.55	48.55	Somatic	0.000	353	311	46.84	46.84	Somatic	0.000	1
8	94,755,932	G	A	FAM92A1	3	5_prime_flanking_region	356	6	1.66	416	387	48.19	48.19	Somatic	0.000	427	335	43.96	43.96	Somatic	0.000	1
8	111,759,719	C	G		0	3	368	5	1.34	436	457	51.18	51.18	Somatic	0.000	320	301	48.47	48.47	Somatic	0.000	1
8	115,838,142	G	A		0	3	315	16	4.83	432	316	42.25	42.25	Somatic	0.000	330	298	47.45	47.45	Somatic	0.000	1
8	128,300,749	C	T		0	2	1013	18	1.75	873	773	46.96	46.96	Somatic	0.000	422	347	45.12	45.12	Somatic	0.000	1
8	129,958,522	A	G		0	2	437	20	4.38	416	356	46.11	46.11	Somatic	0.000	411	400	49.32	49.32	Somatic	0.000	1
8	131,851,617	C	T	ADCY8	3	3_prime_flanking_region	1767	45	2.48	1215	1061	46.62	46.62	Somatic	0.000	465	418	47.34	47.34	Somatic	0.000	1
8	139,813,942	G	A	COL22A1	3	intronic	1021	50	4.67	352	348	49.71	49.71	Somatic	0.000	310	299	49.10	49.10	Somatic	0.000	1
8	142,815,582	G	A		0	3	140	4	2.78	62	57	47.90	47.90	Somatic	0.000	245	256	51.10	51.10	Somatic	0.000	1
8	143,778,000	A	C	LY6K	2	5_prime_flanking_region	219	31	12.40	100	32	24.24	24.24	Somatic	0.296	310	314	50.32	50.32	Somatic	0.000	0
9	7,463,965	A	G	LOC158345	3	3_prime_flanking_region	876	40	4.37	815	743	47.69	47.69	Somatic	0.000	412	366	47.04	47.04	Somatic	0.000	1
9	9,091,885	C	T	PTPRD	3	intronic	59	0	0.00	78	72	48.00	48.00	Somatic	0.000	158	126	44.37	44.37	Somatic	0.000	1
9	24,383,946	T	C		0	3	403	31	7.14	439	368	45.60	45.60	Somatic	0.000	268	268	50.00	50.00	Somatic	0.000	1
9	25,082,867	C	T	ENSG00000222693	3	5_prime_flanking_region	283	15	5.03	439	392	47.17	47.17	Somatic	0.000	318	282	47.00	47.00	Somatic	0.000	1
9	26,311,659	C	T		0	3	712	20	2.73	527	407	43.58	43.58	Somatic	0.000	348	280	44.59	44.59	Somatic	0.000	1
9	28,033,354	A	G	LINGO2	3	intronic	1342	56	4.01	793	709	47.20	47.20	Somatic	0.000	395	303	43.41	43.41	Somatic	0.000	1
9	30,430,017	C	A		0	2	318	12	3.64	602	528	46.73	46.73	Somatic	0.000	339	220	39.36	39.36	Somatic	0.000	0
9	30,927,679	C	T	ENSG00000214020	2	3_prime_flanking_region	370	10	2.63	361	308	46.04	46.04	Somatic	0.000	296	261	46.86	46.86	Somatic	0.000	1
9	32,208,802	T	C		0	3	182	6	3.19	363	299	45.17	45.17	Somatic	0.000	322	290	47.39	47.39	Somatic	0.000	1
9	35,669,773	G	A	CA9	3	intronic	603	16	2.58	57	56	49.56	49.56	Somatic	0.000	184	173	48.46	48.46	Somatic	0.000	1
9	36,409,304	T	A	RNF38	3	5_prime_flanking_region	601	8	1.31	443	279	38.64	38.64	Somatic	0.000	385	311	44.68	44.68	Somatic	0.000	1
9	67,834,918	C	T	ENSG00000219957	3	5_prime_flanking_region	369	6	1.60	327	279	46.04	46.04	Somatic	0.000	240	226	48.50	48.50	Somatic	0.000	1
9	89,535,224	T	G	CTS1L	1	missense	378	6	1.56	303	20	6.19	6.19	Somatic	0.170	487	126	20.55	20.55	Somatic	0.000	0
9	90,908,623	G	A	SHC3	2	intronic	1897	92	4.63	1469	1258	46.13	46.13	Somatic	0.000	414	390	48.51	48.51	Somatic	0.000	1
9	97,330,168	C	T	PTCH1	3	5_prime_flanking_region	787	22	2.72	351	288	45.07	45.07	Somatic	0.000	315	264	45.60	45.60	Somatic	0.000	1
9	100,197,933	C	T	GABBR2	3	intronic	406	5	1.22	127	89	41.20	41.20	Somatic	0.000	207	180	46.51	46.51	Somatic	0.000	1
9	100,343,785	A	G	GABBR2	3	intronic	1249	25	1.96	748	734	49.53	49.53	Somatic	0.000	476	449	48.54	48.54	Somatic	0.000	1
9	105,224,758	G	A		0	3	359	13	3.49	450	336	42.75	42.75	Somatic	0.000	342	284	45.37	45.37	Somatic	0.000	1
9	109,348,264	G	C	ENSG00000216493	3	5_prime_flanking_region	780	11	1.39	326	301	48.01	48.01	Somatic	0.000	383	339	46.95	46.95	Somatic	0.000	1
9	122,056,927	G	A	MIRN147	3	5_prime_flanking_region	794	48	5.70	464	400	46.30	46.30	Somatic	0.000	314	277	46.87	46.87	Somatic	0.000	1
9	122,561,326	T	C	FBXW2	3	3_prime_untranslated_region	393	12	2.96	316	271	46.17	46.17	Somatic	0.000	310	318	50.64	50.64	Somatic	0.000	1
9	131,535,363	A	T	PRRX2	3	3_prime_flanking_region	136	10	6.85	31	27	46.55	46.55	Somatic	0.000	192	176	47.83	47.83	Somatic	0.000	1
9	133,745,510	T	A	MED27	3	intronic	313	9	2.80	226	197	46.57	46.57	Somatic	0.000	330	249	43.01	43.01	Somatic	0.000	1
9	136,875,556	G	A	COL5A1	3	3_prime_untranslated_region	468	13	2.70	64	39	37.86	37.86	Somatic	0.000	199	174	46.65	46.65	Somatic	0.000	1
9	138,867,831	C	T	MAMDC4	1	silent	15	1	6.25	38	25	39.68	39.68	Somatic	0.012	217	186	46.15	46.15	Somatic	0.001	1
10	3,603,767	C	T	LOC100134102	3	5_prime_flanking_region	415	15	3.49	289	212	42.32	42.32	Somatic	0.000	302	273	47.48	47.48	Somatic	0.000	1
10	4,156,953	T	G	LOC731787	3	3_prime_flanking_region	494	12	3.27	1854	63	3.29	3.29	Somatic	0.475	411	119	22.45	22.45	Somatic	0.000	2
10	11,030,362	C	T		0	3	998	37	3.57	675	641	48.71	48.71	Somatic	0.000	335	257	43.41	43.41	Somatic	0.000	1
10	11,670,740	C	A	USP6NL	3	intronic	249	6	2.35	368	252	40.65	40.65	Somatic	0.000	251	187	42.69	42.69	Somatic	0.000	1
10	12,214,774	C	T	SEC61A2	3	intronic	525	17	3.14	311	281	47.47	47.47	Somatic	0.000	316	290	47.85	47.85	Somatic	0.000	1
10	14,654,478	A	C	FAM107B	3	intronic	473	9	1.87	1121	869	43.67	43.67	Somatic	0.000	386	324	45.63	45.63	Somatic	0.000	1
10	19,373,250	T	G	LOC100130846	3	5_prime_flanking_region	326	15	4.40	265	217	45.02	45.02	Somatic	0.000	268	237	46.93	46.93	Somatic	0.000	1
10	21,300,024	C	A	NEBL	2	intronic	186	6	3.13	564	454	44.60	44.60	Somatic	0.000	289	243	45.68	45.68	Somatic	0.000	1
10	34,032,764	G	A		0	3	658	18	2.66	727	617	45.91	45.91	Somatic	0.000	447	412	47.96	47.96	Somatic	0.000	1
10	45,273,565	G	A	MARCH8	3	3_prime_untranslated_region	558	3	0.53	706	9	1.26	1.26	Somatic	0.323	500	118	19.09	19.09	Somatic	0.000	2
10	46,461,650	G	A	PPYR1	2	5_prime_flanking_region	443	4	0.89	1284	13	1.00	1.00	Somatic	0.536	902	61	6.33	6.33	Somatic	0.000	0
10	48,121,263	G	A	ENSG00000221361	3	5_prime_flanking_region	717	32	4.27	307	215	41.19	41.19	Somatic	0.000	305	299	49.50	49.50	Somatic	0.000	1
10	58,815,611	G	A		0	3	243	3	1.22	371	289	43.79	43.79	Somatic	0.000	323	289	47.22	47.22	Somatic	0.000	1
10	59,817,876	G	C	TFAM	3	intronic	198	0	0.00	316	8	2.47	2.47	Somatic	0.021	404	94	18.88	18.88	Somatic	0.000	2
10	65,639,660	C	T	LOC100129267	3	5_prime_flanking_region	509	22	4.14	665	596	47.26	47.26	Somatic	0.000	453	399	46.83	46.83	Somatic	0.000	1
10	82,288,101	G	A	SH2D4B	1																	

11	30,881,639	A	G	DCDC5	1	silent	400	9	2.20	643	545	45.88	45.88	Somatic	0.000	357	320	47.27	47.27	Somatic	0.000	1	
11	65,523,582	C	G	E1F1AD	3	intronic	2168	28	2.34	273	230	45.73	45.73	Somatic	0.000	371	376	50.33	50.33	Somatic	0.000	1	
11	70,869,608	G	A	NADSVN1	3	intronic	105	12	5.53	104	114	52.29	52.29	Somatic	0.000	278	239	46.23	46.23	Somatic	0.000	1	
11	88,164,936	A	T	GRM5	3	intronic	334	11	3.19	412	363	46.84	46.84	Somatic	0.000	246	235	48.86	48.86	Somatic	0.000	1	
11	96,540,220	G	A		0	3	0	750	0	0.00	1293	25	1.90	1.90	Somatic	0.000	567	133	19.00	19.00	Somatic	0.000	2
11	118,994,585	G	T	ENSG000001199217	3	5_prime_flanking_region	464	6	1.28	107	70	39.55	39.55	Somatic	0.000	316	287	47.60	47.60	Somatic	0.000	1	
11	124,600,145	C	T	PKNOX2	2	intronic	1980	71	3.46	380	334	46.78	46.78	Somatic	0.000	287	259	47.44	47.44	Somatic	0.000	1	
12	7,610,680	C	T	LOC100134191	2	3_prime_flanking_region	941	26	2.69	706	562	44.32	44.32	Somatic	0.000	246	253	50.70	50.70	Somatic	0.000	1	
12	10,740,491	A	G	STYK1	3	5_prime_flanking_region	947	35	3.56	610	477	43.88	43.88	Somatic	0.000	315	367	53.81	53.81	Somatic	0.000	1	
12	40,159,842	G	C	PDZRN4	3	intronic	864	29	3.25	491	446	47.60	47.60	Somatic	0.000	386	359	48.19	48.19	Somatic	0.000	1	
12	56,424,837	C	T	TSPAN31	2	5_prime_flanking_region	320	14	4.19	105	102	49.28	49.28	Somatic	0.000	208	170	44.97	44.97	Somatic	0.000	1	
12	71,706,640	T	C		0	3	0	395	16	3.89	663	563	45.92	45.92	Somatic	0.000	365	314	46.24	46.24	Somatic	0.000	1
12	71,787,601	T	A		0	3	0	804	5	0.62	1106	27	2.38	2.38	Somatic	0.001	646	186	22.36	22.36	Somatic	0.000	2
12	76,730,664	C	G	NAV3	3	5_prime_flanking_region	277	10	3.48	334	300	47.32	47.32	Somatic	0.000	339	307	47.52	47.52	Somatic	0.000	1	
12	97,164,212	C	A		0	3	0	780	8	1.02	861	863	50.06	50.06	Somatic	0.000	436	448	50.68	50.68	Somatic	0.000	1
12	99,688,821	T	C	ANO4	2	5_prime_flanking_region	210	8	3.67	353	367	50.97	50.97	Somatic	0.000	265	224	45.81	45.81	Somatic	0.000	1	
12	105,956,296	G	A	CRY1	2	intronic	1387	65	4.48	797	748	48.41	48.41	Somatic	0.000	472	450	48.81	48.81	Somatic	0.000	1	
12	106,519,449	C	T	BTBD11	3	intronic	485	16	3.19	363	348	48.95	48.95	Somatic	0.000	276	281	50.45	50.45	Somatic	0.000	1	
12	113,287,422	C	T	TBX5	3	intronic	409	2	0.49	284	195	40.71	40.71	Somatic	0.000	249	204	45.03	45.03	Somatic	0.000	1	
12	117,961,498	C	A	KIAA1853	2	intronic	2340	75	3.11	744	609	45.01	45.01	Somatic	0.000	327	321	49.54	49.54	Somatic	0.000	1	
12	128,573,952	C	T	TMEM132D	2	intronic	426	13	2.96	343	313	47.71	47.71	Somatic	0.000	394	355	47.40	47.40	Somatic	0.000	1	
13	30,843,489	T	G	B3GALT1	3	3_prime_flanking_region	441	20	4.34	568	35	5.80	5.80	Somatic	0.760	651	1079	14.34	14.34	Somatic	0.000	0	
13	33,827,688	G	A		0	3	0	1310	74	5.35	593	534	47.38	47.38	Somatic	0.000	317	264	45.44	45.44	Somatic	0.000	1
13	35,857,458	T	G	SPG20	3	5_prime_flanking_region	265	8	2.93	237	261	52.41	52.41	Somatic	0.000	269	241	47.25	47.25	Somatic	0.000	1	
13	37,251,407	C	A	TRPC4	3	intronic	496	10	1.98	725	616	45.94	45.94	Somatic	0.000	351	286	44.90	44.90	Somatic	0.000	1	
13	54,761,858	G	A		0	2	0	530	10	1.85	1050	899	46.13	46.13	Somatic	0.000	380	385	50.33	50.33	Somatic	0.000	1
13	55,648,661	C	T	ENSG00000219006	3	5_prime_flanking_region	316	6	1.86	706	563	44.37	44.37	Somatic	0.000	418	375	47.29	47.29	Somatic	0.000	1	
13	57,494,427	G	C	ENSG00000202422	3	3_prime_flanking_region	305	0	0.00	823	15	1.79	1.79	Somatic	0.009	556	146	20.80	20.80	Somatic	0.000	2	
13	59,327,359	C	T	DIAPH3	3	intronic	361	24	6.23	424	367	46.40	46.40	Somatic	0.000	374	309	45.24	45.24	Somatic	0.000	1	
13	60,197,987	T	A		0	3	0	499	24	4.59	433	431	49.88	49.88	Somatic	0.000	354	329	48.17	48.17	Somatic	0.000	1
13	60,264,538	A	G	ENSG00000206854	3	3_prime_flanking_region	371	11	2.88	773	733	48.67	48.67	Somatic	0.000	305	275	47.41	47.41	Somatic	0.000	1	
13	84,248,715	G	A		0	3	0	304	35	5.48	667	538	44.65	44.65	Somatic	0.000	332	294	46.96	46.96	Somatic	0.000	1
13	103,366,966	C	A		0	3	0	152	8	5.00	155	143	47.99	47.99	Somatic	0.000	157	150	48.86	48.86	Somatic	0.000	1
13	105,042,162	A	G		0	2	0	161	1	0.62	196	160	44.94	44.94	Somatic	0.000	214	219	50.58	50.58	Somatic	0.000	1
13	105,714,967	G	T		0	3	0	496	8	1.59	486	437	47.35	47.35	Somatic	0.000	396	348	46.77	46.77	Somatic	0.000	1
13	107,212,497	G	T	FAM155A	3	intronic	518	1	0.19	667	19	2.77	2.77	Somatic	0.000	532	133	20.00	20.00	Somatic	0.000	2	
13	108,489,194	G	T	MYO16	3	intronic	221	8	3.49	147	146	49.83	49.83	Somatic	0.000	118	130	52.42	52.42	Somatic	0.000	1	
14	25,375,341	T	G	ENSG00000212270	2	5_prime_flanking_region	614	24	3.76	1044	112	9.69	9.69	Somatic	0.023	418	186	30.79	30.79	Somatic	0.000	0	
14	27,583,816	G	A		0	3	0	92	0	0.00	203	133	39.58	39.58	Somatic	0.000	130	80	38.10	38.10	Somatic	0.000	0
14	27,583,817	C	A		0	3	0	90	0	0.00	204	133	39.47	39.47	Somatic	0.000	131	80	37.91	37.91	Somatic	0.000	0
14	31,875,476	C	G	AKAP6	3	intronic	748	21	2.73	924	777	45.68	45.68	Somatic	0.000	469	437	48.23	48.23	Somatic	0.000	1	
14	32,863,416	T	C	NPAS3	2	intronic	547	16	2.84	671	616	47.86	47.86	Somatic	0.000	348	318	47.75	47.75	Somatic	0.000	1	
14	42,520,196	A	T		0	3	0	240	12	4.76	233	194	45.43	45.43	Somatic	0.000	232	212	47.75	47.75	Somatic	0.000	1
14	43,259,343	C	A		0	3	0	298	10	3.25	331	242	42.23	42.23	Somatic	0.000	261	252	49.12	49.12	Somatic	0.000	1
14	43,940,576	A	T	LOC729165	3	3_prime_flanking_region	96	5	4.95	103	70	40.46	40.46	Somatic	0.000	173	124	41.75	41.75	Somatic	0.000	1	
14	45,179,799	G	A		0	2	0	497	11	2.17	490	362	42.49	42.49	Somatic	0.000	295	257	46.56	46.56	Somatic	0.000	1
14	47,634,587	C	G		0	2	0	992	32	3.13	1125	899	44.42	44.42	Somatic	0.000	379	389	50.65	50.65	Somatic	0.000	1
14	49,219,295	A	T	POLE2	3	intronic	102	1	0.97	85	88	50.87	50.87	Somatic	0.000	179	187	51.09	51.09	Somatic	0.000	1	
14	69,096,682	A	C	LOC100130174	2	3_prime_flanking_region	443	85	16.10	311	117	27.34	27.34	Somatic	0.000	314	215	40.64	40.64	Somatic	0.000	0	
14	69,239,804	G	A	KIAA0247	3	intronic	567	37	6.13	402	325	44.70	44.70	Somatic	0.000	352	273	43.68	43.68	Somatic	0.000	1	
14	75,418,958	G	A	TTL5	1	missense	303	9	2.88	258	256	49.81	49.81	Somatic	0.000	289	261	47.45	47.45	Somatic	0.000	1	
14	80,669,474	G	A	TSHR	2	intronic	542	28	4.91	408	330	44.72	44.72	Somatic	0.000	375	313	45.49	45.49	Somatic	0.000	1	
14	86,938,324	C	T		0	3	0	255	0	0.00	583	20	3.32	3.32	Somatic	0.001	432	121	21.88	21.88	Somatic	0.000	2
14	91,505,353	G	A	FBLN5	2	5_prime_flanking_region	449	25	6.68	561	56	9.08	9.08	Somatic	0.544	409	136	24.95	24.95	Somatic	0.000	0	
14	95,369,238	C	T	LOC730125	3	5_prime_flanking_region	393	7	1.40	626	522	45.47	45.47	Somatic	0.000	407	358	46.80	46.80	Somatic	0.000	1	
14	98,438,420	A	T		0	3	0	1424	59	3.98	491	450	47.82	47.82	Somatic	0.000	361	363	50.14	50.14	Somatic	0.000	1
14	100,771,639	C	G		0	2	0	242	3	1.22	60	57	48.72	48.72	Somatic	0.000	110	111	50.23	50.23	Somatic	0.000	1
14	103,846,656	T	A	LOC100131034	2	3_prime_flanking_region	180	1	0.55	68	67	49.63	49.63	Somatic	0.000	340	297	46.62	46.62	Somatic	0.000	1	
15	18,435,184	C	T	VSI67	3	5_prime_flanking_region	289	16	5.25	217	114	34.44	34.44	Somatic	0.000	284	224	44.09	44.09	Somatic	0.000	0	
15	25,956,026	G	A	OCA2	3	intronic	982	27	2.68	338	279	45.22	45.22	Somatic	0.000	349	375	51.80	51.80	Somatic	0.000	1	
15	27,762,153	G	A	ENSG00000212769	3	5_prime_flanking_region	123	4	3.15	71	69	49.29	49.29	Somatic	0.000	243	227	48.30	48.30	Somatic	0.000	1	
15	31,122,283	A	T	FMN1	3	intronic	91	3	3.19	104	9	7.96	7.96	Somatic	0.026	256	38	12.93	12.93	Somatic	0.001	0	
15	35,751,548	T	C		0	2	0	607	9	1.46	639	543	45.94	45.94	Somatic	0.000	388	360	48.13	48.13	Somatic	0.000	1
15	35,928,644	G	A		0	3	0	830	18	2.12	1006	914	47.60	47.60</									

16	26,024,776	G	A	HS3T4	3	intronic	942	20	2.08	567	538	48.69	48.69	Somatic	0.000	331	291	46.78	46.78	Somatic	0.000	1	
16	26,732,330	G	T		0	2	0	2087	1	0.05	3254	72	2.16	2.16	Somatic	0.000	773	225	22.55	22.55	Somatic	0.000	2
16	50,030,298	C	T		0	3	0	509	19	3.60	423	331	43.90	43.90	Somatic	0.000	388	333	46.19	46.19	Somatic	0.000	1
16	50,680,429	C	T	LOC388276	3	5_prime_flanking_region	368	15	3.92	295	298	50.25	50.25	Somatic	0.000	284	251	46.92	46.92	Somatic	0.000	1	
16	51,518,521	T	A	LOC390730	3	5_prime_flanking_region	416	11	2.58	223	190	46.00	46.00	Somatic	0.000	232	184	44.23	44.23	Somatic	0.000	1	
16	57,441,427	T	G		0	3	0	251	17	6.34	218	31	12.45	12.45	Somatic	0.005	341	135	28.36	28.36	Somatic	0.000	0
16	57,599,867	C	T		0	3	0	281	4	1.40	247	215	46.54	46.54	Somatic	0.000	282	247	46.69	46.69	Somatic	0.000	1
16	59,855,653	G	C		0	3	0	455	0	0.00	927	27	2.83	2.83	Somatic	0.000	454	130	22.26	22.26	Somatic	0.000	2
16	63,589,987	C	T	CDH11	1	missense	1112	39	3.39	806	689	46.09	46.09	Somatic	0.000	364	333	47.78	47.78	Somatic	0.000	1	
16	74,512,157	C	T		0	2	0	434	7	1.59	366	318	46.49	46.49	Somatic	0.000	337	325	49.09	49.09	Somatic	0.000	1
16	77,161,670	G	A	WVVOX	3	intronic	810	3	1.84	156	115	42.44	42.44	Somatic	0.000	201	177	46.83	46.83	Somatic	0.000	1	
16	77,367,408	G	A	WVVOX	3	intronic	811	40	4.70	637	514	44.66	44.66	Somatic	0.000	404	347	46.21	46.21	Somatic	0.000	1	
16	78,207,037	C	A	MAF	3	5_prime_flanking_region	996	36	3.49	546	483	46.94	46.94	Somatic	0.000	405	425	51.20	51.20	Somatic	0.000	1	
16	81,204,189	C	T	CDH13	2	5_prime_flanking_region	855	36	4.04	593	510	46.24	46.24	Somatic	0.000	425	399	48.42	48.42	Somatic	0.000	1	
16	81,731,917	C	T	CDH13	3	intronic	1040	2	0.19	1218	23	1.85	1.85	Somatic	0.000	519	136	20.76	20.76	Somatic	0.000	2	
16	82,624,963	C	T	SLC38A8	3	intronic	1027	24	2.28	170	160	48.48	48.48	Somatic	0.000	344	327	48.73	48.73	Somatic	0.000	1	
17	4,480,344	C	T	ENSG00000222429	3	5_prime_flanking_region	376	21	5.29	114	99	46.48	46.48	Somatic	0.000	255	234	47.85	47.85	Somatic	0.000	1	
17	5,288,451	T	G	DHX33	1	missense	604	18	2.89	187	158	45.80	45.80	Somatic	0.000	254	193	43.18	43.18	Somatic	0.000	1	
17	7,533,418	A	G	WRAP53	2	intronic	519	24	4.42	125	91	42.13	42.13	Somatic	0.000	248	201	44.77	44.77	Somatic	0.000	1	
17	7,696,343	T	G	JMJ3D	1	missense	114	6	5.00	70	6	7.89	7.89	Somatic	0.135	289	96	24.94	24.94	Somatic	0.005	0	
17	21,950,614	C	T	ENSG00000209244	2	5_prime_flanking_region	85	3	3.41	44	42	48.84	48.84	Somatic	0.000	73	62	45.93	45.93	Somatic	0.000	1	
17	24,186,705	T	C	C17orf63	3	intronic	406	15	3.56	136	144	51.43	51.43	Somatic	0.000	329	289	46.76	46.76	Somatic	0.000	1	
17	25,735,564	A	C	CPD	3	intronic	1078	21	16.28	234	96	29.09	29.09	Somatic	0.025	211	228	51.94	51.94	Somatic	0.000	0	
17	26,580,889	G	C	NF1	3	intronic	477	10	2.05	540	458	45.89	45.89	Somatic	0.000	233	238	50.53	50.53	Somatic	0.000	1	
17	28,127,819	C	T	MYO1D	3	intronic	324	17	4.99	243	180	42.55	42.55	Somatic	0.000	277	221	44.38	44.38	Somatic	0.000	1	
17	29,981,226	C	T	TMEM132E	1	silent	1815	79	4.17	275	343	55.50	55.50	Somatic	0.000	289	248	46.18	46.18	Somatic	0.000	0	
17	35,007,340	T	C	STAR3D	2	5_prime_flanking_region	681	29	4.08	97	80	45.20	45.20	Somatic	0.000	159	115	41.97	41.97	Somatic	0.000	1	
17	36,096,311	T	C	SMARCE1	2	5_prime_flanking_region	128	1	0.78	196	174	47.03	47.03	Somatic	0.000	253	232	47.84	47.84	Somatic	0.000	1	
17	48,968,272	G	A	ENSG00000221534	3	5_prime_flanking_region	329	2	0.60	384	241	38.56	38.56	Somatic	0.000	249	176	41.41	41.41	Somatic	0.000	1	
17	49,442,081	C	T		0	2	0	576	20	3.36	394	342	46.47	46.47	Somatic	0.000	326	241	42.50	42.50	Somatic	0.000	1
17	50,661,710	T	C	HLF	3	5_prime_flanking_region	1225	58	4.52	647	485	42.84	42.84	Somatic	0.000	486	487	50.05	50.05	Somatic	0.000	1	
17	59,597,932	C	G	TEX2	3	intronic	322	5	1.53	441	12	2.65	2.65	Somatic	0.195	380	64	14.41	14.41	Somatic	0.000	0	
17	67,860,759	A	C	ENSG00000211264	3	3_prime_flanking_region	286	4	1.38	185	190	50.67	50.67	Somatic	0.000	291	232	44.36	44.36	Somatic	0.000	1	
17	75,696,250	G	A	GAA	1	silent	23	2	8.00	17	13	43.33	43.33	Somatic	0.016	85	63	42.57	42.57	Somatic	0.001	1	
18	2,228,633	C	T	LOC100131655	2	intronic	1078	46	4.09	1107	952	46.24	46.24	Somatic	0.000	344	333	49.19	49.19	Somatic	0.000	1	
18	5,155,122	G	A	ENSG00000215368	3	5_prime_flanking_region	957	30	3.04	597	492	45.18	45.18	Somatic	0.000	400	338	45.80	45.80	Somatic	0.000	1	
18	11,368,929	G	A		0	3	0	359	17	4.52	443	408	47.94	47.94	Somatic	0.000	318	245	43.52	43.52	Somatic	0.000	1
18	18,624,758	T	C		0	2	0	1300	41	3.06	866	768	47.00	47.00	Somatic	0.000	440	402	47.74	47.74	Somatic	0.000	1
18	26,038,883	G	A		0	3	0	443	0	0.00	736	17	2.26	2.26	Somatic	0.000	524	146	21.79	21.79	Somatic	0.000	2
18	29,585,389	T	G	ASXL3	3	3_prime_flanking_region	162	6	3.57	120	116	49.15	49.15	Somatic	0.000	186	201	51.94	51.94	Somatic	0.000	1	
18	29,798,038	G	A	NOLA	3	intronic	543	21	3.72	860	644	42.82	42.82	Somatic	0.000	441	337	43.32	43.32	Somatic	0.000	1	
18	30,590,076	A	G	DTNA	2	intronic	432	14	3.14	407	314	43.55	43.55	Somatic	0.000	323	329	50.46	50.46	Somatic	0.000	1	
18	36,013,083	C	T		0	3	0	303	11	3.50	426	307	41.88	41.88	Somatic	0.000	279	238	46.03	46.03	Somatic	0.000	1
18	36,630,355	T	G		0	3	0	175	9	4.89	207	186	47.33	47.33	Somatic	0.000	183	174	48.74	48.74	Somatic	0.000	1
18	46,165,704	G	A	C18orf24	1	silent	327	12	3.54	298	242	44.81	44.81	Somatic	0.000	302	315	51.05	51.05	Somatic	0.000	1	
18	47,449,416	C	T		0	2	0	431	6	1.37	496	445	47.29	47.29	Somatic	0.000	434	323	42.67	42.67	Somatic	0.000	1
18	51,514,701	A	G		0	3	0	540	20	3.57	391	381	49.35	49.35	Somatic	0.000	371	347	48.33	48.33	Somatic	0.000	1
18	52,121,185	G	A		0	3	0	393	12	2.96	392	396	50.25	50.25	Somatic	0.000	246	250	50.40	50.40	Somatic	0.000	1
18	52,654,678	A	C	WDR7	3	intronic	533	12	2.20	1205	115	8.71	8.71	Somatic	0.000	393	159	28.80	28.80	Somatic	0.000	0	
18	62,821,154	G	T		0	3	0	283	11	3.74	477	424	47.06	47.06	Somatic	0.000	310	319	50.72	50.72	Somatic	0.000	1
18	63,165,104	T	C		0	3	0	412	1	0.24	1155	33	2.78	2.78	Somatic	0.000	688	185	21.19	21.19	Somatic	0.000	2
18	64,146,279	G	T		0	3	0	348	13	3.60	447	360	44.61	44.61	Somatic	0.000	314	257	45.01	45.01	Somatic	0.000	1
18	64,226,339	T	G		0	3	0	422	41	8.86	925	162	14.90	14.90	Somatic	0.798	394	216	35.41	35.41	Somatic	0.005	0
18	67,103,868	C	T		0	3	0	226	12	5.04	256	251	49.51	49.51	Somatic	0.000	262	232	46.96	46.96	Somatic	0.000	1
18	67,787,857	G	A		0	3	0	142	5	3.40	165	127	43.49	43.49	Somatic	0.000	194	173	47.14	47.14	Somatic	0.000	1
18	69,657,558	A	G		0	3	0	771	52	6.32	682	675	49.74	49.74	Somatic	0.000	372	324	46.55	46.55	Somatic	0.000	1
18	70,624,608	A	G	ZNF407	2	intronic	258	4	1.53	494	472	48.86	48.86	Somatic	0.000	327	311	48.75	48.75	Somatic	0.000	1	
18	74,833,430	C	T	SALL3	3	5_prime_flanking_region	94	5	5.05	17	37	68.52	68.52	Somatic	0.000	242	215	47.05	47.05	Somatic	0.000	0	
19	16,885,826	C	T	CPAMD8	3	intronic	524	11	2.06	146	150	50.68	50.68	Somatic	0.000	293	250	46.04	46.04	Somatic	0.000	1	
19	18,513,222	G	A	FKBP8	3	intronic	396	23	5.49	88	72	45.00	45.00	Somatic	0.000	204	228	52.78	52.78	Somatic	0.000	1	
19	19,045,292	G	A	SLC25A42	3	intronic	1127	38	3.26	119	94	44.13	44.13	Somatic	0.000	391	312	44.38	44.38	Somatic	0.000	1	
19	20,320,043	C	T	ZNF826	3	3_prime_flanking_region	75	4	5.06	58	64	52.46	52.46	Somatic	0.000	86	73	45.91	45.91	Somatic	0.000	1	
19	39,677,436	C	T	WTP1	3	intronic	270	10	3.57	147	107	42.13	42.13	Somatic	0.000	232	223	49.01	49.01	Somatic	0.000	1	
19	48,741,326	C	T	XRC1	3	intronic	1207	60	4.74	248	178	41.78	41.78	Somatic	0.000	358	293	45.01	45.01	Somatic			

21	26,646,726	G	A		0	3		0	215	0	0.00	385	9	2.28	2.28	Somatic	0.018	522	137	20.79	20.79	Somatic	0.000	2
21	30,459,290	C	T	CLDN8	3	3_prime_flanking_region		370	16	4.15	921	766	45.41	45.41	Somatic	0.000	354	330	48.25	48.25	Somatic	0.000	1	
21	41,416,902	G	A	BACE2	3	5_prime_flanking_region		1038	32	2.99	664	560	45.75	45.75	Somatic	0.000	399	339	45.93	45.93	Somatic	0.000	1	
21	42,402,818	A	C	UMODL1	1	missense		84	8	8.70	65	8	10.96	10.96	Somatic	0.577	161	75	31.78	31.78	Somatic	0.000	0	
21	43,397,525	G	A	U2AF1	1	missense		404	30	6.91	91	83	47.70	47.70	Somatic	0.000	343	317	48.03	48.03	Somatic	0.000	1	
21	46,252,211	C	T	COL6A1	2	3_prime_flanking_region		92	1	1.08	89	67	42.95	42.95	Somatic	0.000	213	170	44.39	44.39	Somatic	0.000	1	
21	46,301,845	G	T	COL6A2	2	5_prime_flanking_region		46	0	0.00	11	2	15.38	15.38	Reference	0.041	251	182	42.03	42.03	Somatic	0.000	0	
22	18,187,764	C	T	GNB1L	3	intronic		387	7	1.78	104	92	46.94	46.94	Somatic	0.000	223	211	48.62	48.62	Somatic	0.000	1	
22	19,294,334	C	G	ENSG00000220150	3	5_prime_flanking_region		265	25	8.62	128	119	48.18	48.18	Somatic	0.000	360	298	45.29	45.29	Somatic	0.000	1	
22	20,866,781	G	A	IGLV11-55	3	5_prime_flanking_region		328	21	6.02	185	239	56.37	56.37	Somatic	0.000	165	156	48.60	48.60	Somatic	0.000	0	
22	36,194,879	A	C	ELFN2	3	5_prime_flanking_region		735	74	9.15	291	56	16.14	16.14	Somatic	0.000	502	195	27.98	27.98	Somatic	0.000	0	
22	38,089,713	C	T	SYNGR1	3	intronic		954	46	4.60	97	89	47.85	47.85	Somatic	0.000	145	139	48.94	48.94	Somatic	0.000	1	
22	46,395,195	C	T	FLJ46257	3	3_prime_flanking_region		378	10	2.58	137	111	44.76	44.76	Somatic	0.000	302	308	50.49	50.49	Somatic	0.000	1	
22	46,512,620	A	G		0	3		471	11	2.28	526	17	3.13	3.13	Somatic	0.421	431	115	21.06	21.06	Somatic	0.000	2	
X	3,387,861	G	A		0	3		375	0	0.00	362	21	5.48	2.74	Somatic	0.000	167	112	40.14	20.07	Somatic	0.000	2	
X	29,779,410	G	A	IL1RAPL1	2	intronic		121	6	4.72	21	384	94.81	47.41	Somatic	0.000	19	292	93.89	46.95	Somatic	0.000	1	
X	32,777,396	C	T	DMD	3	intronic		414	38	8.41	31	496	94.12	47.06	Somatic	0.000	21	292	93.29	46.65	Somatic	0.000	1	
X	34,705,765	A	G		0	3		217	29	11.79	7	287	97.62	48.81	Somatic	0.000	12	311	96.28	48.14	Somatic	0.000	1	
X	39,157,340	A	G		0	3		283	22	7.21	22	391	94.67	47.34	Somatic	0.000	22	413	94.94	47.47	Somatic	0.000	1	
X	52,897,186	C	T	FAM156B	3	5_prime_flanking_region		226	11	4.64	18	368	95.34	47.67	Somatic	0.000	14	282	95.27	47.64	Somatic	0.000	1	
X	104,353,541	G	C	IL1RAPL2	3	intronic		141	21	12.96	24	327	93.16	46.58	Somatic	0.000	19	424	95.71	47.86	Somatic	0.000	1	
X	111,088,560	G	A	TRPC5	3	intronic		253	11	4.17	39	584	93.74	46.87	Somatic	0.000	15	321	95.54	47.77	Somatic	0.000	1	
X	116,699,743	C	A		0	3		219	25	10.25	34	461	93.13	46.57	Somatic	0.000	11	396	97.30	48.65	Somatic	0.000	1	
X	118,033,017	G	A	LONRF3	3	intronic		473	37	7.25	18	437	96.04	48.02	Somatic	0.000	16	351	95.64	47.82	Somatic	0.000	1	
X	132,324,635	C	T	GPC4	3	intronic		642	64	9.07	61	841	93.24	46.62	Somatic	0.000	21	412	95.15	47.58	Somatic	0.000	1	
X	142,016,065	C	T	ENSG00000202473	3	3_prime_flanking_region		59	6	9.23	10	139	93.29	46.64	Somatic	0.000	8	164	95.35	47.67	Somatic	0.000	1	
X	151,265,229	C	T	GABRA3	3	intronic		621	28	4.31	50	711	93.43	46.71	Somatic	0.000	14	376	96.41	48.21	Somatic	0.000	1	
Y	9,231,939	G	A	TTTY20	2	intronic		115	8	6.50	6	96	94.12	47.06	Somatic	0.000	17	226	93.00	46.50	Somatic	0.000	1	
Y	15,681,536	T	C		0	3		359	9	2.45	512	41	7.41	3.71	Somatic	0.015	308	110	26.32	13.16	Somatic	0.000	0	
Y	22,378,497	C	T	ENSG00000217635	3	5_prime_flanking_region		217	24	9.96	9	209	95.87	47.94	Somatic	0.000	17	196	92.02	46.01	Somatic	0.000	1	
Y	22,830,969	C	T	RBMV1J	3	5_prime_flanking_region		158	11	6.51	12	215	94.71	47.36	Somatic	0.000	13	321	96.11	48.05	Somatic	0.000	1	

Supplementary Table 3. Annotated Tier 1 Mutations in the MDS and sAML genomes.

Chromosome	Position*	Reference allele	Variant allele	Gene symbol	Transcript ID	Strand	Mutation type	Mutated in MDS	Mutated in sAML	Zygoty	Coding position	Amino acid change
1	112,325,945	G	T	KCND3	NM_004980	-1	synonymous	Y	Y	Het	c.927	I309
1	205,309,412	C	G	PFKFB2	NM_006212	1	synonymous	Y	Y	Het	c.1008	T336
1	229,096,537	T	C	ENSG00000222671	ENST00000410739	1	non-coding RNA	Y	Y	Het	NA	NA
2	20,289,990	C	T	LOC100131373	XM_001720891	1	synonymous	Y	Y	Het	c.261	A87
2	61,569,230	T	A	XPO1	NM_003400	-1	missense	Y	Y	Het	c.2203	N735Y
3	173,529,456	C	A	FNDC3B	NM_022763	1	missense	Y	Y	Het	c.1275	F425L
5	50,152,869	C	G	PARP8	NM_024615	1	missense	Y	Y	Het	c.1734	F578L
5	148,880,083	T	G	CSNK1A1	NM_001025105	-1	missense	Y	Y	Het	c.419	D140A
6	105,713,110	C	A	POPDC3	NM_022361	-1	missense	Y	Y	Het	c.804	M268I
9	89,535,224	T	G	CTSL1	NM_001912	1	missense	Y	Y	Het	c.893	V298G
9	138,867,831	C	T	MAMDC4	NM_206920	1	synonymous	Y	Y	Het	c.342	A114
10	82,288,101	G	A	SH2D4B	NM_207372	1	missense	Y	Y	Het	c.34	D12N
11	12,198,538	C	G	MICAL2	NM_014632	1	missense	Y	Y	Het	c.1163	A388G
11	22,340,860	G	T	SLC17A6	NM_020346	1	splice_site	Y	Y	Het	c.662-1	e6-1
11	30,881,639	A	G	DCDC5	ENST00000406071	-1	synonymous	Y	Y	Het	c.1806	V602
14	75,418,958	G	A	TLL5	NM_015072	1	missense	Y	Y	Het	c.3700	E1234K
16	7,716,860	C	T	ENSG00000209555	ENST00000386820	-1	non-coding RNA	Y	Y	Het	NA	NA
16	63,589,987	C	T	CDH11	NM_001797	-1	missense	Y	Y	Het	c.502	V168M
17	5,288,451	T	G	DHX33	NM_020162	-1	missense	Y	Y	Het	c.1922	Y641S
17	7,696,343	T	G	JMJD3	NM_001080424	1	missense	Y	Y	Het	c.4432	C1478G
17	29,981,226	C	T	TMEM132E	NM_207313	1	synonymous	Y	Y	Het	c.1155	L385
17	75,696,250	G	A	GAA	NM_000152	1	synonymous	Y	Y	Het	c.915	G305
18	46,165,704	G	A	C18orf24	NM_001039535	1	synonymous	Y	Y	Het	c.432	E144
19	53,579,461	T	G	KDELR1	NM_006801	-1	missense	Y	Y	Het	c.442	T148P
19	54,349,852	A	G	HRC	NM_002152	-1	missense	Y	Y	Het	c.455	L152P
20	32,728,482	A	C	PIGU	NM_080476	-1	missense	Y	Y	Het	c.69	S23R
20	32,965,857	A	C	ACSS2	NM_018677	1	missense	Y	Y	Het	c.790	T264P
20	51,626,823	T	C	ZNF217	NM_006526	-1	synonymous	Y	Y	Het	c.1887	R629
21	42,402,818	A	C	UMODL1	NM_173568	1	missense	Y	Y	Het	c.1597	T533P
21	43,397,525	G	A	U2AF1	NM_001025203	-1	missense	Y	Y	Het	c.101	S34F

*build 36

Supplementary Table 4. U2AF1 Somatic Mutations in MDS.

UPN	gene name	chromosome	start	stop	reference allele	variant allele	transcript	strand	mutation type	coding position	amino acid change	SIFT	SIFT score	PolyPhen2	PolyPhen2 score
02-86	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
185295	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
266395	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
300813	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
321864	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
463763	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
571656	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
843623	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
917011	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
947519*	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
989739	U2AF1	21	43397525	43397525	G	A	NM_001025203	-1	missense	c.101	p.S34F	damaging	0	probably damaging	1
137250	U2AF1	21	43397525	43397525	G	T	NM_001025203	-1	missense	c.101	p.S34Y	damaging	0	probably damaging	1
937954	U2AF1	21	43397525	43397525	G	T	NM_001025203	-1	missense	c.101	p.S34Y	damaging	0	probably damaging	1
947519*	U2AF1	21	43387846	43387846	T	C	NM_001025203	-1	missense	c.470	p.Q157R	damaging	0	probably damaging	0.999

UPN, unique patient number

* same UPN

Supplementary Table 5. Characteristics of MDS patients with *U2AF1* Mutations

UPN	Gender	Age at diagnosis	FAB	Karyotype	%BM Blast	IPSS	Therapy after MDS banking (months)	AML Progression?	OS (months)	EFS (months)
266395 ^a	M	65	RAEB	46,XY[20]	7	1	azacytidine (6)	yes	11	2
137250	F	54	RAEB	46,XX[20]	19	2	standard induction, allogeneic SCT	yes	100	1
185295	M	50	RA	46,XY,del(20)(q11.2q13.3)[7]/46,XY,[13]	0	0.5	PTK/ZK (<1), azacytidine (2), allogeneic SCT	no	97	83
300813	F	42	RA	46,XX[20]	2	0	none	no	71	71
321864	F	63	RAEB	46,XX+8[20]	12	2.5	standard induction	no	3	3
571656	M	71	RAEB	46,XY,del(20)(q11.2q13.3)[12]/46XY[8]	4	0.5	standard induction	yes	10	8
843623	M	60	RAEB	46,XY,del(20)(q11.3)[2]/46,XY[18]	6	1	none	yes	5	2
917011	M	79	RA	46,XY,del(20)(q11.2)[12]/ 46,XY[8]	0	0.5	lenalidomide (3)	no	27	25
937954	M	51	RAEB	47,XY,+8[1]/47, idem, del(16)(q12.1)[13]/48, idem,+Y, del(16)(q11.1)[5]/46,XY[1]	12	2.5	azacytidine (3)	no	24	24
947519	M	59	RAEB	46,XY,1-5dms[10]/46,XY[12]	7	1.5	decitabine (3), allogeneic SCT, standard induction, salvage chemotherapy	yes	24	23
02-86	M	53	RAEB-T	92<4n>,XXYY,del(4)(q33)x2,del(5)(q13),add(11)(q23)x2[cp8]/46,XY[12]	21	3	decitabine (2)	yes	10	4
463763	M	54	RA	47,XY,+Y[6]/46,XY[14]	0	0.5	none	no	46	44
989739	M	71	RAEB	46,XY,del(7)(q21)[11]/47,XY,+8[5]/46,XY[4]	18	3	decitabine (8)	yes	15	4

UPN, unique patient number; OS, overall survival; EFS, event-free survival; PTK/ZK (Vatalanib), VEGF inhibitor.

^aindex case

Supplementary Table 7. Clinical Characteristics of MDS replication cohort.

UPN	Gender	Age @ Diagnosis	%BM Blast	FAB	Cytogenetics	[-5, del(5q)] (0=no/1=yes)	[-20, del(20q)] (0=no/1=yes)	[-7, del(7q)] (0=no/1=yes)	[+8] (0=no/1=yes)	IPSS	Disease Status (0 alive/ 1 dead)	Days from Dx to Death or Last Follow-up	Evolution to AML (0=no/1=yes)	Days from Dx to AML or last Follow-up	DNMT3A status (0=wt/1=mut)	U2AF1 status (0=wt/1=mut)
100901	M	70	4	RA	46,XY,del(3)(q21)[16]/46,XY[4]	0	0	0	0	0.5	1	1854	0	1654	0	0
105327	F	54	1	RA	46,XX[20]	0	0	0	0	0.0	1	1647	0	1646	0	0
112848	M	62	5	RAEB	46,XY,del(5)(q13q33)[18]/46,XY[1]/45,XY,-5[1]	1	0	0	0	0.5	1	2116	0	2033	0	0
116960	F	55	2	RA	46,XX,t(11;13)(q23;q14)[14]/47,idem,+2[12]/46,XX[3]/46,XX,inv(14)(q11,q32)[1]	0	0	0	0	1.0	0	2362	0	2246	0	0
123363	M	64	0	RA	46,XY[19]/46,XY,del(20)(q11,q13.1)[1]	0	0	0	0	0.0	0	2421	0	2402	0	0
124124	F	66	18	RAEB	46,XX[20]	0	0	0	0	2.0	1	998	1	891	0	0
127804	M	63	2	RA	46,XY[20]	0	0	0	0	0.5	1	2375	0	2368	0	0
131153	M	78	8	RAEB	44,XY,dup(3)(p21p25),-5,-6,del(12)(q15q22),psu dic(15;14)(p13;q32),add(17)(p11.2),add(19)(q13.3),7i(22;22)(q12.13),+mar[20]	1	0	0	0	2.0	1	621	0	619	0	0
137250	F	54	19	RAEB	46,XX[20]	0	0	0	0	2.0	0	3037	1	38	0	1
137313	F	77	0	RA	47,XX,+8[6]/46,XX[14]	0	0	0	1	1.0	1	320	0	144	0	0
137404	F	70	0	RAR	46,XX,add(1)(p11.1),del(1)(q23),add(3)(p13),del(5)(q13q33),add(8)(p11.2),add(9)(q22),add(10)(q22),add(11)(p14),-12,del(17)(q21),+mar[17]/46,XX[3]	1	0	0	0	1.0	1	674	0	663	0	0
173005	M	77	10	RAEB	45,XY,der(3)(5)(p10;p10),-18,-20,+2mar[12]/46,idem,+2[5]/46,XY[3]	0	0	0	0	2.0	1	800	0	71	0	0
176267	M	56	7	RAEB	43-47,XY,del(5)(q13q33),der(7)(7;11)(q11.2;q21),-9,-12,der(16)t(12;16)(q10;q22),add(16)(q23),+mar1,+mar2[cp12]/44-46,XY,del(5)(q13q33),der(7)t(7;11)(q11.2;q21),-12,del(16)(q10;q22)[cp8]	1	0	0	0	2.0	1	137	1	93	1	0
177356	F	76	2	RA	46,XX[20]	0	0	0	0	0.0	0	1304	0	747	0	0
182896	M	75	0	RA	46,XY,add(4)(p16),t(4;20)(q25;p13),del(5)(q13q33)[13]/46,idem,del(6)(q25)[1]/47,XY,+8[6]	0	0	0	1	1.0	1	1205	1	1047	0	0
184368	F	49	0	RA	46,XX[20]	0	0	0	0	0.0	0	2013	0	81	0	0
184874	F	56	2	RAR	47,XX,+8[10]/46,XX[10]	0	0	0	1	0.5	1	2020	0	2020	0	0
185295	M	50	0	RA	46,XY,del(20)(q11,q13.3)[7]/46,XY[13]	0	1	0	0	0.5	0	2539	0	2536	0	1
189012	M	64	3	RA	46,XY,del(5)(q15q33)[16]/46,XY,del(5)(q12q33),del(7)(q21q32)[1]/46,XY[3]	1	0	0	0	0.5	0	1250	0	1230	0	0
189474	M	61	4	RAEB	46,XY[3]	0	0	0	0	0.0	0	1336	0	907	0	0
194328	M	52	1	RA	46,XY[20]	0	0	0	0	0.5	0	1702	0	809	0	0
197795	M	50	17	RAEB	46,XY[20]	0	0	0	0	1.5	1	1064	0	457	0	0
199019	F	60	0	RA	45-49,XX,+1,add(3)(p11),del(5)(q13q33),del(10)(q24q26),add(18)(q21),+19,-20,+21,+1-2mar[cp20]	1	1	0	0	1.5	1	1491	1	1020	0	0
201641	M	62	14	RAEB	44-45,XY,del(3)(q12),add(5)(q35),del(5)(q12q33),del(7)(q22),-12,der(17)t(7;17)(q22p13)[15]/46,XY[5]	1	0	1	0	3.0	1	220	0	199	0	0
207282	M	76	11	RAEB	46,XY[20]	0	0	0	0	2.0	1	251	1	122	1	0
222841	F	46	17	RAEB	60<3ns>,XX,-X,add(1)(q44),ins(1)(p36)add(1)(q12),-5,del(5)(q11,q23q33),+6,-7,add(7)(p22),-10,-11,del(12)(q13),add(12)(p13),-13,-14,hsr(14)(p11.1),-16,-16,-18,der(19;21)(q10;q10),+21,-22,+mar5/46,XX[15]	1	0	1	0	2.5	1	528	0	528	0	0
266395	M	65	7	RAEB	46,XY[20]	0	0	0	0	1.0	1	321	1	75	0	1
280933	F	31	13	RAEB	46,XX[20]	0	0	0	0	2.0	1	367	0	28	0	0
298273	M	27	18	RAEB-T	46,XY[20]	0	0	0	0	2.0	1	540	1	131	0	0
300813	F	42	2	RA	46,XX[20]	0	0	0	0	0.0	0	2172	0	2172	0	1
312519	F	71	0	RA	46,XX[20]	0	0	0	0	0.5	1	1026	0	436	0	0
313175	M	74	5	RAEB	46,XY[20]	0	0	0	0	0.0	0	961	0	702	0	0
315529	M	56	12	RAEB	46,XY,add(7)(q32),r(8)(p21q24.3),add(11)(p15),t(12;13)(q24.2;q13),-17,r(19)(p13.3q13.4),-20,add(20)(q13.1),-21,+3mar[18]/46,idem,del(5)(q22q34)[1]/46,XY[1]	0	1	0	0	3.0	1	147	0	147	0	0
317598	M	86	11	RAEB	46,XY[20]	0	0	0	0	2.0	1	30	0	30	1	0
319179	F	74	6	RAEB	46,XX[20]	0	0	0	0	0.5	1	483	1	482	1	0
321864	F	63	12	RAEB	47,XX,+8[20]	0	0	0	0	2.5	1	80	0	80	0	1
323977	M	65	0	RA	46,XY[20]	0	0	0	0	0.5	1	249	0	186	0	0
335224	M	65	3	RA	45,XY,del(5)(q22q33),hsr(11)(p15),der(17;22)(q10;q10)[17]/45,XY,idem,hsr(11)(p15)(q23)[3]	1	0	0	0	1.5	1	202	1	104	0	0
335875	F	46	2	RA	46,XX[20]	0	0	0	0	0.0	0	1387	0	1229	0	0
337081	M	67	10	RAEB	46,XY[20]	0	0	0	0	0.5	1	1108	0	31	0	0
342760	F	77	12	RAEB	46,XX[20]	0	0	0	0	1.5	1	1447	0	1447	0	0
343306	F	62	15	RAEB	40-45,XX,-4,-5,add(10)(p15),add(15)(p11.2),-17,+add(19)(p13.3),add(21)(p11.2),-22,+mar[cp3]/46,XX[10]-only 13 analyzed	1	0	0	0	3.0	1	686	0	683	0	0
346313	M	62	6	RAEB	46,XY[20]	0	0	0	0	1.0	1	965	1	787	0	0
350460	M	30	4	RA	46,XY[17]	0	0	0	0	0.5	1	905	0	904	0	0
362699	F	73	2	RA	46,XX[18]/47,XX,+4[1]/47,XX,+9[1]	0	0	0	0	0.5	1	307	0	127	0	0
369682	M	59	ND	RA	45,XY,der(5)t(3;5)(q21;q13)[9]/46,idem,+8[6]/46,idem,+mar[1]/46,XY[4]	0	0	0	1	UK	1	84	1	81	0	0
372440	M	59	2	RA	46,XY[20]	0	0	0	0	0.0	0	1891	0	1894	0	0
379929	F	39	3	RA	46,XX[20]	0	0	0	0	0.5	1	290	1	224	1	0
391828	M	50	3	RA	46,XY,inv(3)(q21q26.2)[6]/46,XY[14]	0	0	0	0	1.0	1	1852	0	1852	0	0
392543	M	74	13	RAEB	55-57,XY,+1,+2,+6,der(7)(7;11)(p22;q13),+9,-10,+11,+13,+14,add(14)(p11.2),add(17)(p11.2),-18,+21,add(21)(p11.2),+22,+1-2r,+1-3mar[cp7]/46,XY[13]	0	0	0	0	2.5	1	599	1	589	0	0
395621	F	35	6	RAEB	46,XX,der(15)t(1;15)(q10;q10)[15]/46,XX[5]	0	0	0	0	1.0	1	541	1	140	0	0
411330	M	83	2	RA	45,X,-Y[3]/46,XY[17]	0	0	0	0	0.0	1	18	0	18	0	0
421452	F	33	10	RAEB	46,XX[20]	0	0	0	0	1.0	0	2016	1	90	0	0
429531	F	38	0	RA	46,XX,del(5)(q12q33)[18]/46,XX[2]	1	0	0	0	0.5	0	2951	0	2577	0	0
430998	M	56	0	RA	47,XY,+14[5],46,XY,t(14;14)(q10;5)/46,XY[10]	0	0	0	0	0.5	0	1067	0	706	0	0
431137	M	55	7	RAEB	46,XY,add(3)(p25),del(3)(p13),del(6)(q15q31),-6,+8,add(12)(p11.2),add(15)(p11.1),+18,+22[cp4]/44,sl,-8,-20,-22,+mar[cp5]/44,sl,-del(3),-7,+2mar[cp5]/43,sl,add(XX)(q28),+add(12p),+12,-mar[cp6]	1	1	1	1	2.0	1	181	0	181	0	0
431586	M	68	6	RAEB	87,XY,+X,+1,+1,+2,+2,+3,+3,+4,+4,+5,+5,+6,+6,+7,+7,+8,+8,+9,+9,+10,+10,+11,+11,+12,+12,+13,+13,+14,+14,+15,+15,+16,+16,+17,+17,+18,+18,+19,+19,+20,+20,+21,+21,+22,+22[3]/46,XY[22]	0	0	0	1	1.5	1	337	1	275	0	0
447189	M	76	0	RA	46,XY[20]	0	0	0	0	0.5	0	1795	0	1436	0	0
455113	F	68	2	RA	46,XY[20]	0	0	0	0	0.0	0	2244	0	1982	0	0
455194	M	62	2	RA	47,XY,+8[19]/46,XY[1]	0	0	0	1	0.5	0	1529	0	1302	0	0
457721	M	63	22	RAEB-T	46,XY[20]	0	0	0	0	2.5	1	398	1	173	1	0
461282	M	65	6	RAEB	46,XY,-17,del(20)(q11.2),r(5)/46,idem,del(5)(q31q33)[10]/46,idem,del(5)(q13q33)[3]/46,idem,-X,add(X)(q22),der(12)t(12;7)(p11.2;7)del(12)(q22)[2]	1	1	0	0	2.0	1	2009	1	1751	0	0
461321	F	78	6	RAEB	46,XX[18]	0	0	0	0	0.5	0	981	1	178	0	0
462596	M	78	0	RA	46,XY[20]	0	0	0	0	0.5	1	395	0	177	0	0
463763	M	54	0	RA	47,XY,+Y[6]/46,XY[14]	0	0	0	0	0.5	0	1385	0	1332	0	1
485523	M	62	11	RAEB	46,XY[20]	0	0	0	0	2.0	0	814	0	804	0	0
497942	M	38	0	RA	46,XY,der(1;22)(q10;q10)[4]/46,XY[16]	0	0	0	0	1.0	1	4626	0	4553	0	0
511734	M	69	4	RAEB-T	46,XY[20]	0	0	0	0	0.0	1	353	0	353	0	0
518747	F	47	6	RAEB	46,XX[11]	0	0	0	0	1.0	0	844	0	816	0	0
530722	M	69	13	RAEB	46,XY[20]	0	0	0	0	2.0	1	311	1	24	0	0
530724	F	67	11	RAEB	Failed	0	0	0	0	UK	1	32	0	12	0	0
537426	F	57	2	RA	45,XX,-7[20]	0	0	1	0	1.5	0	1908	0	1833	0	0
538267	F	49	12	RAEB	42-48,XX,-X,(3;17)(q21;q21),del(5)(q15q33),-10,t(2;18)(q11.2;q11.2),-12,-13,t(17;20)(q21;11.2)add(17)(p11.2),dic(19;21)(p13q22),+r,+mar[cp18]/46,XX[2]	1	0	0	0	3.0	1	407	0	407	0	0
543465	M	75	11	RAEB	46,XY[20]	0	0	0	0	2.0	0	3080	1	160	0	0
547403	M	61	16	RAEB	46,X,-Y,+14[2]/45,X,-Y[2]/45,XY,-20[1]	0	0	0	0	2.5	1	132	0	132	0	0
554562	M	80	0	RA	46,XY,del(20)(q11,q13.3)[8]/46,XY,+8[3]/46,XY[9]	0	1	0	0	0.5	1	1256	1	1314	0	0
557468	M	76	18	RAEB	46,XY[20]	0	0	0	0	2.0	1	712	1	217	0	0
558896	M	73	15	RAEB	47-50,XY,t(2;19)(p16;p13),del(5)(q12q33),-11,-18,-20,+r,+2-6mar[4]/41-50,sl,+5,del(5)(q12q33)[8]/47-50,sl,t,r[8]	1	1	0	0	3.0	1	57	0	57	0	0
571656	M	71	4	RAEB	46,del(20)(q11.2q13.3)[12]/46,XY[8]	0	1	0	0	0.5	1	303	1	231	0	1
574490	M	57	1	RA	46,XY[20]	0	0	0	0	0.0	1	874	0	874	0	0

643607	F	79	3	RA	46.XX[20]	0	0	0	0	0.0	1	897	0	14	0	0
657314	F	28	4	RAEB	45.XX-7[5]	0	0	1	0	1.5	0	1704	0	1681	0	0
667720	F	66	10	RAEB	46.XX[20]	0	0	0	0	1.0	1	724	1	644	0	0
672525	F	73	5	RAEB	46.XX[20]	0	0	0	0	1.0	1	2395	0	2247	0	0
684211	F	62	1	RAR	46.XX.del(5)(q13q33).del(20)(q11.2)[cp5]	1	1	0	0	0.5	1	325	1	231	0	0
690100	M	66	15	RAEB	43-45.XY.del(5)(q22q35).der(16)(16.17)(q22;q21).-17.add(17)(p11.2).-18.dic(18;21)(p11.1;q22).der(22)(1:22)(p13;p11.2).+0-2mar[cp19]/47.XY.+12[1]	1	0	0	0	3.0	1	331	1	324	1	0
693881	F	78	11	RAEB	44.XX.-3.del(5)(q13q33).-7.add(9)(q22).add(13)(p11.2).-18.add(18)(p11.2).-20.+2mar[18]/46.XX[2]	1	1	1	0	3.0	1	383	0	369	0	0
696026	M	48	0	RA	47.XY.+8[2]/47.idem.add(3)(q21)[6]/47.add(3)(q21).del(12)(p11.2p13)[12]	0	0	0	1	1.5	0	4566	0	4498	0	0
697296	M	71	3	RA	46.XY[20]	0	0	0	0	0.5	1	1385	0	87	0	0
698840	F	77	13	RAEB	46.XX.del(5)(q13q23)[1]42-47.si.dic(15;20)(p11.2;q13.3).add(22)(p11.1)[cp3]/44-47.si.dic(3;15)(p11.2;q11.1).der(14;22)(q10;q10).+0-5mar[cp6]/82-87.sbx2.-3.del(4)(q727)x2.-7.add(12)(p11.2)x2.der(15;21)(q10;q10).der(15;22)(q10;q10).-17.-17.+19.-21.add(21)(p11.2).-22.-22.+0-5mar[cp9]/46.XX[1]	0	0	1	0	3.0	1	253	0	7	0	0
703653	F	20	0	RA	46.XY[20]	0	0	0	0	0.5	0	2257	0	2092	0	0
705782	M	34	5	RAEB	46.XY.[3;5](q21;q31)[13]/46.idem.?add(12)(q24.3)[5]/46.XY.?add(12)(q24.3)[2]	0	0	0	0	2.0	0	1711	0	1703	0	0
717045	F	48	2	RA	46.XX.del(5)(q13q33)[20]	1	0	0	0	0.5	1	393	0	393	0	0
732776	F	47	8	RAEB	46.XX[20]	0	0	0	0	1.0	0	2255	0	11	0	0
736895	M	54	2	RA	43-49.XY.-4.-5.-7.add(12)(p11.2).-20.-21.+1-6mar[cp7]	1	1	1	0	1.5	1	38	0	26	0	0
747967	M	41	2	RA	46.XY.+1.der(1:15)(q10;q10)[8]/46.XY.+1.der(1:7)(q10;p10)[4]/46.XY[8]	0	0	0	0	1.0	0	1940	0	1899	0	0
753222	M	79	0	RA	60-61.XXY.add(3)(p21).del(5)(q13q33).add(6)(q25).-7.-8.-9.add(14)(p11.2).-16.-17.+19.add(19)(q13.4)x2.-22[cp2]/46.XY[68]	1	0	1	0	1.5	1	123	0	123	0	0
783437	M	63	8	RAEB	46.XY[20]	0	0	0	0	1.0	1	1095	1	594	0	0
785104	F	44	2	RA	46.XX[20]	0	0	0	0	0.0	0	1317	0	1172	0	0
791334	F	66	3	RA	46.XX[20]	0	0	0	0	0.5	1	923	0	923	0	0
804059	M	69	9	RAEB	47.XY.+8[20]	0	0	0	0	1.5	1	878	1	287	0	0
809605	M	75	14	RAEB	46.XY[12]	0	0	0	0	2.0	0	849	0	849	0	0
817019	F	65	3	RA	46.XX.idel(20)(q11.2)[9]/46.XX[11]	0	1	0	0	0.0	0	1172	0	861	0	0
827584	M	62	8	RAEB	47.XY.+8[15]/46.XY[5]	0	0	0	1	1.0	0	1710	0	1703	0	0
843623	M	60	6	RAEB	46.XY.del(20)(q11.2q13.3)[2]/46.XY[18]	0	1	0	0	1.0	1	139	1	71	0	1
845446	M	73	13	RAEB	47.XY.+8.del(12)(p11.2p13)[18]/47.idem.del(3)(q21)[2]	0	0	0	1	3.0	1	251	0	251	0	0
846155	M	64	0	RA	46.XY[20]	0	0	0	0	0.5	0	1023	0	1026	0	0
854400	M	63	0	RA	46.XY.Yqh+20]	0	0	0	0	0.0	1	945	0	923	0	0
855934	M	62	26	RAEB-T	46.XY[20]	0	0	0	0	2.5	0	453	0	453	0	0
857645	M	60	ND	RA	46.XY[20]	0	0	0	0	UK	1	860	1	821	0	0
859640	F	64	3	RA	46.XX[20]	0	0	0	0	1.5	1	609	1	252	0	0
861691	M	62	13	RAEB	45.XY.-7[15]/46.XY[5]	0	0	1	0	3.0	0	3144	0	118	0	0
875301	F	62	14	RAEB	46.XX[20]	0	0	0	0	1.5	1	463	1	81	0	0
879047	F	74	17	RAEB	46.XX.del(11)(q21q24)[13]/46.XX[7]	0	0	0	0	2.5	1	980	0	980	0	0
882525	M	57	7	RAEB	41-49.XY.+X.+X.del(5)(q22q35).del(7)(q22).+8.-12.add(12)(p12).-13.-18.+1-4mar[cp20]	1	0	1	1	2.0	1	148	1	88	0	0
889352	M	72	14	RAEB	39-44.XY.del(4)(q31)[14].der(5)(5:7)(q21:?) [17].del(6)(q21)[19].-7[19].add(12)(p13)[18].-18[18][cp19]	0	0	1	0	3.0	1	42	0	42	0	0
911999	M	36	4	RA	46.XY[20]	0	0	0	0	0.0	0	3250	0	3232	0	0
912290	M	59	1	RA	46.XY[20]	0	0	0	0	0.5	1	696	0	696	0	0
917011	M	79	0	RA	46.XY.del(20)(q11.2)[12]/46.XY[8]	0	1	0	0	0.5	1	809	0	769	1	1
936943	M	43	ND	RAEB	failed.	0	0	0	0	UK	1	1300	0	1289	0	0
937954	M	51	12	RAEB	47.XY.+8[1]/47.idem.del(16)(q12.1)[13]/48.idem.+Y.del(16)(q11.1)[5]/46.XY[1]	0	0	0	1	2.5	1	729	0	131	0	1
943264	M	65	11	RAEB	46.XY[20]	0	0	0	0	1.5	0	891	0	848	0	0
944541	M	54	20	RAEB	45.XY.-7[19]/46.XY[1]	0	0	1	0	3.0	1	1630	0	1630	0	0
947519	M	59	7	RAEB	46.XY.1-5dms[10]/46.XY[12]	0	0	0	0	1.5	1	734	1	699	0	1
949197	F	60	2	RA	failed	0	0	0	0	UK	1	108	0	89	0	0
958595	F	82	4	RA	46.XX.del(11)(q23)[20]	0	0	0	0	1.0	1	975	0	975	1	0
973424	M	56	3	RA	46.XY[20]	0	0	0	0	0.0	0	1228	0	1027	0	0
975079	M	62	0	RA	46.XY[20]	0	0	0	0	0.0	1	1563	0	1563	1	0
983677	F	60	8	RAEB	46.XX.del(5)(q21q35)[4]/69.XX.+X.+1.+1.+2.+del(5)(q21q35).+5.+6.+8.+9.+9.+10.+11.+11.+13.+13.+14.+14.+16.+16.+19.+19.+20.+21.-22.+mar1.+mar2[7]/46.XX[9]	1	0	0	1	2.0	1	85	1	44	0	0
987089	F	64	3	RAEB	56-58.XX.+X.+del(3)(p12).del(5)(q13q33).+6.+8.+der(8)add(8q22).+10.+11.+13.+14.-17.+19.+21.+mar[cp20]	1	0	0	1	2.0	1	762	1	678	0	0
988428	M	70	2	RAR	47.XY.-9.+2mar[1]/46.XY[19]	0	0	0	0	0.5	1	1975	0	1891	1	0
989739	M	71	18	RAEB	46.XY.del(7)(q21)[11]/47.XY.+8[5]/46.XY[4]	0	0	1	1	3.0	1	468	1	125	1	1
994096	M	72	3	RA	46.XY.del(5)(q14q34).+8.del(11)(q21q23)[19]/46.idem.del(17)(q24)[1]	1	0	0	1	1.0	1	1307	0	1178	0	0
999103	F	66	6	RAEB	46.XX.del(5)(q15q13)[20]	1	0	0	0	1.0	1	4813	0	2839	0	0
01-13	F	41	12	RAEB	46.XX.inv(3)(q21q26)[13]/46.idem.t(14;17)(q13;p13)[1]	0	0	0	0	2.0	1	1649	1	1301	0	0
02-21	M	57	3	RA	46.XY.t(3;12)(q27;p13)[20]	0	0	0	0	1.0	1	709	0	709	0	0
02-28	M	60	1	RA	46.XY.t(5;15)(p10;q10)[2]/46.XY[18]	0	0	0	0	1.0	1	76	0	76	0	0
02-30	M	72	2	RA	46-48.XY.add(5)(q11.2).-7oradd(7)(q11.2).add(8)(p22).del(13)(q21).add(16)(q722).del(22)(q12).+1-3mar[cp19]/46.XY[1]	0	0	1	0	1.5	1	239	1	205	0	0
02-75	F	46	10	RAEB	46.XX[20]	0	0	0	0	0.5	0	3247	0	3014	0	0
02-77	M	67	28	RAEB-T	46.XY[20]	0	0	0	0	2.0	1	1863	1	1662	0	0
02-86	M	53	21	RAEB-T	92-<4n>.XXYY.del(4)(q33)x2.del(5)(q13).add(11)(q23)x2[cp8]/46.XY[12]	1	0	0	0	3.0	1	308	1	133	0	1
02-99	M	66	16	RAEB	46.XY.del(2)(q33).del(5)(q15q33).-7.+del(11)(q273)[cp6]	1	0	1	0	3.0	1	945	0	933	0	0

Supplementary Table 8. Primers used for U2AF1 sequencing.							
Ensembl TranscriptID	Chromosome	Exon#	Exon Start	Exon Stop	Primer 1 Sequence*	Primer 2 Sequence	Ensembl ExonIDs
gDNA primers: targets U2AF1 hotspot only							
ENST00000398137	21	2	43397494	43397581	tgctactctAAACAAACCTGGTAAACGTCG	acatgatgcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	acgactacagAAACAAACCTGGTAAACGTCG	tgctgatgcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	cgtgactagAAACAAACCTGGTAAACGTCG	gcatctgacAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	taacgatgatAAACAAACCTGGTAAACGTCG	atgctcacaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	tactctggtAAACAAACCTGGTAAACGTCG	atgagagcacAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	tagagacagAAACAAACCTGGTAAACGTCG	atctctgctcAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	tcgtctgctAAACAAACCTGGTAAACGTCG	agcagcagcAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	acatacagctAAACAAACCTGGTAAACGTCG	tgatgacgcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	acgcgactatAAACAAACCTGGTAAACGTCG	tgctcgataAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	actactatgtAAACAAACCTGGTAAACGTCG	tgatgatacaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	actgacagtAAACAAACCTGGTAAACGTCG	tgacatgcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	agacataactAAACAAACCTGGTAAACGTCG	tcgtgatgaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	agrcctgctAAACAAACCTGGTAAACGTCG	tcgcagcagaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	agtcactatAAACAAACCTGGTAAACGTCG	tcactcgataAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	atagatgactAAACAAACCTGGTAAACGTCG	tatctcatgaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	caagctacgAAACAAACCTGGTAAACGTCG	gtagcagcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	cgactagcgtAAACAAACCTGGTAAACGTCG	gctcatcgcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	cgactgagcaAAACAAACCTGGTAAACGTCG	gctgcaactgaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	taacacacactAAACAAACCTGGTAAACGTCG	atgtgtgtgaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	taacagtgatAAACAAACCTGGTAAACGTCG	atgtgcaactAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	taacagatgtAAACAAACCTGGTAAACGTCG	atgtctagcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	taacgtgctAAACAAACCTGGTAAACGTCG	atgagcagagaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	taactgtatAAACAAACCTGGTAAACGTCG	atcacatcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	tgactacagAAACAAACCTGGTAAACGTCG	agctagtgaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	tcgcactgtAAACAAACCTGGTAAACGTCG	agcgtgatcaAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
cDNA primers: targets U2AF1 hotspot only							
ENST00000398137	21	1-4	43393632	43400757	acaggtgctGGAGTATCTGGCCTCCATCTTC	tgctcagcaGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	acgctcgacaGGAGTATCTGGCCTCCATCTTC	tgtagctgtGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	agacgcactcGGAGTATCTGGCCTCCATCTTC	tcctgctgagGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	agcactgtgGGAGTATCTGGCCTCCATCTTC	tcgtgacatGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	atcagacagGGAGTATCTGGCCTCCATCTTC	tagctgtgctGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	atatcgcagGGAGTATCTGGCCTCCATCTTC	tatagcctcGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	cgltgctcaGGAGTATCTGGCCTCCATCTTC	gcacagagatGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	ctcgcgtgctGGAGTATCTGGCCTCCATCTTC	gagcgcagGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
ENST00000398137	21	1-4	43393632	43400757	taqtacagGGAGTATCTGGCCTCCATCTTC	atcatatcgGTCAGCAGACTGGGAAGAGTTT	ENSE00001582950, ENSE00001531800, ENSE00001531797, ENSE00001531794
3730 recurrent screening primers: targets all U2AF1 coding exons							
ENST00000398137	21	9	43386136	43386428	tgtaaacgacgcccagITGAGAGAGTGGGTGGTTCTG	caggaaacagctatgaccCCCTTATGAACCTGGTTGGTCAAT	ENSE00001138177
ENST00000398137	21	9	43386136	43386428	tgtaaacgacgcccagIGAGAGTGGGTGGTTCTGTGC	caggaaacagctatgaccTCTTACGATCTCTCGACCG	ENSE00001138177
ENST00000398137	21	8	43387650	43387742	tgtaaacgacgcccagITAACCGTTGGTTAATGGACAGCC	caggaaacagctatgaccGAGGGTTGTGAGGGGAGAAAGAA	ENSE00001268168
ENST00000398137	21	7	43387834	43387967	tgtaaacgacgcccagITAACCGTTGGTTAATGGACAGCC	caggaaacagctatgaccGAGGGTTGTGAGGGGAGAAAGAA	ENSE00001050518
ENST00000398137	21	7	43387834	43387967	tgtaaacgacgcccagITAAAGTCTTATAAAGCGTGGATGCC	caggaaacagctatgaccCACTACTGACGGGAGCAGCC	ENSE00001050518
ENST00000398137	21	6	43388617	43388715	tgtaaacgacgcccagITGATCTTCTGTATCTGTTGGAAAG	caggaaacagctatgaccCACTGCTCGCTTTTCGCTG	ENSE00001050529
ENST00000398137	21	5	43388873	43388922	tgtaaacgacgcccagICTGTGTGTTACTGTAGCAGTTTGAAG	caggaaacagctatgaccACCTCCTGAAGGGAGACCACAG	ENSE00001531791
ENST00000398137	21	4	43393632	43393698	tgtaaacgacgcccagITAACTCAACTGAAGCACTTGCC	caggaaacagctatgaccCCTTGCCACCTCTATGTAACCG	ENSE00001531794
ENST00000398137	21	3	43394545	43394611	tgtaaacgacgcccagIGCCATTTGCAACAATAATGTCTCT	caggaaacagctatgaccCAGCAGCTCAGAACCAACAT	ENSE00001531797
ENST00000398137	21	2	43397494	43397581	tgtaaacgacgcccagITGAACACAAATGGAAATACAACACTACG	caggaaacagctatgaccTTCAAAATGGAGCATGCTGCTC	ENSE00001531800
ENST00000398137	21	2	43397494	43397581	tgtaaacgacgcccagITAAACAAACCTGGTAAACGTCG	caggaaacagctatgaccAATTTGATAGTGTATGTCATGCTGCTG	ENSE00001531800
ENST00000398137	21	1	43400630	43400757	tgtaaacgacgcccagITAGAGCGTCTGCTCGCTCCG	caggaaacagctatgaccCCACCCTCAACCAACC	ENSE00001582950
U2AF1 mRNA isoform expression primers							
					GCCTCCATCTTCCGACCCGA	GGCATGGCTCAGAATCGCCC	
*lower case, bar code; upper case,U2AF1 sequence							

Supplementary Table 9. Expression of Tier 1 mutated genes in sAML sample.

Chromosome	Position*	Gene name	Transcript ID	Exons	Mutant Exon	Total Probes	Present Probes				Probes in Mutant Exon	Present Probes in Mutant Exon			
							in UPN	in > 50% of AML	in > 75% of	in > 90% of AML		in UPN	in > 50% of AML	in > 75% of AML	in > 90% of AML
							266395	samples	AML samples	samples		266395	samples	samples	samples
1	112325945	KCND3	NM_004980	8	2	20	3	2	2	1	3	0	0	0	0
1	205309412	PKFFB2	NM_006212	15	11	29	2	15	9	4	1	0	1	0	0
1	229096537	ENSG00000222671	ENST00000410739	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
2	20289990	LOC100131373	XM_001720891	1	1	3	1	3	1	1	1	1	1	0	0
2	61569230	XPO1	NM_003400	25	18	38	31	33	33	33	1	1	1	1	1
3	173529456	FNDC3B	NM_022763	25	11	43	41	43	43	42	2	2	2	2	2
5	50152869	PARP8	NM_024615	26	16	31	28	30	29	27	1	1	1	1	1
5	148880083	CSNK1A1	NM_001025105	11	4	26	24	25	24	24	1	1	1	1	1
6	105713110	POPDC3	NM_022361	4	4	13	0	1	0	0	3	0	0	0	0
9	89535224	CTSL1	NM_001912	8	7	20	9	11	5	5	1	1	1	1	1
9	138867831	MAMDC4	NM_206920	27	4	49	12	13	11	9	1	0	0	0	0
9	139117274	MAN1B1	ENST00000371587	9	9	45	17	21	17	15	8	5	6	5	5
10	82288101	SH2D4B	NM_207372	7	1	17	4	6	4	1	1	0	0	0	0
11	22340860	SLC17A6	NM_020346	12	splice_site	15	2	2	0	0	NA	NA	NA	NA	NA
11	30881639	DCDC5	ENST00000406071	15	12	54	3	3	3	2	2	0	0	0	0
14	75418958	TTL5	NM_015072	32	30	50	41	45	44	40	2	2	2	2	2
16	7716860	ENSG00000209555	ENST00000386820	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
16	63589987	CDH11	NM_001797	13	4	28	5	8	4	4	2	0	0	0	0
17	5288451	DHX33	NM_020162	12	12	17	6	13	11	7	1	0	0	0	0
17	7696343	JMJD3	NM_001080424	22	19	44	19	27	22	21	1	0	0	0	0
17	29981226	TMEM132E	NM_207313	10	6	13	4	3	2	2	1	0	0	0	0
17	75696250	GAA	NM_000152	20	5	27	5	6	4	2	1	0	0	0	0
18	46165704	C18orf24	NM_001039535	7	5	14	3	6	2	2	1	0	1	0	0
19	53579461	KDELRL1	NM_006801	5	4	12	7	8	8	6	2	1	2	2	1
19	54349852	HRC	NM_002152	6	1	16	1	3	3	2	9	1	2	2	2
20	32728482	PIGU	NM_080476	12	1	18	16	16	15	12	1	1	1	1	0
20	32965857	ACSS2	NM_018677	18	7	28	18	16	11	7	2	1	1	1	0
20	51626823	ZNF217	NM_006526	5	3	15	15	15	15	12	5	5	5	5	5
21	42402818	UMODL1	NM_173568	22	10	28	4	5	4	4	1	0	0	0	0
21	43397525	U2AF1	NM_001025203	8	2	22	18	22	22	20	1	1	1	1	1

*using NCBI Build 36. NA, not available.

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