

Gene	U12-type Intron	RT-PCR	Oligo Forward (5'-3')	Oligo Reverse (5'-3')	Tan (°C)	cDNA product(s) (bp)	cDNA (aberrant) (bp)	Result in patients' blood RNA	Result in controls' blood RNA	gDNA
ARPC5	1	exons 1-3	GAAGGTGGACGTGGATGAAT	GCACCTTCAGACAAATGCTG	59	212	274	normal & aberrant (weak) U12 processing (cryptic)	normal expression and processing	No product
ARPC5L	1	exons 1-3	AATTTGTGGACGACGAGGAG	GTGAGCACITTCAGCACCCAC	59	193	112	normal & aberrant U12 processing (cryptic)	normal expression and processing	No product
ERC5	1 & 13	exons 12-15	GGCCTGAAACCTCTCTCTAA	GGAAATCAATTCGGAGCTGTG	59	333	1196	normal & aberrant U12 processing (intron retention)	normal expression and processing (lower proportion)	No product
HNRPLL	4	exons 3-6	AATCCGCTTATCCAAATACAG	CCAAACTGTGCTATTGCTATCC	59	250 & 148	164 & 67	normal & aberrant U12 processing (cryptic exon)	normal expression and processing	No product
/K	14	exons 12-16	GGACGTTGACAAAGGACCTG	CCCTTCTTGTAACTCCCTGGTC	58	246	none	normal expression and processing	normal expression and processing	No product
/K	14	exon 12-intron 14	GGACGTTGACAAAGGACCTG	GTGCTCTGGAGGAACAAG	58	none	249	U12 intron retention detected in cDNA	U12 intron retention detected in cDNA	No product
/K	14	intron 14-exon 16	TTCATGTTGGGCTCTTAGC	CCCTTCTGTTACCCTGGTC	58	none	248	U12 intron retention detected in cDNA	U12 intron retention detected in cDNA	No product
/MAPK8	6	exons 5-8	GCATGGGC TACAAGGAAAC	ACAAATCCCTTCGCCTGACTG	60	297	216	normal & aberrant U12 processing (cryptic)	normal & aberrant U12 processing (similar)	No product
NUP210	7	exons 6-9	GAAGATCAGCCAAGGGAAA	TCACCAGGTGAACAGTGAA	59	276	none	normal expression and processing	normal expression and processing	No product
NUP210L	7	exons 6-9	TGGTTCAAGGGAGAGTGACA	CCCACTCTAGACTCCATCG	59	292	none	No signal + no reads from RNAseq	No signal + no reads from RNAseq	No product
PSCK1	8	exons 7-10	TAGCC CAGAAGGCTTTTGA	GAGGTCACAGAACCCAGGTG	59	397 & 184	none	No signal + no reads from RNAseq	No signal + no reads from RNAseq	No product
RNIP3	13	exons 12-14	ATCAGAAACACAGCGGATCA	TTCCCTTCTTAGGATCTTGTTTTG	59	200	none	normal expression and processing	normal expression and processing	No product
SLC12A7	10	exons 9-12	TCTACTTCCCTCCCGTGACC	CATGCCGATGACACAGGT	60	218	none	normal expression and processing	normal expression and processing	Product
SPCS2	3	exons 1-5	CGTAGCGGCTTGTGGATAA	ACTTTGTGAACTCGGCTTCC	61	481	274, 245 & 185	normal & aberrant (high proportion) U12 processing	normal & aberrant (low proportion) U12 processing	481 (pseudogene)
SPCS3	2	exons 1-5	ACCGCCTTCAAAGACAGGAG	ACGTTCCAAAGACAGGGTCAA	62	362	414 & 211	normal & aberrant (low proportion) processing	normal expression and processing	No product
SSR3	4	exons 3-5	GAITTAGCCGCAATCTCTC	CATCTCCCTCTCTGTGCT	56	229	none	normal expression and processing	normal expression and processing	No product
UFD1L	5	exons 1-7	GTTGCATTGCCCTGAGAGGAG	CCATCACACGCAAGTTCTGTAG	60	536, 503, 403 & 248	430	normal expression with alternative transcripts	normal & weak aberrant processing	No product
UFD1L	5	exons 5-7	GCCACCTACTCCAAATTCCAA	CCATCACACGCAAGTTCTGTAG	59	160	none	normal expression and processing	normal expression and processing	No product

Supporting Table S3. List of genes with U12-type introns analyzed by RT-PCR, including primers, PCR conditions and final results