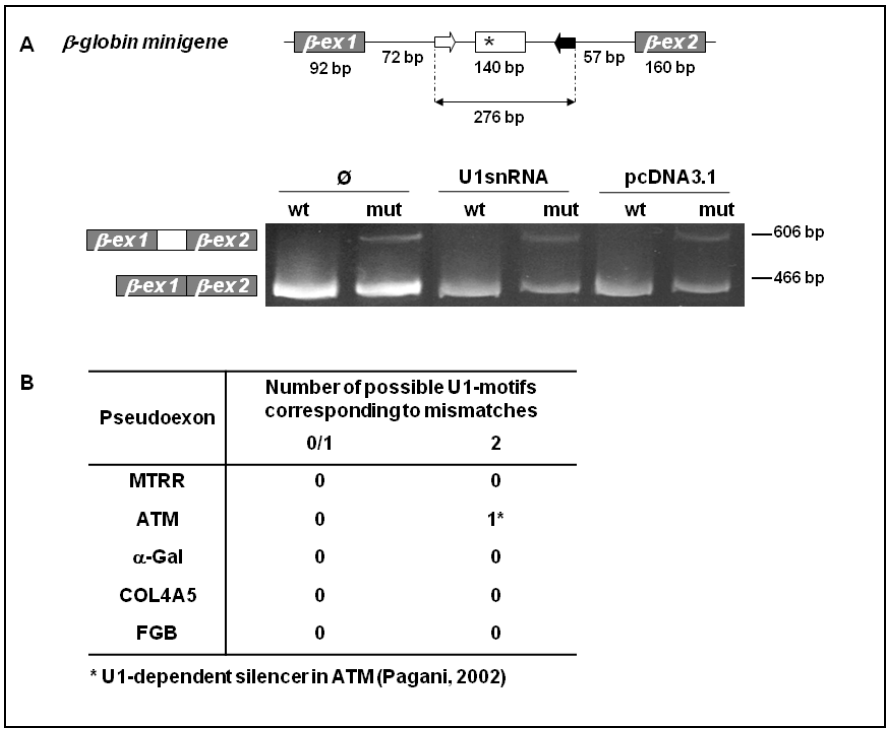

B

Pseudoexon	Number of possible U1-motifs corresponding to mismatches	
	0/1	2
MTRR	0	0
ATM	0	1*
α-Gal	0	0
COL4A5	0	0
FGB	0	0

* U1-dependent silencer in ATM (Pagani, 2002)

Supp. Figure S1. A. U1snRNA overexpression. HEK293 cells were co-transfected by the wild type or c.903+469T>C mutant β-globin minigenes and vector expressing U1 snRNA or empty expression vector *pcDNA3.1*. After RNA isolation the splicing products were analyzed by RT-PCR using minigene specific primers. The lower band represents correctly spliced minigene exons; the upper band represents the *MTRR* pseudoexon inserted between the minigene exons. **B.** U1snRNA binding motif analysis. The sequences of each pseudoexon were scanned for a presence of an U1snRNA binding site (caggtaagtat) with allowance of 0, 1 or 2 mismatches outside of the invariant +1G and +2T. The number of sites identified in the sequences of individual pseudoexons is listed in the table.

Supp. Table S1. In silico analyses of MTRR authentic and pseudoexon splice sites

Program	SpliceSiteFinder			NNSplice		GeneSplicer		NetGene2		SpliceView		MaxEnt	
exon	**5'SS**	**3'SS**	**branch point**	**5'SS**	**3'SS**	**5'SS**	**3'SS**	**5'SS**	**3'SS**	**5'SS**	**3'SS**	**5'SS**	**3'SS**
2[$]	84.3			n.f.		n.f.		0.89		83.0		6.12	
3	73.2	81.8	100.0	0.93	0.95	n.f.	7.018	0.30	0.33	79.00	81.00	5.68	6.16
4	87.6	82.9	81.0	0.99	0.92	n.f.	5.548	0.93	0.77	86.00	82.00	9.16	8.49
5	95.4	92.7	77.5	1.00	0.89	n.f.	5.445	0.67	0.15	90.00	84.00	11.08	6.49
6	79.4	87.4	88.9	0.98	0.73	n.f.	3.262	0.83	0.36	78.00	83.00	9.14	7.54
pseudoexon	84.1	96.9	76.0	0.90	0.98	n.f.	12.582	n.f.	0.94	83.00	92.00	7.09	10.41
7	80.1	81.1	75.1	0.89	0.90	7.318	5.882	0.63	0.07	85.00	86.00	8.92	7.82
8	74.3	77.6	74.0	0.71	n.f.	n.f.	1.498	0.34	0.56	77.00	77.00	7.64	5.39
9	88.5	86.4	71.6	0.99	0.62	9.370	n.f.	0.47	0.16	87.00	82.00	11.11	6.17
10	68.4	86.9	81.0	0.76	0.98	n.f.	n.f.	0.71	0.94	75.00	89.00	7.52	10.41
11	94.4	87.5	98.0	1.00	0.90	4.914	5.540	0.83	0.15	89.00	85.00	10.57	10.37
12	88.7	78.8	88.9	0.99	n.f.	n.f.	5.457	0.54	n.f.	85.00	n.f.	7.76	6.93
13	69.7	86.7	89.2	0.74	0.99	n.f.	7.196	0.55	0.82	74.00	89.00	8.35	9.09
14	88.9	84.3	83.9	0.97	0.62	n.f.	n.f.	0.95	0.16	84.00	79.00	8.73	7.89
15		87.5	68.4		0.98		5.700		0.96		89.00		9.52
median	84.2	86.6	81.0	0.97	0.91	7.32	5.55	0.67	0.36	83.50	84.00	8.54	7.89

SS = splice site; n.f. = not found

[$]The initiation codon of cytosolic isoform is harboured in exon 2, exon 1 is alternatively spliced and not translated.

Reference sequence of the cytosolic isoform of the *MTRR* gene, GenBank Accession No. NC_000005.9 (g.DNA), was used for *in silico* analysis. All nucleotide numbering regarding the *MTRR* gene (NM_002454.2) reflects cDNA numbering with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence, according to journal guidelines (www.hgvs.org/mutnomen). The initiation codon is codon 1.