Supplementary Table 1. Percentage of mutant mRNA (cDNA) with respect to wild-type mRNA in patient cell lines and the effect of different NMD inhibitors

Cell line	Mutation	Amount of mutant PRPF31 mRNA (% of wild type allele)			
		Untreated	Emetine	Cycloheximide	Wortmannin
14523	c.177+1delG	19.60 ± 1.11	9.02 ± 0.53	11.40 ± 2.24	5.21 ± 0.41
14686	c.319C>G	6.69 ± 0.12	18.20 ± 0.42	35.98 ± 2.48	21.73 ± 1.10
14266	c.323-2A>G	6.54 ± 0.94	49.14 ± 1.84	31.83 ± 2.96	29.83 ± 1.36
12688	c.856-2A>G	10.40 ± 0.51	33.60 ± 0.68	29.04 ± 0.68	51.20 ± 2.17
13190	c.856-2A>G	10.20 ± 0.37	31.20 ± 0.49	38.11 ± 0.95	N.D.
14284	c.877_910del	11.96 ± 0.13	48.32 ± 0.27	35.04 ± 2.08	44.84 ± 2.36
AG0293	c.1115_1125del	9.17^{AB} ± 0.30	55.47^{AB} ± 0.64	25.84^{AB} ± 0.53	
		15.41^{AC} ± 0.44	17.85^{AC} ± 0.89	17.07^{AC} ± 0.94	
AG0305	c.1115_1125del	5.38^{AB} ± 0.10	37.05^{AB} ± 0.26	21.00^{AB} ± 0.68	89.50^{AB} ± 2.44^{D}
		13.46^{AC} ± 0.51	16.94^{AC} ± 0.78	15.74^{AC} ± 0.65	N.D.
AG0307	c.1115_1125del	7.80^{AB} ± 0.37	69.93^{AB} ± 3.05	20.22^{AB} ± 0.31	
		12.79^{AC} ± 0.41	16.11^{AC} ± 0.21	15.49^{AC} ± 0.24	

N.D.: not determined. Uncertainties are expressed as standard errors (standard deviations of the means). [A]: measured only with the Biocalculator software, [B]: short form mutant mRNA with exon 11 skipped (NMD-sensitive), [C]: long form mutant mRNA containing the 11 bp deletion (NMD-insensitive), [D]: wortmannin treatment was performed on an equal number of pooled cells.

Supplementary Table 2. PCR primers used in this study

Mutation	Target	Primers (S: sense, A: antisense)	Location
c.177+1delG	cDNA from wild type and mutant mRNA	5'-ACAGTGGTGCGCGGAGAG-3' (S)[A]	Exon 1
		5'-CGGCCTCCACTGGTCCC-3' (A)[A]	Exon 4
c.319C>G	cDNA from wild type and mutant mRNA	5'-CTGAATACCGCGTCATCGTG-3' (S)[A]	Exon4
		5'-GTGGAGATCGAAAACGAGACAT-3' (Supp. S)[A]	Exon junction 4-5
		5'-CCAGTTCAGGGAATCTCTTTGAG-3' (A)[A]	Exon 5
c.323-2A>G	cDNA from wild type and mutant mRNA	5'-CCTGAATACCGCGTCATCGT-3' (S)[A]	Exon 4
		5'-CTTGCACTTGTCCAGGCTGTT-3' (A)[A]	Exon 6
	cDNA from wild type mRNA	5'-TACATCCGCACGGTCAAGG-3' (S)[B]	Exon junction 5-6
		5'-CGACAGCTGCTGCCCCT-3' (A)[B]	Exon junction 6-7
	cDNA from mutant mRNA	5'-TCGAAAACGAGCTGAGAGCTG-3' (S)[B]	Exon junction 4-6
		5'-CGACAGCTGCTGCCCCT-3' (A)[B]	Exon junction 6-7
c.856-2A>G	cDNA from wild type and mutant mRNA	5'-GACATCGTGCAGTCCCTGC-3' (S)[A]	Exon 8
		5'-GTGTGCACTTGGCGGCC-3' (A)[A]	Exon 9
c.877_910del	cDNA from wild type and mutant mRNA	5'-ACATCGTGCAGTCCCTGCC-3' (S)[A]	Exon 8
		5'-CCCTTCTGTGCTCTCGTGG-3' (A)[A]	Exon 9
c.1115_1125del	cDNA from wild type and mutant mRNA	5'-CACGAGAGCACAGAAGGGAAG-3' (S)[A]	Exon 9
		5'-GTGGCCCAGGCTGAATCC-3' (A)[A]	Exon 12
	cDNA from long mutant mRNA form	5'-GGCTGACGGAGATCCAACC-3' (S)[B]	Exon 11 with del
		5'-GTGGCCCAGGCTGAATCC-3' (A)[B]	Exon 12
	cDNA from wild type PRPF31 mRNA	5'-GAGATCCGGAAGCAGGCC-3' (S)[B]	Exon 11
		5'-GTGGCCCAGGCTGAATCC-3' (A)[B]	Exon 12
	cDNA from wild type and mutant pre-mRNA	5'-TGGGGCTGACGGAGATCC-3' (S)[B]	Exon 11
		5'-GGCTGTGGGGTTGAGGAG-3' (A)[B]	Intron 11-12
	cDNA from wild type pre-mRNA	5'-GAGATCCGGAAGCAGGCC-3' (S)[B]	Exon 11
		5'-GGCTGTGGGGTTGAGGAG-3' (A)[B]	Intron 11-12
	cDNA from mutant pre-mRNA	5'-GGCTGACGGAGATCCAACC-3' (S)[B]	Exon 11 with del
		5'-GGCTGTGGGGTTGAGGAG-3' (A)[B]	Intron 11-12
Endogenous control	18s rRNA	5'-CGGCTACCACATCCAAGGAA-3' (S)[B]	
		5'-GCTGGAATTACCGCGGCT-3' (A)[B]	

[A]: Primers used in semi-quantitative RT-PCRs to amplify cDNA regions encompassing mutations, [B]: Primers used in real-time PCR.
(S) = sense oligo, (A) = antisense oligo