

LOF containing genes predicted to undergo NMD

chr	pos	ref_rea	alt_reac	gene	effect	pos_along_cds
12	52863985	2	4	SMUG1	full	0.01
3	51980678	8	1	ABHD14B	full	0.08
11	113776207	20	12	C11orf71	full	0.13
19	54861076	14	13	BCL2L12	full	0.18
6	26077610	6	0	TRIM38	full	0.35
19	4279755	24	0	STAP2	full	0.38
11	2278405	3	3	C11orf21	full	0.39
4	113571937	5	1	ALPK1	full	0.48
16	20699889	11	0	ACSM3	full	0.50
19	17698952	5	3	MAP1S	full	0.55
17	77955236	5	3	C17orf101	full	0.57
1	45735570	6	1	RP11-291L19.	full	0.57
14	23749768	31	0	CHMP4A	full	0.67
4	130250213	11	3	AC093826.1	full	0.72
5	168960999	23	0	CCDC99	full	0.81
1	157084221	17	0	MNDA	full	0.87
19	53429518	56	64	CARD8	partial	0.02
19	53429518	57	60	CARD8	partial	0.02
9	113944533	8	9	SUSD1	partial	0.04
12	120274428	25	25	ANAPC5	partial	0.04
6	31232828	66	39	CCHCR1	partial	0.09
6	74076059	3	2	KHDC1	partial	0.14
15	66284651	12	6	CALML4	partial	0.20
16	88172869	6	5	CPNE7	partial	0.24
22	25192041	4	1	HPS4	partial	0.33
21	43196789	17	1	NDUFV3	partial	0.42
1	234772923	93	3	LGALS8	partial	0.59
17	71589392	67	70	ZACN	partial	0.68
17	71589392	79	61	ZACN	partial	0.68
11	6548630	3	3	DNHD1	partial	0.93

BCL2L12

AAGTAAGTATCCCCACTTTCAGGGTAGCATCCTCTTTCCCTAGTTATTTTCAGTGCAGAATCGAT
TCACGTGTAAGCCTCTACCTCCTGCACTTACCCCAATGTAGACCCCTTCCAAGTCCCCCTAC
AGAAGATCTCCCAAAACATTTCCAACACAGAACTTCCATTCAATTCCCCTCCGACTGCCCGCC
TCCCTCCTCCAGGACAGAGCACGCCCCAGGTTTCGAGACTGAACTACCCCCATCTACGGAGTC
CACCCCTCCCTCTCTAGAACCCCTCAGTGTAGACGCCTCCTCTTTTCTCTTTCCCTCCCTACTT
TTTCTAGTTTTTCTGCAGCCGACCTCCAGCGTCGGCCACTGTAGCTCCTTCCTTGCTCCACTTC
CCCAGAGTACACAGCCTGCCTTTCCACTCTCCGTGCAGACCCATCCTCTTTCCCGCTCCTCGCT
CTCGGACGCCACCAACGCTCTCCCGGGCTCTCCGCGTTACCT **ACCATGG**AAGGTCGGG
GCGTGCGGGCAGCTGGAACCCACCCCTGTCTTGGAGCTCCGGGTAGCTCTCA
AACTCGAGGCTGCGCACCCCTTCCCGTCAGCTGAGCTCTAGAGCGCTGGG
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AACAGACCCAAAAG **CCG**

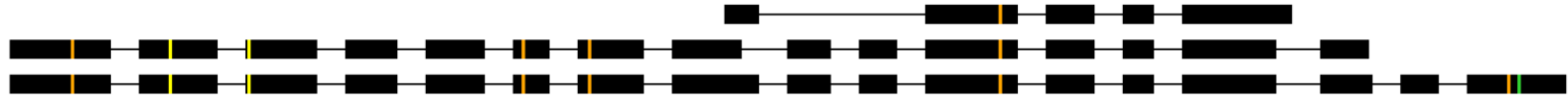
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GGAAAGTTGAACTAATAAAGTTTGTA **C**GAGTTCAGTGGAGGAGACCGCAAGT
TGAGTGGAGGAGGCGGCGGTGGGGCCCCGGACCAGGTGCC **TCCATGG**CAGG
CTCTGAAGAGCTGGGGCTCCGGGAAGACACGCTGAGGGTCTAGCTGCCTTC
CTTAGGCGTGGTGAGGCTGCCGGGTCTCCTGTTCCA ACTCCACCTAGA
AGCCCTGCCAAGAAGAGCCAACAGACTTCCTGAGCCGCCTTCGAAGATGTC
TTCCCTGCTCCCTGGGGCGAGGAGCAGCCCCCTCTGAGTCCCCTCGGCCTTGC
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CCAGAAGCTGGCCTCGGACCCCGCCCTGCGCAGCAAGCTGGTCCGCCTGTCC
TCCGACTCTTTCGCCCGCCTGGTGGAGCTGTTCTGTAGCCGGGATGACAGCTC
TCGCCAAGCCGAGCATGCCCGGGCCCCCGCCTCCTTCCCCGGAGCCCCTG
GCCCGCCTGGCCCTA **GCCATGG**AGCTGAGCCGGCGCGTGGCCGGGCTGGGG
GGCACCTGGCCGACTCAGCGTGGAGCACGTGCACAGCTTCACGCCCTGGA
TCCAGGCCACGGGGGCTGGGAGGGCATCCTGGCTGTTTACCCGTGGACTT
GAACTTGCCATTGGACTGA

SNP indicated in green

Kozak consensus gccgccRccAUGG, R= A/G

ANAPC5

Graphical representation of genetic variants



LEGEND FOR VARIATION TYPES:

spliceOverlap synonymous nonsynonymous prematureStop removedStop insertionNFS insertionFS deletionNFS deletionFS

uORF overlaps with CDS, not strong Kozak

Several strong Kozak after the STOP