

Supplemental Table 1. Predicted miRNA target effects for c.*529A>G variant.

Variant	miRNA	Database	Conservation Score	Functional Effect	Context Score Change
c.*529A>G	hsa-miR-1231	PolymiRTS Database 3.0	10	D	-0.246
		MicroSNiPer	N/A	D	N/A
	hsa-miR-1299	PolymiRTS Database 3.0	11	D	-0.093
		MicroSNiPer	N/A	D	N/A
	hsa-miR-6128	PolymiRTS Database 3.0	11	D	-0.077
	hsa-miR-654-3p	PolymiRTS Database 3.0	12	D	-0.137`
		MicroSNiPer	N/A	D	N/A
	hsa-miR-875-3p	PolymiRTS Database 3.0	11	D	-0.071
		MicroSNiPer	N/A	C	N/A
	hsa-miR-3681-5p	MicroSNiPer	N/A	U	N/A
	hsa-miR-1254	PolymiRTS Database 3.0	10	C	-0.392
		MicroSNiPer	N/A	C	N/A
	hsa-miR-3116	PolymiRTS Database 3.0	10	C	-0.413
		MicroSNiPer	N/A	C	N/A
	hsa-miR-661	PolymiRTS Database 3.0	10	C	-0.15
		MicroSNiPer	N/A	C	N/A
hsa-miR-4633-5p	MicroSNiPer	N/A	C	N/A	
hsa-miR-4254	MicroSNiPer	N/A	C	N/A	

D: variant disrupts the predicted miRNA site; C: variant creates a predicted miRNA site; U: unaffected.

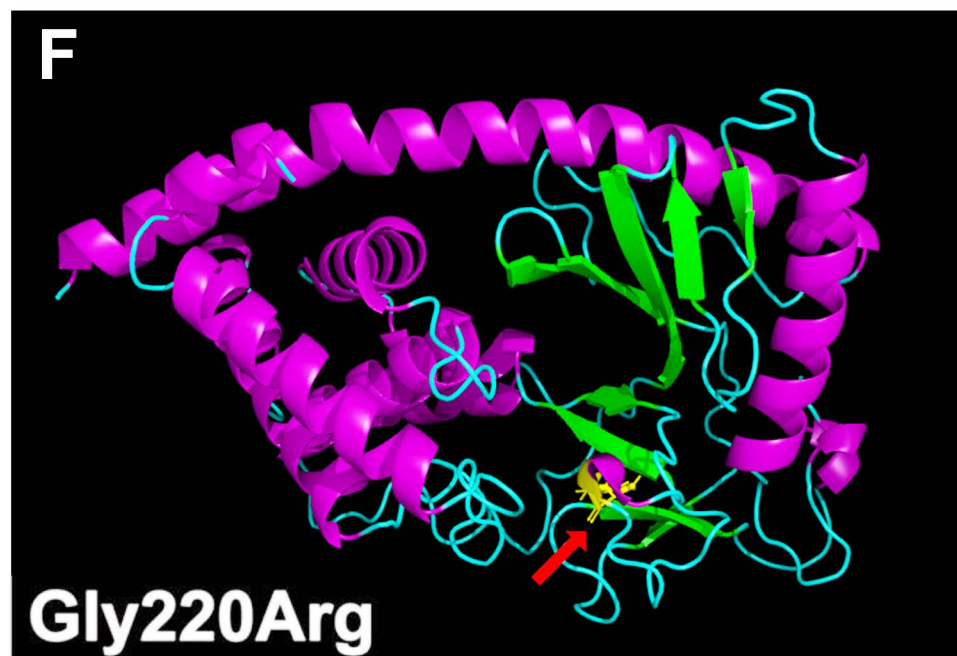
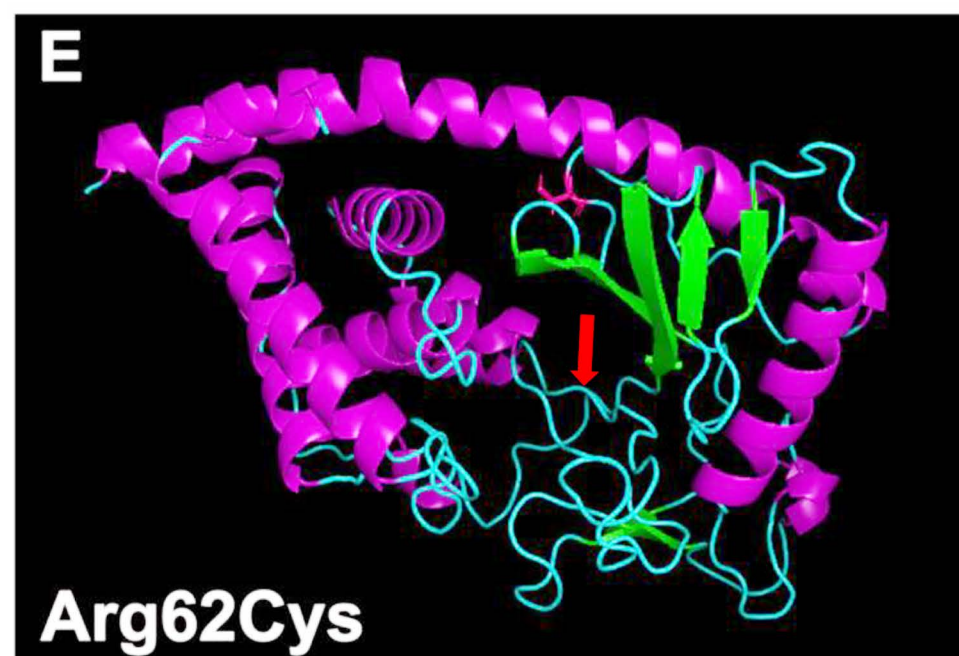
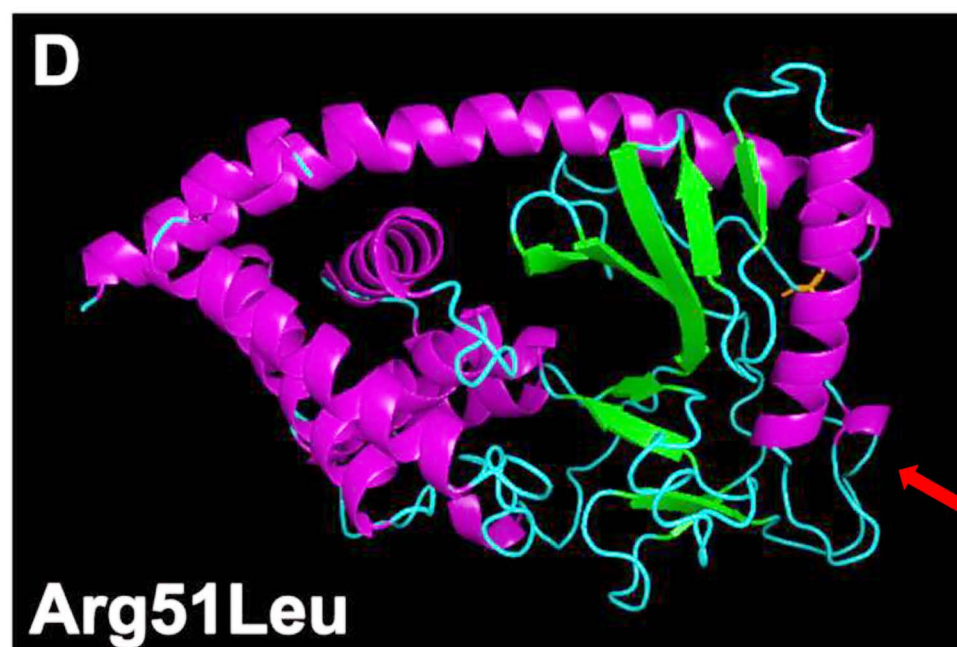
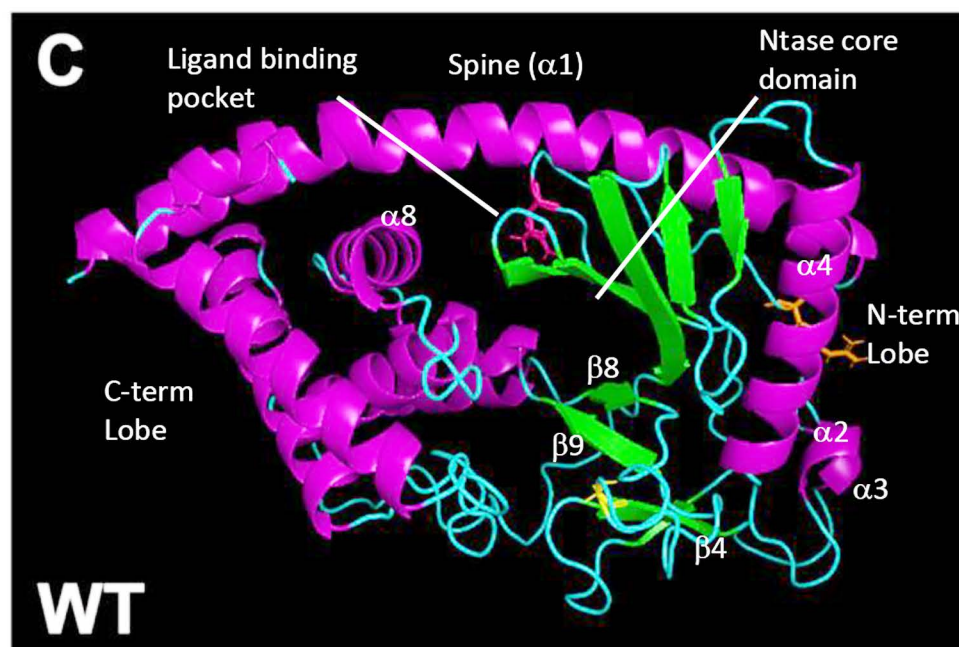
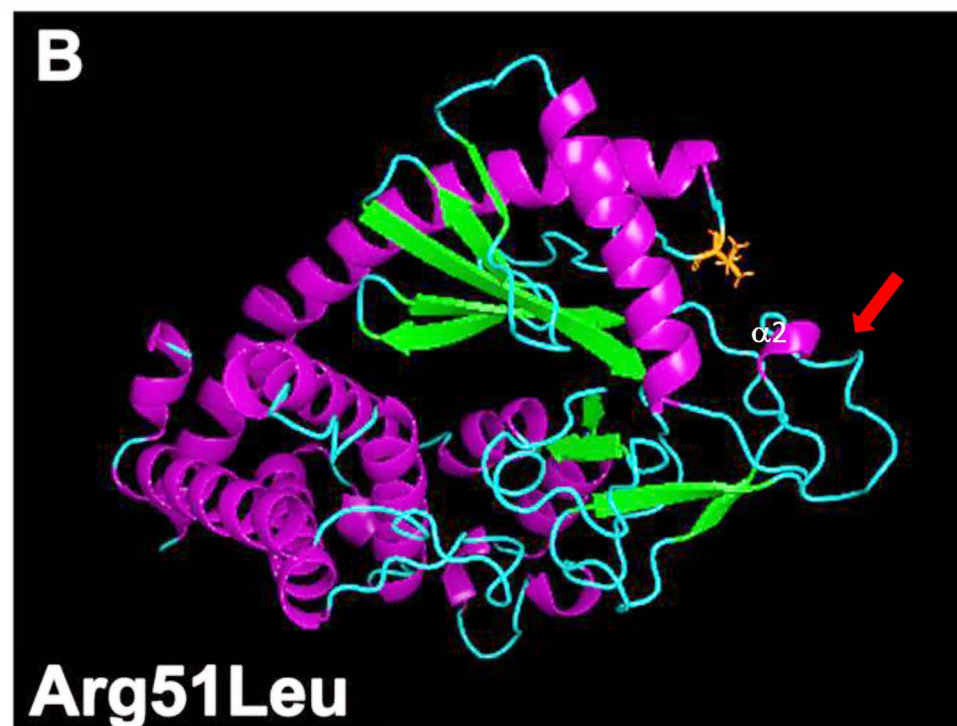
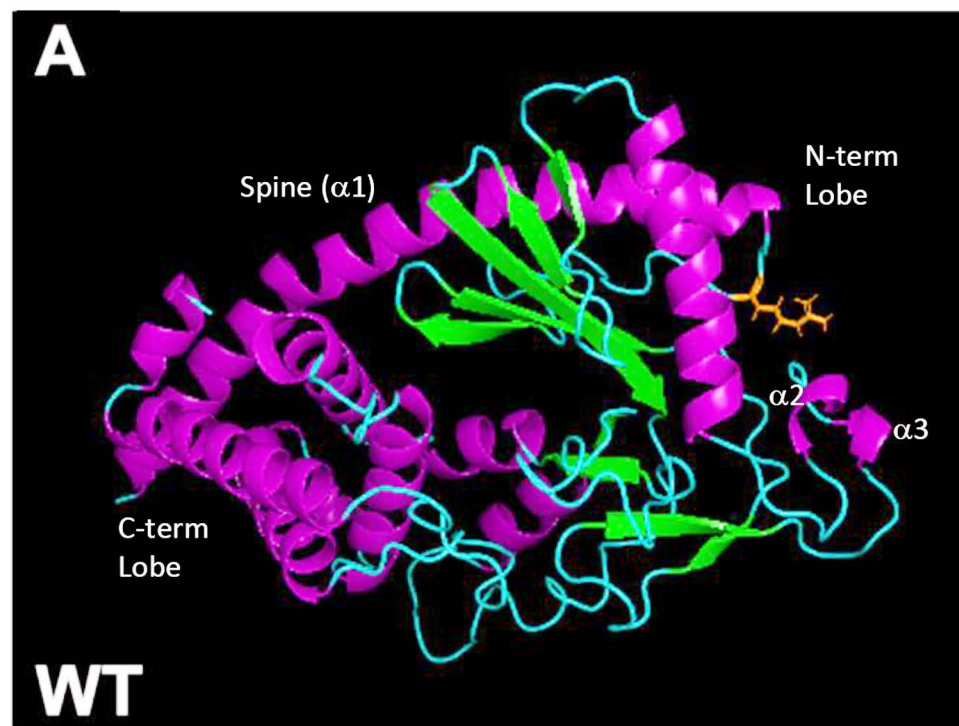
Supplemental Table 2. Predicted RNA binding protein (RBP) effects with non-coding variation.

Untranslated Region	Variant	RNA-Binding Protein	Motif	Functional Effect	Z-score	P-value
5' UTR	WT	SRSF3	cuckucy	D	1.670	4.75e-02
	c.-68T>C				N/A	N/A
	WT	RBM6	haucrar	C	N/A	N/A
	c.-68T>C				2.775	2.76e-03
3' UTR	WT	HNRNPL	amayama	D	2.387	8.49e-03
	c.*529A>G				N/A	N/A
	WT	HNRPLL	rcahaca	U	2.519	5.88e-03
	c.*529A>G				2.025	2.14e-02
	WT	IGF2BP2	vmahwca	D	1.986	2.35e-02
	c.*529A>G				N/A	N/A
	WT	RBM41	wuacwuk	D	2.420	7.76e-03
	c.*529A>G				N/A	N/A
	WT	SRSF3	wcwwc	D	2.402	8.15e-03
	c.*529A>G				N/A	N/A
	WT	SRSF5	yywcwsg	C	N/A	N/A
	c.*529A>G				2.204	1.38e-02

D: variant disrupts the predicted RBP motif site; C: variant creates a predicted RBP motif; U: unaffected.

Z-score and P-value reflect the reliability of the predicted binding site

Supp. Figure 1. *In silico* protein modeling.



Supp. Figure 1. *In silico* protein modeling. Protein structures for the solved MAB21L1 wild-type (5EOG) (de Oliveira Mann et al., 2016) (**A,C**) and the I-TASSER predicted mutants (Arg51Leu **B,D**; Arg62Cys **E**; Gly220Arg **F**). Amino acids (aa) of interest are highlighted as follows: aa51 orange, aa62pink, aa220 yellow. Alpha helices are purple, beta sheets green, linker regions blue. Red arrows (**B,D,E,F**) indicate observed structural changes upon mutation.

Supp. Figure 2. Multi-species sequence comparisons of *MAB21L1* non-coding regions.

A

5' UTR sequence; 13:36050329 – 36050370 (forward sequence)

Human	tttctccaccgaagtggctccagctcttagcagccgcatgg
Individual 2	tttctccaccgaagtggctccagctcttagcagccgcatgg
Chimp	tttctccaccgaagtggctccagctcttagcagccgcatgg
Gorilla	tttctccaccgaagtggctccagctcttagcagccgcatgg
Orangutan	tttctccaccgaagtggctccagctcttagcagccgcatgg
Gibbon	tttctccaccgaagtggctccagctcttagcagccgcatgg
Baboon	tttctccaccgaagtggctccagctcttagcagccgcatgg
Green monkey	tttctccaccgaagtggctccagctcttagcagccgcatgg
Marmoset	tttctccaccgaagtggctccagctcttagcagccgcatgg
Squirrel monkey	tttctccaccgaagtggctccagctcttagcagccgcatgg
Chinese tree shrew	tttctccaccgaagtggctccagctcttagcagccgcatgg
Squirrel	tttctccaccgaagtggctccagctcttagcagccgcatgg
Lesser Egyptian jerboa	ggttctctccgaagtggctcccgcgcagcagccgcatgg
Prairie vole	tttctccaccgaagtggctccagctcttagcagccgcatgg
Chinese hamster	tttctccaccgaagtggctccagctcttagcagccgcatgg
Golden hamster	tttctccaccgaagtggctccagctcttagcagccgcatgg
Mouse	tttctccaccgaagtggctccagctcttagcagccgcatgg
Rat	tttctccaccgaagtggctccagctcttagcagccgcatgg
Naked mole-rat	tttctccaccgaagtggctccagctcttagcagccgcatgg
Guinea pig	tttctccaccgaagtggctccagctcttagcagccgcatgg
Chinchilla	tttctccaccgaagtggctccagctcttagcagccgcatgg
Brush-tailed rat	attctccaccgaagtggctccagctcttagcagccgcatgg
Rabbit	tttctccaccgaagtggctccagctcttagcagccgcatgg
Pika	tttctccaccgaagtggctccagctcttagcagccgcatgg
Pig	tttctccaccgaagtggctccagctcttagcagccgcatgg
Alpaca	tttctccaccgaagtggctccagctcttagcagccgcatgg
Bactrian camel	tttctccaccgaagtggctccagctcttagcagccgcatgg
Killer whale	tttctccaccgaagtggctccagctcttagcagccgcatgg
Tibetan antelope	tttctccaccgaagtggctccagctcttagcagccgcatgg
Cow	tttctccaccgaagtggctccagctcttagcagccgcatgg
Sheep	tttctccaccgaagtggctccagctcttagcagccgcatgg
Domestic goat	tttctccaccgaagtggctccagctcttagcagccgcatgg
Horse	tttctccaccgaagtggctccagctcttagcagccgcatgg
White rhinoceros	tttctccaccgaagtggctccagctcttagcagccgcatgg
Cat	tttctccaccgaagtggctcccgccttagcagccgcatgg
Ferret	tttctccaccgaagtggctcccgccttagcagccgcatgg
Panda	tttctccaccgaagtggctcccgccttagcagccgcatgg
Pacific walrus	tttctccaccgaagtggctcccgccttagcagccgcatgg
Weddell seal	tttctccaccgaagtggctcccgccttagcagccgcatgg
Megabat	tttctgcaac-gaagtggctccagcccagcagccgcatgg
David's myotis (bat)	tttctgccc-gaagtggctccagctcttagcagccgcatgg
Microbat	tttctgcaac-gaagtggctcccgccttagcagccgcatgg
Big brown bat	tttctgcaac-gaagtggctcccgccttagcagccgcatgg
Hedgehog	tttctccaccgaagtggctcccgccttagcagccgcatgg
Shrew	tttctccaccgaagtggctcccgccttagcagccgcatgg
Star-nosed mole	tttctccaccgaagtggctcccgccttagcagccgcatgg
Cape elephant shrew	tttctccaccgaagtggctcccgccttagcagccgcatgg
Manatee	tttctccaccgaagtggctcccgccttagcagccgcatgg
Cape golden mole	tttctccaccgaagtggctcccgccttagcagccgcatgg
Tenrec	gtcctcctccgaagtggctcccgccttagcagccgcatgg
Aardvark	attctgcccgaagtggctcccgccttagcagccgcatgg
Armadillo	cccggcaccgcccgtggctcccgccttagcagccgcatgg
Opossum	tttctcctctgaagtggctcccgccttagcagccgcatgg
Tasmanian devil	tttctcctctgaagtggctcccgccttagcagccgcatgg
Wallaby	tttctcctctgaagtggctcccgccttagcagccgcatgg
Platypus	tttctcctctgaagtggctcccgccttagcagccgcatgg
Saker falcon	tttctcctctgaagtggctcccgccttagcagccgcatgg
Peregrine falcon	tttctcctctgaagtggctcccgccttagcagccgcatgg
Collared flycatcher	tttctcctctgaagtggctcccgccttagcagccgcatgg
White-throated sparrow	tttctcctctgaagtggctcccgccttagcagccgcatgg
Medium ground finch	tttctcctctgaagtggctcccgccttagcagccgcatgg
Zebra finch	tttctcctctgaagtggctcccgccttagcagccgcatgg
Tibetan ground jay	tttctcctctgaagtggctcccgccttagcagccgcatgg
Budgerigar	tttctcctctgaagtggctcccgccttagcagccgcatgg
Parrot	tttctcctctgaagtggctcccgccttagcagccgcatgg
Scarlet macaw	tttctcctctgaagtggctcccgccttagcagccgcatgg
Rock pigeon	tttctcctctgaagtggctcccgccttagcagccgcatgg
Mallard duck	tttctcctctgaagtggctcccgccttagcagccgcatgg
Chicken	attctccc-gaagtggctcccgccttagcagccgcatgg
American alligator	tttctcccctgaagtggctcccgccttagcagccgcatgg
Green sea turtle	tttctcccctgaagtggctcccgccttagcagccgcatgg
Painted turtle	tttctcccctgaagtggctcccgccttagcagccgcatgg
Chinese softshell turtle	tttctcccctgaagtggctcccgccttagcagccgcatgg
Lizard	gattcccgctgcccgcctcccgccttagcagccgcatgg
Coelacanth	tatttttctggaagtggctcccgccttagcagccgcatgg
Tetraodon	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Fugu	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Yellowbelly pufferfish	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Nile tilapia	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Zebra mbuna	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Medaka	cttcttctctaaaaattggctcccgccttagcagccgcatgg
Southern platyfish	cttcttctctaaaaattggctcccgccttagcagccgcatgg
Stickleback	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Atlantic cod	cttcttctctaaaaattggctcccgccttagcagccgcatgg
Zebrafish	attcttctctaaaaattggctcccgccttagcagccgcatgg
Mexican tetra (cavefish)	tttcttctctaaaaattggctcccgccttagcagccgcatgg
Lamprey	ttcttttgctg---cgctgcccgccttagcagccgcatgg

B 3' UTR sequence; 13:36048652 – 36048681 (forward sequence)

Human	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Individual 3	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Chimp	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Gorilla	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Orangutan	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Gibbon	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Baboon	ct-----cga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Green monkey	ct-----cga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Marmoset	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Squirrel monkey	ct-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Chinese tree shrew	tt-----gga----ta-cttcc-a-----gacatttt-----tc--ac-taa--c-a
Squirrel	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Lesser Egyptian jerboa	ct-----ggg----tt-cttcc-a-----gacatttt-----tc--ac-taa--c-a
Prairie vole	t-----gga----aa-tttcc-a-----gacatttt-----cc--ac-gaa--c-a
Chinese hamster	t-----gga----aa-cttcc-a-----gacatttt-----cc--ac-gag--c-a
Golden hamster	t-----gga----aa-cttcc-a-----gacatttt-----cc--ac-gaa--c-a
Mouse	tt-----gga----aa-ctttc-a-----gacattttc-----cc--acagaa--c-a
Rat	tt-----ggg----ga-cttcc-a-----gacatttt-----cc--acagaa--c-a
Naked mole-rat	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Guinea pig	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Chinchilla	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Brush-tailed rat	tt-----gga----ga-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Rabbit	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Pika	ttt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-caa--c-a
Pig	tt-----gga----ta-cttcc-a-----ggcatttt-----cc--ac-taa--c-g
Alpaca	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Bactrian camel	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Dolphin	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Killer whale	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Tibetan antelope	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Cow	tt-----gga----tg--ttcc-a-----gacatttt-----cc--ac-taa--c-a
Sheep	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Domestic goat	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Horse	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
White rhinoceros	tc-----gga----caacttcc-a-----gacatttt-----cc--ac-caa--c-a
Cat	tt-----gga----ta-cttcc-a-----gacatttt-----cc--gc-taa--c-a
Ferret	tt-----gga----ga-ctgcc-a-----gacatttt-----cc--ac-taa--c-a
Panda	tt-----gga----ga-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Pacific walrus	tt-----gga----ga-cttcc-a-----gacagttt-----cc--ac-taa--c-a
Weddell seal	tt-----gga----ga-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Megabat	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-g
Microbat	tt-----gga----ta-cttcc-a-----gacatttt-----cc--tc-taa--c-a
Big brown bat	tt-----gga----ta-cttcc-a-----gacatttt-----cc--tc-tat--c-a
Hedgehog	tttttttaaaaaaaaaa-----ta-cttcc-a-----gctatttt-----cc--ac-taa--c-a
Shrew	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-taa--c-a
Star-nosed mole	tt-----gga----ta-cttcc-a-----gacatttt-----cc--ac-aaa--c-a
Cape elephant shrew	cc-----gga----gg-cccc-a-----gacattttc-----tc--ac-tta--c-a
Manatee	tg-----gga----ga-ctggc-g-----gacatttt-----cc--ac-taa--c-a
Cape golden mole	tt-----ggg----ga-cttcc-a-----gccagttt-----cc--aa-aaa--c-a
Aardvark	tt-----ggg----ga-actcc-a-----gacatttcc-----ccccgc-gaa--c-a
Armadillo	tt-----gaa----ta-cttcc-c-----gacatttt-----cc--tc-taa--c-a
Opossum	ttt-----gga----ta-cttca-g-----gacatttct-----ct--gc-taa--t-g
Tasmanian devil	ttt-----gga----ta-cttca-g-----gacatttct-----ct--gc-taa--t-g
Wallaby	ttg-----gga----ta-cttca-g-----gacatttt-----ct--gc-taa--t-g
Platypus	tt-----gga----ga-cttcc-g-----gacatttt-----ct--gc-tca--a-g
Saker falcon	tt-----gga----ta-cttcaag-----gacagttt-----ct--gc-taaaga-g
Peregrine falcon	tt-----gga----ta-cttcaag-----gacagttt-----ct--gc-taaaga-g
Collared flycatcher	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taaaaa-g
White-throated sparrow	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taaaaa-g
Medium ground finch	tt-----gga----ta-cttcaag-----gacattttt-----ct--gc-taaaaa-g
Zebra finch	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taaaaa-g
Tibetan ground jay	tt-----gga----ta-cttcaag-----aacatttt-----ct--gc-taaaaa-g
Budgerigar	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taaaga-g
Parrot	tt-----gga----ta-cttcaag-----gacattgt-----ct--gc-taaaga-g
Rock pigeon	tt-----gga----ta-cttcaag-----gacaattt-----ct--gc-taaaga-g
Mallard duck	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taaaga-g
Chicken	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taacga-g
Turkey	tt-----gga----ta-cttcaag-----gacatttt-----ct--gc-taacga-g
American alligator	tt-----gga----ta-cttca-g-----gacatttt-----ct--gc-taa--a-g
Green sea turtle	tc-----gga----ta-cttca-g-----gacatttt-----ct--gc-taa--a-g
Painted turtle	tc-----gga----ta-cttca-g-----gacatttt-----ct--gc-taa--a-g
Chinese softshell turtle	tc-----gga----ta-cttca-g-----gacatttt-----ct--gc-taa--a-g
Lizard	tt-----gga----tg-cttct-g-----gacatttt-----cc--ac-tga--a-g
Coelacanth	ct-----gga----ta-cttga-g-----gacattttttt-----ct--gc-taa--g-g
Tetraodon	tt-----tgc--tcatc-ttttt-g-----aaagtttacat-----t-ct--ga-tag--gcg
Nile tilapia	tt-----ttt--tc--tt-tatct-g-----aactttct-----ctttat--ct--ga-tag--a-a
Medaka	tt-----aaaaatc-ca-cgtcg-g-----aactttttt-----ctttat--ct--ga-tag--a-a
Southern platyfish	tt-----aaa-----tct-g-----aactttttt-----ctttat--ct--ga-tag--a-a
Stickleback	tt-----aca--tt--tt-gatag-gcgtacaacatttg-----ccacat-----tga--a-a
Atlantic cod	tt-----ggaaatc--tc-atcct-c-----ggcctctc-----tttctt-gt--ga-tat--a-a
Zebrafish	tt-----taa--ca--tt-cgcct-g-----gattattt-----ctgaagaga--ga-aag--a-a
Spotted gar	tt-----atg--aa--ta-acttc-a-----ggattttt-----ct--gc-tag--aaa

Supp. Figure 2. Multi-species sequence comparisons of *MAB21L1* non-coding regions.

Nucleotide alignments for 5'UTR (**A**) and 3'UTR (**B**) are shown using data generated by UCSC Multiz Alignments of 100 Vertebrates. Identical nucleotides are shaded in grey; positions of variant nucleotides are indicated in bold; non-coding variants in Individuals 2 (**A**) and 3 (**B**) are shown in red.

Supp. Figure 3. Alignment of full-length MAB21L1 and related proteins.

MAB21L1	MIAAQAKLVYHLNKYYNEKCOARKAAIAKTIREVCKVVS	DVLKEVEVOEPRRFISSLNE-M	59
MAB21L2	MIAAQAKLVYQLNKYYTERCOARKAAIAKTIREVCKVVS	DVLKEVEVOEPRRFISSLSE-I	
mMab2111	MIAAQAKLVYHLNKYYNEKCOARKAAIAKTIREVCKVVS	DVLKEVEVOEPRRFISSLNE-M	
cMab2111	MIAAQAKLVYHLNKYYNEKCOARKAAIAKTIREVCKVVS	DVLKEVEVOEPRRFISSLNE-M	
mab2111	MIAAQAKLVYHLNKYYNEKCOARKAAIAKTIREVCKVVS	DVLKEVEVOEPRRFISSLNE-M	
mab-21	MLGHNQNVVYQVNNYFNEKVQHRKVRVTKTVQRIAKVVQEILKEVEAQEPRRFINTLSETT		
Individuals 1a, 1b		L	
MAB21L1	DNRRYEGLEVI-SPTFEFVVLYLNQMGVFNFDVDDGSLPGCAVLKLS	DGRKRSM	118
MAB21L2	DARRYEGLEVI-SPTFEFVVLYLNQMGVFNFDVDDGSLPGCAVLKLS	DGRKRSM	
mMab2111	DNRRYEGLEVI-SPTFEFVVLYLNQMGVFNFDVDDGSLPGCAVLKLS	DGRKRSM	
cMab2111	DNRRYEGLEVI-SPTFEFVVLYLNQMGVFNFDVDDGSLPGCAVLKLS	DGRKRSM	
mab2111	DNRRFEGLEVI-SPTFEFVVLYLNQMGVFNFDVDDGSLPGCAVLKLS	DGRKRSM	
mab-21	TGRFDGI-VVHSPSEYEAVLYLNQMGVFNFDVDDGTIQGCAVLKLS	DGRKRSM	
Individual 2	C		
MAB21L1	ASGYLSARKIRSRFOTLVAQAV-----DKCSYRDVVKMVADTSEVKLRIRDRYVVQITP		172
MAB21L2	ASGYLSARKIRSRFOTLVAQAV-----DKCSYRDVVKMIADTSEVKLRIRERYVVQITP		
mMab2111	ASGYLSARKIRSRFOTLVAQAV-----DKCSYRDVVKMVADTSEVKLRIRDRYVVQITP		
cMab2111	ASGYLSARKIRSRFOTLVAQAV-----DKCSYRDVVKMVADTSEVKLRIRDRYVVQITP		
mab2111	ASGYLSARKIRSRFOTLVAQAV-----DKCSYRDVVKMVADTSEVKLRIRDRYVVQITP		
mab-21	ASGYLSARKIRHRFQNIQAQVLQTPQFSDYC-----KLLQDNTDVRVRVDDKYTVQITC		
MAB21L1	AFKCTGIWPRSAAHWPLPHIPWPGPNR-VA-EVKAEGFNLLSKECHSLA-----GKQSSA		225
MAB21L2	AFKCTGIWPRSAAQWPMPHIPWPGPNR-VA-EVKAEGFNLLSKECYSLT-----GKQSSA		
mMab2111	AFKCTGIWPRSAAHWPLPHIPWPGPNR-VA-EVKAEGFNLLSKECHSLA-----GKQSSA		
cMab2111	AFKCTGIWPRSAAHWPLPHIPWPGPNR-VA-EVKAEGFNLLSKECHSLA-----GKQSSA		
mab2111	AFKCTGIWPRSAAHWPLPHIPWPGPNR-VA-EVKAEGFNLLSKECYSLN-----GKQSSA		
mab-21	AFRCNGIWPRSAHWPIAAGLPWP--NAALANQTKAEGFDLTSRET-AITQQNNPNKQASS	R	
Individual 3			
MAB21L1	-ESDAWVLOFAEAENRLOMGCCRKKCLSILKTLRDRHLELPGQPLNNYHMKTLLVSYECEK		284
MAB21L2	-ESDAWVLOFGEAENRLLMGCCRKNKCLSVLKTLRDRHLELPGQPLNNYHMKTLLLYECEK		
mMab2111	-ESDAWVLOFAEAENRLOMGCCRKKCLSILKTLRDRHLELPGQPLNNYHMKTLLVSYECEK		
cMab2111	-ESDAWVLOFAEAENRLOMGCCRKKCLSILKTLRDRHLELPGQPLNNYHMKTLLVSYECEK		
mab2111	-ESDAWVLOFAEAENRLLGCCRKKCLSLLKTLRDRHLELPGQPLNNYHMKTLLVSYECEK		
mab-21	MEADAWAMKMHGAENMLLTGG-RRKTLNILKCLRDAHMDFPGTPTVTNYILKTLVLYECEK		
MAB21L1	HPRESDWDESCLGDRLNGILLQLISCLQCRRCPHYFLPNLDFOGKP-HSALENAAKQTW		343
MAB21L2	HPRETDWDESCLGDRLNGILLQLISCLQCRRCPHYFLPNLDFOGKP-HSALESAAKQTW		
mMab2111	HPRESDWDESCLGDRLNGILLQLISCLQCRRCPHYFLPNLDFOGKP-HSALENAAKQTW		
cMab2111	HPRESDWDESCLGDRLNGILLQLISCLQCRRCPHYFLPNLDFOGKP-HSALENAAKQTW		
mab2111	HPRESDWDENCLGDRLNGILLQLISCLQCRRCPHYFLPNLDFOGKP-HSALENAAKQTW		
mab-21	HCSEYEWEDPNIGDRLVGIILLQLVSLQCRRCAPHYFLPSLDLLRSKPVHS-IEHSAQLAW		
MAB21L1	RLAREILTNPKSLEKL	359	
MAB21L2	RLAREILTNPKSLEKL		
mMab2111	RLAREILTNPKSLEKL		
cMab2111	RLAREILTNPKSLEKL		
mab2111	RLAREILTNPKSLEKL		
mab-21	HLVRKLMIDPNALQSL		

Supp. Figure 3. Alignment of full-length MAB21L1 and related proteins. Identical amino acids are shaded in grey; positions of variant amino acids are indicated in bold and variants identified in Individuals 1a, 1b, 2 and 3 are shown in red. Note high conservation between different vertebrate MAB21L1/Mab21l1/mab21l1 proteins as well as with its close human homolog MAB21L2 and *C. elegans* mab-21. Human MAB21L1 (NP_005575.1), human MAB21L2 (NP_006430.1), mouse Mab21l1 (NP_034880.1), chicken Mab21l1 (NP_989864.1), zebrafish mab21l1 (NP_694506.2) and *C. elegans* mab-21 (NP_497940.2).