Gene: PMS1 - Sequence: NG\_008648.1 Date : January 11, 2015

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gctggtgc	ggtactc	atccta	ttccaa	ccagaaa	acctt	ttccc	agcag	gtttgo	cgcgc	
• •	•			•					•	
tgcccacg	actggca	aacaat	gacctt	acagaco	gtgaa	gaggc	tgttg	gctctg	gccctc	
gccggtgc	ctccgaa	 gacggc	gacccg	cagcggo	ccctc	agggg	agtcc	cccctg	gctaga	
0 00 0		0 00	0 0	0 00		0000	J			
ccgacagc	ccgagcg	 ctctgg	gccaga	gaaaact	cagca	laaagc	· cgccg	ctgcta	accacc	
tcagcgcc	tccccca	acccgc	cccttc	cttctcg	geggee	gcacg	tcacg	tcagga	accctg	
GGCAAGAC									-46	9
ddonnano										
CACGCAGG									-40 CCTAGG	)9
CCGCCAGC	-399 AGTTCCC								-34 GCCCAC	ŀ9
									-28	39
TTACCCGC	CGCCCCA	CTCCGG	GCCGCC	GGCTCGC	CAGCAG	GACCA	GCCCG	GCTGCT	TACGGC	
									-22	29
CGCGGATA	CACGCCC	TCAGGC	CCGGCG	CTGCGC	GCTTG	CGGAA	GCTTT	CCCGG	ACAGAC	
									-16	39
TCGCTGCC	AGCGGAT	TGGCTG	CGAGCA	GCGCCAA	TCTCA	.CGTTG	CCCCC	GGGCG <i>I</i>	AGGCGG	
G A GTTG A GTT									-10	)9
GACTCAGT	JCCGCGC	TCTCTG	CACCCG	CTCTGC(	GCGCG	CGTGC	G I GCT	GGGTG(	GGGTG	
									-49	)
CGGGTGCG	GGGTTGG	GCCTGC	GCATCG	GGTGAGA	CGCTG	GCTGC	TTGCG	GCTAGT	TGGATG	

$\tt GTAATTGCCTGCCTCGCGCTAGCAGGAAGgtagtgtgtgtgtgactaacgggtatttatt$	a
tatcctcgatggctcgagggccccgtcccctctactcgtcctggtccgggcgcgaacgc	t
	;c
	g
	;c
agatttaacccatggcaactgtaactcat	
Exon 2   Start: 510   End: 661   Length: 151	
	.g
cacctgtaatcccagctactctggaggctggggcaggggtggctgaacctgggaggcagggtggctgaacctgggaggcagggtggctgaacctgggaggcagggtggctgaacctgggaggcagggtgacagcgaggtgacagcgagatttcaggctgagtgag	
	a
	a g
aggttgcagtgagtcgagatcacgccactgcactccagcctgggtgacagcgagatttc	a g
aggttgcagtgagtcgagatcacgccactgcactccagcctgggtgacagcgagatttc  tctccaaaaaaaaaa	a g

GTTC'	TCA	GAT	CAT	CAC'	TTCG	GTGG'	TCAG	TGT	TGT.	AAA	AGA	GCT	TAT	TGA	AAAC'	TCCTT	GG
S	Q	Ι	Ι	Т	S	V V  21	S	V	V	K	Е	L	Ι	E	N  31	S L	D
						121  GATG										aatac	ct
A	G	A	Т	S	V	D V  41	K	L	ug o	546	ogo	000	uga	.gaa	ccca	aavac	CU
						tgga										tattt	ta
						attc										taaat	cc
						aaat										aaaag	aa
						catca										caaat	at
						gtttt											
Exon	3	l S	tar	t: (	662	En	d: 8	44	L	eng	th:	18	2				
						tgta										caacc	tt
						tatt										aaggg	ag
						agaa										atatc	ca
						nagta										ttctt	tc
						itatt										ttata	gg
		1	41		1	51		16	1		1	71		1	181		191

GA	GAA(	CTA	TGG.	ATT:	ΓGΑΊ	TAAAAT	TGA(	GGT(	GCG	AGAT	ΓΑΑ	CGG(	GGA(	GGG1	TATCA	AG	GCT	'GTT
E	N	Y	G	F	D	K I  51	E	V	R	D	N	G	E	G	I K  61		A	V
GA	TGC					211 AATGAA												
D	A	P	V	М	A	M K  71	Y	Y	Т	S	K	I	N	S	H E  81	. 1	D	L
		TTT	GAC	AAC	ГТАС	271 CGGTTT	TCG	TGG	AGA	AGC	CTT	GGG	GTC	AATI	TGTT	GT.	ATA	.GCT
E	N	L	Т	Т	Y	G F  91	R	G	Е	A	L	G	S	Ι	C C		Ι	A
GA E	Ggt	aag	gtaa			tggtt												gtta
aa	acta	aac	cca			ggtgaag											ata	tac
						aggtgg												
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Ex	on 4	4 l	Sta	art	: 84	15   E1	nd:	94	7	Ler	ngtl	h: 1	102					
						catata:												acg
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 ttatta	aggtaaa	uatacgcta	 ittgagta	aaatata	ittatgo	:aataat	 cattto	caaatct	ttg
	 gaaccca	itcgcaata	 itctaaag	gtgttag	gttttat	tagat@	 gtttttt	 tcccc	agg
GTTTTA		331 ACAAGAAC T R T  111	GGCTGC					TTAGAT	371 TGGC G
AGTGGC		391 CTTTCTCA L S Q  131		TTCACAT		CAAGgt	Jaagaaa	agtagct	;ttg
tataca	aaacatt	 ctttacct	tttctgt	 tcttaat	tgttgg	gtttaaa	naaaaaa	naagtta	ictc
ggtgaa	atacaaa	 itatattgo	tttggg	ttggto	ctgata	ıaaggct	agttaa	acttcca	lagt
aaacat	ctcagcc	 tttattct	agccaco	caattto	ttaaat	atttag	gaattgt	stctcaa	ıaag
gaactg	gtatgag	 gagactgga	cacttco	 catggac	tagtta	actccto	ctgtaaa	aaactaa	igag
tattta	attttac	 cagtgctg	gattac	 cagtgtc	atctto	catat			
Exon 5	5   Sta	ırt: 948	End:	1111	Lengt	sh: 163	3		
		ttatgata						ngtgata	ıgaa
		:ttctttag							gtt

421  431  441  451  461  471 GTACAACTGTAACTGCTTTAAGATTATTTAAGAATCTACCTGTAAGAAAGCAGTTTTACT
T T V T A L R L F K N L P V R K Q F Y S
481  491  501  511  521  531 CAACTGCAAAAAAATGTAAAGATGAAATAAAAAAAGATCCCAAGATCTCCTCATGAGCTTTG
T A K K C K D E I K K I Q D L L M S F G
541   551   561   571   581 GTATCCTTAAACCTGACTTAAGGATTGTCTTTGTACATAACAAGgtaaacattattttat I L K P D L R I V F V H N K   181     191
agaggcaccacctatgaaccagaaagtgggacctcactagatac
Exon 6   Start: 1112   End: 1228   Length: 116

${\tt aggcatatgtgcttgtctccatatggatttttatcttccaacaactctgagaaatcttgt}$
aaagaggagaaaaccaaggtaaagggaagttaaatagatggtacactgcaacatatacag
591   601   611   621   631   641  GCAGTTATTTGGCAGAAAAGCAGAGTATCAGATCACAAGATGGCTCTCATGTCAGTTCTG  A V I W Q K S R V S D H K M A L M S V L   201   211
651   661   671   681   691   GGGACTGCTGTTATGAACAATATGGAATCCTTTCAGTACCACTCTGAAGAATCTCAGgta G T A V M N N M E S F Q Y H S E E S Q   1221   1231
Exon 7   Start: 1229   End: 1351   Length: 122

${\tt aatgctctatgtcctatgacagttaaatatctgaaagtataaccattcatgaagatttt}$	a
	a
	t
	a
	a
701  711  721  731  741  751 ATTTATCTCAGTGGATTTCTTCCAAAGTGTGATGCAGACCACTCTTTCACTAGTCTTTC I Y L S G F L P K C D A D H S F T S L S  241  251	A
761   771   781   791   801   811   ACACCAGAAAGAAGATTTCATCATAAACAGTCGACCAGTACATCAAAAAGATATCTT T P E R S F I F I N S R P V H Q K D I L   261   271	A
$ \begin{array}{cccccccccccccccccccccccccccccccccccc$	a
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gga	

 ${\tt caagtatctgggattacatgttcctgccactatgcccggctaatttttgtactttttagt}$ agagatggggtttcatcatgttggccaggctggtcttgaactcctgacctcaagtgatcc $\verb|ccccaccttggcctcccaaagtgctgggatcccaggcatgagccactgtgccgggtcatg|$  $at a at g \verb|cttttccta| at a a g \verb|cagttgcatcta| ctca at ttctca g \verb|ttgaatttgctgggt|$ |841 |851 |861 TTAATCCGACATCATTACAATCTGAAATGCCTAAAGGAATCTACTCGTTTGTATCCTGTT LIRHHYNLKCLKESTRLYPV 281 891 1901 |911 1921 931  $\tt TTCTTTCTGAAAATCGATGTTCCTACAGCTGATGTTGATGTAAATTTAACACCAGATAAA$ F F L K I D V P T A D V D V N L T P D K 301 |311 |961 . . 1951  ${\tt AGCCAAGTATTATTACAAAATAAGgtaactcttttcagataattttttcttatgctattt}$ SQVLLQNK 1321 ataaacatatattactgttttcatataaaaagatttgtttatattttatctttttattat $\verb|ttaaaatcttgtctattattttcataatttcccctaacatttgttaatttgtatttttag|\\$ gaatctgttttaattgctcttgaaaatctgatgacgacttgttatggaccattacctagt

Exon 8 | Start: 1352 | End: 1495 | Length: 143

 ${\tt acaa} {\tt attcttatgaa} {\tt aaa} {\tt ataa} {\tt aaa} {\tt acagatgtttccgcagctgacatcgttcttagtaa}$ 

acagcagaaacagatgtgcttttt

Exon 9 | Start: 1496 | End: 2385 | Length: 889

ta atccga cat catta ca atctga a atgccta a agga atctactcg ttt gtatcctg ttt $\verb|tctttctgaaaatcgatgttcctacagctgatgttgatgtaaatttaacaccagataaaa| \\$  $\tt gccaagtattattacaaaataaggtaactcttttcagataattttttcttatgctattta$ taaaatcttgtctattattttcataatttcccctaacatttgttaatttgtatttttagg981 1991 1001 1011 GAATCTGTTTTAATTGCTCTTGAAAATCTGATGACGACTTGTTATGGACCATTACCTAGT E S V L I A L E N L M T T C Y G P L P S 1331 1341 |1031 |1041 |1051 |1061 |1071 I 1081 ACAAATTCTTATGAAAATAATAAAACAGATGTTTCCGCAGCTGACATCGTTCTTAGTAAA T N S Y E N N K T D V S A A D I V L S K 1351 1361 |1101 |1111 |1121 1091 |1131 ACAGCAGAAACAGATGTGCTTTTTAATAAAGTGGAATCATCTGGAAAGAATTATTCAAAT T A E T D V L F N K V E S S G K N Y S N 371 |381 11151 |1161 |1171 |1181 |1191 GTTGATACTTCAGTCATTCCAAAATGATATGCATAATGATGAATCTGGAAAAAAC V D T S V I P F Q N D M H N D E S G K N 391 1401 1221 |1231 |1241 |1251 1261 1211

 ${\tt ACTGATGATTGTTTAAATCACCAGATAAGTATTGGTGACTTTGGTTATGGTCATTGTAGT}$ 

- T D D C L N H Q I S I G D F G Y G H C S |411 | 421
- | 1271 | 1281 | 1291 | 1301 | 1311 | 1321 AGTGAAATTTCTAACATTGATAAAAACACTAAGAATGCATTTCAGGACATTTCAATGAGT S E I S N I D K N T K N A F Q D I S M S | 441
- | 1331 | 1341 | 1351 | 1361 | 1371 | 1381 | AATGTATCATGGGAGAACTCTCAGACGGAATATAGTAAAACTTGTTTTATAAGTTCCGTT N V S W E N S Q T E Y S K T C F I S S V | 461
- | 1451 | 1461 | 1471 | 1481 | 1491 | 1501 | GAAGAAGCAGGTCTTGAAAACTCTTCGGAAATTTCTGCAGATGAGTGGAGCAGGGGAAAT E E A G L E N S S E I S A D E W S R G N | 491 | 501
- | 1511 | 1521 | 1531 | 1541 | 1551 | 1561 | ATACTTAAAAATTCAGTGGGAGAGAATATTGAACCTGTGAAAAATTTTAGTGCCTGAAAAA I L K N S V G E N I E P V K I L V P E K | 511 | 521

|591 |601

		181			18			183			184			18				
CT	ACA	AAT'	ΓGAA	GA.	ACT	GTG(	GAA(	GACAT:	rgag'	TGA.	AGA(	GGA	AAA	ACT(	GAAg	gtaa	gtttc	С
L	Q	Ι	E	Е	L	W	K	T L  611	S	E	E	E	K	L	K			
aga	agc	ttg	cate	gtg	act	tgaa	atgt	ttcago	ctat	ttc	cat	tct	ata	tga	ctca	actg	tttaa	а
																<b>.</b>		
aaį	gaa	666		,Ca		C 6 6	1661	Jaugg	aaaa	gca	gau	cag	aaa	ala		ııgg	cttgg	a
ta	gag†	tac	cttt	tt	tgg	taa	cact	ttttt	tttt:	ctg	cta	att	tag	gtti	ttti	tgtt	ggggc	g
gg	ggc†	ttg	tatt	at <sup>.</sup>	ttt:	gtt1	tttt	cttttg	gttt:	aag	ccti	taa	ctg	ctt	tctį	gata	ttatt	a
aa <sup>-</sup>	ttt†	tttį	gcta	ıaa	ggt	ttt	ccat	ctcato	cttc	tca	att <sup>1</sup>	tca	aac	cac	cat			
Ex	on :	10	St	ar	t: :	2386	6	End:	287	1	Lei	ngt	h:	485				
gt	gtti	tcg	ccat	tt.	tgg	cca	ggct	tagtc1	ttga:	act	gctį	gac	ctc	agg	tgat	tctg	ccaac	С
tc	agc	ctc	ctaa	ıaa <sup>.</sup>	tgc	tggg	gatt	cacagg	gtgtį	gaa	cca	cca	cta	tgc	ccg	gcca	ataat	t
ga	ttt	tta	tatt	ttį	gtt	cagt	tgtt	Etgtca	atag	tta	tatį	gca	gga	gagʻ	ttaį	gtct	aatac	a
tg	ttc	ctt	tgtc	at.	tac	tata	atct	Eggaa	cacca	agt	act	tta	ttt	tta	taaa	aatt	tactt	С
aga	atg	gtta	aata	ıta	ttt	ctga	atta	atgato	ctca	tta	gtt	cta	ttt	tati	ttc	tatt	cttag	a
	ATG		AGAA	GG		CTA			GAAC	GAT.		ATA			ΓGΑ		1911 AGCCA	T

|621 |631

TGA	1921 ACAGGA								TGG								19 CAG	
E	Q E  641	S	Q	M	S	L	K	D	G	R  69		K	Ι	K	P	Т	S	A
	1981 GAATTT	GGC	CCA	GAA	GCA	CAA	GTT	AAA		CTC	ATT.	ATC'	TAA	TCA	ACC	AAA	ACT	TGA
W	N L  661	A	Q	K	Н	K	L	K	T	S  67		S	N	Q	Р	K	L	D
	2041																	
	ACTCCT																	
Е	L L  681	Q	S	Q	Ι	Е	K	R	R	S  69		N	Ι	K	М	V	Q	Ι
	2101		1:	211	1		21	21		12:	131			214	1		21	51
CCCC	CTTTTC	TAT	GAA.	AAA	CTT	'AAA	AAT	AAA	TTT'	TAAC	GAA.	ACA.	AAA	CAA	AGT	TGA	CTT	AGA
Р	F S  701	М	K	N	L	K	Ι	N	F	K  71		Q	N	K	V	D	L	E
	2161		1:	217	1		21	81		12:	191		1	220	1		122	11
AGAG	GAAGGA	TGA	ACC'	TTG	CTT	GAT	CCA	CAA	TCT	CAG	GTT'	TCC'	TGA	TGC	ATG	GCT	AAT	GAC
E	K D  721	E	P	С	L	Ι	Н	N	L	R  73		P	D	A	W	L	M	Т
	2221		1:	223	1		122	41		122	251		ı	226	1		122	71
ATC	CAAAAC	AGA(	GGT.	AAT	GTT	TTA	'AAA'	TCC	ATA'	TAGA	AGT.	AGA	AGA	AGC	CCT	GCT	ATT	TAA
S	K T  741	E	V	M	L	L	N	P	Y	R  79		E	E	A	L	L	F	K
	2281																	
	ACTTCT																	AAC T
R	L L  761	Е	IN	п	ĸ	ь	Р	А	Е	77		£	ĸ	Р	1	М	Ь	1
	2341																	
AGAG	GAGgta	tgat	tga	tac	aat	act	ttt	taa	gag	taaa	aaa	tat <sup>.</sup>	tta	tta	taa	tag	ttc	ata
E	S  781																	
ctag	gattaa	aato	cga	agg	tag	aag	gtt	ggg	aat	gtti	tcc <sup>.</sup>	tag	taa	gga	tta	gtg	gta	ttt

${\tt atttattttaatcttttaaacatttgatttctaggccgggcgcagtggctcatgcctgta}$
gcacgc
Exon 11   Start: 2872   End: 3002   Length: 130
2351  2361  2371  2381  2391  2401
TCTTTTTAATGGATCTCATTATTTAGACGTTTTATATAAAATGACAGCAGATGACCAAAG
L F N G S H Y L D V L Y K M T A D D Q R  791   801
2411

tcca	agas	rt.t.;	acat	t.t.c	røøt.	ttc	at.ø	att	tac	ເລດຂ	atat	t.øøt	.oot	tata	at.t.1	ttt	ot.ct	cac	:t.
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ataa	gta	cagt	tgtc	ago	tat	gtt	aaa	aca	ttt	ttt	tca	aaaa	acat	tcad	cct	ctg	cctg	gaaa	ıc
	•	-++	~~~	+ -+	+ +	+	- 		+ ~ -	.++4	•	.++		.+++	+	.+		+ .	
aagt	aag	UUU	ggca		, C L L	lla	gic	Caa	.ugc		gc	166	Jgca	1661		ala	acto	iau	Ja
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aata	gtti	tta	ttgt	aat	ttt	tgt	cat	cct	ttg	gaca	acaa	atat	tca	aaa	ctg	cct	gctt	tgt	t
·																			
taaa	tta	tta	a																
Exon	12	1 4	3+2r	+ •	300	13 I	Fn	۸.	216	32	ΙΤ	an ort	-h·	160	1				
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ggga	ctc	-2+1	+++c	+ c+	·cca		C 2 2	ma a	2+0	ra+ <i>c</i>	+ = :	220		+++	2 2 00	מבים	ຕລູດເລ	200	a cr
ggga	CUC	Jau			cca	aag	caa	gaa	aug	sauc	Juan	aacc	iaai		agg	gaa	cago	iacc	<b>1</b> 8
•	•			•		•	•		•		•					•			
caat	aact	tggg	gatt	aag	gtta	aat	tat	ata	aag	gcad	cate	gtgg	gtgt	tcag	gtga	agc	actt	ate	ga
	•	- ~~	•		+ ~~	•	· ~++	20+		+ ~+	•	+ ~ ~ .			~~~·	•		.+++	۰. ۰۰
ctta	alg	Lga	gacc	Cla	ırgg	gaa	all	acı	all	, tg i		LCCa	acı	Laat	gga	aca	aaac		,g
tcct	gact	tgta	atta	aat	ttg	aat	gag	ggt	tca	cta	aaat	tgt1	ttta	aaca	agca	aat	tttt	gtt	t
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ttaa	tgga	aca	tgtt	ctg	ggaa	tat	gaa	ctt	aac	ctat	gtg	gtti	ttt	attt	ttt	ttt	cttt	cag	gg
		248	31		124	91		12	501	_		25:	l1		12	521		12	2531
GAGT"	ттс	ΔΑΤΊ	ГАСТ	GAA											гтсг	гст	CCC	ттс	T.
			T																
V	S	_	1	E			ь	E	_	Ŀ	G	M	А	IA			г	Г	1
					183	31									84	41			
		254	41		125	51		12	561			25	71		12	581		12	2591
ATGG.																			
			D											R		A			
G	V	А	ע	ь			т	ь	ΤΛ	А	Т	ь	IA	n			N	Ε	V
					185	1									186	51			

		126				311			262			1263		•				
TTTA													Ggta	cgc	att	att	ttat	tata
Y	Ε	С	R	P			V	Ι	S	Y	L	E						
					187	<b>7</b> 1												
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tatg	tta	attg	tata	aca	gggg	gtco	cca	act	CCC	ggg	сса	agco	cggt	tag	gaa	cca	ggc	caca
	•		+ ~ ~ ~	•	mc ct		- a ~	•		+ 63		ccto		· + c+	жсс.	++ c+	· - ~+ /	- 2 ~ 2
cago	ag	gagg	rgag	SCE	gcci	,,,,,	ag	CBB	gca	cca	cca		gagc	CC C	gcc	666	ugu	Jaga
tcag	te	cag	catt	ag	atto	tca	ate	gaa:	gca	caa	acc	ctat	ttet	oaa	cte	tgt:	atgo	gag
0000	-00	5		6				D(	6	-				5	0	-6	~~6`	-6-6
ggat	cga	aggt	tgtg	gca	ctto	ctte	gtg	gaa	caa	ttt	cct	gaaa	acca	tcc	cca	ccc	gc C 8	aact
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ggaa	aaa	actc	ttcc	caca	aaaa	acce	ggt	ccc	tgg	tgc	caa	aaag	3					
Exon	13	3	Star	ct:	316	64	E:	nd:	35	21	L	engt	th:	357				
•	•		•	•		•		•	•		•		•	•		•	•	
aatc	act	tcta	tcat	gt	ggc	cag	gtt	taa	ttt	tac	gca	ttat	ttta	ttg	act	aaaa	atgt	ttct
			· + ~++	•	a+ a+	•	.+	+ ~ ~	•		•	+				•	•	+
gtct	LLI	Lgrc	tgtt		ctgi		CC	rgc:	act	aga	atg	taaa	actg	cac	aag	agca	aagu	CCLL
· ~+++	++1	- aat	+cac		ctar	•				tas		cata	artt		act:	· taat	:	+2+
gttt		rggu	ccac	.ca	Cual	auc		gag	agc	uga	Caa	Catc	ag c c	gau	act	uaa	Jaac	luau
ttgt	τ.σ:	agt.g	· aatt	.cai	taaa	· aat.g	รลล	· aat:	· aat.	caa	· gat	tcts	· rt.ct:	t.c.c	ttt	· t.t.ø	ccas	ogat.
0050	960	-6 -6	uuo	Jou	ouuc	عاسم	Saa	uu o	aao	caa	540	3000	5000	000	000	006	عماد	5540
aaca	tgt	tttt	cago	ctt	cage	ggta	ata	tca <sup>.</sup>	tgg	taa	aat	gtgt	tgac	ttt	ccc <sup>.</sup>	ttts	ggad	cagg
					00	50			00			0.0	0				30	- 66
		264	1		265	51		12	661		ı	2671	1	1:	268	1		2691
GGAG																		
G E	i	A V	R	L	S	R	Q	L	Р	M	Y	L	S	K	Ε	D	Ι	Q
		881									- 1	891						

GACAT		2711 AGAATGAAGCAC				
		R M K H				
	ATTTTTTC	2771 CATCATTTAACC H H L T	CTATCTTCCAGA	AACTACATG		
AAGAT		+31 CATTGAAATTGG				
ATCTT		+91  GTGTCACATGG				+131 TTTTA
TATTG		+151 FTCCACGTATTC				
tgatt	ctcaagaa	 accaaccatcat	 cataattatga	.gttgtaggct	 .caaaattgga	aaata
ggtag	 tatttttg	 gggtggttattt	 actaggtcctc	tgtgcaggga	 agaaatcaaac	atcac
acata	ctggcata	 atctgacaaaca	 Igtgtcaatatt	 ttttaaagta	acaatggagag	tcaga
gtggg	 agaaaatg	 gagaaaacgaag	 tttagaggaag	 gaagaaaacca	 ngaaatccttc	cttta
acttc						++a