Gene: SCN1A - Sequence: NG_011906.1 Date : January 11, 2015

Exon 1 | Start: 1 | End: 282 | Length: 281

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gt	gtt	aaa	att	ttt	tct	tta	gaa	atc	acc	ttt	caa	ttt	aag	gag	aaa	aca	gac	ttt	aaat	
сс	tct	agc	tca	tgt	ttc	atga	aca	aga	att	tat	tta	tat	taa	cat	ctc	tta	gtc	cac	tctt	
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ΑA	TGT	GCA			CAA			GCA				TGT			AGG			CAG	CTTC	
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gcatatt	Luguug	Claalle	gcctac	igca	LLCCL	iggac	Lgilg	gragea	iccaac	Jaca	itge
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caaatta	agcacaa	aatctta	agtat	cctt	ctgtt	taaga	aatga	aacac	atttg	gtgg	ggtg
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gtttatg	gcatata	catgaga	aatga	attga	attat	atcat	aacat	atgca	actg1	tccc	aaa
 tgcacca	atgacaa	ataaaaa		·cagg	taraa	tøøct	caago	·ctota	atcc	ລອດ	act
ugcacca	rugacaa	avaaaac	Rappe	cage	6668	CEECU	caage	,cugua	aucci	ug	acu
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ttgaggg	gccgag	gcaggca	agatca	cgag	gttaa	gagat	tga				
Exon 2	Star	t: 283	End	l: 40	1 L	ength	: 118	3			
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atttaac	ctcagac	acttaaa	atato	gaga	aaata	aatga	cttct	cttgg	ctaai	ttgt	CCT
	•	•									
atgtaca	aataaaa [.]	ttgaggg	gtaagt	atta	ggtaa	aaggt	aagtg	gtato	attg	gatt	tcc
tctctaa	attaaga	agagato	cagtg	gacag	tttgt	tttca	tgggg	gcactt	tagaa	aatt	gtg
attgtgd	rtaattt	ctcattt				++2++					rtam
arregige	reggere	Cucauui	aacu	taca	ataat	Coacc	augac	aagua	acago	عممع	suag
		•									
ataacag	gagttta	agtggtt	tatac	tttc	atact	tctat	gttgt	gttcc	tgtc1	ttac	aga
ı	271	281	L	129:	1	30	1	l31	.1	ı	321
ACTTTTA	TAGTAT										
	V L	N K	G K	. A	I F			A T	S	Α	L
l	91					10	1				
I	331	341	L	35:	1	36	1	37	1	ı	381
TACATTT										ACAT	TCa
Y I L	. ТР	F N	PΙ	. R	K I	Α	I K	ΙI	. V	Η	S

|111 121 ${\tt tatcctttttcaagtgattaatattaactatttgtacatgatctgtaagcactttatagc}$ taaatatcaaattaagttgggaaatgtccatattatatagttttcatcactctcattttg $\verb|catctttgtcatattagcctcattcttaaagttcattaatcacatagacattactgaaac| \\$ $\verb|tttctaaaaaacacttatatgtattgaatcctacaaatctaaagtaagataaccatgttt|\\$ Exon 3 | Start: 402 | End: 491 | Length: 89 $\verb|ttctctgggaggtgaatatattgatttaataatagttgtaaaaacaacaccttatactga|\\$ ${\tt tataatgctttcatatacattacctcatttaatctatacaaatactcagtgaaggtgata}$ $\verb|ttattacccacattttacacatgaagaaattgaaatgtaaggagattagaagacttgccc|$ ${\tt aca} {\tt atg} {\tt catttatccctg} {\tt aattttggctaagctgcagtttgggcttttcaatgttagctt$ $\verb|tttgta| a ta ta a cacttgg at ttt g at ttt cttttgtgtgttcctta a caata accta ca$ 391 401 |411 421 431 441 ATTATTCAGCATGCTAATTATGTGCACTATTTTGACAAACTGTGTGTTTTATGACAATGAG |131 1141 |461 | 471 451

 ${\tt TAACCCTCCTGATTGGACAAAGAATGTAGAgtaagttcaacttatatttttaataacata}$

tata			ggga																
tttt	tatt	aaa	igto							cgca								ggc	cct
cagg	gcct	gct	Eggo							tcto						gtg	gtto	ac	ctg
aaga			acto																
atgt	:ggt	caa	aago	aat	tgc	aga	laaa	ıga	gcc										
Exor	n 4	5	Star	t:	492	I	End	l: (620	I	eng	gth:	12	28					
ctaa	nata	ttt	tctc	agg	gaga	cta	tat	tt:	aca [.]	taaa	atac	caa	igac	att	tca	igad	caat	;gg	cta
aata	ıgat	tto	cata	tac	ctt	gta	ttt	ct	cac	acta	acto	ctta	laga	icac	ttt	ace	gaaa	aca	act
cttt	gtg	gtta	agga	ago	tga	att	taa	lat	tta	ggg	ctac	gtt	tca	ittt	gta	itga	aaat	ta	aaa
tcca	itct	gct	ttag	gttt	tct	ttt	tta	ıgt	att [.]	tato	ctat	tcc	act	gat	gga	ıgte	gata	aag	aaa
ttgg	gtat	gct	tatg	gaaa	ıaaa	cac	tgt	ta	ctt [.]	tato	caaa	attt	ttt	gga	tgc	ttg	gttt	tc	aga
ATAC Y	CACC	TT(F 16	CACA T 51	G G	ATA I	TAT Y	'ACT T	TT F	TGA. E	ATC <i>i</i> S	L L	I I 17	AAAA K '1	ITA.	'ATT I	GC <i>I</i> A	AAGO R	G G	F
CTGT	TT	54 GA <i>I</i>																	591 CAT
С	L	Ε	D	F	Т	F	L	R	D	P	W	N	W	L	D	F	T	V	I

	181						191				
TACATT T F	TTGCgta		ctttt								
	tatgct										
	caaacto										
	aggcttt										
	atatgco									acaca	acac
aatcta	natg										
Exon 5	5 Sta	ırt: 6	21 E	ind:	712	Leng	gth: 9	1			
	5 Sta										
 taccac		itattt	atcatt	taaa	atcac	caaga	aggto	tttat	aaaag	accca	atgc
	cttataa	tattt .tcctg	atcatt ctaaaa	taaa .tctt	atcac gaata	caaga attct	aaggto	tttat .aattc	aaaag tacag	accca gtttg	ntgc
taccac aagtaa cagaat	cttataa ntataaa	.tcctg .accta	atcatt gctaaaa	. taaa . tctt . tttt	atcac . gaata . cagct	.attct .cttcg	aaggtc ggattt gcactt	tttat aattc tcaga	aaaag tacag gtctt	accca gtttg gagag	atgc gtaa gctt
aagtaa	cttataa atataaa	tattt .tcctg .accta	atcatt	. taaa . tctt . tttt	atcac . gaata . cagct . agaag	. caaga . attct . cttcg . tgatt	aaggto	. tttat . aattc . tcaga . aaaagg	tacag . tactt . gtctt . atagg	accca gtttg gagag ctctt	gtaa gctt ctgt

211 221 |681 671 691 ${\tt CCGAGCATTGAAGACGATTTCAGTCATTCCAGgtgagagcaaggttagataatgagacgg}$ ${\tt acccatcatgtgattcagcatccttctctgcttgacattcagttttacagaaaatcagga}$ $at cata agact agg t \verb|gtttcaaagaaaatgattcttatt \verb|gtttagacataagctttatca|$ $\tt gccttggagtttaacaatgtcttatgtgagatactgtaatttgaggtcaaggtgacctgc$ ${\tt acttacaaatatcatgggttttttttttttttcaccatgttagttttatcgagaatttgct}$ $\verb|cccttttaaatgtaccaatctgtaatttctta|\\$ Exon 6 | Start: 713 | End: 982 | Length: 269 $\tt gtagcaaaaaggaaaaagacaaggccctttccttaatataggctccatgaagaacatgaa$ $\verb|ccttccattactctatctcgacataatgggatatgaatcttcatttcattgtttaaatca|\\$ $\verb|ttacatttctggagtttatcttcataagttcttccccttcaatgaagtgttgcttctcca|\\$ $\verb|ctagcgttgcaaacattcttggatttatgaatgaaatctatgttttcaagaaaattgtgt|\\$

|721 |731

|741

|711

								124	11									251
тсст																		811 GGGCA
L								F 26	Α									
	GAG	GAA	TAA	ATG	TAT	ACA	ATG	84 GCCT P 28	rcc P	CAC	CAA	TGC	TTC	CTT	GGA	.GGA	ACA	TAGTA
TAGA.	AAA		TAT	AAC	TGT	GAA		TAAT	rgg'	ТАС	ACT		AAA	TGA	AAC	TGT		931 TGAGT E F
ь	IX	14	_	1	V	14	1	30		1		_	14		1	v	ľ	311
	CTG	GAA	GTC.	ATA	TAT	TCA	AGA'	96 TTC# S 32	AAg R	taa	.gaa	itta	ttg	tta	tgt.	aca	ttt	cctta
aaaa	gta	gaa	ttg	gat	tgt	ttg	taa	caca	aaaį	gga	taa	lata	ctt	gag	ggg	ctg	gat	atccc
attt [.]	tac	atg	atg [.]	tga	ıtta	Igca		atte								tca	tgt	acccc
atat	ata	gaa	aca	cct				ccad										taatg
aaat	aaa	taa	aac	acc	ctc	taa	aaa	agaa	atta	act	ato	atg	tat	att	tgg	tct	ttt	cattg
ctaa	tgt	gcc	ttc	cat	cta	ıgga	atg	aaaa	aa									
Exon	7	l S	tar [.]	t:	983	3	End	: 10)46	I	Len	ıgth	: 6	3				
tatc	act	aac	ttt:	aca	itto	ata	atg	aaga	aaa	tac	ttt	gca	ctg	taa	.agt	gtc	taa	agtat

ctttgcactgtatctaatctaatgtcatttcttcataatgaagaaatactttgcactgta
971 981 991 1001 1011 1021 GATATCATTATTTCCTGGAGGGTTTTTTAGATGCACTACTATGTGGAAATAGCTCTGATG Y H Y F L E G F L D A L L C G N S S D A 331 341
aaag
Exon 8 Start: 1047 End: 1188 Length: 141

gacaccaagtagctaataagactatatgcagtcaaaagttgggaaatagattagttactt														
gtatcagagtaaaaaggcagcagaacgacttgtaatactttcttt														
1031														
1091														
1151 1161														

Exon 9 | Start: 1189 | End: 1395 | Length: 206

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gtto	ct	tg	ac†	ttt	CC	taa	aag	gct	cag	ggto	gtt	aat	gga	gga	ctc	cat	ggc	att	tta	ttga
								_												
ctgt	tt	ca	tt	tt,	gg	tga	aat	aca	aaa	att	ctg	cca	tag	aaa	tca	tag	tgt	gaa	tgc	ataa
gcta	ıga	aag	aga	aag	gaa	aga	aag	ctg	aag	gtta	ıaag	aaa	caa	taa	aaa	aaa	agt	aca	ggg	tgct
atga	ıco	aa	cat	taa	tta	act	tat	tta	tat	cte	gagt	ttt	gct	aga	cac	atg	agt	aaa	ttg	aaag
	•																			
ttga	aag	gcc	ac	cac	tta	agt	tga	aat	aat	att	gag	aaa	taa	ttc	tga	tat	ttt	tgt	ttg	caga
1117		יככ		111		מממ				СЛТ			ጥጥጥ		121		יר א די	12		GGGC
		R	A			G		Т				F		V				F	L	G
123 TCAT		ΔΤ.		12 12		ΓΔΔ		12 GAT		raac		261 сст			127 GGC		CGA	12 GGA		GAAT
S F	7	Y				N	L	I	L	A		V	A	М	A	Y	E	E	Q	N
129 CAGO Q #	CC	CAC T		ΓGG	AA			AGA	ACA	AGAA K	AGA		CGA E				GAT M	13 GAT I		ACAG Q
431	_										4	41								
135 CTT <i>A</i> L k 451	Α <i>Ι</i>	AAA K		13 AAC Q	AG	GAC E				lGgt	aaa	gct	gcc	ctg	tct	cat	ggc	act	gac	cttt
tato	gt	tc	tga	att	gti	tac	cgt	aat	att	gaa	ıgag	aaa	gtg	agt	acc	taa	ıgag	cag	gtg	gttg
tatg	gag	gga	at:	taa	ta	atg	gat	ttc	tat	att	ttg	atg	atc	cac	tta	ctt	tta	ttt	agt	ttaa
ttat	tt	ta	tti	tga	ga	tgt	tgt	ata	ata	aata	acc	aca	ata	gaa	ctc	tct	cat	gct	att	ttaa

${\tt tcatcttaagattctcaaagtaaagggaaaatgttattttggggtattttcaagtgagagaga$
acttttattcaatgagtagactggcat
Exon 10 Start: 1396 End: 1680 Length: 284
ggagcattaacagagacataataaatgttaccatggagcaaactaaattatctccaaaag
1381 1391 1401 1411 1421 1431 CAGGCAGCAACGGCAACTGCCTCAGAACATTCCAGAGAGCCCAGTGCAGCAGGCAG
1441 1451 1461 1471 1481 1491 TCAGACAGCTCATCTGAAGCCTCTAAGTTGAGTTCCAAGAGTGCTAAGGAAAGAAGAAAT S D S S S E A S K L S S K S A K E R R N 481 491
1501 1511 1521 1531 1541 1551 CGGAGGAAGAAAAGAAAACAGAAAGAGAGAGAGAGAGAAGA
1561 1571 1581 1591 1601 1611 TTCCAAAAATCTGAATCTGAGGACAGCATCAGGAGGAAAGGTTTTCGCTTCTCCATTGAAFF Q K S E S E D S I R R K G F R F S I E 521 531

		621			163		•	641			651			1661				
GG	GAA	CCG.	ATT	GAC	ATA	TGAA	AAGA	GGTA	CTC	CTC	CCC	ACAC	CCA	Ggta	atgg	cacte	gctga	
G	N	R 41	L				K R				P	Н	Q					
gt	tta	ctg	atg	cat	ggt [.]	tgaaa	aatt	aaaa	cat	ggg	aga	gage	ggg	gaga	att	agaaa	aatgg	
ac	tca	.gga	att	ttt	atc	aactį	gaat	caac	cac	tgt	tgt	gtta	ata	ttta	aaac	ccato	ccctt	
ct	tca	cat	agt	tat	gca	aaaa	cttt	acto	cac	aga	tat	gtaa	agt	ctac	cagc	tcggt	tgtag	
tt	aag	ata	aca	cca	agt	tgaca	agtc	agga	ıgaa	ctg	gac	tgto	cat	tctg	gata	ctgt	ctgct	
ga	tat	ttg	tct	ata	act	aatga	acat	ttaa	itct	ttt	tga	tcct	tcaį	g				
Ex	on	11	l S	tar	t:	1681	E:	nd:	2028	8	Le	ngth	n: 3	347				
СС	ttt	gaa	ctt	gag	aaa [.]	tggt	cgtt	cagg	gtg	tgt	gtg	tate	gtgt	tgtg	gtgt	gtgtg	gtttc	
aa	tat	gtt	aag	gtt	gca	atcta	atct	cctc	att	ctt [.]	taa	tccc	caa	gggo	tag	aaact	tttct	
tt	tat	caa	ggt	aat	tta	attta	aatg	tgaa	itgc	aca [.]	taa	aatg	gaga	aatg	gata	atcaa	aaagg	
aa	tga	acc	ata	ttc	tgt	tatga	aatg	ctga	laat	ctc	ctt	ctac	cata	aato	cttg	caaaa	atgaa	
																actto	ctagt	
TC	TTT				'CCG'		81 TCCC'										17 GCCTT	'21
S	L	L	S	Ι	R	G S		F	S	P	R	R	N	S	R '	T S	L	

			173	31		11	741		- 1	175	1		17	61		1	771		1	781
TT	CAG	CTT	TAG	AGG	GCG	AGC	AAA	GGA	TGT	GGG	ATC	TGA	GAA	.CGA	CTT	CGC	AGA	TGA	TGAG	
F	S	F	R	G	R	Α	K	D	V	G	S	E	N	D	F	Α	D	D	E	
						15	81									5	91			
			179	91		1	801		- 1	181	1		18	21		1	831		1	841
CA	CAG	CAC	CTT	ΓGΑ	GGA														ACAC	
Н	S	Т	F	E	D	N		S	R			S		F	V			R.	Н	
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			18	51		11	861		- 1	187	1		118	81		l 1	891		11	901
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						10	21									10	31			
			140	4 4		14	004			100	4		140	.11		14	054		14	001
	шаа		119:		~ ^ ^		921							41			951			961
																			GGTT	
F	P	A	N	G	K		Н	S	T	V	ט	С	N	G	V	٧		L	V	
						16	41									16	51			
			19				981			199			120				•		•	
																Ggt	gat	aat	agat	
G	G	P	S	V	P			Р	V	G	Q	L	L	P	E					
						16	61													
aa	gcc	agc	tac	tga [.]	tga	caa	tgt	aag	gaa	gtt	tta	aat	agt	tca	ggc	atg	gct	ggc	tcac	
ta	ttg	ctg	cac	cag	cca	gtg	ttt	cta	cag	aac	ggc	aac	ctt	gag	aat	gat	tcc	tgg	ttgg	
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tc	acg	ctg	tga	atg	cac	ctg	cat	ctt	gta	ata	tct	tta	ata	gac	taa	cca	act	aaa	actt	
									0					0 -						
aa	· aac	ctt.	age	· aota	ctc	· cto	cac	ลลล	cct	_ຜ ຸລ ລ	· toc	att	tac	tta	tta	aaa	oto	cta	agga	
aa	aac		agc	ag u		CUB	cac	aaa	.000	gaa	ugu	auu	, cac	, c ca		aaa	8 6	Cua	agga	
++	ma+:	~c+	aca		+22	++^	c+~	cc+		m++	•	٠.	+++		2+0	+				
UU	gai	uag	aca	Jaa	uaa	uud	cug		cca	gul	gga	gga		gct	aua	. 0				

Exon 12 | Start: 2029 | End: 2161 | Length: 132

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gtaata	aactg	ttaaa	atata	atctc	ttaa	gag	tttta	tcaa	acta	tatt	taaaa	atta [.]	ttt	tgta
tttta	Faaaa	++a+a	raaat		aagt	taa	catct	+ aat		tact	totat	- ແລ ຕ	taa:	at or o
cocca	uaaa	.0000	saaa	Juage	aagu	vaa	Cauco	68		ugu	guar	gac	vaa	1088
ttaaca	agttt	gaaca	attco	caggc	taat	gat	acaat	aagt	cag	aaat	tatct	Secc	atc	acca
	-6	8				6		6	0			- 6		
attga	atatg	gaaagt	tgcat	tgatg	catg	tgt	ttcat	gaaa	attc	actg	gtgto	cacc	att	tggt
J			Ü	0 0				•						
tgtttg	gcttg	· ctcata	· attgo	ctcaa	atta	att	gttta	.atgo	catt	agca	atttt	· :ttt	tta	cagg
2011		12023	1	20	21		2041		12	051		120	61	
GGAACA	AACCA	-									TTCC <i>I</i>	-		CATG
G T 671	т т	` E	T I	E M	R		R R 1681	S	S	S I	FH	V	S	M
1071							1001							
2071 GACTT	ГСТАС	2081 AAGA			91 AAGG		2101 CGAGC			111 ATA(21 CAT		AACA
D F	L E			3 Q	R	Q :	R A	М			A S	I	L	T
691							701							
2131		2143												
AATACA N T	AGTAG V E		ttggt	taaca	aatt	cta	ttttc	gtti	caa	ttat	ttttc	cacc	aaa	ctta
711														
tattg	tctca	tttca	aaaca	aaata	tatt	tgt	gagtt	ggga	aata	gtgo	catto	ctaa	tga	aaag
acagto	ctaat	tcaag	gagct	tgtta	tttc	tta	tatct	acto	caga	tatt	tctag	gaag	cct	taac
				•								•		
aattta	atttt	aaaat	tgagt	tgata	ttgg	gac	taaga	atg1	tttt	ccta	aactg	gtgt	agc	aact
		•		•								•		
ctttga	aaaat	gcaga	aacaa	agca	tttc	tgt	taggg	gtti	tcta	atct	tcatt	tagt	cata	agtg
accct		Lgt												

Exon 13 | Start: 2162 | End: 2400 | Length: 238

gttc	cca	ttg	aga	cgt	cac	atg	aaa	gac	cac	tca	ttt	aac	ttg	gagg	cca	agt	gct	gag	cc
acaa	tgt	cag	tgc	ata	aga	caa	aaa	act	acc	cat	tgt	tac	ctg	ggc	tct	atg	tgt	gtg	tc
tgat	gaa	ata	tcc	ttgį	gga	ggt	tta	gag	taa	act	gta	att	ttt	tta	aca	agt	aca	aaa	aa
ggtt	gtc	tct	gta	acaa	aaa	atg	tgt	tga	tta	ctg	aaa	ıata	agt	tta	gtg	gat	atg	aaa	ta
aatg	tgt	gtg	tat	aaag	gta	tac	ctt	ttg	gtg	gtt	ctt	ttt	ttt	ttt	ttt	ctt	aat	cta	ga
AACT L			ATC	CAG(R	GCA Q	K	ATG	CCC	217 ACC P	CTG			TAA	ATT F	TTC	N	CAT.		2201 CT L
TAAT	CTG	122 GGA		TTC'	12	21 221 ATA			223 'AAA			22 ACA		TGT		251			2261 GA
Ι	W	D		S	P 7	Y 41	W	L	K	V	K	Н	V	V	N 7	L 51	V	V	M
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D	Р	F	V	D	L 7	A 61	Ι	1	Ι	C	1	V	L	N	T 7	_	F	M	A
CCAT	GGA	23 .GCA		TCC	AAT				235 TTT	CAA		23 TGT		TAC		371 AGG.		-	2381 Gg
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taag	cat	att	gga	agg	taa	atg	tgt	tta	gtc	ttc	aaa	ittt	tct	gct	tga:	aaa	act	gtt	ta
catt	taa	ttg	tgt	ata	gca	gtc	ttt	caa	.cca	tcc	ttc	atg	ctt	cct	ggc	ccc [.]	tgc	aaa	at

Exon 14 Start: 2401 End: 2574 Length: 173
2391 2401 2411 2421 2431 2441 GTTTTCACTGGGATCTTTACAGCAGAAATGTTTCTGAAAATTATTGCCATGGATCCTTAC
V F T G I F T A E M F L K I I A M D P Y 801
2451 2461 2471 2481 2491 2501
TATTATTTCCAAGAAGGCTGGAATATCTTTGACGGTTTTATTGTGACGCTTAGCCTGGTA Y Y F Q E G W N I F D G F I V T L S L V 821 831
2511 2521 2531 2541 2551 .
GAACTTGGACTCGCCAATGTGGAAGGATTATCTGTTCTCCGTTCATTTCGATTGgtaaaa E L G L A N V E G L S V L R S F R L
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Exon 15 Start: 2575 End: 2931 Length: 356	
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2561 2571 2581 2591 2601 2611 CTGCGAGTTTTCAAGTTGGCCAAAATCTTGGCCAACGTTAAATATGCTAATAAAGATCATGLRVFKLLRVFKLLRVFKLLRVFKLLRVFKLRVFKLRVFKL	
2621 2631 2641 2651 2661 2671 GGCAATTCCGTGGGGGGCTCTGGGAAATTTAACCCTCGTCTTGGCCATCATCGTCTTCAT	Г
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GC	CAG'	TGA	ГТG	TCA	ACT	CCC.	ACG	CTG	GCA	CAT	GAA	TGA	CTT	CTT	CCA	CTC	CTT	CCTGATT
A	S	D	С	Q	L	P	R		H 21	M	N	D	F	F	Η	S	F	L I 931
	1:	280:	1		28	11		12	821		ı	283	1		28	41		2851
GT	GTT	CCG	CGT	GCT	GTG'	TGG	GGA	GTG	GAT	AGA	GAC	CAT	GTG	GGA	CTG	TAT	'GGA	GGTTGCT
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AA	GAT'	TTT.	AGA'	TGA	AAT	TAA	ACC	ACT:	ΓGA7	rga'	ГСТ	AAA	CAA	CAA	GAA	AGA	CAG	TTG	ΓATG
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		- 13	316	1		31	71		3:	181		- 1	319	1		32	01		3211
TC	CAA'	TCA'	TAC	AGC	AGA	AAT'	TGG	GAA	AGA:	rct'	ΓGΑ	CTA	TCT	TAA	AGA	TGT	AAA	TGG	AACT
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		- 13	322	1		132	31		132	241		- 1	325	1		132	61		3271
AC.	AAG'	TGG'	TAT.	AGG	AAC	TGG	CAG	CAG	rgt:	ΓGA.	AAA	ATA	CAT	TAT	TGA	TGA	AAG	TGA	ΓΤΑC
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		1:	328	1		132	91		33	301		- 1	331	1		33	21		3331
AT	GTC.	ATT	CAT.	AAA	CAA	.CCC	CAG	TCT:	rac:	rgt(GAC	TGT	ACC	AAT	TGC	TGT	AGG.	AGA	ATCT
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		- 13	334	1		133	51		133	361		- 1	337	1		133	81		3391

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Ex	on	17	St	art	: 3	415	1	End	d: 3	353	5	Le	ngtl	h: :	120				
				tct												tgtg	ggc1	Egat	ggc
				cat													ttai	zggt	aaa
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	1:	340:	1	1	341	1		34	121		13	343	1		34	41		34	151

AA	ACT	GAA'	ΓGA.	AAG	CAG	TAG	CTCA	TC	AGA.	AGG'	ΓAG	CAC	rgt(GGA	CAT	CGG	CGC	ACCT	GTA
K	L	N	Ε	S	S	S	S	S	E	G	S	Т	V	D	I	G	Α	P	V
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gc	cca	ata	tgt	gga	aga	gat	taag	gaad	cat	gga	gga	aaaa	atat	ttt	tagt	taco	cato	caaa	act
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tc	taa	ctt	ata	ttt	tta	aag	taag	gtag	gtg	att [.]	tca	caat	taag	gtt	ggca	atct	ttag	gaca	cta
С																			
Ex	on	18	l S	tar	t:	353	6	End	l:	369) (Lei	ngth	1:	154				
tg	ata	tgg	tac	aag	gaa	aag	ctac	cto	cac	att	tat	ttgt	taat	tt	acaa	aagt	ttt	ctct	tgg
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3581
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caccaaaggatgtgaatattcaattactcattgtt
Exon 19 Start: 3691 End: 3864 Length: 173

	ggaaaaaaaaaagtaacagtaac
tactgtttctctgccctcctattccaatgaaatgtca	
3681 3691 3701 GCATTTGAAGATATATATTGATCAGCGAAAGACGA	3711 3721 3731
A F E D I Y I D Q R K T I	
3741 3751 3761	
GACAAGGTTTTCACTTACATTTTCATTCTGGAAATGC D K V F T Y I F I L E M I 1251	
3801 3811 3821 TATCAAACATATTTCACCAATGCCTGGTGTTGGCTG	GACTTCTTAATTGTTGATgtaggt
Y Q T Y F T N A W C W L I	D F L I V D 1281
atcgttcatatttttgtctctgttcaaggtagcttg	tcttatttatattcaaattctaca
. atagtgagtctcagaccactatgttatgttgacagac	
	aatttaattctgatttaaaataac
Exon 20 Start: 3865 End: 3987 I	Length: 122

${\tt ttgatttctttttttttttattttactaatttttactttgctttttttt$.gtt
	aaa
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gcaactacacattgaatgatgattctgtttattaattgttattatttttgtgtgtg	agg
3851 3861 3871 3881 3891 39 GTTTCATTGGTCAGTTTAACAGCAAATGCCTTGGGTTACTCAGAACTTGGAGCCATC V S L V S L T A N A L G Y S E L G A I 1291 13	SAAA K
3911 3921 3931 3941 3951 39 TCTCTCAGGACACTAAGAGCTCTGAGACCTCTAAGAGCCTTATCTCGATTTGAAGGG S L R T L R A L R P L R A L S R F E G 1311 13	ATG M
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	ctc
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Exon 21	Start:	3988	End:	4269	Length:	281
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gca	agca	aag	ttc	ctc	tga	tttt										ctc		ccc1	tttt
tti	tta	aaa	aca	ttt	ttg	ttto	cat	ttc	ttg	gtt	ata	ttg	cct	ata	gtt	gtt	ttc	ctaa	agtg
tai	ttg	ctt:	aag	aaa	aaa	aaat	zga:	att	tta	aga	ttt	ttt	tga	acc	ttg	ctt	tta	cata	atcc
tag	gaat	taa:	ata	gca	ttg	atag	gaa	aaa	aag	aat	gga	aag	acc	aga	gat	tac	tag	ggg	aatt
tti	ttt	tct	tta	tta	aca						ttt					ttt	gtg	tati	tagg
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				TGC A			G				S S			GAA N			L	V 341	TTGT C
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CT	ΓΑΤ	ATT(CTG	GCT	AAT	TTTC											CAA.	ATT(CTAC
L	Ι	F	W	L	Ι	F		I 351	M	G	V	N	L	F	A	G		F 361	Y
4	109:	1		41	01		14	111		- 1	412	1		141	31		14	141	
CAC	CTG	ΓΑΤ															TAA'	TCAT	ГАСТ
Н	С	Ι	N	Т	T	Т		D 371	R	F	D	Ι	Ε	D	V	N		Н 381	Т
4	115:	1		41	61		14	171		1	418	1		41	91		4:	201	
GA	ΓTG	CCT	AAA	ACT	AAT	AGA/	AAG.	AAA'	TGA	GAC	TGC	TCG	ATG	GAA	AAA	TGT	GAA.	AGT	AAAC
D	С	L	K	L	Ι	E		N 391	Е	T	A	R	W	K	N	V		V 401	N
4	121:	1		42	21		14:	231		1	424	1		142	51.				
TT	ΓGA:	ΓΑΑΊ															aca	ctat	tttt
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tgaccagaccaaacaaagacctctttgtctttttttttt
gctctttctatataaacaagtaaattaataaatcagtaactt
Exon 22 Start: 4270 End: 4323 Length: 53
4261 4271 4281 4291 4301 . GCCACATTCAAAGGATGGATGTATAATGTATGCAGCAGTTGATTCCAGAAATgtaagt A T F K G W M D I M Y A A V D S R N 1421 1431

${\tt caggtaaatattcaagcatagcaatgtttatcagaaagatcttactaagataattcaaca}$									
tccttttaaatattttctattttgtttcctatttatccacacattatataatta									
Exon 23 Start: 4324 End: 4461 Length: 137									
4311									
4371									
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aaaactagaaagcaaccg
Exon 24 Start: 4462 End: 4566 Length: 104
tttgggggtgtttgtcttcataagataataataataataagtcaattttgaattatttaggg
4451 4461 4471 4481 4491 4501 TTTGGAGGTCAAGACATCTTTATGACAGAAGAACAGAAGAAATACTATAATGCAATGAAA F G G Q D I F M T E E Q K K Y Y N A M K 1491 1501
4511 4521 4531 4541 AAATTAGGATCGAAAAAACCGCAAAAGCCTATACCTCGACCAGGAgtaagaagtatcaaa K L G S K K P Q K P I P R P G 1511
tgatatgggggaaaaatacaaaaacaaaaacttgcatgcttgtctcacaaaaaagaaag

agctaacatttccagtagaaaaaatttctttcacttgtagagcatcatctaaaagtgatg											
tttgtatcttttatgataatggtaacaacaaaaaaaattaaagaccaaaaaagaacaaga											
Exon 25 Start: 4567 End: 4837 Length: 270											
4551 4561 4571 4581 4591 4601											
AACAAATTTCAAGGAATGGTCTTTGACTTCGTAACCAGACAAGTTTTTGACATAAGCATC											
N K F Q G M V F D F V T R Q V F D I S I 1521 1531											
4611 4621 4631 4641 4651 4661											
ATGATTCTCATCTGTCTTAACATGGTCACAATGATGGTGGAAACAGATGACCAGAGTGAA											
M I L I C L N M V T M M V E T D D Q S E 1541 1551											
4671 4681 4691 4701 4711 4721 TATGTGACTACCATTTTGTCACGCATCAATCTGGTGTTCATTGTGCTATTTACTGGAGAG											

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ga	ıgat	aat	cac	tgt	aaa	tat	· ttt	gat	aca	ttt	tat	cca	agtg	gtg	tttį	gtt	ttt	tta:	acaa
aa	ıtgt	ctc	cta	cta	tatį	gtc						tct				gaa	caa	tat	actt
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Ex	on :	26	l S¹	tar	t: 4	483	8	En	d: :	810	0	Le	ngtl	n:	326:	2			
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aa	ıaca [.]	tct	ctt	aaa	gata	aaa	aaa	aag	tac	tat	aat	cct	tata	aat	tga	aat	tat	tgc	tgaa
tt	tca	aaa	tta	aga	ttt:	ata	ata	tat	tat	gta	caa	itta	tata	ata	tat	tat	ata	tag	ttgt

 $\verb|catatttaatataactgggttcaggactctgaaccttaccttggagctttagaagaaaca|\\$

 ${\tt tatgtttattttaacgcatgatttcttcactggttggtattctcattgtttattcatagg}$ 4821 4831 4841 |4851 4861 GTATGTTTCTTGCCGAGCTGATAGAAAAGTATTTCGTGTCCCCTACCCTGTTCCGAGTGA M F L A E L I E K Y F V S P T L F R V I |1611 1621 14881 14891 4901 4911 4921 TCCGTCTTGCTAGGATTGGCCGAATCCTACGTCTGATCAAAGGAGCAAAGGGGATCCGCA R L A R I G R I L R L I K G A K G I R T |1631 l 1641 14941 14951 14961 14971 14981 14991 $\tt CGCTGCTCTTTGCTTTGATGATGTCCCTTCCTGCGTTGTTTAACATCGGCCTCCTACTCT$ LLFALMMSLPALFNIGLLLF |1651 |1661 15001 |5011 15021 15031 15051 15041 TCCTAGTCATGTTCATCTACGCCATCTTTGGGATGTCCAACTTTGCCTATGTTAAGAGGG LVMFIYAIFGMSNFAYVKRE 1671 1681 15061 |5071 |5081 |5091 |5101 AAGTTGGGATCGATGACATGTTCAACTTTGAGACCTTTGGCAACAGCATGATCTGCCTAT V G I D D M F N F E T F G N S M I C L F 1691 |1701 |5121 |5131 |5141 |5151 |5161 |5171 TCCAAATTACAACCTCTGCTGGCTGGGATGGATTGCTAGCACCCATTCTCAACAGTAAGC Q I T T S A G W D G L L A P I L N S K P |1711 |1721 l5191 I5181 15201 |5211 |5221 15231 CACCCGACTGTGACCCTAATAAAGTTAACCCTGGAAGCTCAGTTAAGGGAGACTGTGGGA P D C D P N K V N P G S S V K G D C G N 1731 11741 |5241 |5251 |5261 |5271 |5281 PSVGIFFFVSYIIISFLVVV 1751 1761 |5301 |5311 |5321 |5331 |5341 |5351

|1771 |1781

|5721 |5731 |5741 |5751 |5761 |5771 |
CTGCTGTCATTATTCAGCGTGCTTACAGACGCCACCTTTTAAAGCGAACTGTAAAACAAG
A V I I Q R A Y R R H L L K R T V K Q A |1911 |1921

|5781 |5791 |5801 |5811 |5821 |5831 CTTCCTTTACGTACAATAAAAACAAAATCAAAGGTGGGGCTAATCTTCTTATAAAAGAAG S F T Y N K N K I K G G A N L L I K E D |1931 |1941

|5841 |5851 |5861 |5871 |5881 |5891
ACATGATAATTGACAGAATAAATGAAAACTCTATTACAGAAAAAACTGATCTGACCATGT
M I D R I N E N S I T E K T D L T M S
|1951 |1961

CCAC	TGC	AGC	TTGTCC.	ACC	TTC	CTA	TGA	CCG	GGT	GAC	AAA	GCC	'AAT	TGT	'GGA	AAA	ACA	TG
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			•															
150	61		15071		- 1	508	1		150	Q1	1	+ 1			l ± 1	1		L+21
5961 5971 5981 5991 +1 +11 +21 AGCAAGAAGGCAAAGATGAAAAAAGCCAAAGGGAAATAAAT																		
											AAI	GAR	AAI.	AAA	IAA	AAA	.IAA	.11
Ų	Ł	G	K D		K	A	K	G	K	*								
			1991															
		- 1	+31		+4	1		+	51			+61			+7	1		+81
GGGT	GAC	AAA	TTGTTT.	ACA	.GCC	TGT	'GAA	GGT	GAT	GTA	TTT	'TTA	TCA.	ACA	GGA	CTC	CTT	'TA
		- 1	+91		+1	01		+	111		- 1	+12	1		+1	31		+141
GGAG	GTC		GCCAAA															
		- 1	+151		l + 1	61		1+	171		1	+ 18	:1		l ± 1	Q1		+201
A A C A	CAC																	.CA
AAGA	CAG	IGA	CCCCTT	GIU	AGO	AAA	CIG	11 GA	CIC	161	GIA	AAC	uuu.	AGA	IGA	.001	IGA	CA
						٠.												1.004
		-																+261
GGAG	GTT	ACT	GTTCTC.	ACT	ACC	AGC	TGA	CAC	TGC	TGA	AGA	TAA	GAT.	GCA	CAA	TGG	CTA	.GT
		- 1	+271		1+2	81		+	291			+30	1		1+3	11		+321
CAGA	CTG	TAG	GGACCA	GTT	TCA	AGG	GGT	GCA	AAC	CTG	TGA	TTT	TGG	GGT	'TGT	'TTA	ACA	TG
		- 1	+331		1+3	41		1+	351		- 1	+36	1		1+3	71		+381
ΔΔΔα	ΔСТ																	.CA
	1101		ararna	1 1111		1111	0011	.010		u 011		0111	.014	001	.0111		1 011	.011
		- 1	±201		١٨٨	Λ1		14	111			Τ // C	1		1 _ 1	21		+441
mmmm	т л т																	
1111	TTTTTATGGAATCTGTTAGTGGATTCATCTTTTTGTTAATCCATGTGTTTATTATATGTG																	
																		+501
ACTA	TTT	TTG	TAAACG	AAG	TTT	CTG	TTG	AGA	AAT	AGG	CTA	AGC	ACC'	TCT	'ATA	ACA	GGT	'AT
		- 1	+511		+5	21		+	531			+54	1		+5	51		+561
GCCA	CCT	GGG	GGGTAT	GGC	AAC	CAC	ATG	GCC	CTC	CCA	GCT	'ACA	CAA.	AGT	'CGT	'GGT	'TTG	CA
		- 1	+571		1+5	21		1+	591		- 1	+60	1		1+6	11		+621
тслс	ccc																	CT
IGAG	GGC	AIG	CIGCAC	111	GAG	AIC	AIG	ICHI	GAG	HHH	HAG	TOF	CAA	GHH	AAA	CAA	.All	CI
									054						1.0			1.004
																		+681
TAAA	TTT	CAC	CATATT	TCT	GGG	AGG	GGT	TAAT	TGG	GTG	ATA	AGT	GGA	GGT	GCT	"TTG	TTG	AΤ
			+691		+7	01		+	711			+72	1		+7	31		+741
CTTG	TTT	TGC	GAAATC	CAG	CCC	CTA	GAC	CAA	GTA	GAT	TAT	TTC	TGG	GTA	GGC	CAG	TAA	AT

|5931

|5941

|5951

|5901

|5911 |5921

+811 +821 +831 +841 +851 +861 TGTATTAAAAAAAAACCTGAATAGTGAATATTGCCCCTCACCCTCCACCGCCAGAAGAC +871 l+881 |+891 l+901 +921 +911 TGAATTGACCAAAATTACTCTTTATAAATTTCTGCTTTTTCCTGCACTTTGTTTAGCCAT I+931 l+941 l+951 |+961 l+971 l+981 l+991 l+1001 1+1011 l+1021 l+1031 1+1041 ${\tt TAGTCATTTTACCCTGTGGTGCACGTTTGAGCAAACAAATAATGACCTAAGCACAGTATT}$ +1051 |+1061 +1071 +1081 +1091 |+1101 TATTGCATCAAATATGTACCACAAGAAATGTAGAGTGCAAGCTTTACACAGGTAATAAAA 1+1111 1+1121 l+1131 1+1141 l+1151 l+1161 TGTATTCTGTACCATTTATAGATAGTTTGGATGCTATCAATGCATGTTTATATTACCATG +1171 +1181 +1191 +1201 +1211 |+1221 $\tt CTGCTGTATCTGGTTTCTCTCACTGCTCAGAATCTCATTTATGAGAAACCATATGTCAGT$ 1+1231 +1241 +1251 +1261 +1271 |+1281 +1291 |+1301 |+1311 +1321 +1331 |+1341 $\tt GCAGCACTTTAACAGCTTTTTGGTTATTTTTACATTTTAAGTGGATAACATATGGTATAT$ l+1351 |+1361 |+1371 |+1381 l+1391 l+1401 AGCCAGACTGTACAGACATGTTTAAAAAAAACACACTGCTTAACCTATTAAATATGTGTTT +1411 +1421 +1431 +1441 +1451 +1461 AGAATTTTATAAGCAAATATAAATACTGTAAAAAGTCACTTTATTTTATTTTCAGCATT |+1521 +1471 |+1481 +1491 |+1501 +1511 ATGTACATAAATATGAAGAGGAAATTATCTTCAGGTTGATATCACAATCACTTTTCTTAC l+1531 l+1541 l+1551 l+1561 l+1571 l+1581 $\tt TTTCTGTCCATAGTACTTTTCATGAAAGAAATTTGCTAAATAAGACATGAAAACAAGAC$ +1591 +1601 +1611 |+1631 +1621 +1641

+751

|+761 |+771

+781

l+791

+801

 $\tt TGGGTAGTTGTAGATTTCTGCTTTTTAAATTACATTTGCTAATTTTAGATTATTTCACAA$

TTTTAAGG					+1691 CAATTGGAAA	
GGTTTAAA					+1751 TGAAGGTAGT	
TTATGTTA					+1811 TCTTTGGAGT	
TGCTGCTC					+1871 TTCCACAAAA	
AGAGTAGT					+1931 TTACAGAAGC	
ACCATAGG					+1991 TGAACTGCAT	
TGGAAAATO					+2051 GTGCAAAACT	
TAAAGATTA	•	+2081 . TTTTAttgtt		 ctttttgtta	acagaatatt	ct
	 ggagatttt	 tacattacaa	 attgggggag	 aaggggagcg	 cgcgcgcaca	ca
cacacaca	 cacacacaca	cacacacaca	cacacagagg	catacccacg	 ttgacaacaa	aa
cctagggta	 agatatgtca	ctggaggtag	 ggggtaatga	 cctcccagaa	 ttacaagcag	ca
· · · ggtgtgtt	ctctgttagg	aggaagaact	ggtgtcagag	gatagctagt	 gattctagga	gg
	 tatggaagcc					