Gene: GJB2 - Sequence: NG_008358.1 Transcript: NM_004004.5 - Protein: NP_003995.2 Date : February 26, 2015

1^{st} line: Base numbering. Full stops for intronic $+/-$ 5, 10, 15 2^{nd} line: Base sequence. lower case Introns, upper case Exons 3^{rd} line: Amino acid sequence. Printed on FIRST base of codon 4^{th} line: Amino acid numbering. Numbered on 1^{st} and increments of 10
Exon 1 Start: 5001 End: 5193 Length: 192
-209 -199 -189 -179 -169 -159 GGGGTGCGGTTAAAAGGCGCCACGGCGGGAGACAGGTGTTGCGGCCCCGCAGCGCCCGCG
-149 -139 -129 -119 -109 -99 CGCTCCTCTCCCCGACTCGGAGCCCCTCGGCGGCCCCAGGACCCGCCTAGGAGC
-89 -79 -69 -59 -49 -39 GCAGGAGCCCCAGCGCAGACCCCCAACGCCGAGACCCCCGGCCCCGGCCCCGCCCCGCCCTT
-29

cggccg	agctc	cggag	ctgg	aatgg	ggcgg	ccggg	gaagt	ggacg	cgatg	gcacc	gcccg	
gggtgo					cgggag			gcgcg	ggcga	gccgc	cagcg	
cgaggt	ttgtg	gt										

Exc	n 2	I	Sta	rt:	83	/3	Enc	1:	108	013	Ι.	Len	gth	: 2	140				
gac	act	ccc	cag	cac	agc	aaat	tttt	tat	gat	tgt	gtt	taa	aga	ttg	ggt	gaat	ttac [,]	tca	ıgg
U			J		J				0		0		O	J	00	0			00
tga	.aca	agc	tac	ttt	tta	tcag	gagaa	aca	.cc1	taa	aaa	cac	gtt	caa	gag	ggt:	ttgg	gaa	ıct
		_											_					_	
ata	.cat	tta	atc	cta	tga	caaa	actaa	agt	tgg	gtt	ctg	tct	tca	.cct	gtt	ttgg	gtga	ggt	tg
tgt	aag	agt	tgg	tgt	ttg	ctca	aggaa	aga	gat	ttt	aag	cat	gct	tgc	tta	ccca	agac	tca	ıga
										•									
gaa	gtc	tcc	ctg	ttc	tgt	ccta	agcta	agt	gat	ttc	ctg	tgt	tgt	gtg	cat	tcg	tctt	ttc	ca
	-19	9		-	9		1			I	11			21			31		
AGC	AAA	CCG	CCC	AGA	GTA	GAAC	GATG(M I		TG(W			GCT L	GCA Q	GAC T	GAT I	CCT(GGGG G		GT V
							11	,	w	u	1	ь	ų	1	1	ь	111	ď	V
ı	41			51			61			1	71			81			91		
GAA	CAA			CAC	CAG		rgga <i>i</i>	AAG	AT(CTG	GCT			CCT	CTT		TTTT		
N	K	H	S	T	S	Ι	G F	X	Ι	W	L	T	V	L	F	Ι	F :	R	Ι
1	101			11	1		121	1		ı	131			14	1		115	1	
		CCT	CGT			TGC#	AAAG							-		CGA	•		TG
M	Ι	L	V	V	A	A	K F 41	Ξ	V	W	G	D	E	Q	A	D	F 51	V	C
																	101		
	161 CAC	ССТ	GCA	17 GCC		CTGO	181 CAAG <i>I</i>				191 CTA		тса	120 CTA		CCC	21 CATC		:CA
N	T	L	Q				K 1							Y	F	P	I		Н
							61										71		
	221			123			1241				251			126			127		
							GCTG <i>I</i> L												
1	16		w	А	ь	Ų	81	L	r	V	b	1	Г	А	ь	ь	91		II.
ı	281			29	1		301	1		1	311			32	1		33	1	
							rgag <i>i</i>										GATA.	AAG	AG
Н	V	A	Y	R	R	Н	E H		K	R	K	F	Ι	K	G	E	I :		S

	341			35															
TGA.	ATT	TAA	GGA	CAT	CGA	GGA			AAC	CCA	GAA	GGT	CCC	CAT	'CGA			CCT	GTG
E	F	K	D	Ι	Ε	E	Ι	K	T	Q	K	V	R	Ι	Ε	G	S	L	W
							1	21									13	31	
- 1	401			41	1		4	21		- 1	431			44	1		4	51	
GTG	GAC	CTA	CAC	AAG	CAG	CAT	CTT	CTT	'CCG	GGT	CAT	CTT	CGA	AGC	CGC	CTT	CAT	GTA	CGT
W	T	Y	T	S	S	Ι	F	F	R	V	Ι	F	Ε	Α	Α	F	М	Y	V
							1	41									1	51	
- 1-	461			47	1		4	81		- 1	491			150	1		5	11	
CTT	CTA	TGT	CAT	GTA	CGA	CGG										CAA	CGC	CTG	GCC
F	Y	V	M	Y	D	G	F	S	M	Q	R	L	V	K	C	N	Α	W	P
							1	61									1'	71	
1.	521			53	1		5	41		- 1	551			156	1		5	71	
TTG	TCC	CAA	CAC	TGT	GGA	CTG	CTT	TGT	GTC	CCG	GCC	CAC	GGA	GAA	GAC	TGT	CTT	CAC.	AGT
C	P	N	T	V	D	C	F	V	S	R	Р	T	Ε	K	T	V	F	T	V
							1	81									19	91	
1.	581			59	1		16	01		- 1	611			162	1		163	31	
GTT	CAT	GAT'	TGC	AGT	GTC	TGG	AAT	TTG	CAT	CCT	GCT	GAA	TGT	'CAC	TGA	ATT	GTG	ГТА	TTT
F	M	Ι	Α	V	S	G	Ι	С	Ι	L	L	N	V	Т	E	L	C	Y	L
							12	01									12	11	
1	641			65	1		16	61		- 1	671			168	1		:	*11	
GCT.	AAT'	TAG.	ATA	TTG	TTC	TGG	GAA	GTC	AAA	AAA	GCC	AGT	TTA	ACG	CAT	TGC	CCA	GTT	GTT
L	Ι	R	Y	C	S	G	K	S	K	K	Р	V	*						
							12	21											
	*2	1		*	31		- 1	*41			 *5	1		*	61		:	*71	
AGA'	ГТА	AGA.	AAT	AGA	CAG	CAT	GAG	AGG	GAT	GAG	GCA	ACC	CGT	GCT	'CAG	CTG	TCA	AGG	CTC
	*8	1		*	91		- 1	*10	1		*1	11		*	121		:	*13	1
AGT	CGC'	TAG	CAT	TTC	CCA	ACA	CAA	AGA	TTC	TGA	CCT	TAA	ATG	CAA	.CCA	TTT	GAA	ACC	CCT
	*1	41		*	151		- 1	*16	1		*1	71		*	181		:	*19	1
GTA	GGC	CTC.	AGG	TGA	AAC	TCC	AGA	TGC	CAC	AAT	GGA	GCT	CTC	CTC	CCC	TAA	AGC	CTC.	AAA
		Λ 1																	
	*2	OΙ		*	211			*22	1		*2	31		*	241		:	*25	1
ACA.	•																		
ACA.	•																		
	•	GCC'	TAA	TTC	TAT	GCC'	TGT	CTT	'AAT	TTT	CTT	TCA	CTT	'AAG	TTA	GTT		CTG.	AGA

l*321 l*331 l*341 |*351 l*361 l*371 GCATTTGTTTCTCTGAGGACAAGAGAAAAAAGCCAGGTTCCACAGAGGACACAGAG **|***381 |*391 **|***401 **|***411 **|***421 |*431 AAGGTTTGGGTGTCCTCCTGGGGTTCTTTTTGCCAACTTTCCCCACGTTAAAGGTGAACA |*451 |*461 **|***471 |*481 |*491 l*441 TTGGTTCTTTCATTTGCTTTGGAAGTTTTAATCTCTAACAGTGGACAAAGTTACCAGTGC l*511 l*521 l*531 l*541 l *551 $\tt CTTAAACTCTGTTACACTTTTTGGAAGTGAAAACTTTGTAGTATGATAGGTTATTTTGAT$ l*561 |*571 |*****581 **|***591 l*601 l*611 GTAAAGATGTTCTGGATACCATTATATGTTCCCCCTGTTTCAGAGGCTCAGATTGTAATA |*621 **|***631 **|***641 **|***651 |*661 |*671 TGTAAATGGTATGTCATTCGCTACTATGATTTAATTTGAAATATGGTCTTTTGGTTATGA |*731 l*681 l*691 l*701 l*711 l*721 ATACTTTGCAGCACAGCTGAGAGGCTGTCTGTTGTATTCATTGTGGTCATAGCACCTAAC l*741 l*751 l*761 |*771 l*781 l*791 AACATTGTAGCCTCAATCGAGTGAGACAGACTAGAAGTTCCTAGTGATGGCTTATGATAG |*801 |*811 **|** *821 |*831 **|***841 |*****851 CAAATGGCCTCATGTCAAATATTTAGATGTAATTTTGTGTAAGAAATACAGACTGGATGT **|***861 |*****871 |*****881 |*891 |*901 |*911 ACCACCAACTACTACCTGTAATGACAGGCCTGTCCAACACATCTCCCTTTTCCATGACTG |*921 l*931 l*941 l*951 l*961 l*971 TGGTAGCCAGCATCGGAAAGAACGCTGATTTAAAGAGGTCGCTTGGGAATTTTATTGACA |*991 |*1001 |*1011 l*1021 l*1031 l*981 CAGTACCATTTAATGGGGAGGACAAAATGGGGCAGGGGAGGAGAAGTTTCTGTCGTTAA I*1041 l*1051 l*1061 l*1071 l*1081 l*1091 AAACAGATTTGGAAAGACTGGACTCTAAAGTCTGTTGATTAAAGATGAGCTTTGTCTACT |*1111 |*1121 |*1131 |*1141 TCAAAAGTTTGTTTGCTTACCCCTTCAGCCTCCAATTTTTTAAGTGAAAATATAGCTAAT | *1161 |*1171 **|***1181 |*1191 |*1201 | *1211 AACATGTGAAAAGAATAGAAGCTAAGGTTTAGATAAATATTGAGCAGATCTATAGGAAGA

|*1271

|*1231 |*1241 |*1251 |*1261

|*1221

TTGAACCTGAATATTGCCATTATGCTTGACATGGTTTCCAAAAAATGGTACTCCACATAT
*1281 *1291 *1301 *1311 *1321 *1331 TTCAGTGAGGGTAAGTATTTTCCTGTTGTCAAGAATAGCATTGTAAAAGCATTTTGTAAT
*1341 *1351 *1361 *1371 *1381 *1391 AATAAAGAATAGCTTTAATGATATGCTTGTAACTAAAATAATTTTGTAATGTATCAAATA
*1401 *1411 *1421 *1431

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