

Exon 2

Intron 1

gccttat caacagtaaa acaatgaatc accatagtac atgggtcttt tctgaaatac atatttctcc cttttaaatc

	26	27	28	29	30	31	32	33	34	35	36	37	38	39	40	41	42	43	44
	Arg	Cys	Gly	Gln	Pro	Leu	Gln	Asn	Lys	Val	Gln	Leu	Lys	Gly	Arg	Asp	Leu	Leu	Thr
tctttttacag	G	TGT	GGA	CAA	CCA	CTA	CAA	AAT	AAA	GTG	CAG	CTG	AAG	GGC	CGT	GAC	CTT	CTC	ACT

45	46	47	48	49	50	51	52	53	54	55	56	57	58	59	60	61	62	63	64	65	66
Leu	Lys	Asn	Phe	Thr	Gly	Glu	Glu	Ile	Lys	Tyr	Met	Leu	Trp	Leu	Ser	Ala	Asp	Leu	Lys	Phe	Arg
CTA	AAA	AAC	TTT	ACC	GGA	GAA	GAA	ATT	AAA	TAT	ATG	CTA	TGG	CTA	TCA	GCA	GAT	CTG	AAA	TTT	AGG

G
Arg

67 68 69 70 71 72

Ile Lys Gln Lys Gly Glu

Intron 2

ATA AAA CAG AAA GGA GAG gtatgtaaca ttttcttttt acgttccatt actaccagtc cccttttttt aaaggcagcc

ttcccaaaga aagagggaaa aaaatacatt aaaattctta aacagtagct aagtaatgaa acctcacagt caaggtaatt

gattatcatg gagcatacag gcatacaaaa aagcataaaa

Exon 3

tgaaccaccacac ctggcctaaa ttcacttttt aaacaatatt ttaaactt atttgggggt agttattact

Intron 2

tattttctaa taaagaatat gttttaaaac ataatttata tataagatat attttaattc tattcttgtc cttgatttatag

a

IVS2-54 insA

73	74	75	76	77	78	79	80	81	82	83	84	85	86	87	88	89	90	91	92	93	94
Tyr	Leu	Pro	Leu	Leu	Gln	Gly	Lys	Ser	Leu	Gly	Met	Ile	Phe	Glu	Lys	Arg	Ser	Thr	Arg	Thr	Arg
TAT	TTG	CCT	TTA	TTG	CAA	GGG	AAG	TCC	TTA	GGC	ATG	ATT	TTT	GAG	AAA	AGA	AGT	ACT	CGA	ACA	AGA

95 96 97 98 99 100

Leu Ser Thr Glu Thr Gly

Intron 3

TTG TCT ACA GAA ACA G gtaagt cactgccaa attcacactt gtgttgaaga gagggattga aggtgaagac

tttggagggg taac

Exon 4

Intron 3

ccctcaattcct cattttgctt tgcattggaa ttctgtatca gagaagttgg agagctttaa agattttgac ttttcagttg

agatgatgcc aattccttgt aattttggtt tccactttag ttgttttttc aaaatgattt ttttcttttt tttttattgtag

t t a
IVS3-34insT IVS3-17insT IVS3-8A/T

100	101	102	103	104	105	106	107	108	109	110	111	112	113	114	115	116	117	118	119	120	121
Gly	Leu	Ala	Leu	Leu	Gly	Gly	His	Pro	Cys	Phe	Leu	Thr	Thr	Gln	Asp	Ile	His	Leu	Gly	Val	Asn
GC	TTT	GCA	CTT	CTG	GGA	GGA	CAT	CCT	TGT	TTT	CTT	ACC	ACA	CAA	GAT	ATT	CAT	TTG	GGT	GTG	AAT
	A										C										
	Leu										Pro										

122 123 124 125 126 127 128 129

Glu Ser Leu Thr Asp Thr Ala Arg

Intron 4

GAA AGT CTC ACG GAC ACG GCC CG gtttgtaaatt attttcttct ctccaaagct gatttcagaa tctgatggat

g
IVS4+8A/G

aaatttcaaa aataaaacat aattctcttt aaataatggt tttccctcta attgttctgt tctccatttt ccctctttcc

ctgatattct gaatacctct ccctcatcct gtggccactt tccatcggtt gaaataat

Exon 5

Intron 4

gttggtaggttttcatg tagtaaaaat tctatatttatt agtattttcctt gttattattt ctgatcttat attaaataa

129 130 131 132 133 134 135 136
Arg Val Leu Ser Ser Met Ala Asp
ggcattattaa gcataattat cttagattat ctttttcttg gtttgccacag T GTA TTG TCT AGC ATG GCA GAT

a
IVS4-7A/G

137 138 139 140 141 142 143 144 145 146 147 148 149 150 151 152 153 154 155 156 157 158
Ala Val Leu Ala Arg Val Tyr Lys Gln Ser Asp Leu Asp Thr Leu Ala Lys Glu Ala Ser Ile Pro
GCA GTA TTG GCT CGA GTG TAT AAA CAA TCA GAT TTG GAC ACC CTG GCT AAA GAA GCA TCC ATC CCA
C C
Tyr Leu

159 160 161 162 163 164 165 166 167 168 169 170 171 172 173 174 175 176 177 178 179 180
Ile Ile Asn Gly Leu Ser Asp Leu Tyr His Pro Ile Gln Ile Leu Ala Asp Tyr Leu Thr Leu Gln
ATT ATC AAT GGG CTG TCA GAT TTG TAC CAT CCT ATC CAG ATC CTG GCT GAT TAC CTC ACG CTC CAG

Intron 5

gttggtttatttatt tgtcttataa aagagcaaaa tcaaataatt cctgacttgc ttttaagttaa acagtttact

taaatacatgg tattgg

Exon 6

Intron 5

attggg aattcattcc ttctaacaag gcactaatac tgagattaag actatatttatt ttaaccttag taattgctac

a
IVS5-63G/A

181 182 183 184 185 186 187 188
Glu His Tyr Ser Ser Leu Lys Gly
acataagcga atttacgcct ggatttcatac tccttcatcc cgtgccttttag GAA CAC TAT AGC TCT CTG AAA GGT

189 190 191 192 193 194 195 196 197 198 199 200 201 202 203 204 205 206 207 208 209 210
Leu Thr Leu Ser Trp Ile Gly Asp Gly Asn Asn Ile Leu His Ser Ile Met Met Ser Ala Ala Lys
CTT ACC CTC AGC TGG ATC GGG GAT GGG AAC AAT ATC CTG CAC TCC ATC ATG ATG AGC GCA GCG AAA
C T A
Cys Phe Gly

211 212 213 214 215 216 217 218 219 220 221
Phe Gly Met His Leu Gln Ala Ala Thr Pro Lys
TTC GGA ATG CAC CTT CAG GCA GCT ACT CCA AAG gtagg gaaacttttt gccttgaaac taaccctct

Intron 6

cttaaatacat cctcagatgc aattacccag tgacaaaaaa agctctcttc cattaagta tagcacaggt gaggccacag

aatttaggtt acgttaccag

Exon 7

Intron 6

atcata ttctactgaa catggtggga ccacatcttg aaaaagggaa ggagacgcga tattgaaaag aaaataatat

222 223 224 225 226
Gly Tyr Glu Pro Asp
atatttaaga aaacatgtat aataaaatta cctaaataag atttaaattc ttcctcctttag GGT TAT GAG CCG GAT

227 228 229 230 231 232 233 234 235 236 237 238 239
Ala Ser Val Thr Lys Leu Ala Glu Gln Tyr Ala Lys Glu
GCT AGT GTA ACC AAG TTG GCA GAG CAG TAT GCC AAA GAG gtat gctctttacatgtaaagcta ttattgccttt

Intron 7

tactgtccc atgaagttat ttaaccagcg tgtttatgta tgctagaatg gtaccaagct gttgctgaca aatgatccat

tggaagcagc gcatggaggc aatgtattaa ttacagacac ttggataagc atgggacaag aagaggagaa gaaaaagcgg

ctccaggctt tccaaggtta ccaggttaca atgaaggtac aaattgatgc ctctctgaag gttcattaat tccattcatg

aaggccagaa ccatctaate acttattcac tttagggggag

Exon 8

aatcata ttctactgaa catggtggga ccacatcttg aaaaagggaa ggagacgcga tattgaaaag aaaataatat

atatttaaga aaacatgtat aataaaatta cctaaataag atttaaattc ttctctcttt agggttatga gccggatgct

agtgtaacca agttggcaga gcagtatgcc aaagaggtat gctctttaca tgtaaagcta ttattgcctt ttactgtccc

240 241 242 243 244 245 246 247 248 249 250 251
Asn Gly Thr Lys Leu Leu Leu Thr Asn Asp Pro Leu
atgaagttat ttaaccagcg tgtttatgta tgctag AAT GGT ACC AAG CTG TTG CTG ACA AAT GAT CCA TTG

252 253 254 255 256 257 258 259 260 261 262 263 264 265 266 267 268 269 270 271 272 273
Glu Ala Ala His Gly Glu Asn Val Leu Ile Thr Asp Thr Trp Ile Ser Met Gly Gln Glu Glu Glu
GAA GCA GCG CAT GGA GGC AAT GTA TTA ATT ACA GAC ACT TGG ATA AGC ATG GGA CAA GAA GAG GAG

G
Arg

274 275 276 277 278 279 280 281 282 283 284 285 286 287 289 290
Lys Lys Lys Arg Leu Gln Ala Phe Gln Gly Tyr Gln Val Thr Met Lys
AAG AAA AAG CGG CTC CAG GCT TTC CAA GGT TAC CAG GTT ACA ATG AAG gtacaaattgatgc ctctctgaag
A A
Gln Gln

Intron 8

gttcattaat tccattcatg aaggccagaa ccatctaadc acttattcac tttagggggag

g
IVS8+35G/T

Exon 9

Intron 8

aaaccat cactctgctc ctttgtctct ccctttgcat tgatgtctga ctactggcca tgtgtgtttt tagatactga

290 291 292 293 294
Thr Ala Lys Val Ala
agaaaacaaa tatgactgcc acatataata gtcaaaaagt ggtccttatcc ccatctcttttag ACT GCT AAA GTT GCT

295 296 297 298 299 300 301 302 303 304 305 306 307 308 309 310 311 312 313 314 315 316
Ala Ser Asp Trp Thr Phe Leu His Cys Leu Pro Arg Lys Pro Glu Glu Val Asp Asp Glu Val Phe
GCC TCT GAC TGG ACA TTT TTA CAC TGC TTG CCC AGA AAG CCA GAA GAA GTG GAT GAT GAA GTC TTT

317 318 319 320 321 322 323 324 325 326 327 328 329 330 331 332 333 334 335
Tyr Ser Pro Arg Ser Leu Val Phe Pro Glu Ala Glu Asn Arg Lys Trp Thr Ile Met
TAT TCT CCT CGA TCA CTA GTG TTC CCA GAG GCA GAA AAC AGA AAG TGG ACA ATC ATG gtaagcaaga
G
Ala

Intron 9

aacaaggaat ggaggataag ttctttgtgt ggttccaact tggtcattcatgc

Exon 10

Intron 9

ctgcaaagc tgaaacctaa ttcaccttct ctaggtccct aagcagactg tcgctaattgt ttatccattt ctttctttct

^c
IVS9-70T/C

		336	337	338	339	340	341	342	343	344	345	346	347	348	349	350	351	352	353
		Ala	Val	Met	Val	Ser	Leu	Leu	Thr	Asp	Tyr	Ser	Pro	Gln	Leu	Gln	Lys	Pro	Lys
ttgtttgtg	atcag	GCT	GTC	ATG	GTG	TCC	CTG	CTG	ACA	GAT	TAC	TCA	CCT	CAG	CTC	CAG	AAG	CCT	AAA

^g
IVS9-12G/T

354

Phe ***

TTT TGATgttg tgttacttgt caagaaagaa gcaatgttct tcagtaacag aatgagttgg tttatgggga aaagagaaga

gaatctaaaa aataaaciaa tccctaacac gtggtatggg tgaaccgtat gatatgcttt gccattgtga aactttcctt

aagcctttaa ttaagtgt gatgcactgt aatacgtgct taactttg