

SMN

IPB010304, IPB008191, and IPB005062

Genomic Sequence



Coding Sequence



For help interpreting these results, view the [PARSENP Introduction](#) page.

#	View On Sequence	Nucleotide Change	Effect	Restriction Enzyme Differences from REBASE		PSSM Difference	SIFT Score	Description	Zygoty
				Gained in Variant	Lost from Reference				
1	G C	T711G	S38A	BbvI , BsmI , Fnu4HI , MwoI , TseI	SfaNI	6.6	0.27	5058	Homo
2	G	C793T	Intron		MnII			7840	Homo
3	G	C944T	Intron		BseMII , DdeI			7526	Homo
4	G C	G1115A	G118E	TspDTI				7962	Homo
5	G C	C1143T	D127=	DpnI , MboI , MboII	MaeIII , Tsp45I			8020	Homo
6	G C	C1143T	D127=	DpnI , MboI , MboII	MaeIII , Tsp45I			4281	Homo
7	G C	C1143T	D127=	DpnI , MboI , MboII	MaeIII , Tsp45I			1088	Homo
8	A G C	A1165G	M135V	BtrI , MaeII	NlaIII	7.7	0.22	8305	Homo
	B G C	C1295T	T148I						
9	G	T1255A	Intron					1559	Homo
10	G C	T1399G	F183V	NlaIV				6583	Homo
11	G C	C1402G	P184A		BfiI			8511	Homo
12	G	T1457A	Intron					8371	Homo

13	G	T1587A	Intron		MseI , VspI			195	Homo
14	G	G5894C	Intron	AlwNI , BseMII , DdeI , MsiI	Tsp4CI			6397	Homo
15	G	G5894A	Intron		Tsp4CI , TspRI			6846	Homo
16	G	T5915C	Intron	BsrDI , CviRI				6770	Homo
17	G C	T5963A	S233R	BsmAI , Eco31I	HinfI , Hpy178III , PleI	7.5	0.17	5068	Homo
18	G C	C5984T	D240=		Hpy99I			4211	Homo
19	G C	T6044A	Y260*					8394	Homo
20	G C	G6136A	G262D			9.8	0.22	6507	Homo
21	G C	G6136A	G262D			9.8	0.22	3332	Homo
22	G C	T6139A	L263*					7292	Homo
23	G C	T6168A	S273T		FokI , SfaNI			1589	Homo

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[View Variants on 3D Structure](#)

No [protein homology model](#) was submitted. You may add one using any or all of the fields below.

Blocks Families:

Blocks File: no file selected

Sequence Alignment: no file selected

-OR-

Genomic Sequence

```

gcaaggcaagcgtcgcgtgatttgtacgtcatcagcagctgcgttcatcagagtggcgcacgttggacttg      0
                                                                                       72
                                                                                       IPB010304A (2.7e-34) IC 3.32
                                                                                       M A N G A E D V V F C 11
gaaatattttaaattttcatacatcccc atg gcc aat ggt gca gaa gat gtc gta ttt tgt      133
    
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R G T G Q 16
 cgt ggg act ggt caa gtatgtattgagccttttaaaagaaaagaaaattcgcctatgtagatttagc 200

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 560

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 632

S D D S D I W D D T A L I K 30
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A Y D K A V A S F K 40
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 gS38A[1]

ttaccttgTTTTcttttagtaaaattgatgttgtttttcacctcttttgttcaacag N A L K 44
 t[2] aat gct ttg aaa 818

G E D G A T P Q E N D N P G K K R K 62
 ggt gag gat ggg gcg acc cca caa gaa aac gac aac cca ggg aag aag agg aaa 872

IPB010304B (3.1e-41) IC 3.17
 N N K K N K S R K R C N A A P D K E 80
 aac aac aaa aag aat aag agc agg aag aga tgc aac gca gca ccg gat aaa gag g 927

taattacaaccatctctcagtttgtttggagttataatacaagctgatgaactgaatgtgTTTTgtagtg 80
 t[3] 999

V G D S C Y A F W S E D G N L Y T 97
 cag gtt gga gac tcc tgt tat gcg ttc tgg tct gaa gat ggc aac ctg tac act 1053

A T I T S V D O E K G T C V V F Y T 115
gcc acc att acc tca gtc gac cag gag aag ggc acc tgt gtg gtc ttt tac aca 1107

D Y G N E E E Q N L S D L L T E P P 133
gat tat gga aat gag gag gag cag aac ctc agt gac ctt ctg act gag cct cca 1161
aG118E[4] tD127=[5]
tD127=[6]
tD127=[7]

IPB010304C (1.4e-07) IC 2.65

D M D E D A L K T A N 144
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gM135V[8A]

V K 146
tagatgtttaagcattgaattatgcagttgcataatattgacgtttgtttgctttaacacag gtc aaa 1290
a[9]

E T E S S T E E S D R S F T P Q K S 164
gaa aca gag tcc tcc aca gaa gag agt gat cgc tct ttc acc cca cag aag tcc 1344
tT148I[8B]

G H A K H K S K S N F P M G P P S W 182
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F P S F P P G P P P P P P H F K K 199
ttt ccc agt ttc cca cct gga ccg cca cca cca cca ccc cac ttt aaa aag gtga 1453
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gP184A[11]

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a[12] 1525

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a[13] 1597

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5891

IPB010304D (2.2e-41) IC 3.35
M I P P P P 230
cagtgagatgagtttattattttgtaatggtgataaactggtgcag atg atc cca cca ccg cca 5954
c[14] c[16]
a[15]

P M S P D F G E D D E A L G S M L I 248
cct atg agt cca gac ttt ggg gag gac gac gag gct ttg ggc agt atg ctg att 6008

aS233R[17]

tD240=[18]

S W Y M S G Y H T G Y Y M 261
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 aY260*[19]

atttacaagcatctctttgtctacctgctgttttcactttttactttaattatatttttttaaacag G 262
 ggt 6137
 aG262D[20]
 aG262D[21]

L R Q G R K E A A A S K K S H R K * 280
 tta aga cag ggc cgt aaa gag gct gct gca tcc aag aaa tcg cat cgg aaa taa a 6192
 aL263*[22] aS273T[23]

aaccaacgggtttgttttttttcagtctcataatttcacatggatcactgttaaatttgaatttctagatat 280
 6264

agggaggggtttgggttgagtagatgtg 280
 6291

Coding Sequence

IPB010304A (2.7e-34) IC 3.32
M A N G A E D V V F C R G T G Q | S 17
 atg gcc aat ggt gca gaa gat gtc gta ttt tgt cgt ggg act ggt caa | agt 51

D D S D I W D D T A L I K A Y D K A 35
 gat gat tct gac att tgg gat gat aca gct ttg att aaa gca tac gat aaa gct 105

V A S F K | N A L K G E D G A T P Q 52
 gtt gca tca ttc aag | aat gct ttg aaa ggt gag gat ggg gcg acc cca caa 156
 gS38A[1]

IPB010304B (3.1e-41) IC 3.17
 E N D N P G K K R K N N K K N K S R 70
 gaa aac gac aac cca ggg aag aag agg aaa aac aac aaa aag aat aag agc agg 210

K R C N A A P D K E | V G D S C Y A 87
 aag aga tgc aac gca gca ccg gat aaa gag | gtt gga gac tcc tgt tat gcg 261

F W S E D G N L Y T A T I T S V D O 105
 ttc tgg tct gaa gat ggc aac ctg tac act gcc acc att acc tca gtc gac cag 315

E K G T C V V F Y T D Y G N E E E Q 123
 gag aag ggc acc tgt gtg gtc ttt tac aca gat tat gga aat gag gag gag cag 369
 aG118E[4]

N L S D L L T E P P D M D E D A L K 141
aac ctc agt gac ctt ctg act gag cct cca gac atg gat gaa gat gct ctg aag 423
tD127=[5] gM135V[8A]
tD127=[6]
tD127=[7]

IPB010304C (1.4e-07) IC 2.65
T A N | V K E T E S S T E E S D R S 158
aca gca aat | gtc aaa gaa aca gag tcc tcc aca gaa gag agt gat cgc tct 474
tT148I[8B]

F T P Q K S G H A K H K S K S N F P 176
ttc acc cca cag aag tcc ggt cat gca aag cac aaa tct aaa agc aat ttt cct 528

M G P P S W F P S F P P G P P P P P 194
atg gga ccc cca tca tgg ttt ccc agt ttc cca cct gga ccg cca cca cca cca 582
gF183V[10]
gP184A[11]

P H F K K | M D G R R G E G P G P S 211
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IPB010304D (2.2e-41) IC 3.35
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ttt cct gga tgg cct ccc atg att cca ctc ggt cca ccg | atg atc cca cca 684

P P P M S P D F G E D D E A L G S M 246
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aS233R[17] tD240=[18]

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aG262D[20]
aG262D[21]

R Q G R K E A A A S K K S H R K * 280
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aS273T[23]