

SNP Annotation is a "Snap"

This article was featured in [Library Notes #48](#) (Fall 2008).

A common question asked of bioinformatics consultants in libraries is "What can I use for SNP annotation?" The discouraging answer is that there are very few tools that satisfy most users' demands for features in SNP annotation. There are a few, though, that come close. One of these is Snap.



Snap stands for "SNP Annotation Platform", and is hosted by the Institute for Human Genetics at Aarhus University of Denmark. While there are other online and desktop SNP annotation tools, this one is good because it is updated regularly and draws its information from Ensembl, KEGG, UniProt, UCSC, OMIM and Pfam databases.

Genes can be searched in Snap by gene name, marker, clone or SNP ID. Search results are returned with known features and characteristics listed for the gene under investigation, complete with links to the appropriate database from which the evidence was taken.

Introduction:

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Data source:

[Ensembl \(49_36k\)](#)
[UCSC \(hg18\)](#)
[UniProt \(9.4\)](#)
[KEGG \(0.6.1\)](#)
[BIND](#)
[MINT](#)
[OMIM \(20070110\)](#)
[Pfam \(20061106\)](#)
[DAS-CBS](#)
[TreeFam](#)

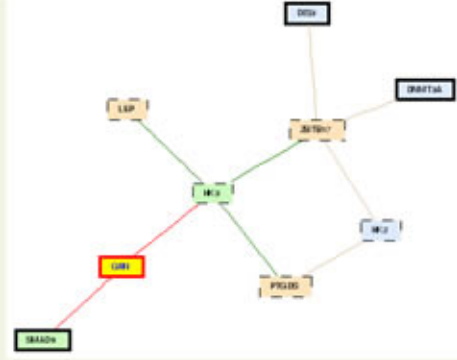
Other:

[Jian Li's PhD thesis supplementary files](#)




Search genes by

Gene : ENSG00000030582
Symbol : GRN
Description : Granulins pre G); Granulin-2 (Granulin F); (Granulin D); Granulin-7 (Gr
Position : Chr17 : 3977801
Transcripts :
GRN(ENST00000053867)
Q9H8S1_HUMAN(ENST0000
P28799-2(ENST0000035735
Clones : RP11-812N09(); B



Relation View :



Related Disease : GRN [C] Frontotemporal deme

Polymorphism statistics :

- Coding**

Frameshift Coding (2) ▲

RS_ID	Position	Class	Peptide Allele	Protein Feature
rs34975779	G/- 39785088	indel		
rs34100802	G/- 39785440	indel		

Non Synonymous Coding (4) ▲

RS_ID	Position	Class	Peptide Allele	Protein Feature
rs11547442	T/C [Y] 39782195	snp Splice site	I/P	
rs25646	T/C [Y] 39783156	snp	D/I	
rs1141754	C/A [M] 39785256	snp	Q/K	
rs25647	G/C [S] 39785365	snp	G/A	VARIANT: G -> A (in dbSNP:rs25647). /FTId=VAR_014830;
- Non Coding**

[] = AND, [] = OR

Granulin-1 (Granulin
Granulin C); Granulin-6

Features of the search result screen include a "relation view" that opens in a new window, illustrating the network of interactions between your gene of interest and other genes. The graphic display also includes all known polymorphism classes, the residues for which the SNPs code and any resultant protein variants.

Snap includes a now-familiar graphic style of gene representation, with the sequence color-coded by region (5' and 3' UTRs, coding region, etc.), but this platform allows users to redraw the sequence graphic by changing features of the introns, phosphorylation and N-glycosylation sites and other features in the gene.

Transcript : GRN (ENST0000053867) [Color Desc](#)
 [chr17 : 39778017 - 39785996 (1)] 2305bp

Numbering: genomic coding region

Features: use class source
 [Click feature bars to highlight the snp]

enlarge in common use

ENST0000053867

CARBOHYD

DISULFID

PAm (click for links to motifs)

SIGNAL

VARIANT

enlarge seldom used

ENST0000053867

MATURE POLY(TET)PEIDE

N-GLYCOSYLATION

NES-SIGNAL

NON_TER

PEPTIDE

PHOSPHORYLATION (S)

PHOSPHORYLATION (T)

PHOSPHORYLATION (Y)

STRAND

TURN

VAR_SEQ

redraw

Transcripts :

with introns (size: 10 bp)

extend 50 bp

GRN (ENST0000053867)
 [chr17 : 39778017 - 39785996 (1)] 2305bp

highlight codons peptide

Q9H8S1_HUMAN (ENST00000393566)
 [chr17 : 39778017 - 39785996 (1)] 2045bp

highlight codons peptide

P28799-2 (ENST00000357351)
 [chr17 : 39778017 - 39785996 (1)] 1840bp

highlight codons peptide

Show transcripts beside or after each other

redraw

```

1  GCGGAGAGGAAGCAGGGAGGAGAGTATTGATAGAAAAGAAACACAGCATTCCAGGCT
61  GGCCCCACCTCTATATTGATAGCAATGGGAGCGGGTAGCCCTGATCCCTGGCCAA
121  TGGAAACTGAGGTAGGCGGGTTCATCGCGCTGGGGTCTGTAGTCTGAGCGCTACCCGGTGG
181  CTGCTGCCCAAGGACCGCGGAGTCGGACGCGAGGCAGACCATGTGGACCCCTGGTGAAGTGG
241  GTGGCCTTAACAGCAGGGCTGGTGGCTGGAACCGGGTGGCCAGATGGTCAGTCTGCCCCT
301  GTGGCCTGCTGCCTGGACCCCGGAGGAGCCAGCTACAGCTGCTGCCGTCCCTTCGGAC
361  AAATGGCCCAACAACACTGAGCAGGCATCTGGGTGGCCCTGCCAGGTGATGCCACTGC
421  TCTGGCGGCCACTCCTGCATCTTTACCGTCTCAGGGACTTCCAGTTGCTGCCCTTCCCA
481  GAGCCCGTGGCATGGGGGATGGCCATCACTGCTGCCACCGGGCTTCCACTGCAGTGCA
541  GACGGCGGATCCTGCTTCCAAAGATCAGGTAACAACCTCCGTGGGTGCCATCCAGTGCCCT
601  GAAAGTCAGTTGCAATGCCCGGACTTCTCCACGTGCTGTGTTATGGTCGATGGCTCTGG
661  GGGTGTGCCCCATGCCCGAGGCTTCTGCTGTGAAGACAGGGTGCACCTGCTGTCCGCAC
721  GGTGCCTTCTGGGACCTGGTTCACACCCCGTGCATCACACCCAGGGCACCACCCCGCTG
781  GCAAAGAGCTCCCTGCCCAGAGGACTAACAGGGCAGTGGCCCTGTCCAGCTCGGTCAAG
841  TGTCGGAGCAGCGTCCCGGTGCCCTGATGGTTCACCTGCTGTGAGCTGCCAGTGGG
901  AAGTATGGCTGCTGCCCAATGCCCAACGCCACCTGCTGCTCCGATCACCTGCAGTCTGTC
961  CCCCAGACACTGTGTGTGACCTGATCCAGAGTAAAGTGCCTTCCAAAGGAGAACCCATCC
1021  ACGGACCTCCTCACTAAGCTGCCCTGCCACACAGTGGGGATGTGAAATGTGACATGGAG
1081  GTGAGCTGCCCAGATGGCTATACCTGCTGCCGTCTACAGTCGGGGGCTGGGGCTGCTGC
1141  CCTTTTACCCAGGCTGTGTGCTGTGAGGACACATACACTGCTGTCCCGCGGGTTTACG
1201  TGTGACACGCAAGGGTACCTGTGAACAGGGGCCCAACAGGTGCCCTGGATGGAGAAG
1261  GCCCCAGCTCAGCTCAGCTGCCAGACCCACAAGCCTTGAAGAGAGATGTCCTCTGTGAT
1321  AATGTCAGCAGCTGTCCTCCTCCGATACCTGCTGCCAACTCAGCTGTGGGAGTGGGGC
1381  TGCTGTCCAAATCCAGAGGCTGTGCTGCTCGGACCCAGCACTGCTGCCCCAGGGC
1441  TACAGTGTGTAGCTGAGGGGAGTGTGAGGAGGAGGAGAGATCGTGGCTGGACTGGAG
1501  AAGATGCTGCCCGCGGGCTTCCCTTATCCACCCAGAGACATCGGCTGTGACCCAGCAC
1561  ACCAGCTGCCCGGTGGGCGAGACCTGCTGCCCGAGCCTGGGTGGGAGCTGGCCCTGCTGC
1621  CAGTTGCCCATGCTGTGTGCTGCGAGGATCGCGAGCACTGCTGCCCGGTGGCTACACC
1681  TGCAACGTGAAGGCTCGATCCTGCGGAGAGGAAAGTGGTCTGTGCCAGCCTGCCACCTTC
1741  CTGGCCGTAGCCCTCAGTGGGTGTGAAGGACGTGGAGTGTGGGAAAGGACACTTCTGC
1801  CATGATAACAGACCTGCTGCCGAGACAACCCAGCGCTGGGCTGCTGTCCCTACCGC
1861  CAGGGCTGTGTTGTGCTGATCGGGCGCACTGCTGTCCCTGCTGGCTTCCGCTGCCAGCC
1921  AGGGGTACCAAGTGTGTCGCGAGGGAGGCCCGGCTGGGAGCCCTTTGAGGGACCCA
1981  GCCTTGAGACAGTGTGTGAAGGACAGTACTGAAGACTGTGACGCCCTCGGGACCCAC
2041  TCGGAGGGTGGCCCTCTGCTCAGGCCCTCCCTAGCACCTCCCTAACCAAAATCTCCCTGG
2101  ACCCCATTGTGAGCTCCCCATCACCATGGGAGGTGGGGCCTCAATGAAGGCCTTCCCTG
2161  TCAGAAGGGGGTGTGGCAAAGGCCAATTACAAGTGCOCATCCCTCCCGCTTTCAGTGG
2221  GACCTGTGGCCAGGTGCTTTTCCCTATCCACAGGGGTGTTTGTGTGTGCGCGTGTGC
2281  GTTTCATAAAGTTTGTACACTTTC
  
```

Finally, Snap allows users to view forward or reverse primer sequences for the most significant regions of the gene, or to select a portion of the sequence, by clicking and dragging, for which Snap will recommend primers in a separate browser window.

Resequencing
Gene : GRN ([ENSG0000030582](#))
Transcript : GRN ([ENST0000053867](#)) [length: 2305]
 With introns (10)
 Primer size: - Primer TM value: - Product size: -
 Resequencing overlap:
 only exons (extend with 0 bp in introns)

Primer parameters :
 in transcript: [ENST0000053867](#)
 Primer size: -
 Primer TM value: -
 Product size: -

Resequencing:
 with introns overlap:
 only exons (extend with 0 bp in introns)
 surround intron: intron_1 (RT-PCR)

Sequence Primer Design:
 - - :
 (Click and drag in the sequence)
[For selected sequence](#)
[Start of left primer](#), Length
[Start of right primer](#), Length

Exon1-13 :
Forward Primer 1: [AGGGAGGAGAGTGA](#)
 LEN: 20 GC: 50.000 TM: 55.881 POS: 14
Reverse Primer 1: [ATAACACAGCAGTGGAGA](#)
 LEN: 19 GC: 47.368 TM: 55.016 POS: 644
 Primer product size: 631
 Overlap: 202

Forward Primer 2: [TTACCGTCTCAGGGACTTC](#)
 LEN: 19 GC: 52.632 TM: 55.177 POS: 443
Reverse Primer 2: [GTAGACGGCAGCAGGTATAG](#)
 LEN: 20 GC: 55.000 TM: 55.194 POS: 1117
 Primer product size: 675
 Overlap: 152

Forward Primer 3: [AGACACTGTGTGTGACCTGA](#)
 LEN: 20 GC: 50.000 TM: 54.748 POS: 966
Reverse Primer 3: [GGACAGCTGCTGACATTATC](#)
 LEN: 20 GC: 50.000 TM: 55.832 POS: 1337
 Primer product size: 372
 Overlap: 176

Forward Primer 4: [TGTGAGGACCACATACACTG](#)
 LEN: 20 GC: 50.000 TM: 55.225 POS: 1162
Reverse Primer 4: [GGTCTGGTTATCATGGCA](#)
 LEN: 18 GC: 50.000 TM: 55.105 POS: 1815
 Primer product size: 654
 Overlap: 118

Forward Primer 5: [ATCCTGCGAGAAGGAAGT](#)
 LEN: 18 GC: 50.000 TM: 54.782 POS: 1698
Reverse Primer 5: [ACAAACTTTATTGAAACGCAC](#)
 LEN: 21 GC: 33.333 TM: 54.677 POS: 2297
 Primer product size: 600

Gap region(s):
 Start: 1 - End: 14
 Start: 2297 - End: 2305

```

1  GGGAGGAGGAGCAGGAGGAGTGA
61  GGGCCCACTCTATATTGATAAGTAGO
121 TGGAAACTGAGGTAGGGGGTCAATGG
181 CTGCTGCCAAGGACCGGGAGTGGGA
241 GTGGCTTAACAGCAGGGCTGGTGGCT
301 GTGGCTGTGCTGGACCCCGGAGGA
361 AAATGGCCCAACAACACTGAGCAGGCAD
421 TCTGGCGGCACTCCTGCACTTTAAG
481 GAGGCGGTGGCATGGGGGATGGCCAT
541 GAGGGCGATCTGCTTCCAAAGATCA
601 GAGTACAGTTCGAATGCCGGACT
661 GGGTGTGCCCATGCCCCAGGCTTC
721 GGTGCTTCTGGCACTGGTTCACACC
781 GCAAAGAAGTCCCTGCCAGAGGACT
841 TGTCCGACGCAAGGTCCCGGTGCCCT
901 AAGTATGGCTGCTGCCCAATGCCCAAD
961 CCCCAGGAGTGTGTGTGACCTGATG
1021 ACGAGCTCTCTCACTAAGCTGCTGG
1081 GTGAGCTGCCAGATGG
1141 CCTTTACCCAGGCTGTGTGCTGGAG
1201 TGTGACAGCAGAAAGGTAACCTGTAA
1261 GCCCAGCTCACCTCAGSCTGCCAGAG
1321 AATGAGGAGGCTTCTCTCCGATACTGCTGCCA
1381 TGCTTCCAATCCAGAGGCTGTCTGCTGGAC
1441 TACACGTGTGAGCTGAGGGGCACTGTCAGC
1501 AAGATGCTGCCCCGCGGGCTTCTTATCCCA
1561 ACCAGTGCCTGGGTGGGACAGACTGCTGG
1621 CAGTGGCCCCATGCTGTGTGCTGGCAGGAT
1681 TGCAAGCTGAAGGCTCGTCTGCGAAGGAA
1741 CTGGCCGTAGCCCTCACGTGGTGTGAGGAG
1801 TGGTATGAGGAGTTCCTGCTGCTGCTGCTG
1861 CAGGGGCTGTGTGTGCTGATCGGGGCACT
1921 AGGGGTACCAAGTGTGTGGCAGGGAGGCC
1981 GCCTTGAGCAGCTGCTGTGAGGGACAGT
2041 TGGAGGGTGGCCCTCTGCTCAGGGCTCC
2101 ACCCCATGTGAGCTCCCATCACCATGGG
2161 TCAGAAGGGGGTGTGGCAAAGCCACATT
2221 GACCTGTGGCCAGGTGCTTTTCCCTATCC
2281 TGGTATGAGGAGTACACTTC

```

Mis-splicing :
 Nucleotide : - A/G[R]
 (Click on the nucleotide)

Sequence tool box:
 GC% : 0 % Length : 0

Exon	No.	Forward Primer	LEN	GC	TM	POS	Reverse Primer	LEN	GC	TM	POS	Size	Overlap
1-13	1	AGGGAGGAGAGTGA	20	50.000	55.881	14	ATAACACAGCAGTGGAGA	19	47.368	55.016	644	631	0
1-13	2	TTACCGTCTCAGGGACTTC	19	52.632	55.177	443	GTAGACGGCAGCAGGTATAG	20	55.000	55.194	1117	675	202

This flexibility, combined with links to sources of evidence in [OMIM](#), [KEGG](#) and Pfam, provides one of the most complete free-access SNP annotation platforms available.

Try it out!

Do you have suggestions for other sites you'd like to see featured in the Biosciences Blog? Send a recommendation or a comment to me:

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 312-503-8689

[e-mail Pamela](#)

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