Northwestern University Feinberg School of Medicine
Galter Health Sciences Library & Learning Center

SNP Annotation is a "Snap"

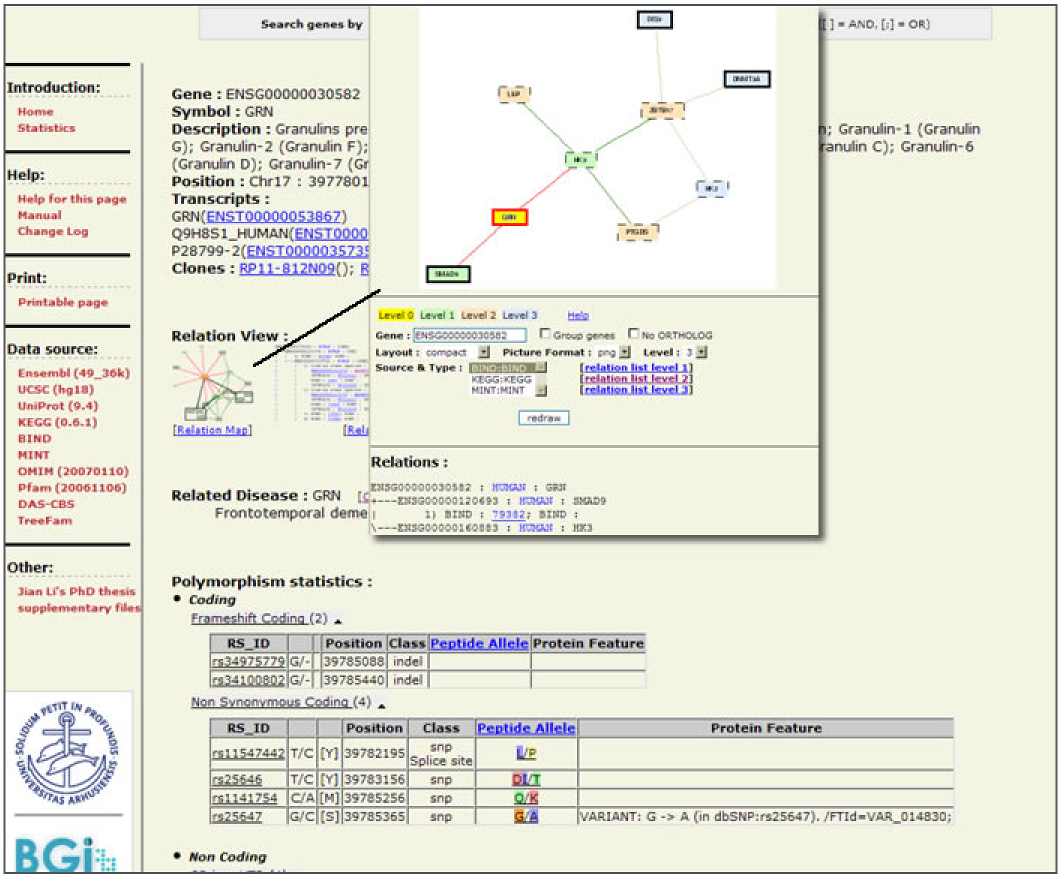
This article was featured in *Library Notes #48* (Fall 2008).

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 - a SNP Annotation Platform v3.4

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.



Gene : ENSG00000030582
Symbol : GRN
Description : Granulins pre… n; Granulin-1 (Granulin
G); Granulin-2 (Granulin F); … ranulin C); Granulin-6
(Granulin D); Granulin-7 (Gr
Position : Chr17 : 3977801
Transcripts :
GRN(ENST00000053867)
Q9H8S1_HUMAN(ENST0000
P28799-2(ENST00000357
Clones : RP11-812N09(); R

Gene : ENSG00000030582 ☐ Group genes ☐ No ORTHOLOG
Layout : compact Picture Format : png Level : 3
Source & Type : BIND:BIND KEGG:KEGG MINT:MINT

Relations :

```
ENSG00000030582 : HUMAN : GRN
+---ENSG00000120693 : HUMAN : SMAD9
|        1) BIND : 79382; BIND :
\---ENSG00000160883 : HUMAN : HK3
```

Related Disease : GRN
Frontotemporal deme

Polymorphism statistics :

- *Coding*

Frameshift Coding (2)

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

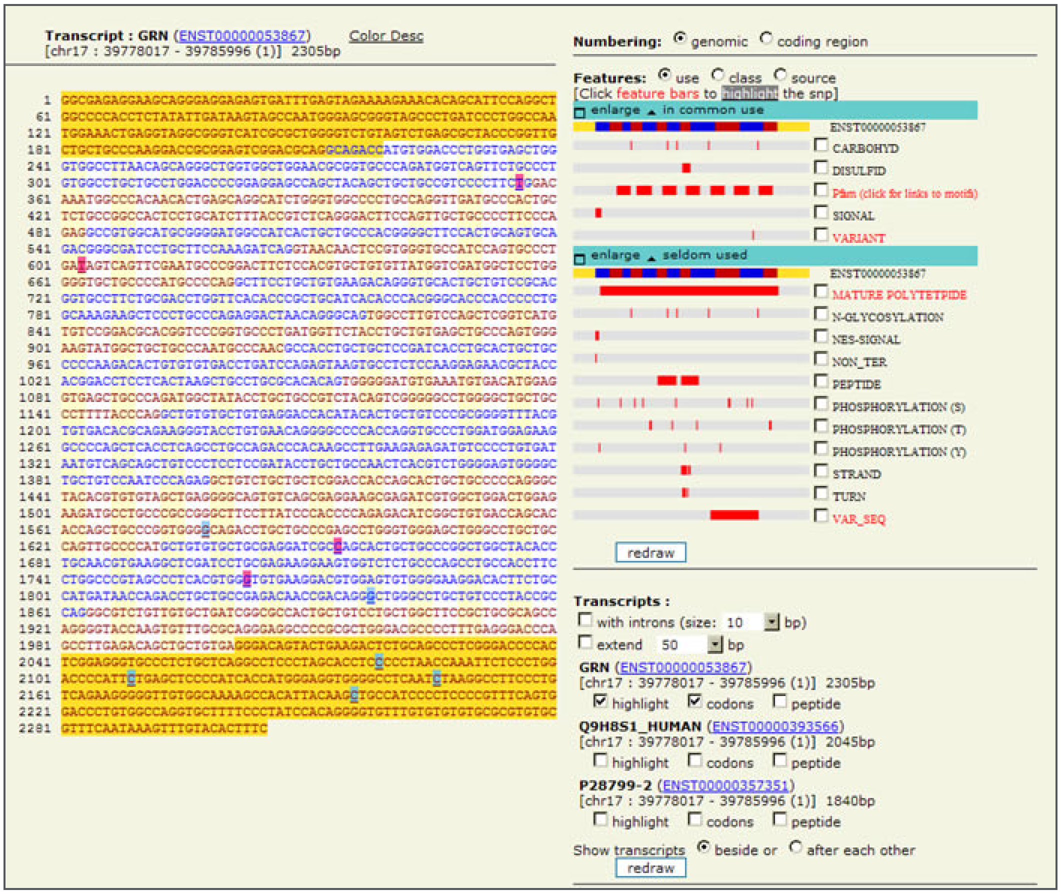
Non Synonymous Coding (4)

RS_ID			Position	Class	Peptide Allele	Protein Feature
rs11547442	T/C	[Y]	39782195	snp Splice site	L/P	
rs25646	T/C	[Y]	39783156	snp	D/T	
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*

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 (ENST00000053867) Color Desc
[chr17 : 39778017 - 39785996 (1)] 2305bp

Numbering: ● genomic ○ coding region

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

Transcripts :
☐ with introns (size: 10 bp)
☐ extend 50 bp

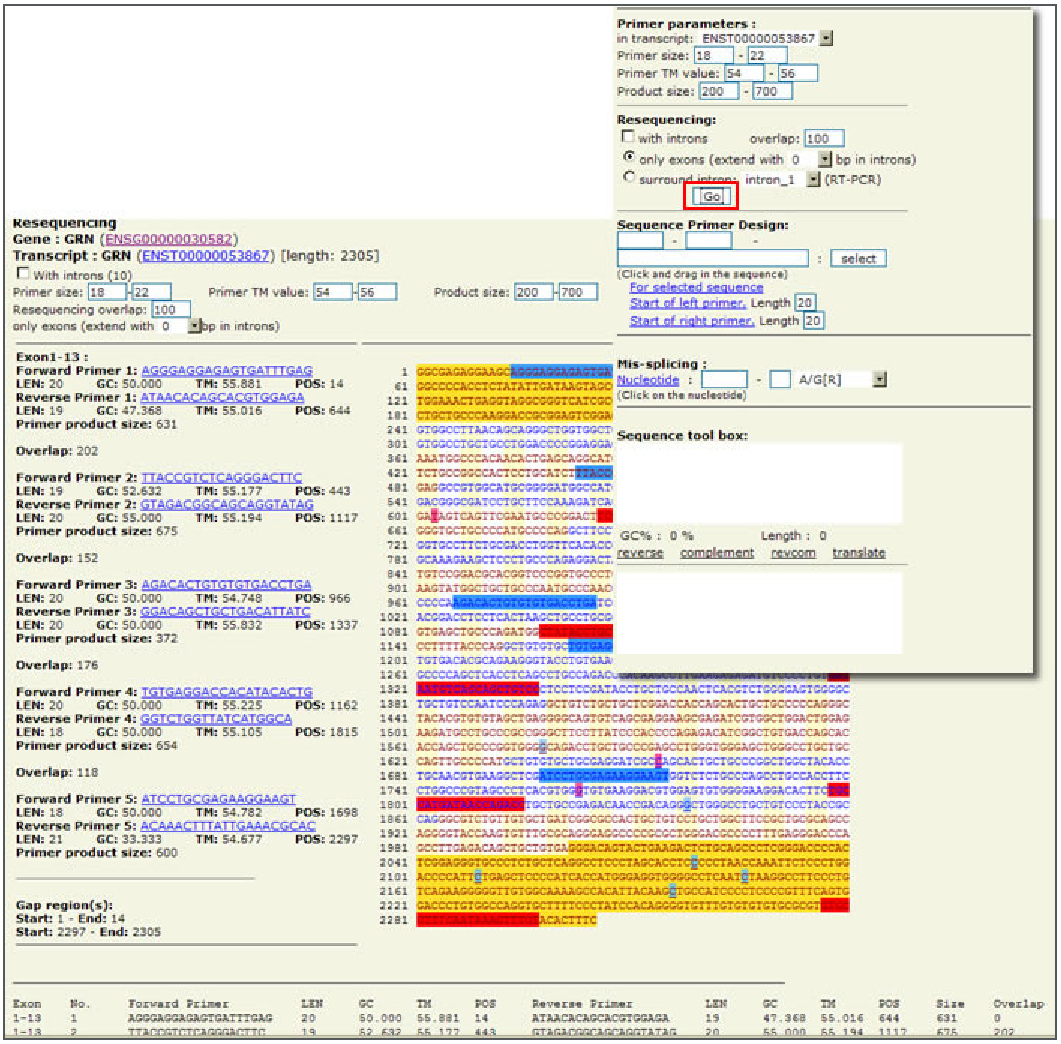
GRN (ENST00000053867)
[chr17 : 39778017 - 39785996 (1)] 2305bp
☑ highlight ☑ codons ☐ peptide

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

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.



Primer parameters :
in transcript: ENST00000053867
Primer size: 18 - 22
Primer TM value: 54 - 56
Product size: 200 - 700

Resequencing:
☐ with introns overlap: 100
● 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 20
Start of right primer, Length 20

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

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

Resequencing
Gene : GRN (ENSG00000030582)
Transcript : GRN (ENST00000053867) [length: 2305]
☐ With introns (10)
Primer size: 18 - 22 Primer TM value: 54 - 56 Product size: 200 - 700
Resequencing overlap: 100
only exons (extend with 0 bp in introns)

Exon1-13 :
Forward Primer 1: AGGGAGGAGAGTGATTTGAG
LEN: 20 **GC:** 50.000 **TM:** 55.881 **POS:** 14
Reverse Primer 1: ATAACACAGCACGTGGAGA
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

```
Exon   No.   Forward Primer          LEN   GC      TM      POS    Reverse Primer          LEN   GC      TM      POS    Size   Overlap
1-13   1     AGGGAGGAGAGTGATTTGAG    20    50.000  55.881  14     ATAACACAGCACGTGGAGA     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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