Single Nucleotide Polymorphism

- Concept
- Classification
- Search method
- Assay method
- Clinical application

Polymorphism

Single nucleotide polymorphism

Tandam repeat polymorphism

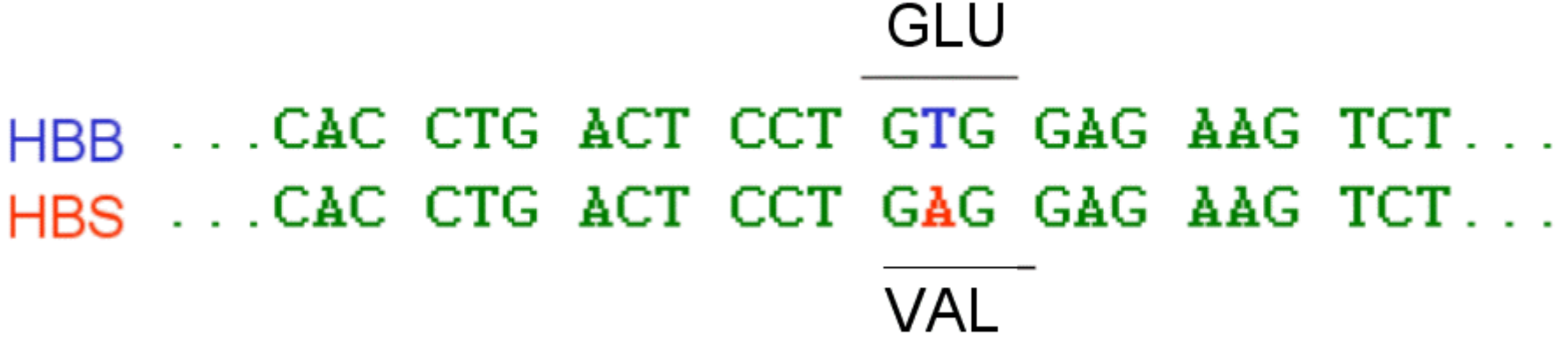
Insert/deletion polymorphism

Single Nucleotide Polymorphisms

DNA sequence variations
(A,T,C,or G)

AAGGCTAA -> ATGGCTAA

Sickle cell anemia:

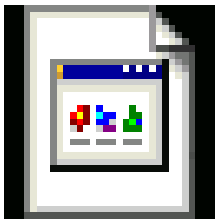


SNPs vs. Mutations

- by allele frequency

- A single base change, occurring at a frequency of >1%
  - a single nucleotide polymorphism (SNP)

- When a single base change occurs at <1%
  - a mutation.



DNA.rep.movie.swf

패키지

Recombination

Tandem Repeat Polymorphisms

- A very common class of polymorphism variable length of sequence motifs

  -Microsatellites or Short Tandem Repeat (STR)
  repeat unit: 1-6
  (dinucleotide repeat: CACACACACAC)

  -Minisatellites
  repeat unit: 14-100

Insertion/Deletion Polymorphisms

Sequence repetitiveness (direct or inverted)
Predispose DNA to rearrangements

Example

Association between coronary heart disease & a 287 bp **Indel Polymorphism**

Located in intron 16 of the angiotensin converting enzyme (ACE) (OMIM 106180)

Responsible for 50% of the inter individual variability of plasma ACE concentration.

Estimated Numbers of SNP

- 0.3-1.0 kb average intervals
  entire human genome - $3X10^7$ bp
  number: up to 5-10 million

- short insertion/deletions
  in between SNPs and VNTR

Polymorphisms & Disease Markers

-Very few of SNP

Direct impact on deleterious phenotype

non-disease-causing polymorphisms

-Markers to identify & map other genes

Terminologies

Allele: Alternative form of a genetic locus
a single allele from each parent

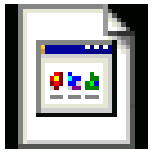
Polymorphism: Difference in DNA sequence
among individuals.

Linkage Disequilibrium (LD): 2 alleles inherited together more often than would be predicted

Haplotype : set of alleles on one particular chromosome
Each person has two haplotypes

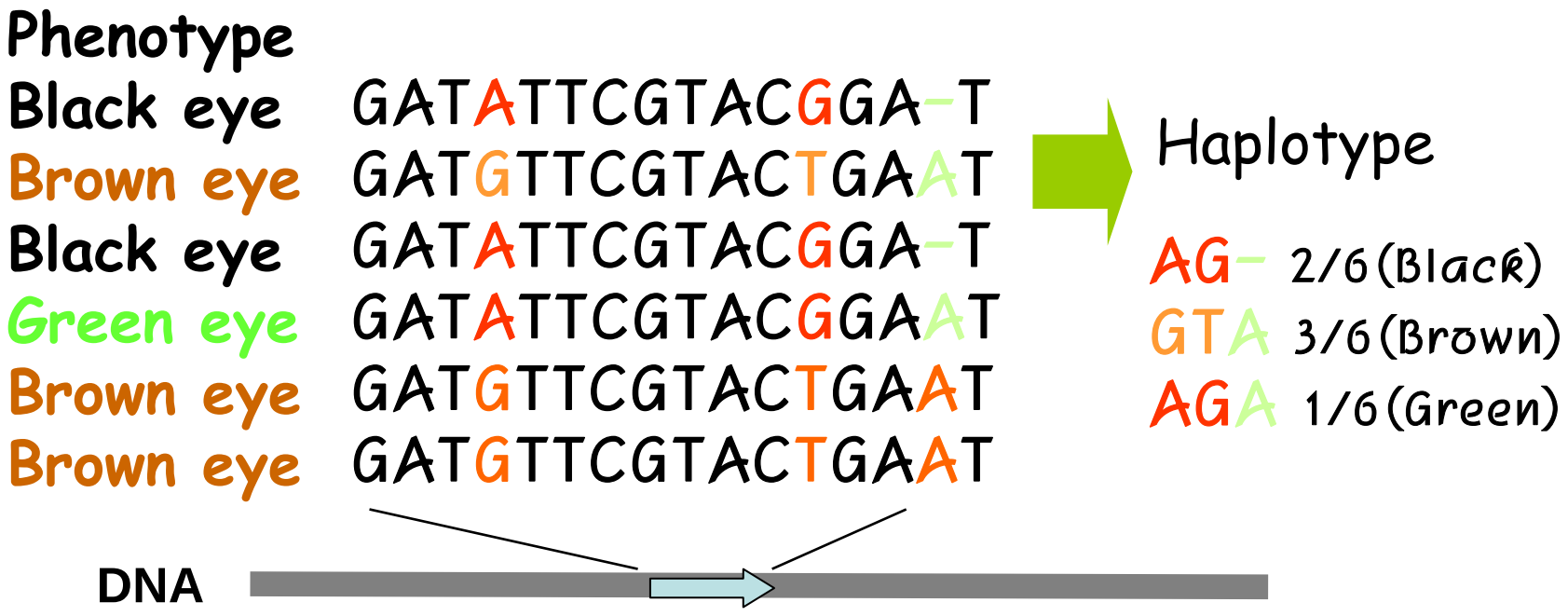
SNP – Binary

ACGGGTACCGT
ACGGGGACCGT



haplotype.mov

From SNP to Haplotype



human genome is organized in blocks ~50 kb in size

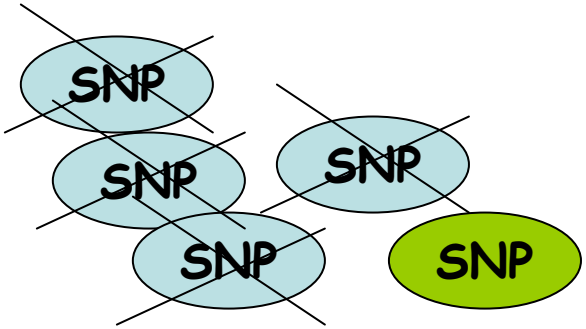
Haplotypes with high LD

regions with a high level of concomitant inheritance

Separated by regions of low LD

regions with a low level of concomitant inheritance

Life Cycle of SNPs & Mutations



Apprearance of
New variants
By mutation

SNP

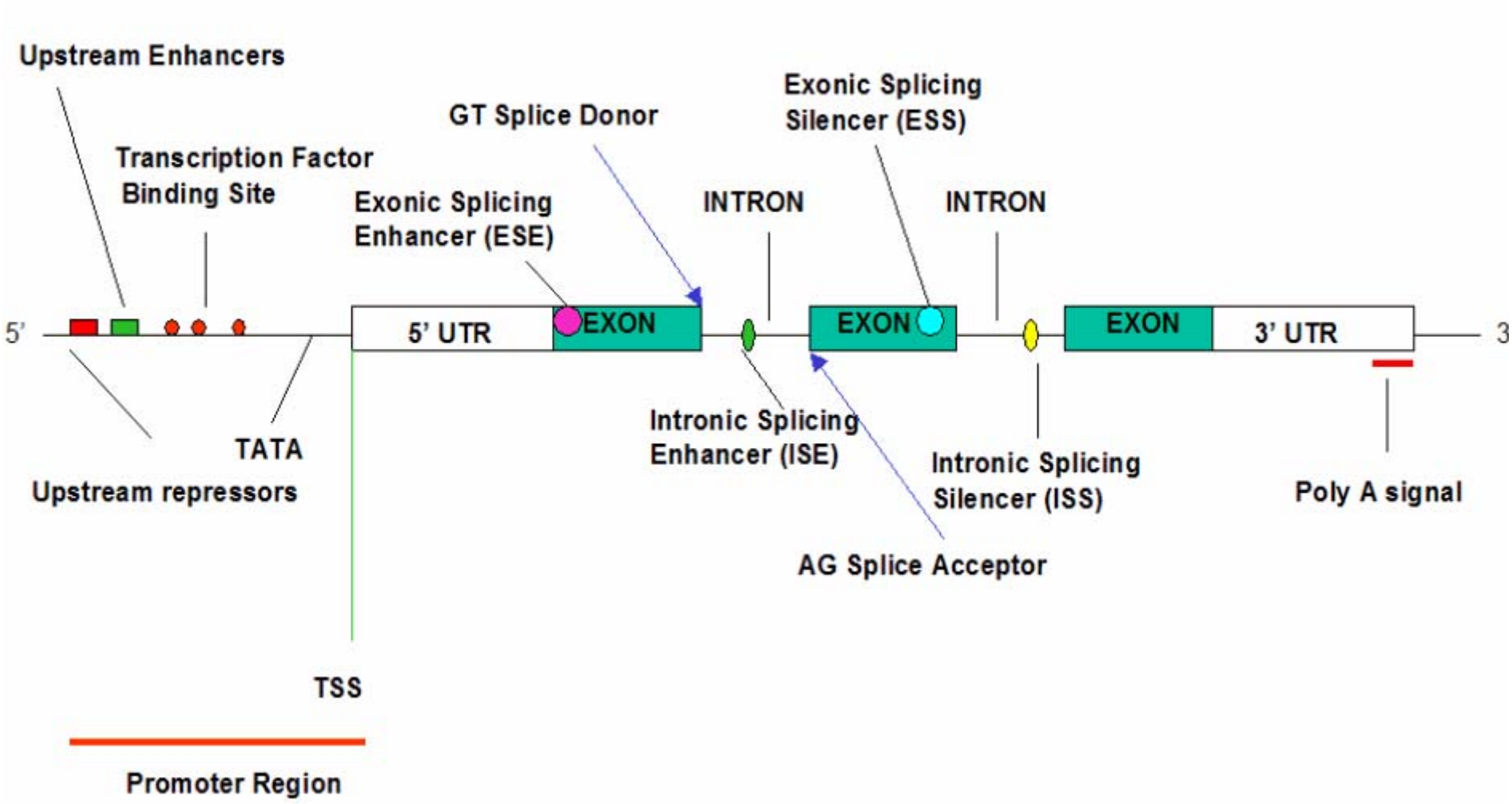
Survival of alleles

SNP SNP SNP

Increase of the allele
To a substantial
frequency

Species
Differentiation

Fixation of the allele
In a population


Upstream Enhancers
Transcription Factor Binding Site
GT Splice Donor
Exonic Splicing Silencer (ESS)
Exonic Splicing Enhancer (ESE)
INTRON
INTRON
5'
5' UTR
EXON
EXON
EXON
3' UTR
3
TATA
Upstream repressors
Intronic Splicing Enhancer (ISE)
Intronic Splicing Silencer (ISS)
Poly A signal
AG Splice Acceptor
TSS
Promoter Region

Classification of SNP

SNPs : intronic, exonic, promoter region

Coding SNPs

- Synonymous: no change in a.a
  silent mutation
- Non-synonymous: a change in a.a.
  conservative vs.
  nonconsevative

Genetic Variations Databases

SNP Databases

- dbSNP
  http://www.ncbi.nlm.nih.gov/SNP/index.html

- Human Genome Variation Database (HGVbase)
  http://hgvbase.cgb.ki.se/

- TSC: The SNP Consortium
  http://snp.cshl.org/

dbSNP

http://www.ncbi.nlm.nih.gov/SNP/index.html

- Single-base nucleotide substitutions (SNPs)
- Small-scale multi-base deletions or insertions (deletion insertion polymorphisms- DIPs)
- Microsatellite repeat variations (short tandem repeats - STRs)

dbSNP Data Type

1.Submitted data from original observation

submitted SNPs (SS) with
ss# (ss 5586300)

2. Computed data

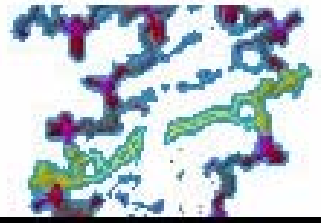
reference SNP Clusters (Ref SNP) with
rs# (rs 4986582)

Submitted SNP Details Page

- collection methods (assay technique)
- submitter information
  - contact data, individual submitter
- variation data
  - frequencies, genotypes

NCBI

Single Nucleotide Polymorphism



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DOCUMENTATION
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updated 2006

dbSNP Search Options

Entrez SNP	ID Numbers	Submission Info	Batch	Locus Info	Between Markers

ANNOUNCEMENT

- 04/06/2006: SNP FAQ Archive updated with 2006 contents
- 03/06/2006: dbSNP Mouse Build 126 is now available
- 10/26/2005: Accessioned Haplotype Content Now Available in dbSNP
- 10/20/2005: Schema Changes
- 10/31/2005: 1 or 0 Based Mapping Position

Search by IDs

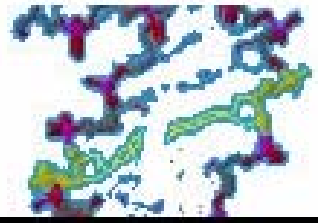
Note: rs# and ss# must be prefixed with "rs" or "ss", respectively (i.e. rs25, ss25)

Reference cluster ID(rs#)

Search Reset

NCBI

Single Nucleotide Polymorphism



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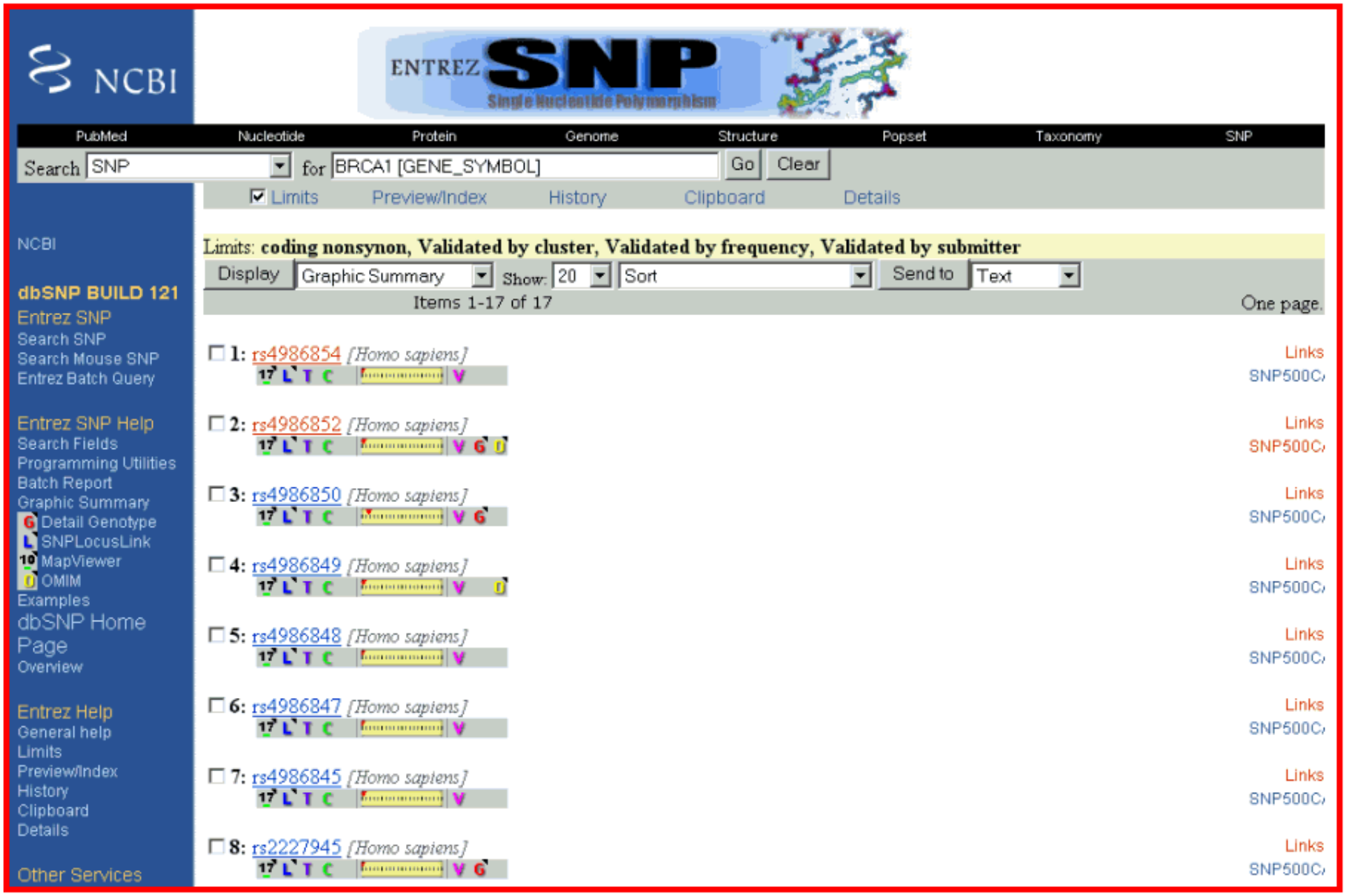
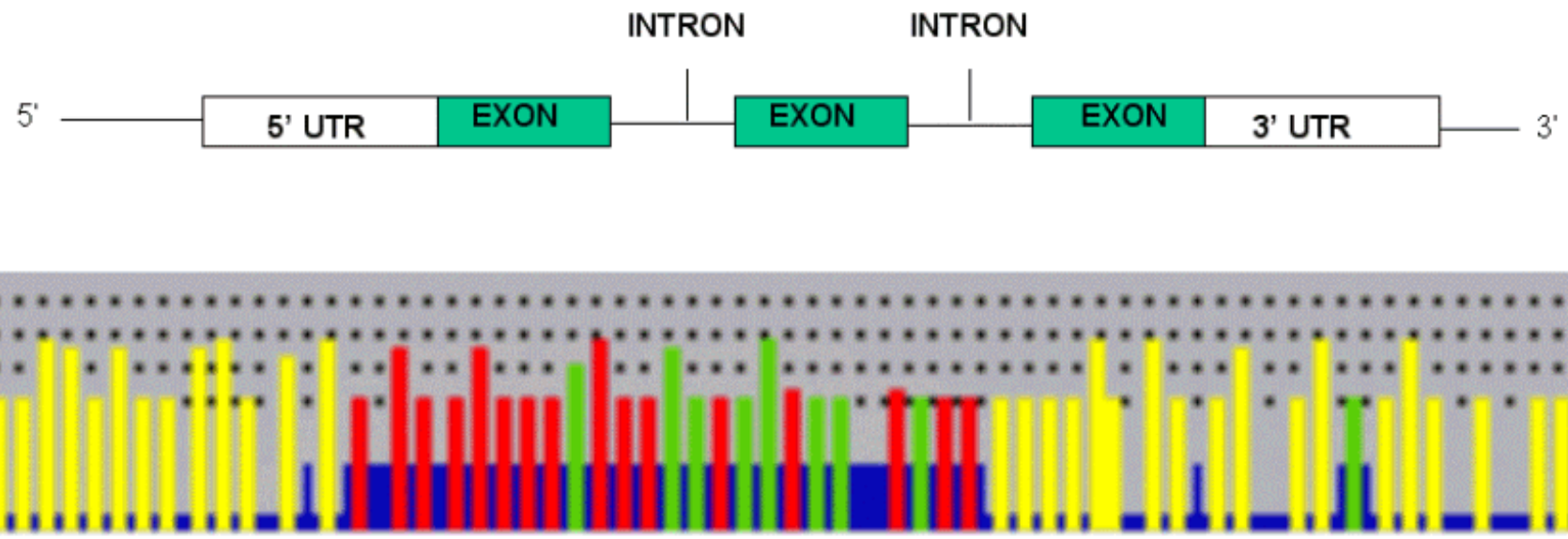
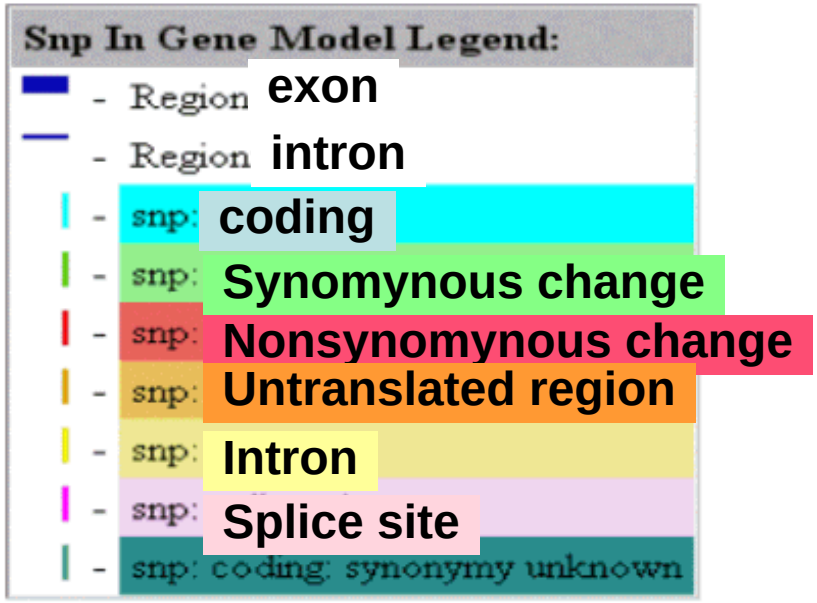
Reference cluster ID(rs#)

Search Reset

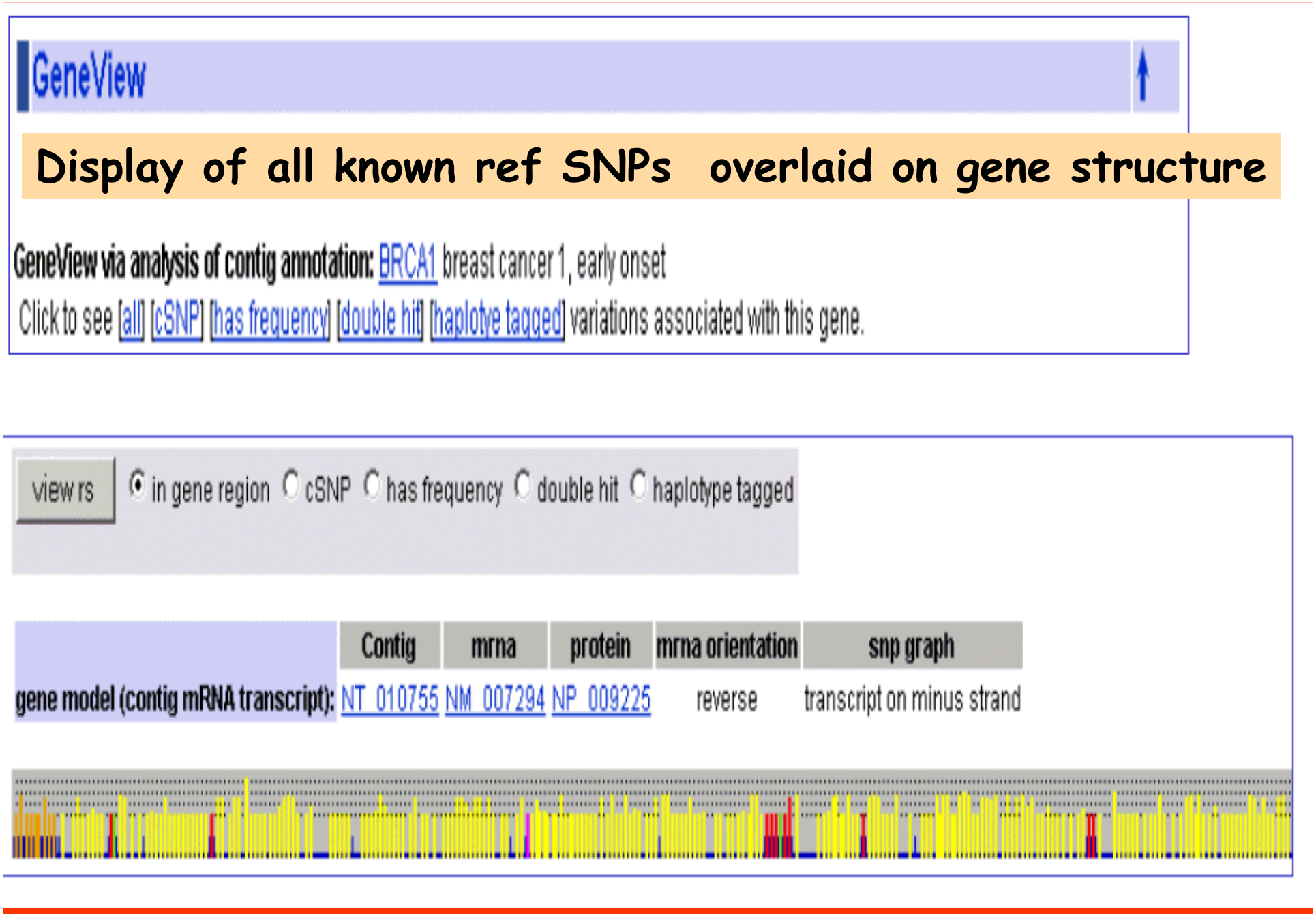
dbSNP Search Result







Gene-Oriented SNP Visualization



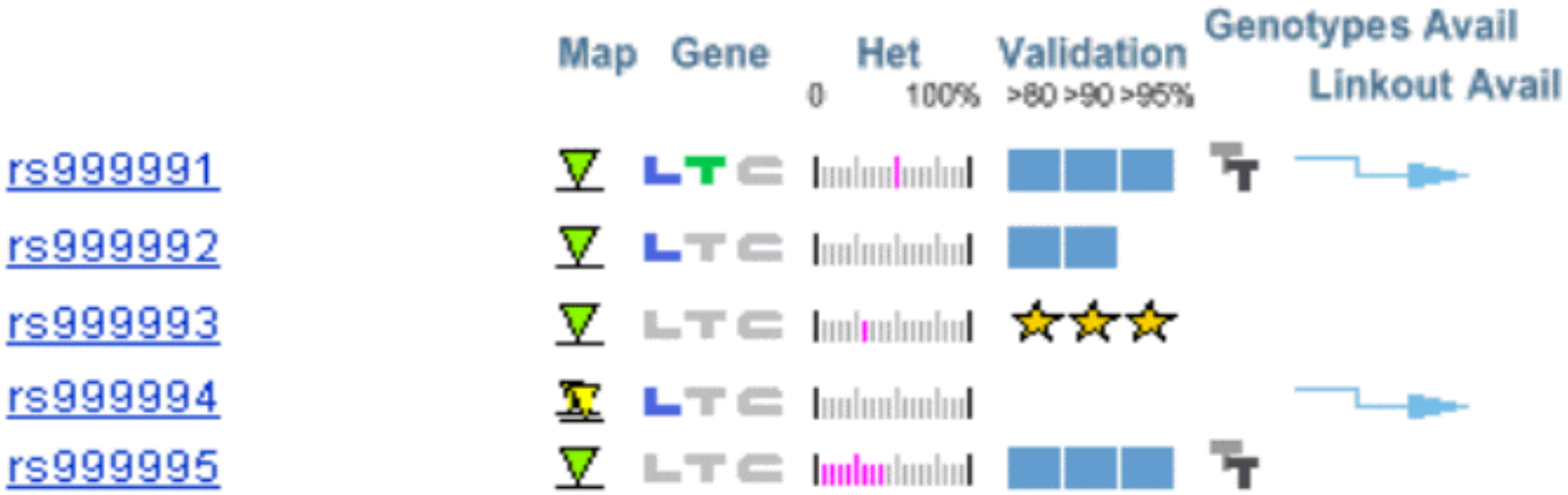
GeneView for All SNPs

RefSNP Summary Info

Summary Information for refSNPs

The NCBI genome map viewer can be configured to supply additional information about elements in the master map by selecting **verbose mode** in the display settings dialog box.

When the "SNP" map is selected as the master map, the user is presented with a graphical summary of several properties of the SNP in a display that looks like this:



In silico map results

The quality of the SNP is computed from mapping and validation data. The current set of map icons and their meanings are:

Map	Meaning
	Marker mapped to **Unique position** n the genome. Markers mapped to two contigs on the same chromosome will also receive this icon, on the assumption that the contigs might not have been joined during sequence assembly.
	Marker mapped to **Multiple position** in the genome. Markers mapped to 2 to 10 of the genome will receive this warning icon. Markers mapped to more than 10 locations will not be annotated on human genome sequence, nor will they be included in the genome map views.
	Markers that have b **Withdrawn marker** dbSNP as a duplicated gene re artifact will be flagged with this WITHDRAWN icon. Evidence for withdrawing the marker can be found in the dbSNP refSNP report for the marker.
	This marker is not **Not mapped to any genomic location** on.

Mouseover Text

MAP COUNTS: reports the number of hits to each of the following categories:

- Total number of chromosomes [CH]
- Total number of contigs [NT]
- Total distinct locations [TOT]

Map Viewer icon for SNPs

Gene region / functional classification

Markers placed on the sequence map are examined for neighboring gene features (gene, mRNA, CDS, and exon). Markers that satisfy the conditions below are considered a member of the set of markers associated the the gene, and each is assigned a functional classification depending on its particular location in the gene region:

Gene	Meaning
L	**Locus** S: Any part of the marker position on sequence map is a 2kb interval 5' of the most 5' feature of gene (CDS, mRNA, gene), OR the marker position is within a 500 base interval 3' of the most 3' feature of the gene. Both strands of sequence are examined for gene features, so a marker can potentially be a variation on multiple genes at a single location.
T	**Transcript** T: Any part of marker position overlaps with mRNA verlaps with UTR/intron and mRNA feature is missing), BUT marker position is not within the coding region of the transcript.
C	**Coding** : Any part of the marker position overlaps with a equence (CDS) region (or overlaps with exon region in the unlikely case an exon is annotated but CDS is missing).
L	**Locus not within known gene locus** s (as defined above) for
T	**Transcript not within known trancript region** annotated
C	**Locus not within known coding region** or any annotated

Mouseover Text

Mouseover on any letter will show the set of gene symbols for each respective functional category

Map Viewer Icon for SNPs

Quality Information

This scale distinguishes validated markers (★) from unvalidated ones ( ). Viewed as a vertically stacked histogram, the width of each scale reports the likelihood that a marker is real as defined by the submitter. This likelihood is based on validation experiments conducted by the submitter for each batch of reported markers, where the success rate for a batch is defined as 1 minus the false positive rate. Scale probabilities are:

Validation	Meaning
★★★	Marker has been validated by imdependent resequencing
	Unvalidated marker – success rate >95%
	Unvalidated marker – success rate >90%
	Unvalidated marker – success rate >80%

The success rate for the marker is reported. If several submissions have been clustered into a single refSNP, then the reported figure is the maximum success rate for the cluster.

Other Information

Success rate = 1-false positive rate

	Meaning
	Individual genotypes have been submitted to dbSNP for this marker.
	Links have been established between the dbSNP marker and a submitter website or database. These resources can provide additional information about the quality of the marker or its phenotypic effects for the organism or gene.

Map Viewer Icon for SNPs

Marker heterozygosity

This 0 – 100% scale indicates the average heterozygosity as the range avg_het±2 [SE(avg_het)], where SE(avg_het) is the standard error of the estimator. Thus the graph shows an approximate 95% confidence interval for the marker.

Het	Meaning
	**No allele frequencies** measures of observed heterozygosity were submitted for this marker.
	**Average heterozygosity 0.26-0.30** % confidence interval (0.26 – 0.30). The estimate has a small standard error.
	**Average heterozygosity 0.00-0.40** confidence interval (0.00 – 0.40) The estimate has a large standard error.

Mouseover Text

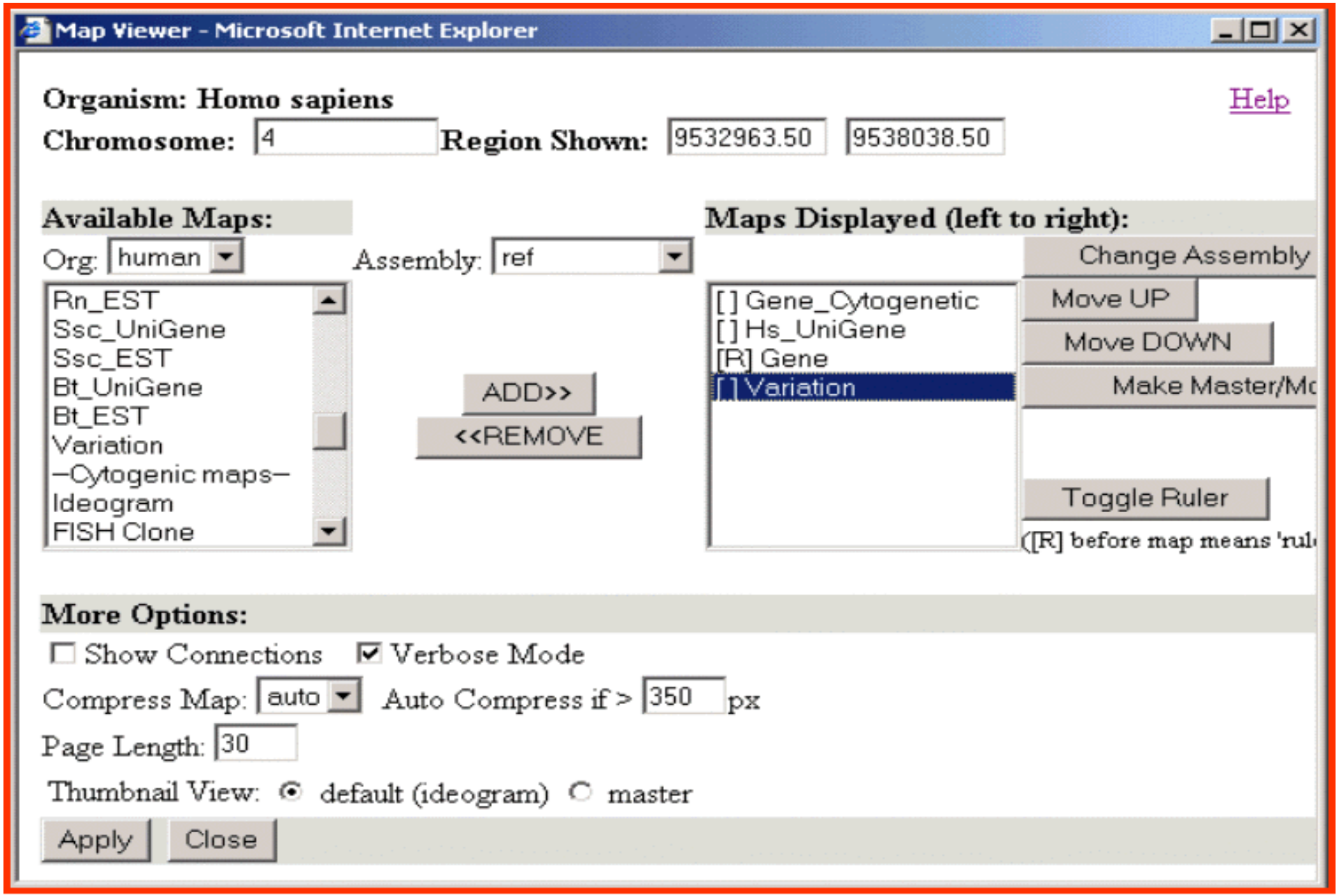
Mouseover on the graph will report average heterozygosity and standard error in the form Het (S.E.). E.g. Het (S.E.)=0.32 (0.008)

Map Viewer Icon for SNPs

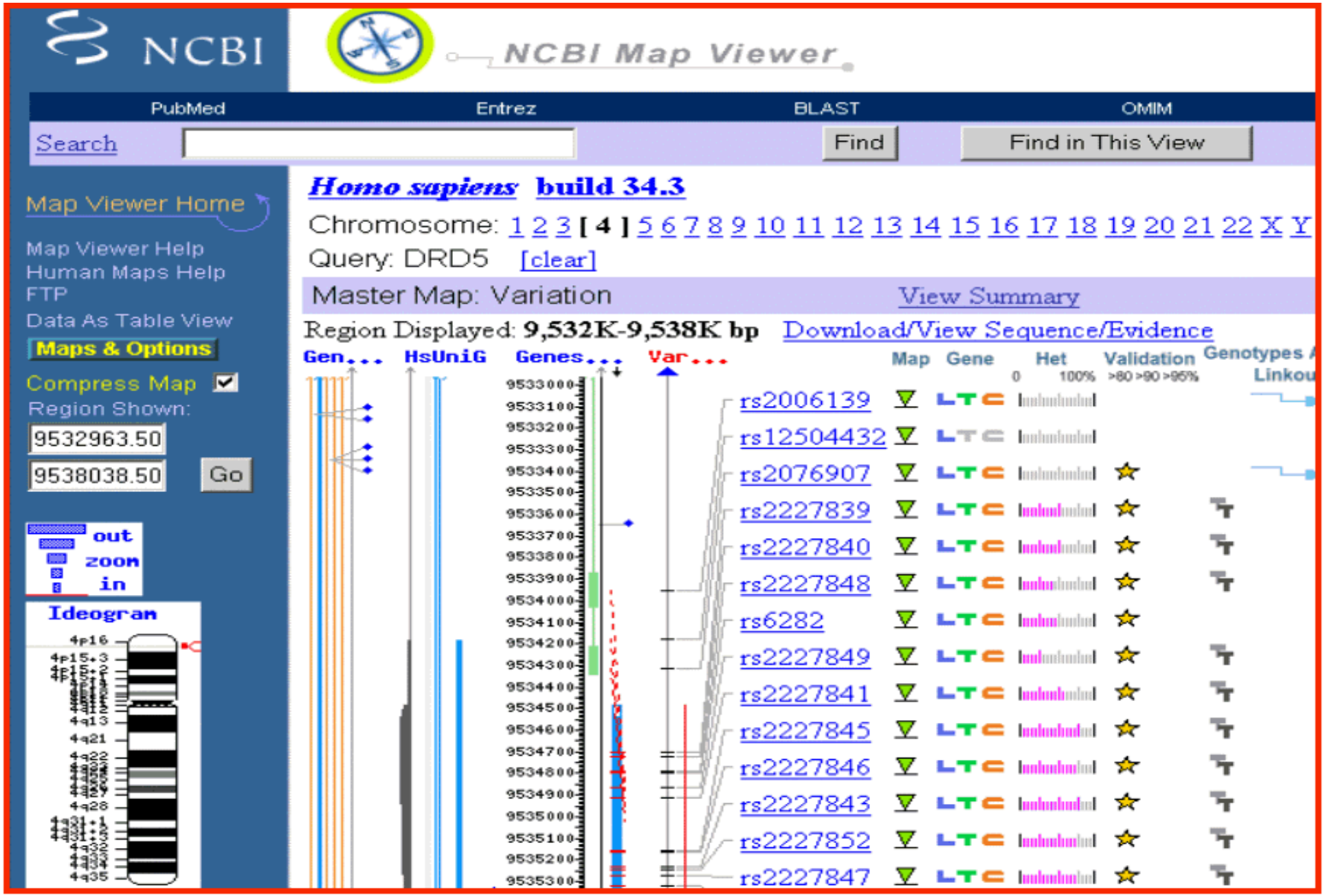
Validation Methods

	validated by multiple, independent submissions to the refSNP cluster
	validated by frequency or genotype data: minor alleles observed in at least two chromosomes.
	validated by submitter confirmation
	all alleles have been observed in at least two chromosomes apiece
	validated by HapMap project

Map Viewer Display Option



SNP: Genome View



Online Mendelian Inheritance in Man (OMIM)

OMIM Allelic Variants

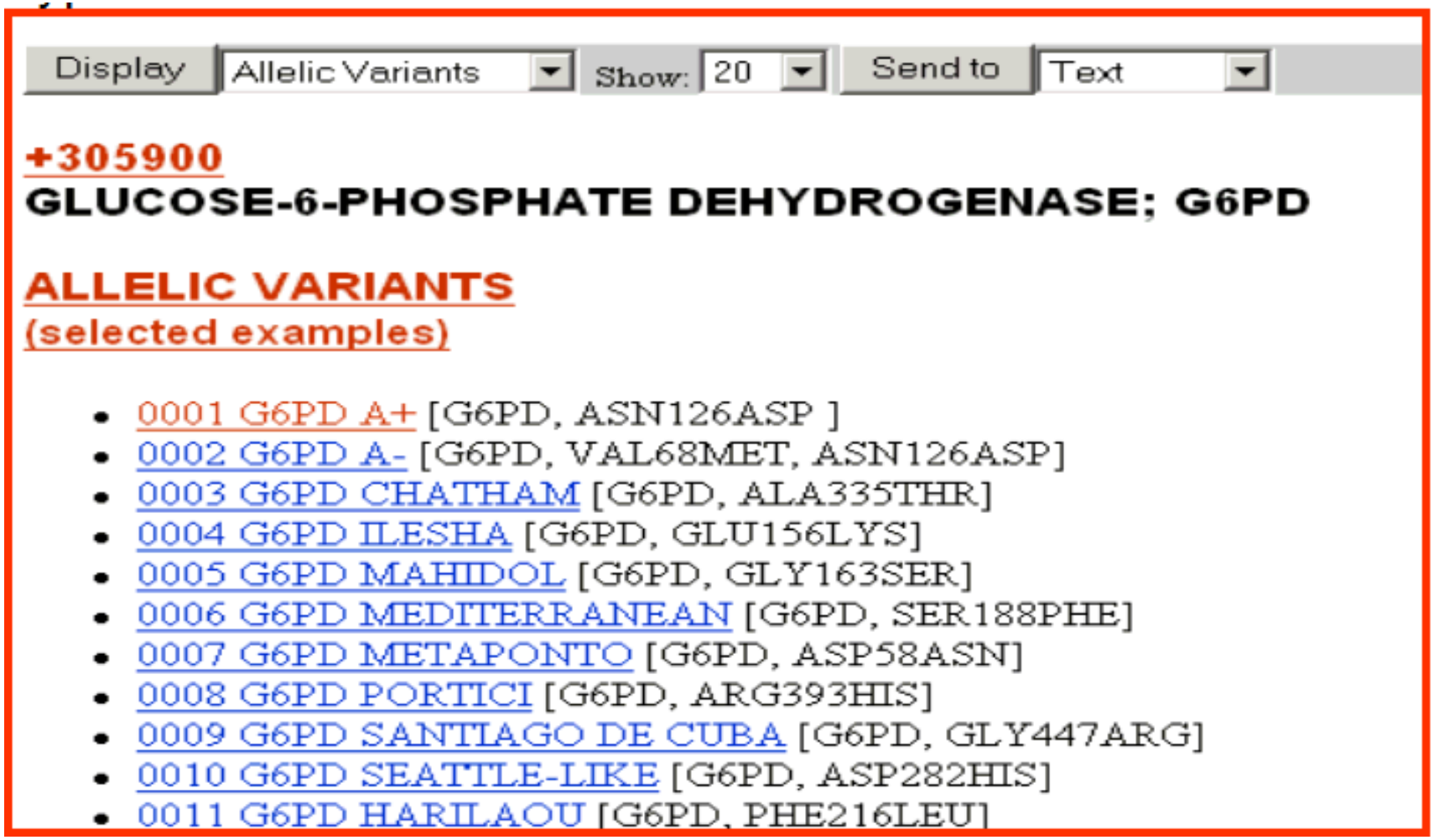
- OMIM : genetic disorders by all levels of mutation/variation
  nucleotide substitutions
  to large-scale chromosomal abnormalities.

- "Allelic Variants"
  nucleotide substitutions;
  - small insertions and deletions (indels)

- 10-digit OMIM number (e.g., 141900.0003)
- 6 digit OMIM number of the parent locus
- + decimal point + 4-digit variant number.

Viewing the List of Allelic Variants

OMIM allelic variant number (4 digit)
name of associated disorder or condition
gene symbol
type/location of variant



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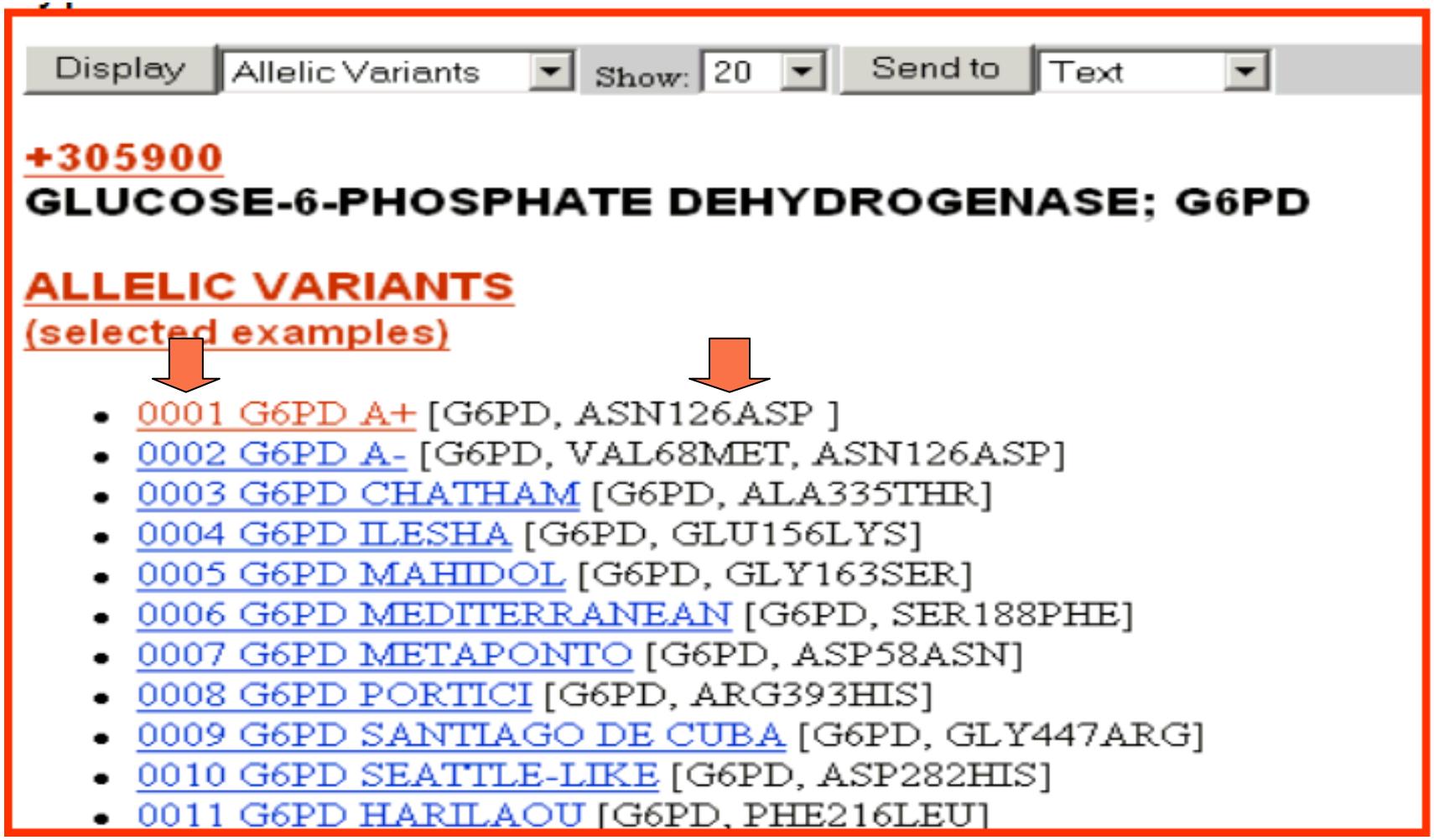
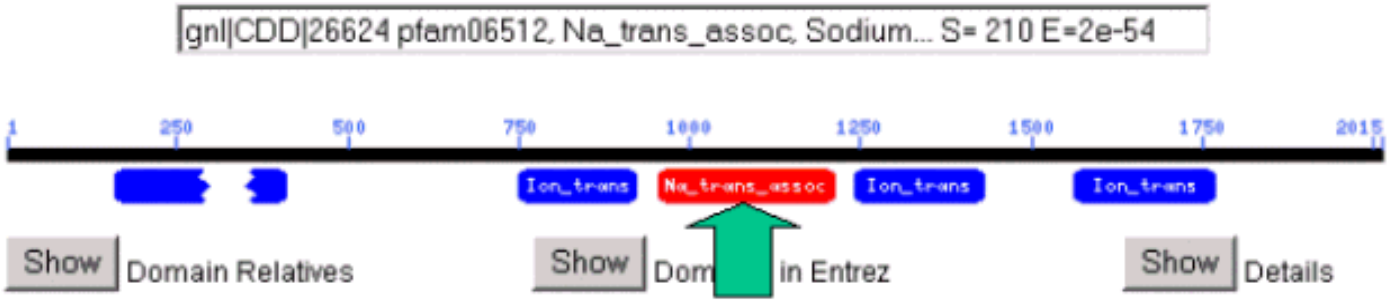


Functional Analysis of Polymorphisms

Sequence Alignment

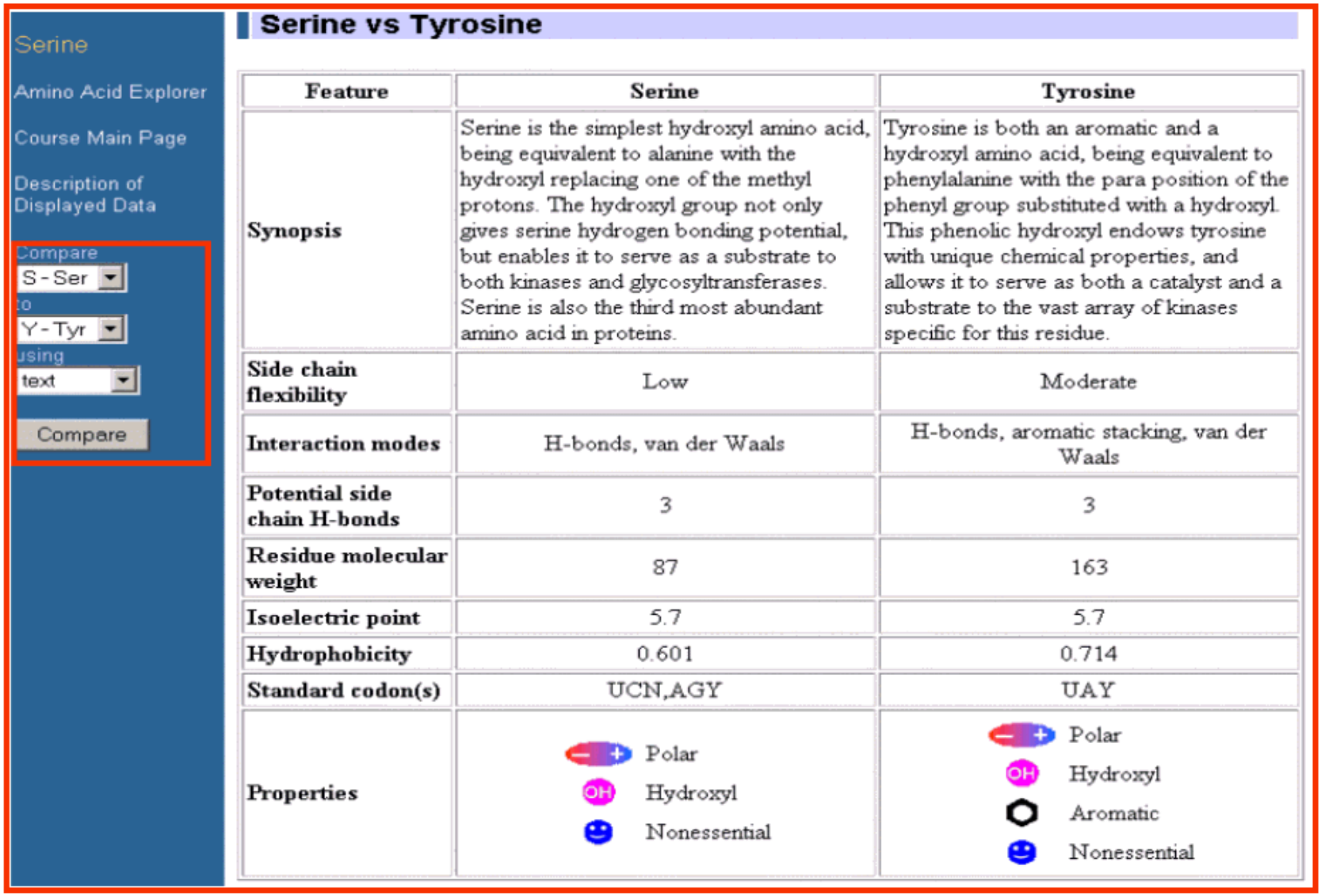



```
                          190       200       210       220       230       240
                    ....*....|....*....|....*....|....*....|....*....|....*....|
consensus     104 QPSE--E--SQFEDLPTE-TKR--LTSPDDESVETEVFSE-E-DLNLTIQDSRKK----- 149
query        1071 EESS--K--QESQPVSGGpEAP--PDSRTWSQVSATASSE-A-EASASQADWRQQ----- 1117
gi 4033376   1071 EESS--K--QQESQPVSG-WPRgpPDSRTWSQVSATASSE-A-EASASQADWRQQ----- 1118
gi 13774490  1062 QEED--E--ENSLGTEEE-SSK--QTPEDS-------YSE-G-ST---------------- 1090
gi 116452    1074 EESS--K--QESQVVSGG-HEP--YQEPRAWSQVSETTSS-E-AGASTSQADWQQ----- 1119
gi 1209467   1023 WQEE--DpkGQQEQLPQV-QKC--ENHQAARSPASMMSSE-DlAPYLGESWKRKDspqvp 1076
gi 116449    1054 VPIAvgE--SDFENLNTE-----------------EFSSE-S-ELE----ESKEK----- 1083
gi 27263190  1087 VPIAvgE--SDFENLNTE-----------------DFSSE-S-DLE----ESKEK----- 1116
gi 3087876    918 PIAS--E--ESDLEMPTE-E-------------ETDAPSEpE-DIKKPLQPL-------- 950
gi 14165234   309 ----------------------------DDESVDL---ST-E-ENMIKLKGA--L----- 328
```

```
S1102  Human  RTWSQVSATASSEAEASASQA
       Mouse  RAWSQVSETTSSEAEASTSQA
       Rat    RAWSQVSETTSSEAGASTSQA
```

Amino Acid Comparison Text View

NCBI Amino Acid Explorer



Serine

Amino Acid Explorer

Course Main Page

Description of Displayed Data

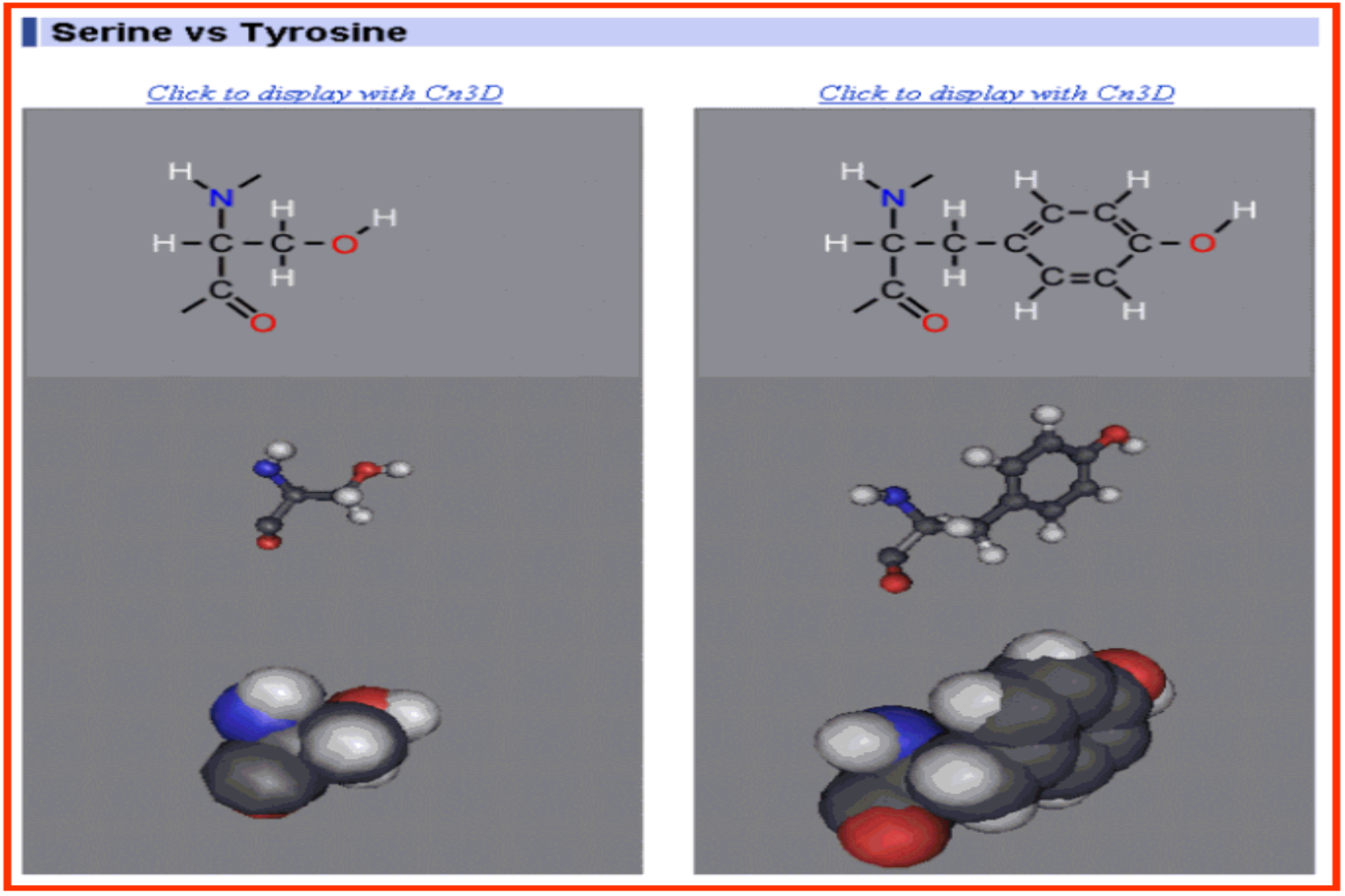
Compare
S - Ser
to
Y - Tyr
using
text
Compare

Serine vs Tyrosine

Feature	Serine	Tyrosine
Synopsis	Serine is the simplest hydroxyl amino acid, being equivalent to alanine with the hydroxyl replacing one of the methyl protons. The hydroxyl group not only gives serine hydrogen bonding potential, but enables it to serve as a substrate to both kinases and glycosyltransferases. Serine is also the third most abundant amino acid in proteins.	Tyrosine is both an aromatic and a hydroxyl amino acid, being equivalent to phenylalanine with the para position of the phenyl group substituted with a hydroxyl. This phenolic hydroxyl endows tyrosine with unique chemical properties, and allows it to serve as both a catalyst and a substrate to the vast array of kinases specific for this residue.
Side chain flexibility	Low	Moderate
Interaction modes	H-bonds, van der Waals	H-bonds, aromatic stacking, van der Waals
Potential side chain H-bonds	3	3
Residue molecular weight	87	163
Isoelectric point	5.7	5.7
Hydrophobicity	0.601	0.714
Standard codon(s)	UCN,AGY	UAY
Properties	Polar Hydroxyl Nonessential	Polar Hydroxyl Aromatic Nonessential

Amino Acid Comparison Graphic View

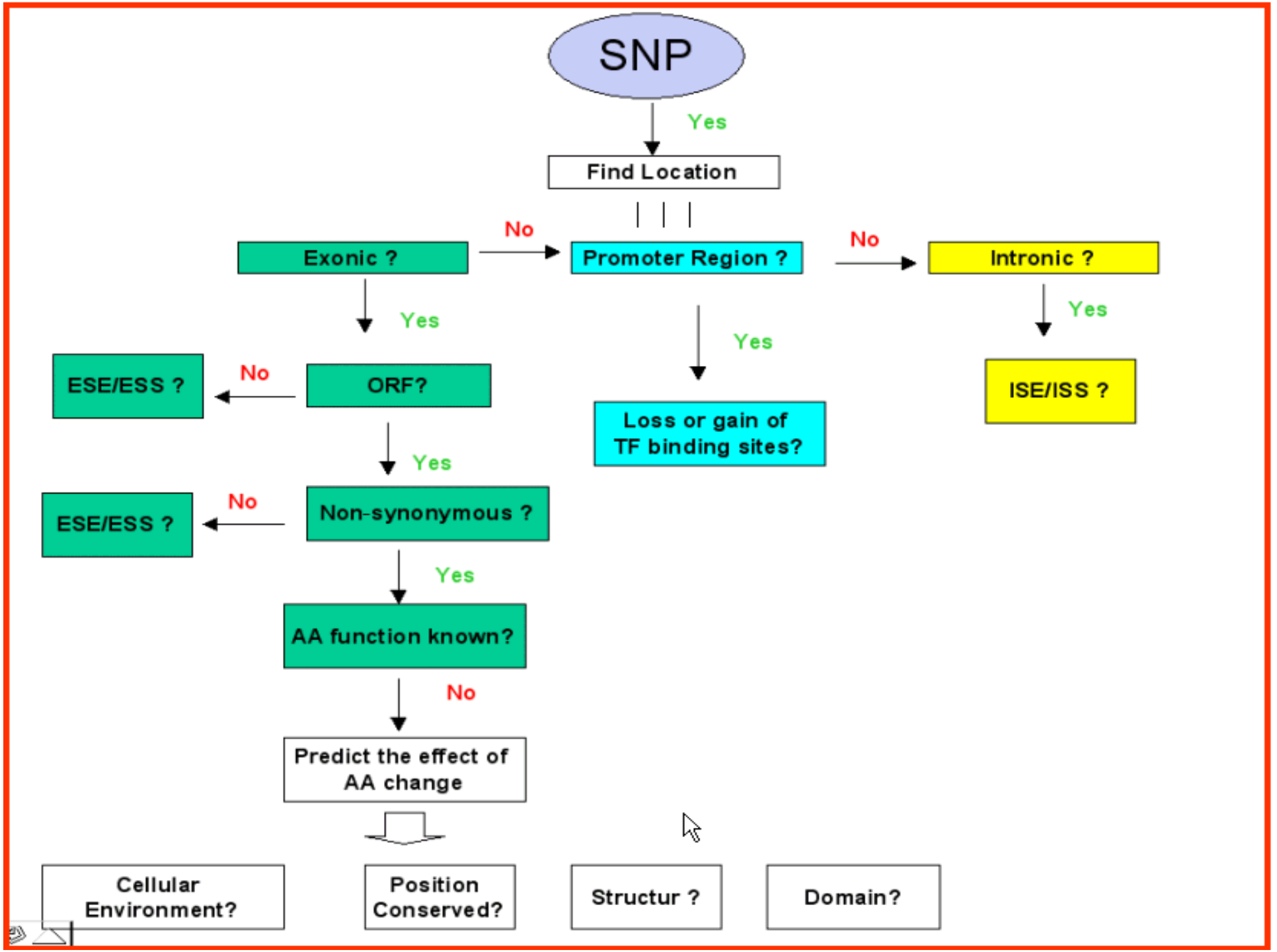
NCBI Amino Acid Explorer



Mutation Database

- Nucleic Acids Research Database Issue
  http://nar.oupjournals.org/content/vol32/suppl_1/

- HUGO Mutation Database Initiative
http://www2.ebi.ac.uk/mutations/cotton/dblist/dblist.html

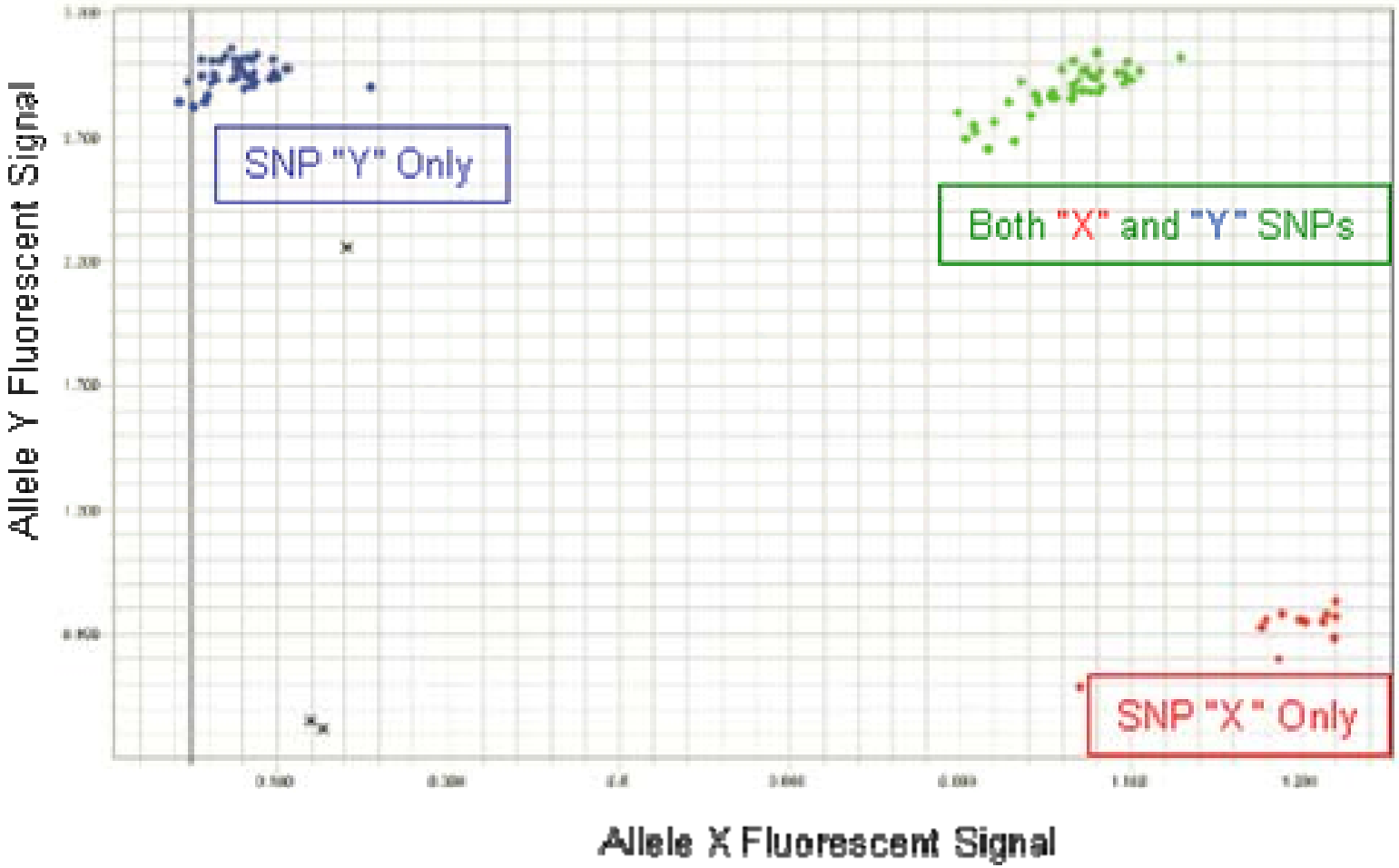

SNP
Yes
Find Location
Exonic ?
No
Promoter Region ?
No
Intronic ?
Yes
Yes
Yes
ESE/ESS ?
No
ORF?
Loss or gain of TF binding sites?
ISE/ISS ?
Yes
ESE/ESS ?
No
Non-synonymous ?
Yes
AA function known?
No
Predict the effect of AA change
Cellular Environment?
Position Conserved?
Structur ?
Domain?

SNP class:	
het	variation has unknown sequence composition, but is observed to be heterozygous
in del	insertion deletion polymorophism, deletions represented by '-' in allele string
microsat	microsatellite / simple sequence repeat
mixed	
mnp	multiple nucleotide polymorphism (all alleles same length where length>1)
named	allele sequences defined by name tag instead of raw sequence, e.g. (Alu)/-
no variation	submission reports invariant region in surveyed sequence
snp	true single nucleotide polymorphism

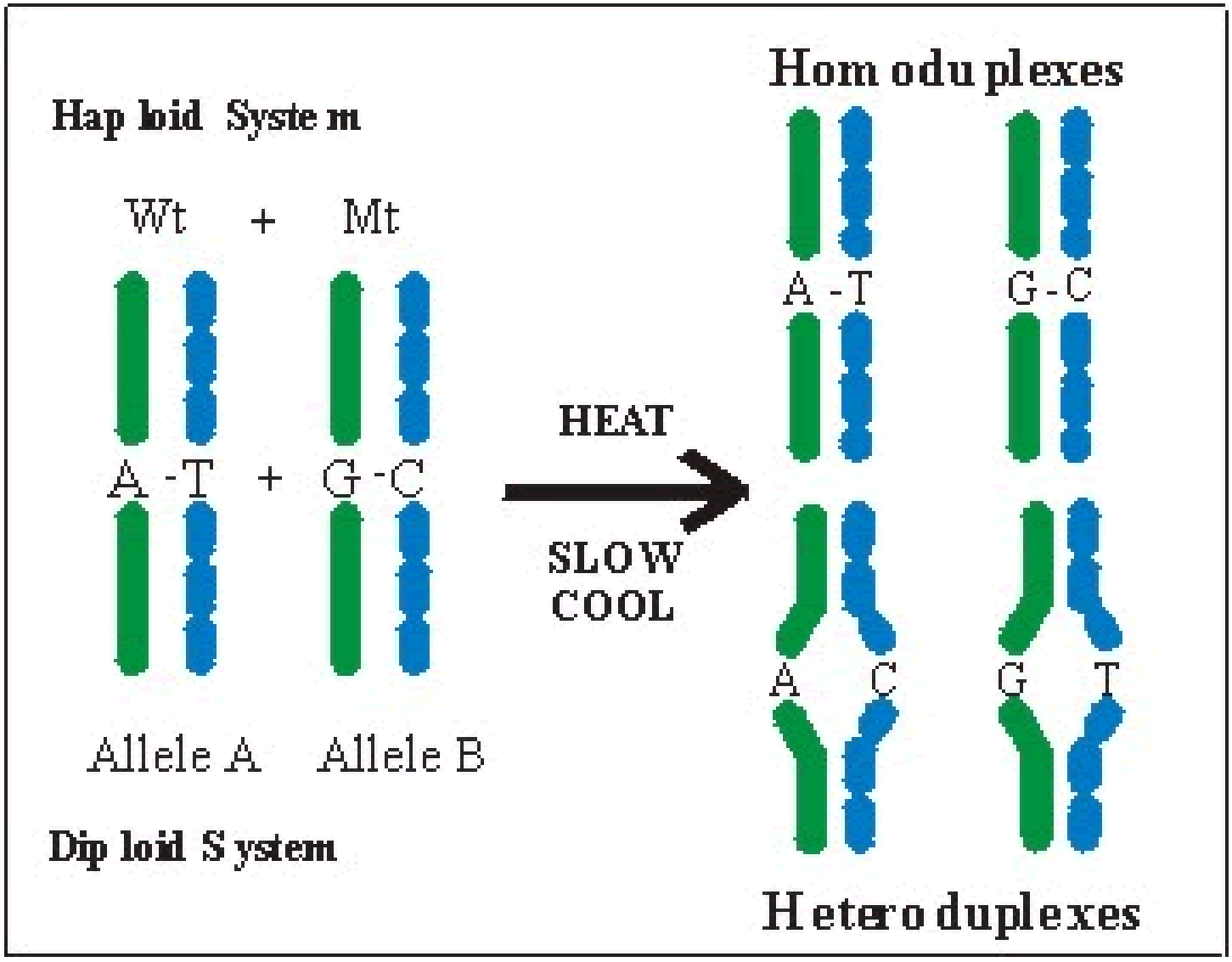
Method class

computed	variation was mined from sequence alignment with software
dhplc	Denaturing High Pressure Liquid Chromatography used to detect SNP
hybridize	hybridization method (e.g. chip) was used to assay for variation
other	other method used to detect variation
rflp	variation in enzyme restriction site used to detect variation
sequence	samples were sequenced and resulting alignment used to define variation
sscp	single stranded conformational polymorphism used to detect variation
unknown	

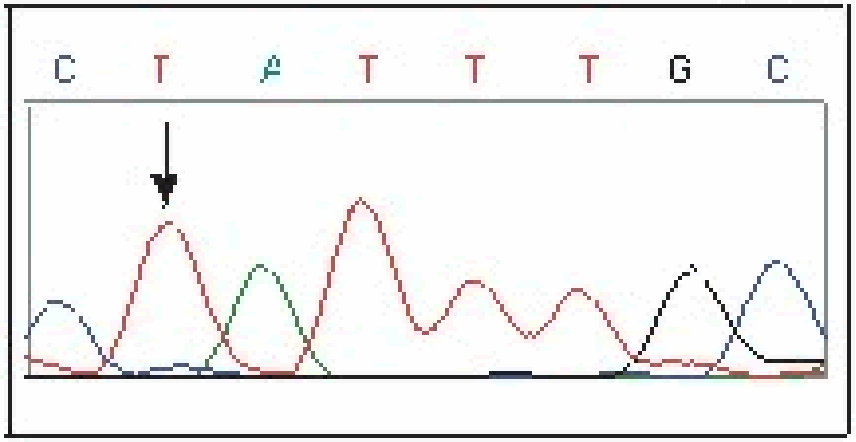
TaqMan Assay

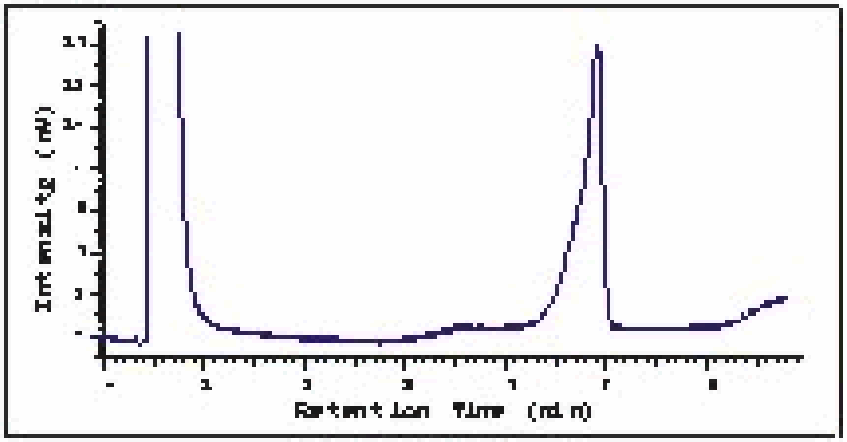


Denaturing High Performance Liquid Chromatography (DHPLC)

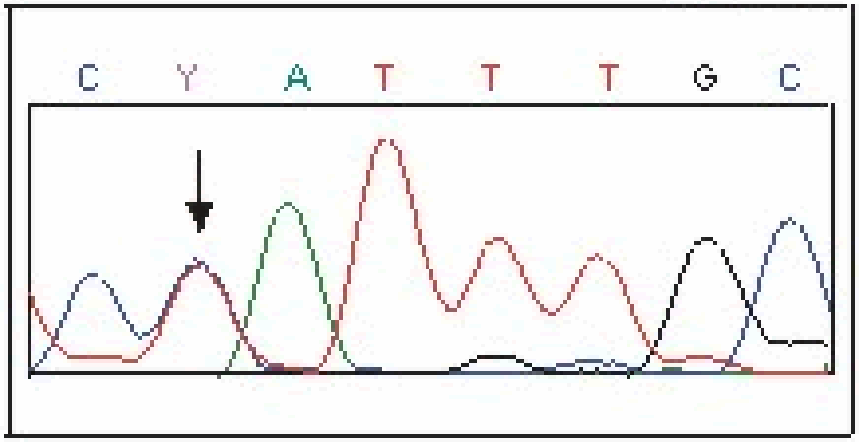


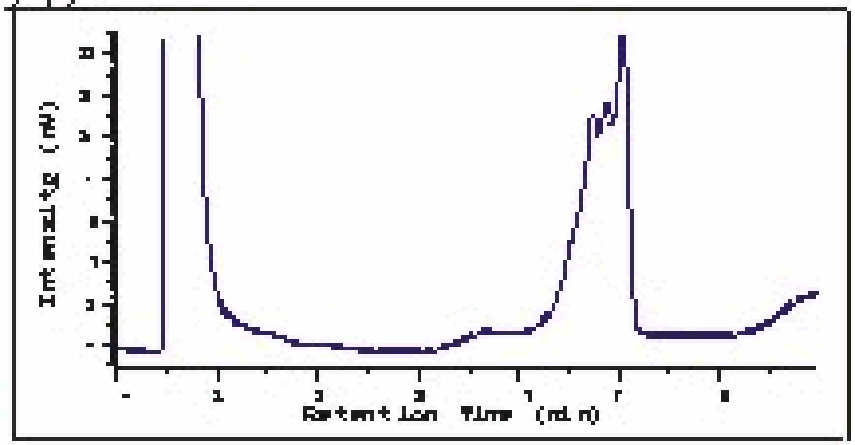
Homozygote



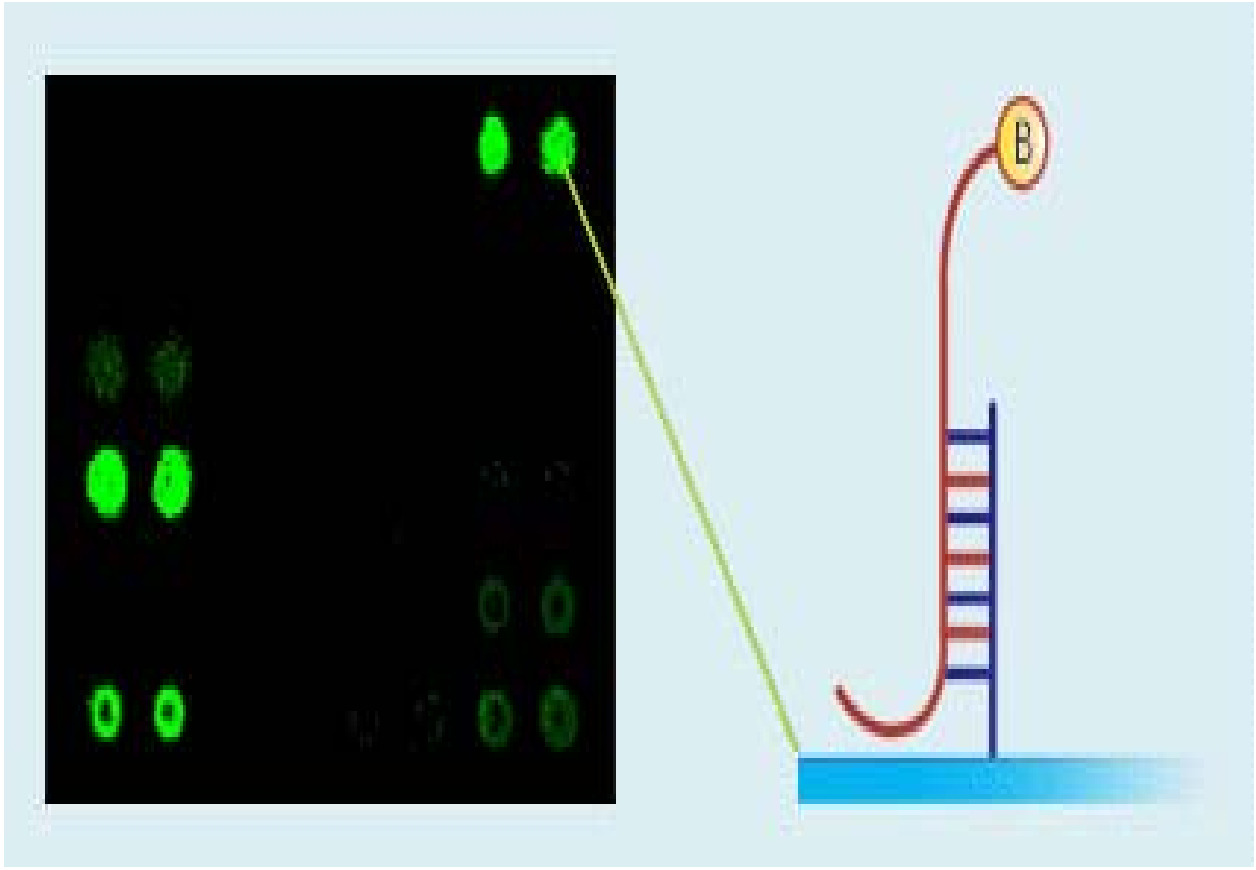


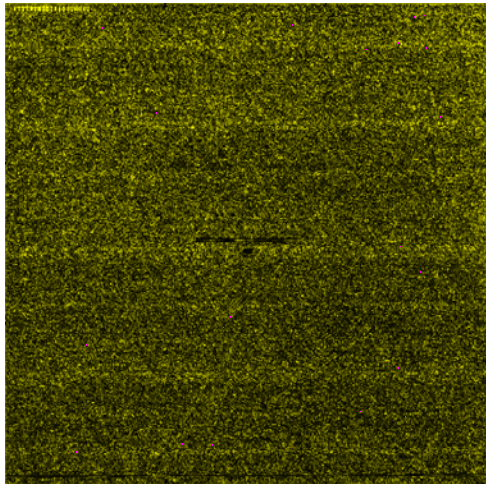
Heterozygote





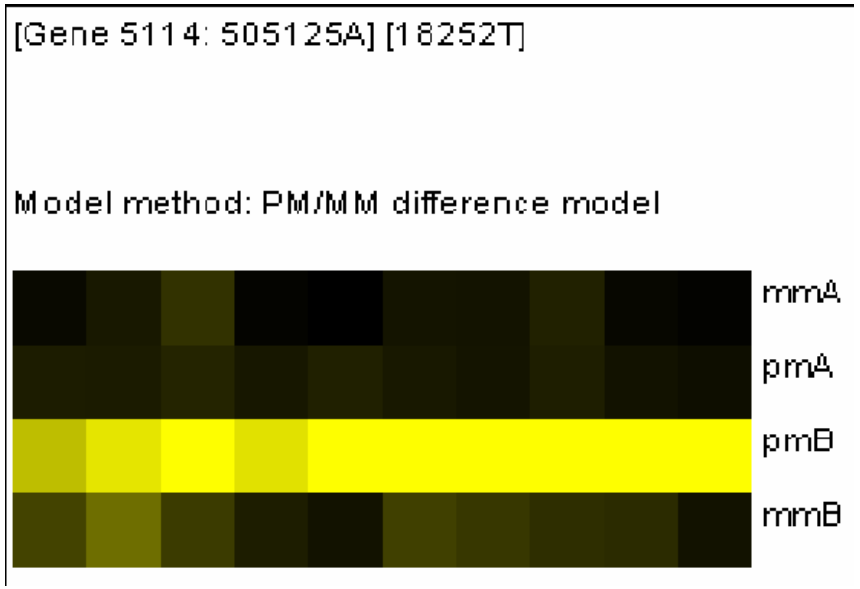
SNP Microarray Chip





Affymetrix Probe Layout

- Two alleles, A & B
- PerfectMatch (Signal)
  MisMatch (Background)



Pharmacogenomics

- Inherited variations in genes @ drug response
- Predict whether a good or bad response to a drug

pharmacogenomic.swf

Chemosensitivity & Genomics

phase I enzymes
activation or inactivation of the drug

Quinone oxidoreductase (NQO1, NADH)

carcinogenic quinones -> less toxic hydroxyquinones

inactivate the antileukemic agent doxorubicin

Lower activity GSTP1 1578A >G (*GSTP1*B*)
a.a. change Val105Ile

- associated with higher etoposide clearance in African Americans treated with steroids

GeneCard for protein-coding RUNX1 GC21M035081

runt-related transcription factor 1 (acute myeloid leukemia 1; aml1 oncogene)

Symbol approved by the HUGO Gene Nomenclature Committee (HGNC) database
(Previous symbols: **AML1, CBFA2**)

Services Jump to Section... NEW

Aliases and Descriptions
(According to [1]HGNC, [2]Entrez Gene, [3]UniProt/Swiss-Prot, [4]UniProt/TrEMBL, [5]GDB, [6]OMIM, [8]Nature:405,311-319 and CroW21, and/or [7]GeneLoc)
About This Section

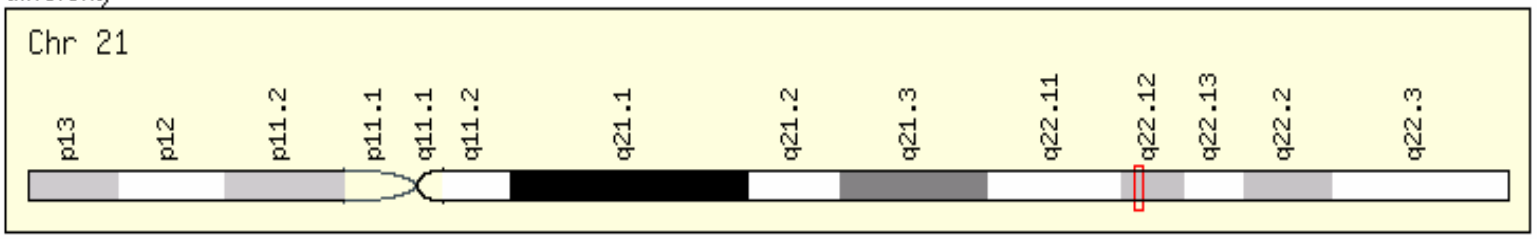
Aliases	Descriptions
AML1 [2,3,5,6]	Acute myeloid leukemia 1 protein [3]
AML1-EVI-1 [2]	CBF-alpha 2 [3]
AMLCR1 [1,2,5]	Core-binding factor, alpha 2 subunit [3]
CBFA2 [2,3,5,6]	Oncogene AML-1 [3]
EVI-1 [2]	PEA2-alpha B [3]
PEBP2A2 [1,2,5]	PEBP2-alpha B [3]
PEBP2aB [2]	Polyomavirus enhancer binding protein 2 alpha B subunit [3]
	Runt-related transcription factor 1 [3]
	SL3-3 enhancer factor 1 alpha B subunit [3]
	SL3/AKV core-binding factor alpha B subunit [3]
	acute myeloid leukemia 1 protein (oncogene AML-1), core-binding factor, alpha subunit [8]
	core-binding factor, runt domain, alpha subunit 2 (acute myeloid leukemia 1; aml1 oncogene) [5]

Genomic Location
(According to GeneLoc and/or HGNC, and/or Entrez Gene (NCBI build 35), and/or miRBase, Genomic Views According to UCSC and Ensembl, Whole Chromsome Sequence According to Nature (Cited Here with Permission):405,311-319 and CroW21)
About This Section

Jump to Section...

Chromosome: **21**
Entrez Gene cytogenetic band: **21q22.3** *Ensembl cytogenetic band:* **21q22.12**
Nature(405: 311-319) cytogenetic band: **21q22.12**

Gene in genomic location: bands according to Ensembl, locations according to GeneLoc *(and/or* ***Entrez Gene*** *and/or* ***Ense***
different)



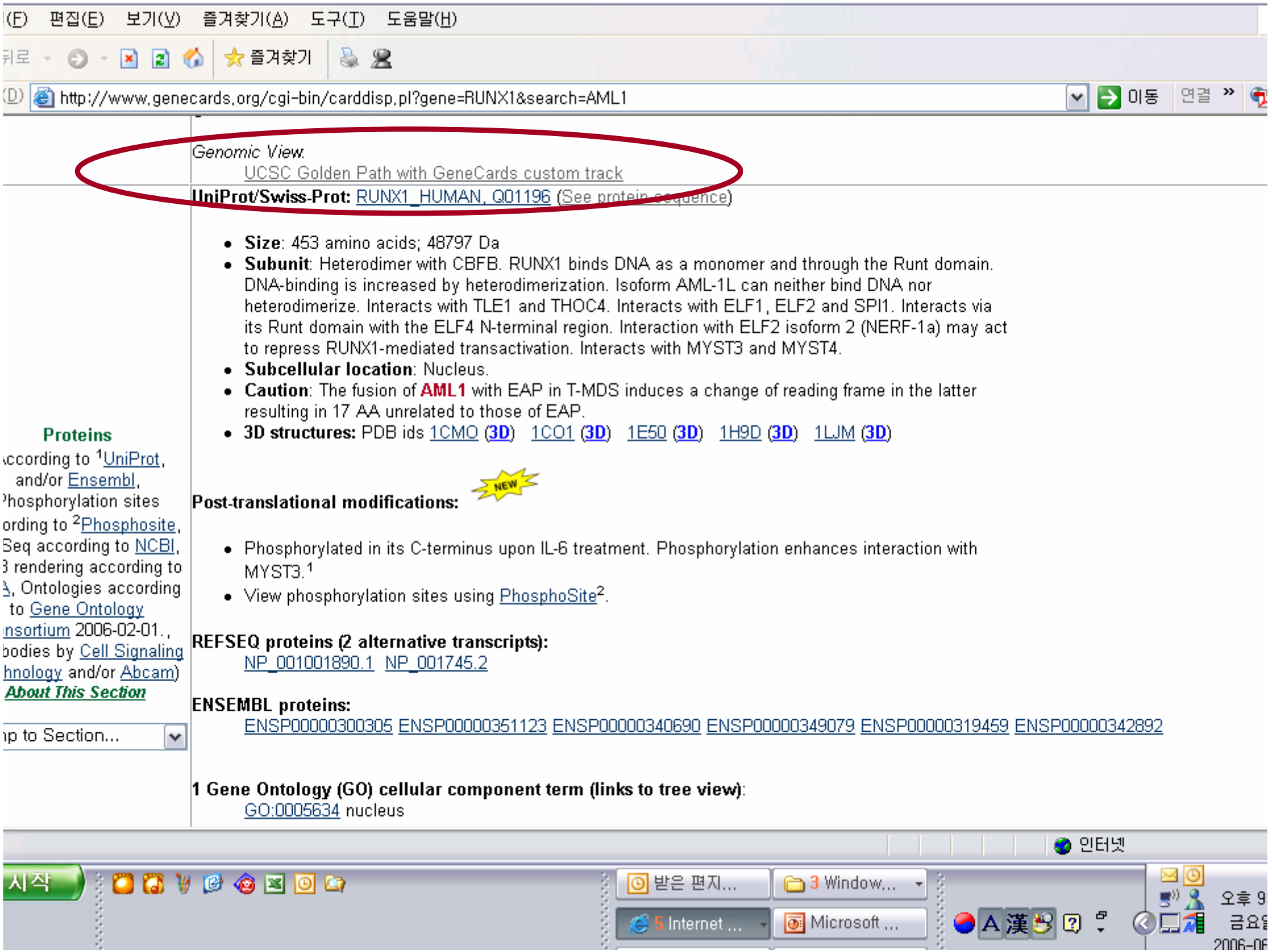
GeneLoc gene densities for chromosome 21

(about GC identifiers)

	GeneLoc	Nature:405,311-319
GC21M035081:		
Start:	**35,081,971** bp from *pter*	**21,770,223** bp from *centromere*
End:	**35,343,511** bp from *pter*	**21,837,636** bp from *centromere*
Size:	**261,540** bases	**67,414** bases
Orientation:	**minus** strand	**minus** strand

RefSeq genomic assemblies:
NC_000021.7 NT_011512.10 NT_086913.1

Whole chromosome sequencing:
cDNA sequence: D43967
genomic clones: PPQ140K16 to P499A22


(F) 편집(E) 보기(V) 즐겨찾기(A) 도구(T) 도움말(H)
즐겨찾기
http://www.genecards.org/cgi-bin/carddisp.pl?gene=RUNX1&search=AML1
이동
연결
Genomic View.
UCSC Golden Path with GeneCards custom track
UniProt/Swiss-Prot: RUNX1_HUMAN, Q01196 (See protein sequence)
Size: 453 amino acids; 48797 Da
Subunit: Heterodimer with CBFB. RUNX1 binds DNA as a monomer and through the Runt domain. DNA-binding is increased by heterodimerization. Isoform AML-1L can neither bind DNA nor heterodimerize. Interacts with TLE1 and THOC4. Interacts with ELF1, ELF2 and SPI1. Interacts via its Runt domain with the ELF4 N-terminal region. Interaction with ELF2 isoform 2 (NERF-1a) may act to repress RUNX1-mediated transactivation. Interacts with MYST3 and MYST4.
Subcellular location: Nucleus.
Caution: The fusion of AML1 with EAP in T-MDS induces a change of reading frame in the latter resulting in 17 AA unrelated to those of EAP.
3D structures: PDB ids 1CMO (3D) 1CO1 (3D) 1E50 (3D) 1H9D (3D) 1LJM (3D)
Proteins
ccording to 1UniProt, and/or Ensembl, hosphorylation sites ording to 2Phosphosite, Seq according to NCBI, rendering according to , Ontologies according to Gene Ontology nsortium 2006-02-01., bodies by Cell Signaling hnology and/or Abcam)
About This Section
p to Section...
NEW
Post-translational modifications:
Phosphorylated in its C-terminus upon IL-6 treatment. Phosphorylation enhances interaction with MYST3.1
View phosphorylation sites using PhosphoSite2.
REFSEQ proteins (2 alternative transcripts):
NP_001001890.1 NP_001745.2
ENSEMBL proteins:
ENSP00000300305 ENSP00000351123 ENSP00000340690 ENSP00000349079 ENSP00000319459 ENSP00000342892
1 Gene Ontology (GO) cellular component term (links to tree view):
GO:0005634 nucleus
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받은 편지...
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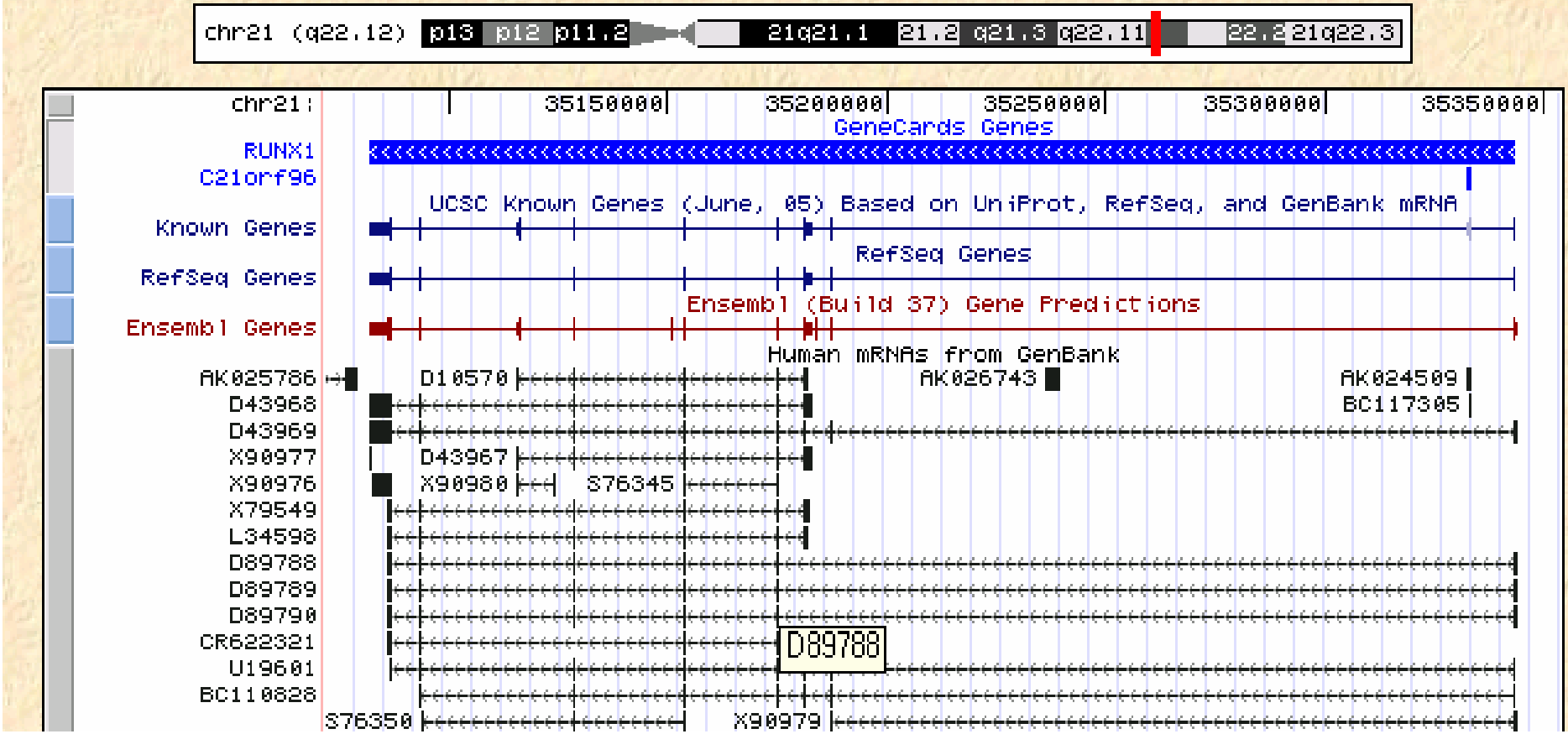
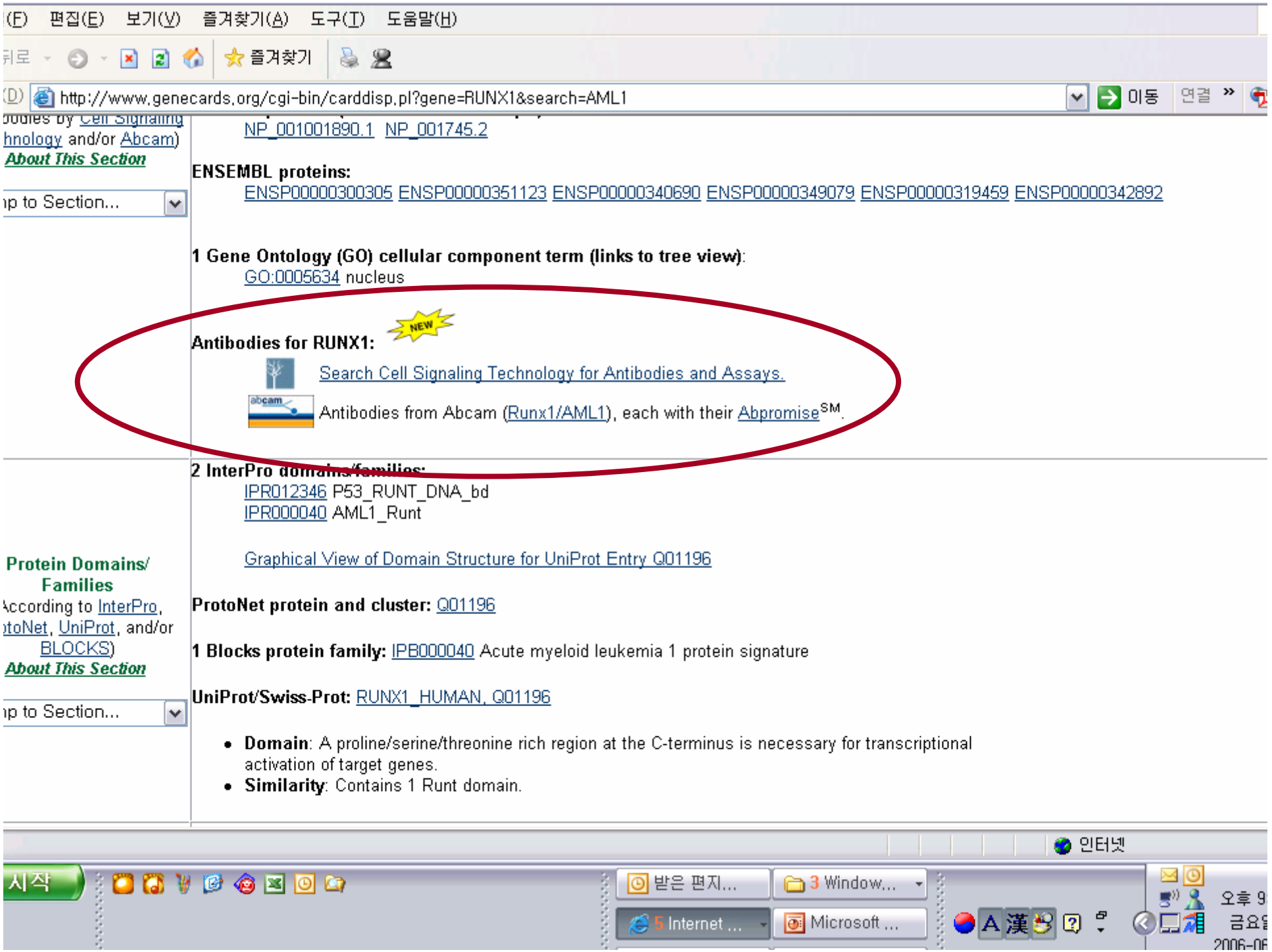
Microsoft ...
오후 9
금요일
2006-06

UCSC Genome Browser on Human May 2004 Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chr21:35,071,971-35,353,511 jump clear size 281,541 bp. configure




(F) 편집(E) 보기(V) 즐겨찾기(A) 도구(T) 도움말(H)
즐겨찾기
http://www.genecards.org/cgi-bin/carddisp.pl?gene=RUNX1&search=AML1
이동
연결
hnology and/or Abcam)
About This Section
NP_001001890.1 NP_001745.2
ENSEMBL proteins:
ENSP00000300305 ENSP00000351123 ENSP00000340690 ENSP00000349079 ENSP00000319459 ENSP00000342892
1 Gene Ontology (GO) cellular component term (links to tree view):
GO:0005634 nucleus
NEW
Antibodies for RUNX1:
Search Cell Signaling Technology for Antibodies and Assays.
Antibodies from Abcam (Runx1/AML1), each with their Abpromise℠.
Protein Domains/ Families
According to InterPro, toNet, UniProt, and/or BLOCKS)
About This Section
2 InterPro domains/families:
IPR012346 P53_RUNT_DNA_bd
IPR000040 AML1_Runt
Graphical View of Domain Structure for UniProt Entry Q01196
ProtoNet protein and cluster: Q01196
1 Blocks protein family: IPB000040 Acute myeloid leukemia 1 protein signature
UniProt/Swiss-Prot: RUNX1_HUMAN, Q01196
Domain: A proline/serine/threonine rich region at the C-terminus is necessary for transcriptional activation of target genes.
Similarity: Contains 1 Runt domain.
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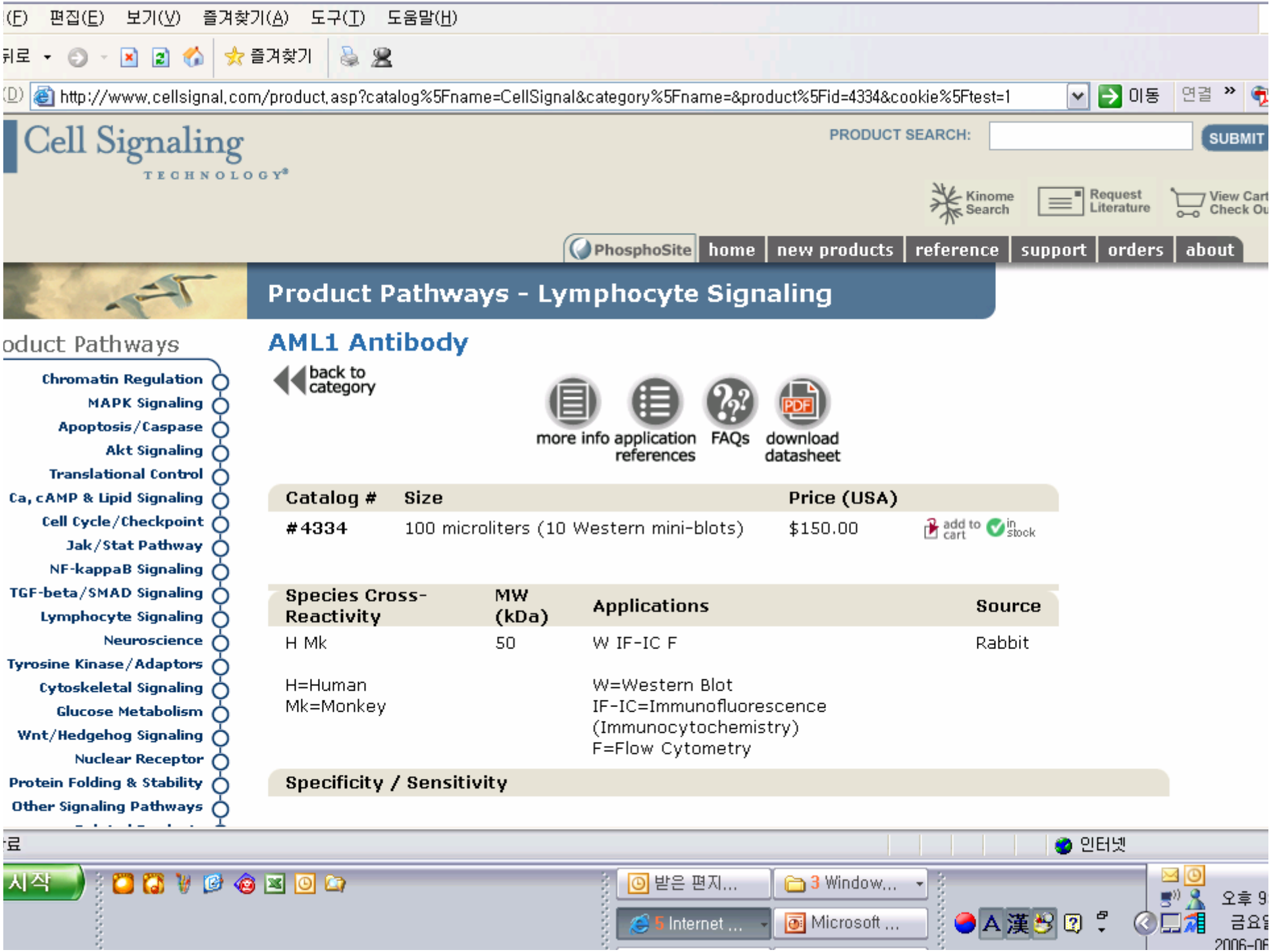
2006-06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즐겨찾기
http://www.cellsignal.com/product.asp?catalog%5Fname=CellSignal&category%5Fname=&product%5Fid=4334&cookie%5Ftest=1
이동
연결
Cell Signaling TECHNOLOGY
PRODUCT SEARCH:
SUBMIT
Kinome Search
Request Literature
View Cart
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Product Pathways - Lymphocyte Signaling
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Chromatin Regulation
MAPK Signaling
Apoptosis/Caspase
Akt Signaling
Translational Control
Ca, cAMP & Lipid Signaling
Cell Cycle/Checkpoint
Jak/Stat Pathway
NF-kappaB Signaling
TGF-beta/SMAD Signaling
Lymphocyte Signaling
Neuroscience
Tyrosine Kinase/Adaptors
Cytoskeletal Signaling
Glucose Metabolism
Wnt/Hedgehog Signaling
Nuclear Receptor
Protein Folding & Stability
Other Signaling Pathways
AML1 Antibody
back to category
more info
application references
FAQs
download datasheet
Catalog # Size Price (USA)
#4334 100 microliters (10 Western mini-blots) $150.00
add to cart
in stock
Species Cross-Reactivity
MW (kDa)
Applications
Source
H Mk
50
W IF-IC F
Rabbit
H=Human
Mk=Monkey
W=Western Blot
IF-IC=Immunofluorescence (Immunocytochemistry)
F=Flow Cytometry
Specificity / Sensitivity
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ug Discovery Tools
Screening Technologies
Active Kinases & Kits
PathScan ELISA
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Rabbit Monoclonals
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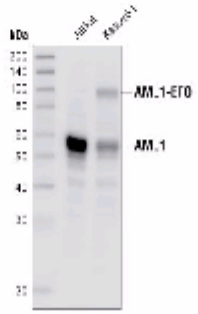
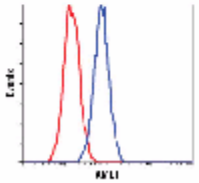
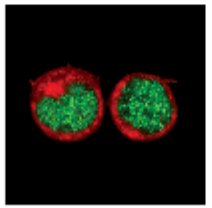
protein.

Click on any image below to Zoom

Western Blotting	Flow Cytometry	IF-IC
		
Western blot analysis of extracts from Jurkat and Kasumi-1 cells, using AML1 Antibody.	Flow cytometric analysis of Jurkat cells, using AML1 Antibody (blue) compared to a nonspecific negative control antibody (red).	Confocal immunofluorescent image of Jurkat cells labeled with AML1 Antibody (green). Mitochondria have been labeled with MitoTracker Red CMXRos.

Source / Purification

(F) 편집(E) 보기(V) 즐겨찾기(A) 도구(T) 도움말(H)

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(D) http://www.dsi.univ-paris5.fr/genatlas/ 이동 연결 »

ENATLAS
versité René Descartes - Paris

20861 genes / 3883 phenotypes / 61367 citations
release Fri 2 Jun 2006

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GENATLAS : GENE Database

omim HGNC | sequences genelynx | swissprot genecards | citations Ensembl | locuslink Unigene | source linkage

FLASH GENE

Symbol	MPO
HGNC name	myeloperoxidase
HGNC id	7218
Corresponding disease	MPO
Location	17q22-q23.1
EC.number	1.11.1.7

last update : 11/03/2006

DNA | RNA | EXP/sub-loc | PROTEIN | PATHOLOGY

DNA

TYPE	functioning gene
SPECIAL FEATURE	
text	tail to tail with LPO
STRUCTURE	11,08 kb 12 Exon(s)

present in the contig : NT_010783 of Genbank in reverse/complement

DNA size 11.08 Kb mRNA size 3215 bp 12 exons NM_000250 see the exons

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(D) http://www.dsi.univ-paris5.fr/genatlas/ 이동 연결 »

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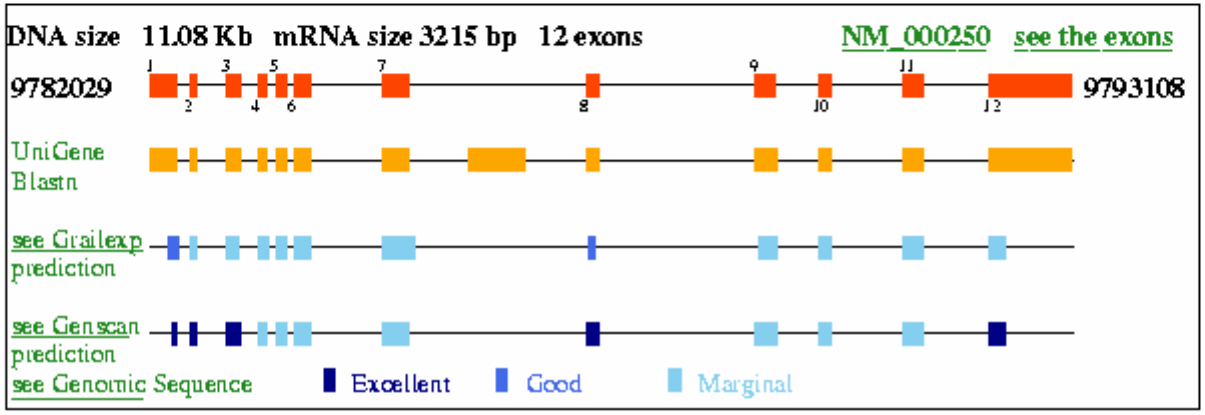
present in the contig : NT_010783 of Genbank in reverse/complement



10 Kb 5' upstream gene genomic sequence study

MAPPING	cloned	Y	linked	status	confirmed

mode in situ hybridation, recombinant DNA, somatic cell hybrid

Map cen – MPO *MPO* – LPO – EPX – qter

Authors Sakamaki (00)

Physical map

MPO Physical Map Genatlas link Locuslink link

5'-->3'

EPX
LOC

인터넷

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(D) http://www.infobiogen.fr/services/chromcancer/ 이동 연결 »

Atlas of Genetics and Cytogenetics in Oncology and Haematology

How to contribute: **Instructions to Authors**

ENTITIES: by THEME

GENES
LEUKAEMIAS
SOLID TUMOURS
CANCER PRONE DISEASES

ENTITIES: by CHROMOSOME

X Y 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 Not Assigned

REVIEWS and CASE REPORTS

DEEP INSIGHTS
CASE REPORTS (see also a list of Rare Translocations and Authors Instructions)
11p15/NUP98 Project: Authors Instructions

PORTAL

DATABASES AND LINKS

료 인터넷

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(D) http://canceratlas.spis.co.uk/login.aspx 이동 연결 »

Atlas | Cancer

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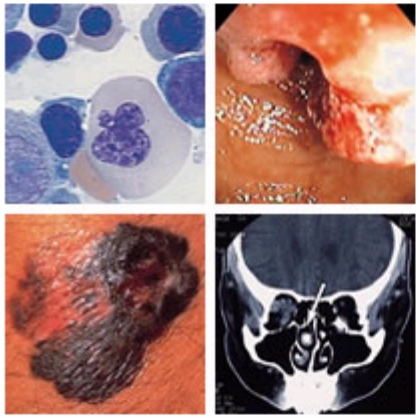

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