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Bioinformatics

Common tools, useful databases, and tricks of the trade.



bioteach.ubc.ca/bioinfo2008

Workshop Schedule

- Laptops, available here for your use 9am - 4:30pm
- wireless login

mslguest

4myguest
- Vancouver guide books available



Today's Topics

- **BLAST** - Finding Function by Sequence Similarity
- **GUIDED TOUR** - Advanced Tips & Tricks for Using BLAST
- **PRACTICAL EXERCISES** - The Jurassic Park Detective Story
- **Genome Browsers** - Accessing Genome Annotations
- **PRACTICAL EXERCISES** - Three different views of the BRCA1 gene

BLAST

Finding Function By Sequence Similarity



Concepts of Sequence Similarity Searching

- The premise:

One sequence by itself is not informative; it must be analyzed by comparative methods against existing sequence databases to develop hypothesis concerning relatives and function.

The BLAST algorithm

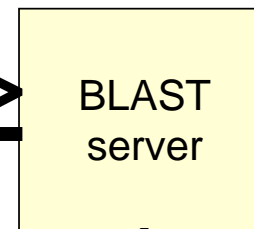
- The BLAST programs (Basic Local Alignment Search Tools) are a set of sequence comparison algorithms introduced in 1990 that are used to search sequence databases for optimal local alignments to a query.
 - Altschul SF, Gish W, Miller W, Myers EW, Lipman DJ (1990) “Basic local alignment search tool.” J. Mol. Biol. 215:403-410.
 - Altschul SF, Madden TL, Schaeffer AA, Zhang J, Zhang Z, Miller W, Lipman DJ (1997) “Gapped BLAST and PSI-BLAST: a new generation of protein database search programs.” NAR 25:3389-3402.

```
>gi|15237380|ref|NP_197163.1| myb family transcription factor (MYB43) [Arabidopsis thaliana]
MGRQPCCKVGLKKGPMTEEDKKLINFILTNHCCWRALPKLSGLLRGKSCRLRWLYLRPDLKRGLL
SEYEEQKVINLHAQLGNRWSTIASHLPGRTONEIKHWNTHIKKLRMGIDPLTHKPLSEGEASQQAQG
RKXSLVPHDKNPKQDQQTQDEGEQHLQALEKNNTSVSGDGPCEVPLLNPHIELIDTSSSHHHHSN
DDNVAINTSKFTSPSSSSSTSSCISVWPGDEFKFFDEMEILDLKWLSSDQSLGDDISKDGKFNSTV
IDTNLWDINDLSSLDPMFNEHDDGFIQNGNGCRMVLDQDSWTFDLL
```

Submit Query

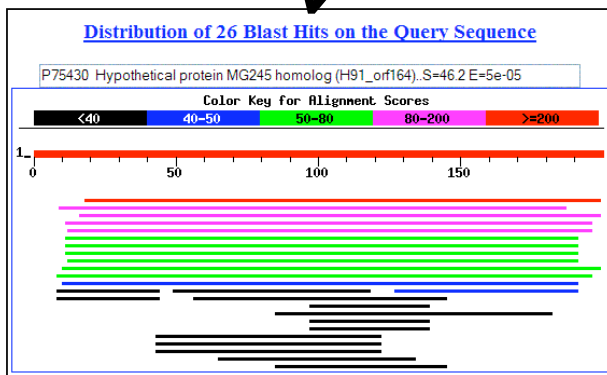


Request Results



Return Formatted Results

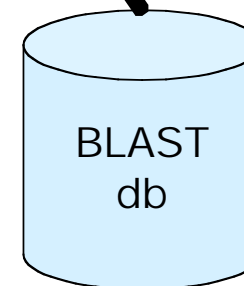
Display Results



fetch ASN.1



fetch sequence



What BLAST tells you ...

- BLAST reports surprising alignments
 - Different than chance
- Assumptions
 - Random sequences
 - Constant composition
- Conclusions
 - Surprising similarities imply evolutionary homology

Evolutionary Homology: descent from a common ancestor
Does not always imply similar function

Basic Local Alignment Search Tool

- Widely used similarity search tool
- Heuristic approach based on Smith Waterman algorithm
- Finds best local alignments
- Provides statistical significance
- www, standalone, and network clients

BLAST programs

Program	Description
blastp	Compares an amino acid query sequence against a protein sequence database.
blastn	Compares a nucleotide query sequence against a nucleotide sequence database.
blastx	Compares a nucleotide query sequence translated in all reading frames against a protein sequence database. You could use this option to find potential translation products of an unknown nucleotide sequence.
tblastn	Compares a protein query sequence against a nucleotide sequence database dynamically translated in all reading frames.
tblastx	Compares the six-frame translations of a nucleotide query sequence against the six-frame translations of a nucleotide sequence database.

more BLAST programs

Program		Notes
Megablast	Contiguous	Nearly identical sequences
	Discontiguous	Cross-species comparison
Position Specific	PSI-BLAST	Automatically generates a position specific score matrix (PSSM)
	RPS-BLAST	Searches a database of PSI-BLAST PSSMs



nucleotide only



protein only

BLAST Algorithm

- Scoring of matches done using scoring matrices
- Sequences are split into words (default n=3)
 - Speed, computational efficiency
- BLAST algorithm extends the initial “seed” hit into an HSP
 - HSP = high scoring segment pair = Local optimal alignment

Sequence Similarity Searching – The statistics are important

Discriminating between real and artifactual matches is done using an estimate of probability that the match might occur by chance.

We'll talk more about the meaning of the scores (S) and e-values (E) that are associated with BLAST hits

Where does the score (S) come from?

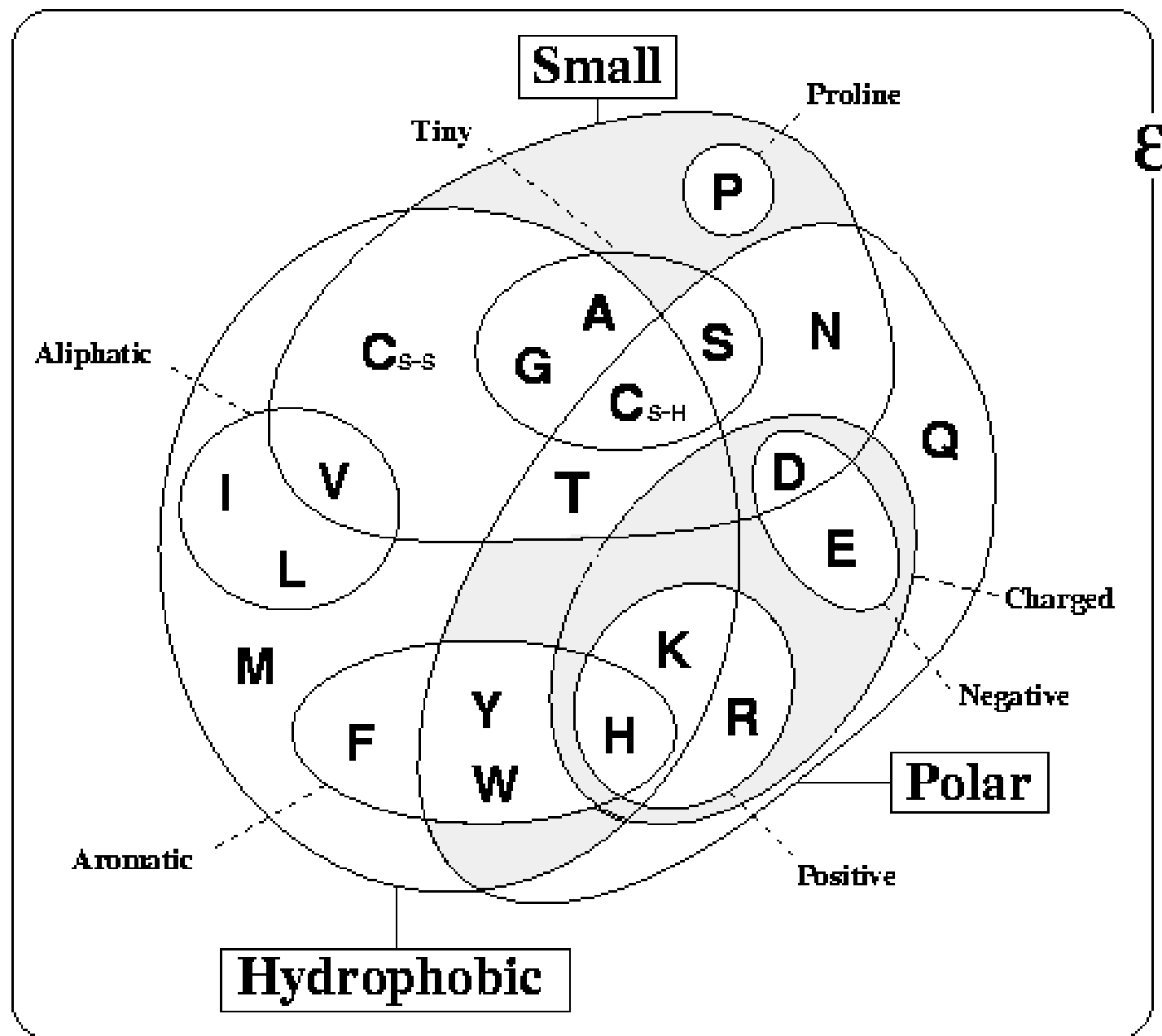
- The quality of each pair-wise alignment is represented as a score and the scores are ranked.
- **Scoring matrices** are used to calculate the score of the alignment base by base (DNA) or amino acid by amino acid (protein).
- **The alignment score will be the sum of the scores for each position.**

What's a scoring matrix?

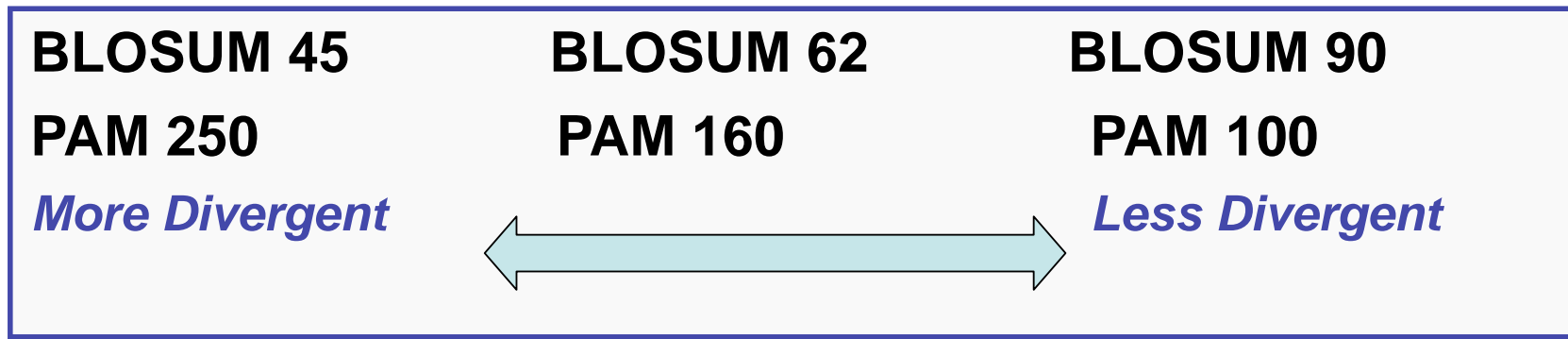
- Substitution matrices are used for amino acid alignments.
- each possible residue substitution is given a score
- A simpler unitary matrix is used for DNA pairs (+1 for match, -2 mismatch)

	A	C	D	E	F	G	H	
A	4	0	-2	-1	-2	0	-2	
C	0	9	-3	-4	-2	-3	-3	
D	-2	-3	6	2	-3	-1	-1	
E	-1	-4	2	5	-3	-2	0	
F	-2	-2	-3	-3	6	-3	-1	
G	0	-3	-1	-2	-3	6	-1	
H	-2	-3	-1	0	-1	-1	0	

BLOSUM 62



BLOSUM vs PAM



- BLOSUM 62 is the default matrix in BLAST 2.0. Though it is tailored for comparisons of moderately distant proteins, it performs well in detecting closer relationships. A search for distant relatives may be more sensitive with a different matrix.

What do the Score and the e-value really mean?

- The quality of the alignment is represented by the **Score (S)**.

The score of an alignment is calculated as the sum of substitution and gap scores. Substitution scores are given by a look-up table (PAM, BLOSUM) whereas gap scores are assigned empirically .

- The significance of each alignment is computed as an **E value (E)**.

Expectation value. The number of different alignments with scores equivalent to or better than S that are expected to occur in a database search by chance. The lower the E value, the more significant the score.

Notes on E-values

- Low E-values suggest that sequences are homologous
 - ⊙ Can't show non-homology
- Statistical significance depends on both the size of the alignments and the size of the sequence database
 - ▶ Important consideration for comparing results across different searches
 - ▶ E-value increases as database gets bigger
 - ▶ E-value decreases as alignments get longer

Homology: Some Guidelines

- Similarity can be indicative of homology
- Generally, if two sequences are significantly similar over entire length they are likely homologous
- Low complexity regions can be highly similar without being homologous
- Homologous sequences not always highly similar

Suggested Reading

Take Home Message:
Always look at your alignments

SCOTT

- Source: Chapter 11 – Bioinformatics: A Practical Guide to the Analysis of Genes and Proteins
- For nucleotide based searches, one should look for hits with E-values of 10^{-6} or less and sequence identity of 70% or more
- For protein based searches, one should look for hits with E-values of 10^{-3} or less and sequence identity of 25% or more

BLAST Algorithm

- Scoring of matches done using scoring matrices
- Sequences are split into words (default $n=3$)
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How Does BLAST Really Work?

- The BLAST programs improved the overall speed of searches while retaining good sensitivity (important as databases continue to grow) by breaking the query and database sequences into fragments ("words"), and initially seeking matches between fragments.
- Word hits are then extended in either direction in an attempt to generate an alignment with a score exceeding the threshold of "S".

BLAST Algorithm

Query Word ($W = 3$)

TLSHAWRLSNETDKRPFIEAERL**RDQ**HKKDYPEYKYQPRRRKNGKPGSSSEADAHSE

Determine neighborhood

RDQ 16	QDQ 12	EDQ 11	RDN 11	RDB 11	BDQ 10	RDP 10
RBQ 14	REQ 12	HDQ 11	RDD 11	ADQ 10	XDQ 10	RDT 10
RDZ 14	RDR 12	ZDQ 11	RDH 11	MDQ 10	RQQ 10	RDY 10
KDQ 13	RDK 12	RNQ 11	RDM 11	SDQ 10	RSQ 10	RDX 10
RDE 13	NDQ 11	RZQ 11	RDS 11	TDQ 10	RDA 10	DDQ 9 ...

How Does BLAST Really Work?

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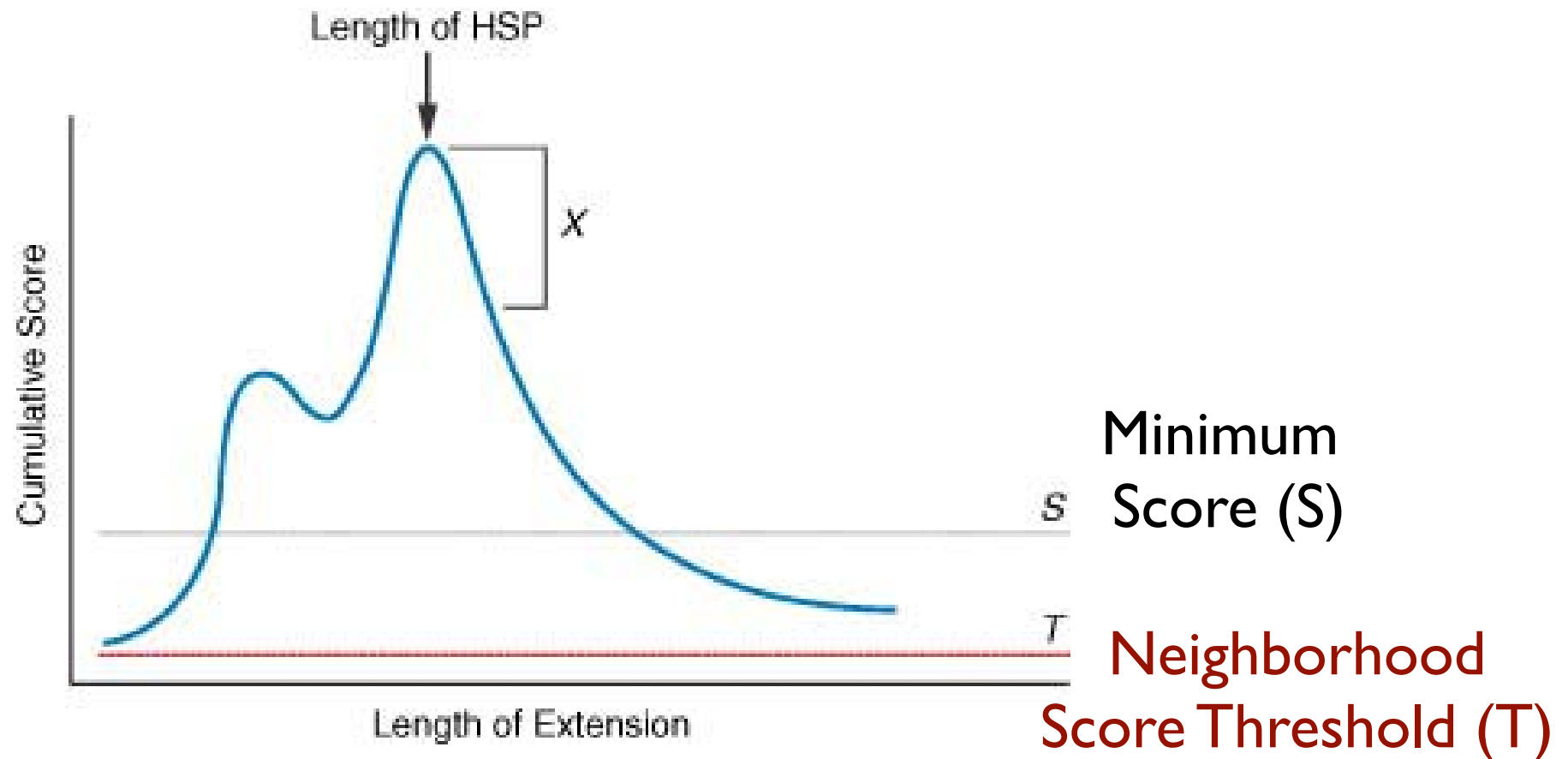
BLAST Algorithm

RDQ 16	QDQ 12	EDQ 11	RDN 11	RDB 11	BDQ 10	RDP 10
RBQ 14	REQ 12	HDQ 11	RDD 11	ADQ 10	XDQ 10	RDT 10
RDZ 14	RDR 12	ZDQ 11	RDH 11	MDQ 10	RQQ 10	RDY 10
KDQ 13	RDK 12	RNQ 11	RDM 11	SDQ 10	RSQ 10	RDX 10
RDE 13	NDQ 11	RZQ 11	RDS 11	TDQ 10	RDA 10	DDQ 9 ...

*Extension using neighborhood words
greater than neighborhood score
threshold ($T = 11$)*

Query: 1 T L S H A W R L S N E T D K R P F I E T A E R L **RDQ** H K K D Y P E Y K Y Q P R R R K N G K P G S S E A D A H S E 58
 T L W R L N + K R P F + E A E R L R + Q H K K D + P + Y K Y Q P R R R K + K G S D +
 Sbjct: 140 T L E S G W R L E N P G E K R P F V E G A E R L **REQ** H K K D H P D Y K Y Q P R R R K S V K N G Q S E P E D G S E Q 197

Extending the High Scoring Segment Pair (HSP)



> [gb|AAL08419.1](#) PTEN [Takifugu rubripes]
Length=412

Score = 197 bits (501), Expect = 2e-49, Method: Composition-based stats.
Identities = 95/100 (95%), Positives = 98/100 (98%), Gaps = 0/100 (0%)

```
Query 2 IVSRNKRRYQEDGFDLDLTYYIPNIIAMGFPAERLEGVYRNNIDDVVRFLDSKHKNHYKI 61
      +VSRNKRRYQEDGFDLDLTYYIPNIIAMGFPAERLEGVYRNNIDDVVRFLDSKHKNHYKI
Sbjct 8 MVS RNKRRYQEDGFDLDLTYYIPNIIAMGFPAERLEGVYRNNIDDVVRFLDSKHKNHYKI 67

Query 62 YNLCAERHYDTAKFNCRVAQYPPFEDHNPPQLELIKPFKQN 101
      YNLCAERHYD AKFNCRVAQYPPFEDHNPPQLELIKPF ++
Sbjct 68 YNLCAERHYDAAKFNCRVAQYPPFEDHNPPQLELIKPFCE 107
```

Score = 83.6 bits (205), Expect = 4e-15, Method: Composition-based stats.
Identities = 60/103 (58%), Positives = 68/103 (66%), Gaps = 32/103 (31%)

```
Query 99 KQNKMLKKDKMFHFWVNTFFIPGPEEV-----D 126
      KQNKMK+KKDKMFHFWVNTFFIPGPEE +
Sbjct 260 KQNKMMKKDKMFHFWVNTFFIPGPEESRDKLENGAVNNADSQQGVPA PGQGQPQSAECRE 319

Query 127 NDKEYLVLTLTkndldkankdkanRYFSPNFKVKLYFTKTVEE 169
      +D++YL+LTL+KND DKANKDKANRYFSPNFKVKL F+KTVEE
Sbjct 320 SDRDY LILTL SKNDRDKANKDKANRYFSPNFKVKLCFSKTVEE 362
```

> [gb|AAH93110.1](#) **UG** Ptenb protein [Danio rerio]
Length=289

Score = 197 bits (500), Expect = 2e-49, Method: Composition-based stats.
Identities = 95/99 (95%), Positives = 98/99 (98%), Gaps = 0/99 (0%)

```
Query 3 VSRNKRRYQEDGFDLDLTYYIPNIIAMGFPAERLEGVYRNNIDDVVRFLDSKHKNHYKIY 62
      VSRNKRRYQEDGFDLDLTYYIPNIIAMGFPAERLEGVYRNNIDDVVRFLDSKHK+HYKIY
Sbjct 9 VSRNKRRYQEDGFDLDLTYYIPNIIAMGFPAERLEGVYRNNIDDVVRFLDSKHKDHYKIY 68

Query 63 NLCAERHYDTAKFNCRVAQYPPFEDHNPPQLELIKPFKQN 101
      NLCAERHYDTAKFNCRVAQYPPFEDHNPPQLELIKPF ++
Sbjct 69 NLCAERHYDTAKFNCRVAQYPPFEDHNPPQLELIKPFCE 107
```

BLAST Algorithm

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Credits

- Materials for this presentation have been adapted from the following sources:

NCBI HelpDesk - Field Guide Course Materials

Bioinformatics: A practical guide to the analysis of genes and proteins

- Questions? Please contact:

Dr. Joanne Fox

Michael Smith Laboratories

joanne@msl.ubc.ca




BLAST

GUIDED TOUR: Advanced Tips & Tricks for Using BLAST



<http://www.ncbi.nlm.nih.gov/BLAST/>

**BLAST**
Basic Local Alignment Search Tool

HomeRecent ResultsSaved StrategiesHelp

► NCBI/ BLAST Home

BLAST finds regions of similarity between biological sequences. [more...](#)

[Learn more](#) about how to use the new BLAST design

BLAST Assembled Genomes

Choose a species genome to search, or [list all genomic BLAST databases](#).

- [Human](#)
- [Mouse](#)
- [Rat](#)
- [Arabidopsis thaliana](#)

- [Oryza sativa](#)
- [Bos taurus](#)
- [Danio rerio](#)
- [Drosophila melanogaster](#)

- [Gallus gallus](#)
- [Pan troglodytes](#)
- [Microbes](#)
- [Apis mellifera](#)

Basic BLAST

Choose a BLAST program to run.

nucleotide blast	Search a nucleotide database using a nucleotide query <i>Algorithms:</i> blastn, megablast, discontinuous megablast
protein blast	Search protein database using a protein query <i>Algorithms:</i> blastp, psi-blast, phi-blast
blastx	Search protein database using a translated nucleotide query
tblastn	Search translated nucleotide database using a protein query
tblastx	Search translated nucleotide database using a translated nucleotide query


Specialized BLAST

News

[Old BLAST Web Pages to be deleted June 11th 2007](#)

As previously announced access to the old pages will be removed on June 11, 2007.

2007-06-01 12:15:00

 [More BLAST news...](#)

find human sequences in a database that can be amplified with a particular primer pair.

A frequent use of nucleotide-nucleotide BLAST is to check the specificity of oligonucleotides for hybridization in PCR. The goal is usually to make sure that the primers will give a unique product from the target genome or cDNA population. Because BLAST is local and

New BLAST homepage

NCBI/BLAST Home

BLAST finds regions of similarity between biological sequences. [more...](#)

[Learn more](#) about how to use the new BLAST design (beta) [Old blast](#)

BLAST Assembled Genomes

Choose a species genome to search, or [list all genomic BLAST databases](#).

- [Human](#)
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Basic BLAST

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protein blast	Search protein database using a protein query <i>Algorithms:</i> blastp, psi-blast, phi-blast
blastx	Search protein database using a translated nucleotide query
tblastn	Search translated nucleotide database using a protein query
tblastx	Search translated nucleotide database using a translated nucleotide query

Specialized BLAST

Choose a type of specialized search (or database name in parentheses.)

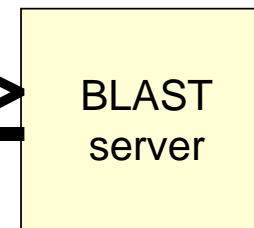
- Search [trace archives](#)
- Find [conserved domains](#) in your sequence (cds)
- Find sequences with similar [conserved domain architecture](#) (cdart)
- Search sequences that have [gene expression profiles](#) (GEO)
- Search [immunoglobulins](#) (IgBLAST)
- Search for [SNPs](#) (snp)
- Screen sequence for [vector contamination](#) (vecscreen)
- [Align two sequences](#) using BLAST (bl2seq)

```
>gi|15237380|ref|NP_197163.1| myb family transcription factor (MYB43) [Arabidopsis thaliana]
MGROPCCDKVGLKKGPMTEEDKKLINFILTNHCCWRALPKLSGLLRGKSCRLRWYLRPDLKRGLL
SEYEEQKVINLHAQLGNRWSTIASHLPGRTONEIKHWNTHIKKLRMGIDPLTHKPLSEGEASQAQG
RKXSLVPMDKPNKQDQQTQDEGEQHLQALEKNNTSVSGDGPCEDEVPLLNPHIELIDTSSSHHHNSN
DDNVAINTSKFTSPSSSSSTSSCISVWPGDEFKFFDEMEILDLNWLSSDQSLGDDISKDGKFNSTV
IDTNLWDINDLSSLDPMFNEHDDGFIQNGNGCRMVLDQDSWTFDLL
```

Submit Query

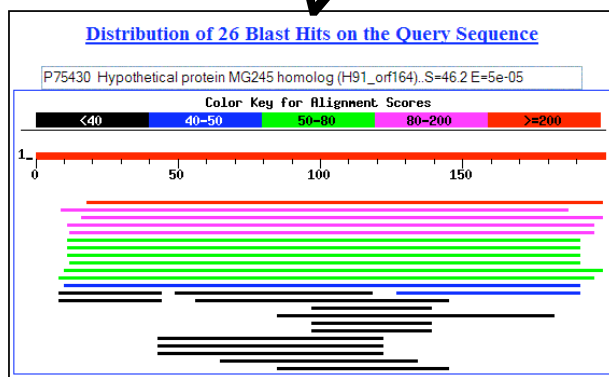


Request Results



Return Formatted Results

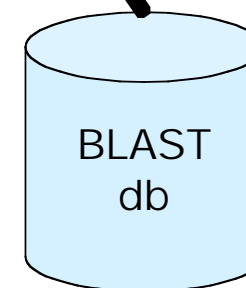
Display Results



fetch ASN.1



fetch sequence



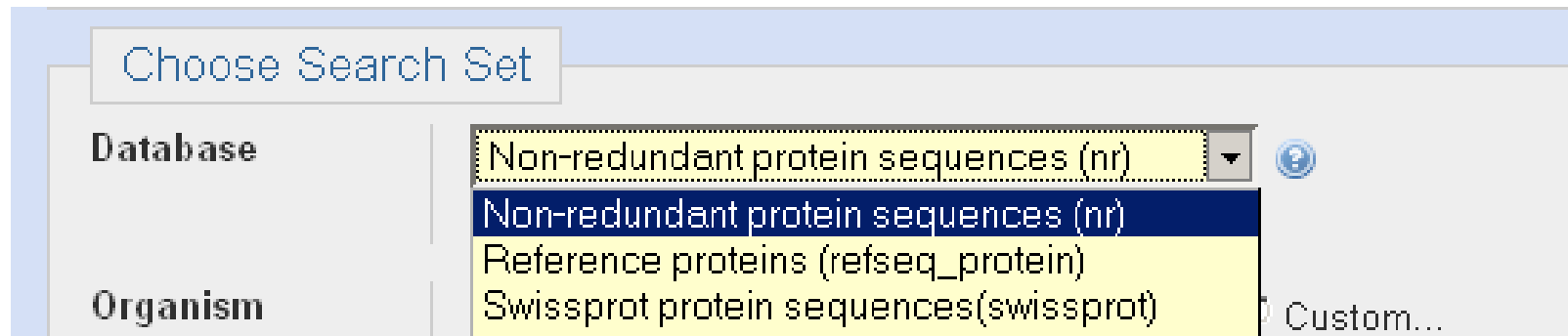
Consider your research question ...

- Are you looking for a particular gene in a particular species?
- Are you looking for additional members of a protein family across all species?
- Are you looking to annotate genes in your species of interest?

Know your reagents

- Changing your choice of database is changing your search space
- Database size affects the BLAST statistics
- Databases change rapidly and are updated frequently

Protein Databases: nr



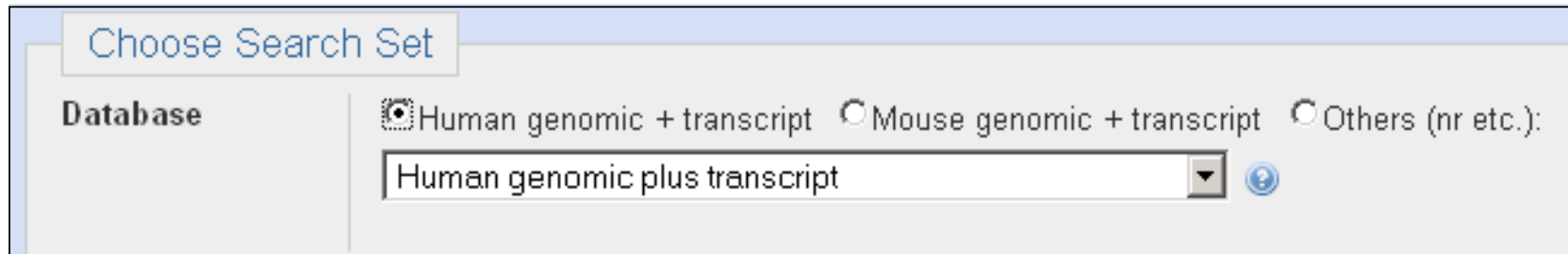
- nr (non-redundant protein sequences)
 - GenBank CDS translations
 - NP_ RefSeqs
 - Outside Protein
 - PIR, Swiss-Prot, PRF
 - PDB (sequences from structures)
- pat protein patents
- env_nr environmental samples

Services

blastp

blastx

Nucleotide Databases: Human and Mouse



Choose Search Set

Database

☒ Human genomic + transcript ☐ Mouse genomic + transcript ☐ Others (nr etc.):

Human genomic plus transcript

- Human and mouse genomic + transcript default
- Separate sections in output for mRNA and genomic
- Direct links to Map Viewer for genomic sequences

Megablast, blastn service

Nucleotide Databases: Traditional

Choose Search Set

Database	Nucleotide collection (nr/nt)
Organism Optional	
Entrez Query Optional	

BLAST

- Nucleotide collection (nr/nt)
- Reference mRNA sequences (refseq_mna)
- Reference genomic sequences (refseq_genomic)
- Expressed sequence tags (est)
- Non-human, non-mouse ESTs (est_others)
- Genomic survey sequences (gss)
- High throughput genomic sequences (HTGS)
- Patent sequences(pat)
- Protein Data Bank (pdb)
- Human ALU repeat elements (alu_repeats)
- Sequence tagged sites (dbsts)
- Whole-genome shotgun reads (wgs)
- Environmental samples (env_nt)

Services

blastn
tblastn
tblastx

Nucleotide Databases: Traditional

- **nr (nt)**
 - Traditional GenBank
 - NM_ and XM_ RefSeqs
 - [refseq_rna](#)
- **refseq_genomic**
 - NC_ RefSeqs
- **dbest**
 - EST Division
 - [est_human](#), [mouse](#), [others](#)
- **htgs**
 - HTG division
- **gss**
 - GSS division
- **wgs**
 - whole genome shotgun
- **env_nt**
 - environmental samples

Databases are mostly non-overlapping

<http://www.ncbi.nlm.nih.gov/BLAST/>

The screenshot shows the NCBI BLAST website interface. At the top, there is a navigation bar with links: Home, Recent Results, Saved States, and Help. The Help link is highlighted with a blue circle. To the right of the navigation bar, there is a 'My NCBI' section with a welcome message for 'joannealisonfox' and a 'Sign Out' link. Below the navigation bar, the main content area is titled 'NCBI/BLAST/Help' and contains a search box for 'Browse BLAST documentation.' The content is organized into several sections: 'Getting Started' with links to 'BLAST short course' and 'BLAST program selection guide'; 'About BLAST' with links to 'Frequently Asked Questions', 'NCBI Handbook: BLAST', 'The Statistics of Sequence Similarity Scores', 'NAR 2004 Web server issue', 'NAR 2006 Web server issue', 'BLAST glossary', and 'References'; 'Getting Help' with links to 'Email blast-help' and 'Mailing list'; and 'BLAST information' with links to 'Download BLAST Software and Databases' and 'Developer information'. A large white arrow with the text 'Program Selection Guide' points to the 'BLAST program selection guide' link. At the bottom, there is a 'BLAST News' section with a link to 'BLAST News directory'.

BLAST Alignment Search Tool

My NCBI
Welcome joannealisonfox. [Sign Out]

Home Recent Results Saved States **Help**

► NCBI/BLAST/Help

Browse BLAST documentation.

Getting Started

- BLAST short course
- BLAST program selection guide**

About BLAST

- Frequently Asked Questions
- NCBI Handbook: BLAST
- The Statistics of Sequence Similarity Scores
- NAR 2004 Web server issue
- NAR 2006 Web server issue
- BLAST glossary
- References

Getting Help

- Email blast-help
- Mailing list

BLAST information

- Download BLAST Software and Databases
- Developer information

BLAST News

BLAST News directory

Program Selection Guide

If your sequence is NUCLEOTIDE



	Database	Purpose	BLAST Program	
20 bp or longer	Nucleotide	Identify the query sequence	MEGABLAST (accept batch queries) Standard BLAST (blastn)	Learn More Learn More
		Find sequences similar to query sequence	Standard BLAST (blastn)	Learn More
		Find similar proteins to translated query in a translated database	Translated BLAST (tblastx)	Learn More
	Protein	Find similar proteins to translated query in a protein database	Translated BLAST (blastx)	Learn More
7 - 20 bp	Nucleotide	Find primer binding sites or map short contiguous motifs	Search for short, nearly exact matches	Learn More

If your sequence is PROTEIN



	Database	Purpose	BLAST program	
15 residues or longer	Protein	Identify the query sequence or find protein sequences similar to query	Standard Protein BLAST (blastp)	Learn More
		Find members of a protein family or build a custom position-specific score matrix	PSI-BLAST	Learn More
		Find proteins similar to the query around a given pattern	PHI-BLAST	Learn More
	Conserved Domains	Find conserved domains in the query	CD-search (RPS-BLAST)	Learn More
	Conserved Domains	Find conserved domains in the query and identify other proteins with similar domain architectures	Conserved Domain Architecture Retrieval Tool (CDART)	Learn More
5-15 residues	Nucleotide	Find similar proteins in a translated nucleotide database	Translated BLAST (tblastn)	Learn More
	Protein	Search for peptide motifs	Search for short, nearly exact matches	Learn More

Specialized Database Searches



Query	Database	Purpose	BLAST Program	
Nucleotide or Protein	None	Compare the query and second sequence directly	BLAST 2 Sequences	Learn more
	The NCBI Draft Human Genome	Map the query sequence. Determine the genomic structure. Identify novel genes.	Human Genome BLAST	Learn More
	Mouse Genome	Map the query sequence. Determine the genomic structure. Identify novel genes.	Mouse Genome BLAST	Learn More
	Rat	Map the query sequence. Determine the genomic structure. Identify novel genes.	Rat Genome BLAST page	Learn More
	Fugu (Pufferfish)	Map the query sequence. Determine the genomic structure. Identify novel genes.	Fugu rubripes Genome BLAST page	Learn More
	Zebrafish	Map the query sequence. Determine the genomic structure. Identify novel genes.	Zebrafish Genome BLAST page	Learn More
	Arabidopsis thaliana	Map the query sequence. Determine the genomic structure.	Arabidopsis thaliana BLAST page	Learn More



► NCBI/ BLAST Home

BLAST finds regions of similarity between biological sequences. [more...](#)

[Learn more](#) about how to use the new BLAST design

BLAST Assembled Genomes

Choose a species genome to search, or [list all genomic BLAST databases](#).

- | | | |
|--|---|-----------------------------------|
| ▣ Human | ▣ Oryza sativa | ▣ Gallus gallus |
| ▣ Mouse | ▣ Bos taurus | ▣ Pan troglodytes |
| ▣ Rat | ▣ Danio rerio | ▣ Microbes |
| ▣ Arabidopsis thaliana | ▣ Drosophila melanogaster | ▣ Apis mellifera |

Basic BLAST

Choose a BLAST program to run.

[nucleotide blast](#)

Search a **nucleotide** database using a **nucleotide** query
Algorithms: blastn, megablast, discontinuous megablast

[protein blast](#)

Search **protein** database using a **protein** query
Algorithms: blastp, psi-blast, phi-blast

[blastx](#)

Search **protein** database using a **translated nucleotide** query

[tblastn](#)

Search **translated nucleotide** database using a **protein** query

[tblastx](#)

Search **translated nucleotide** database using a **translated nucleotide** query

News

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2007-06-01 12:15:00

[More BLAST news...](#)

Tip of the Day

How to use BLAST to find human sequences in a database that can be amplified with a particular primer pair.

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Enter Query Sequence

Enter accession number, gi, or FASTA sequence ?

[Clear](#)

Query subrange ?

231571

231571

From

To

Or, upload file

[Browse...](#)

Job Title

Q02067:Achaete-scute homolog 1 (Mash-1)

Enter a descriptive title for your BLAST search ?

Choose Search Set

Database

Swissprot protein sequences(swissprot) ?

Organism
Optional

Enter organism name or id--completions will be suggested

Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown. ?

Entrez Query
Optional

Enter an Entrez query to limit search ?

Program Selection

Algorithm

- ☒ blastp (protein-protein BLAST)
- ☐ PSI-BLAST (Position-Specific Iterated BLAST)
- ☐ PHI-BLAST (Pattern Hit Initiated BLAST)

Choose a BLAST algorithm ?

BLAST

Search database **swissprot** using **Blastp (protein-protein BLAST)**



☐ Show results in a new window

▼ [Algorithm parameters](#)

Note: Parameter values that differ from the default are highlighted in yellow

Context Specific Help

Choose Search Set

Database	Swissprot protein sequences(swissprot) 
	Select the sequence database to run searches against. No BLAST database contains all the sequences at NCBI. BLAST databases are organized by informational content (nr, RefSeq, etc.) or by sequencing technique (WGS, EST, etc.). more...
Organism Optional	<input type="text" value="Enter organism name or id--completions will be suggested"/> Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown. 
	Select from the list or choose "Custom" to enter the name of an organism. The search will be restricted to the sequences in the database which are from the organism selected.
Entrez Query Optional	<input type="text"/> Enter an Entrez query to limit search 
	You can use Entrez query syntax to search a subset of the selected BLAST database. This can be helpful to limit searches to molecule types, sequence lengths or to exclude organisms. more...

Limiting Database: Organism

Organism
Optional

☐ Any ☐ Human ☐ *A.thaliana* ☐ Mouse ☒ Custom...

Search

bacter

- CFB group **bacter**ia (taxid:976)
- GNS **bacter**ia (taxid:200795)
- green sulfur **bacter**ia (taxid:1090)
- Bacter**ia (taxid:2)
- purple **bacter**ia and relatives (taxid:1224)
- purple non-sulfur **bacter**ia (taxid:1224)
- purple photosynthetic **bacter**ia (taxid:1224)
- purple photosynthetic **bacter**ia and relatives (taxid:1224)
- purple **bacter**ia (taxid:1224)
- low G+C Gram-positive **bacter**ia (taxid:1239)

taxa will be shown.

Organism autocomplete

Limiting Database: Entrez Query

Entrez Query
Optional

Enter an Entrez query to limit search ?

all[filter] NOT mammals[organism]

gene_in_mitochondrion[Properties]
2006:2007 [Modification Date]

Nucleotide
biomol_mrna[Properties]
biomol_genomic[Properties]

BLAST

Search database **swissprot** using **Blastp (protein-protein BLAST)**

☐ Show results in a new window

▼ Algorithm parameters

Note: Parameter values that differ from the default

General Parameters

Max target
sequences

100 ▼

Select the maximum number of aligned sequences to display ⓘ

Short queries

☒ Automatically adjust parameters for short input sequences ⓘ

Expect threshold

10 ⓘ

Word size

3 ▼ ⓘ

Scoring Parameters

Matrix

BLOSUM62 ▼ ⓘ

Gap Costs

Existence: 11 Extension: 1 ▼ ⓘ

Algorithm parameters: Protein

The image shows a web interface for protein algorithm parameters, divided into three sections: General Parameters, Scoring Parameters, and Filters and Masking. Annotations highlight specific settings and their effects.

General Parameters

- Max target sequences:** A dropdown menu is open, showing options: 100, 10, 50, 100 (highlighted), 250, 500, 1000, 5000, 10000, 20000. An annotation "Expand" points to the dropdown arrow. Another annotation "May limit results" points to the value 100.
- Short queries:** A checkbox labeled "Automatically adjust parameters for short input" is checked.
- Expect threshold:** A text input field containing the value 10. An annotation "Adjust to set stringency" points to this field.
- Word size:** A dropdown menu showing the value 3.

Scoring Parameters

- Matrix:** A dropdown menu showing "BLOSUM62".
- Gap Costs:** A text input field showing "Existence: 11 Extension: 1".
- Compositional adjustments:** A dropdown menu showing "Composition-based statistics". An annotation "Default statistics adjustment for compositional bias" points to this field.

Filters and Masking

- Filter:** A checkbox labeled "Low complexity regions" is unchecked. An annotation "Off now by default. Conflicts with comp-based stats" points to this checkbox.
- Mask:** Two checkboxes are present: "Mask for lookup table only" (unchecked) and "Mask lower case letters" (unchecked).

Automatic Short Sequence Adjustment

Job Title: Elvis Lives!

No putative conserved domains have been detected

Your search parameters were adjusted to search for a short input sequence.

WAITING

Request ID 1WSB0FX012

Status Searching

Subr

Curre

Time

This p

e-value	20000
Word Size	2
Matrix	PAM30
Comp Stats	Off
Low Comp Filter	Off

>[ref|ZP_01712014.1|](#) conserved hypothetical protein [Pseudomonas putida] Length=245


Score = 18.5 bits (36), Expect = 15305
Identities = 5/5 (100%), Positives = 5/5 (100%), Gaps = 0/5 (0%)

Query 1 ELVIS 5
ELVIS
Sbjct 126 ELVIS 130

>[ref|ZP_01712512.1|](#) Substrate-binding region of ABC-type glycine betaine system [Pseudomonas putida GB-1] Length=342

Score = 18.5 bits (36), Expect = 15305
Identities = 5/5 (100%), Positives = 5/5 (100%), Gaps = 0/5 (0%)

Query 1 ELVIS 5
ELVIS
Sbjct 172 ELVIS 176

>[ref|XP_001366374.1|](#)  PREDICTED: similar to R7 binding protein [Mycobacterium tuberculosis] Length=257

Score = 18.5 bits (36), Expect = 15305
Identities = 5/5 (100%), Positives = 5/5 (100%), Gaps = 0/5 (0%)

Query 1 ELVIS 5
ELVIS
Sbjct 69 ELVIS 73

>[ref|ZP_01711731.1|](#) GCN5-related N-acetyltransferase [Caldivirga maritima] Length=166

Score = 18.5 bits (36), Expect = 15305
Identities = 5/5 (100%), Positives = 5/5 (100%), Gaps = 0/5 (0%)

Query 1 ELVIS 5
ELVIS
Sbjct 20 ELVIS 24

Enter Query Sequence

Enter accession number, gi, or FASTA sequence ?

Clear

```
>gi|231571|sp|Q02067|ASCL1_MOUSE Achaete-scute homolog 1  
(Mash-1)  
MESSGKMEAGAGQQPQQPFLPPAACFFATAAAAAAAAAAQAQQQQPQAPPQQAPQLS  
CGGHKSAAKQDKRQRSSSPPELMRCKRRLNFGSGFGYSLPQQQPAAVARRNERERNRVKLVNLG  
PNGAANKKMSKVETLRSAVQYIPALQQLLEHDAVSAAFQAGVLSPTISPNYSNDLNSMAGS
```

Query subrange ?

From

To

Or, upload file

Browse...

Job Title

MASH1 BLAST for CBW

Enter a descriptive title for your BLAST search ?

Choose Search Set

Database

Swissprot protein sequences(swissprot) ?

Organism
Optional

Enter organism name or id--completions will be suggested

Enter organism common name, binomial, or tax id. Only 20 top taxa will be shown. ?

Entrez Query
Optional

Enter an Entrez query to limit search ?

Program Selection

Algorithm

- ☒ blastp (protein-protein BLAST)
☐ PSI-BLAST (Position-Specific Iterated BLAST)
☐ PHI-BLAST (Pattern Hit Initiated BLAST)

Choose a BLAST algorithm ?

BLAST

Search database **swissprot** using **Blastp (protein-protein BLAST)**

☐ Show results in a new window

**BLAST***Basic Local Alignment Search Tool*[Home](#)[Recent Results](#)[Saved Strategies](#)[Help](#)**My NCBI**Welcome joannealisonfox. [\[Sign Out\]](#)[► NCBI/ BLAST/ blastp/ Formatting Results - 75AMH5J9015](#)[\[Formatting options\]](#)**Job Title: gij231571 (231 letters)**

Putative conserved domains have been detected, click on the image below for detailed results.



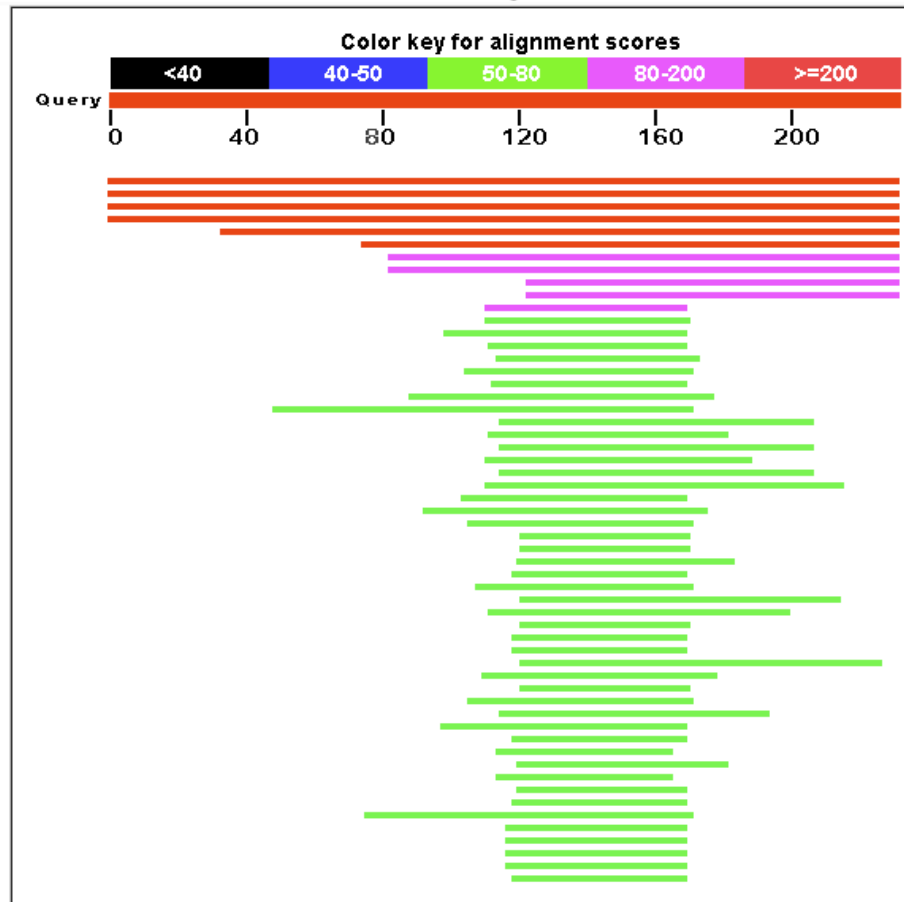
Request ID	75AMH5J9015
Status	Searching
Submitted at	Fri Jun 15 11:04:49 2007
Current time	Fri Jun 15 11:04:54 2007
Time since submission	00:00:04

This page will be automatically updated in 3 seconds

A graphical view

Distribution of 100 Blast Hits on the Query Sequence

Mouse-over to show define and scores, click to show alignments



The BLAST hit list

[Distance tree of results](#) **NEW**

Sequences producing significant alignments:

		Score (Bits)	E Value	
sp P50553 ASCL1 HUMAN	Achaete-scute homolog 1 (HASH1)	269	9e-73	G
sp Q99929 ASCL2 HUMAN	Achaete-scute homolog 2 (Mash2) (HASH2)	100	4e-22	G
sp Q9NQ33 ASCL3 HUMAN	Achaete-scute homolog 3 (bHLH transcriptio	47.4	6e-06	G
sp P61296 HAND2 HUMAN	Heart- and neural crest derivatives-exp...	46.6	1e-05	G
sp Q02575 HEN1 HUMAN	Helix-loop-helix protein 1 (HEN1) (Nesci...	40.0	9e-04	G
sp Q96004 HAND1 HUMAN	Heart- and neural crest derivatives-exp...	39.3	0.001	G
sp Q16559 TAL2 HUMAN	T-cell acute lymphocytic leukemia-2 protein	38.9	0.002	G
sp Q02577 HEN2 HUMAN	Helix-loop-helix protein 2 (HEN2) (Nesci...	38.1	0.004	G
sp P17542 TAL1 HUMAN	T-cell acute lymphocytic leukemia-1 prot...	36.6	0.010	G
sp P47928 ID4 HUMAN	DNA-binding protein inhibitor ID-4 (Inhibito	36.6	0.011	G
sp Q7RTU7 SCX HUMAN	Basic helix-loop-helix transcription factor	36.2	0.013	G
sp P15172 MYOD1 HUMAN	Myoblast determination protein 1 (Myogenic	36.2	0.015	G
sp Q7RTS3 PTF1A HUMAN	Pancreas transcription factor 1 subunit...	36.2	0.015	G
sp P41134 ID1 HUMAN	DNA-binding protein inhibitor ID-1 (Inhibito	35.8	0.018	G
sp P13349 MYF5 HUMAN	Myogenic factor 5 (Myf-5)	35.4	0.024	G
sp P04198 MYCN HUMAN	N-myc proto-oncogene protein	35.4	0.025	G
sp Q02363 ID2 HUMAN	DNA-binding protein inhibitor ID-2 (Inhibito	35.4	0.025	G
sp P12980 LYL1 HUMAN	Protein lyl-1 (Lymphoblastic leukemia-deriv	35.4	0.025	G
sp Q12870 TCF15 HUMAN	Transcription factor 15 (bHLH-EC2 protein)	34.7	0.037	G
sp P23409 MYF6 HUMAN	Myogenic factor 6 (Myf-6)	33.9	0.072	G
sp Q7RTS1 BHLH8 HUMAN	Class B basic helix-loop-helix protein ...	33.5	0.082	G
sp Q15784 NDF2 HUMAN	Neurogenic differentiation factor 2 (Neu...	33.5	0.088	G
sp Q60HK4 FIGLA HUMAN	Factor in the germline 57kDa (Transcrip...	33.5	0.094	G

BLAST Alignments

```
>sp|P20389|MYC2\_MARMO N-myc 2 proto-oncogene protein  
Length=454
```

```
Score = 35.8 bits (81), Expect = 0.14, Method: Composition-based stats.  
Identities = 22/52 (42%), Positives = 30/52 (57%), Gaps = 4/52 (7%)
```

```
Query 133 FATLREHVPNGAANKKMSKVETLRSVQYIRALQ----QLLDEHDAVSAAFQ 180  
          F TLR+HVP      N+K +KV  L+ A +Y+  LQ      QLL E + + A  Q  
Sbjct 391 FTTLRDHVPPELVKNEKAAKVVLKKACEYVHYLQAKEHQLLMEKEKLQARQQ 442
```

Identical match

positive score
(conservative)

gap


Negative or zero

BLAST Alignments

>[sp|P04198|MYCN HUMAN](#)  N-myc proto-oncogene protein
Length=464


Score = 35.4 bits (80), Expect = 0.025, Method: Composition-based stats.
Identities = 22/52 (42%), Positives = 31/52 (59%), Gaps = 4/52 (7%)

```
Query 133 FATLREHVPNGAANKKMSKVETLRSAVQYIRALQ---QLLDEHDAVSAAFQ 180
          F TLR+HVP      N+K +KV  L+ A +Y+ +LQ      QLL E + + A  Q
Sbjct 401 FLTLRDHVPPELVKNEKAAKVVLKKATEYVHSLQAEHQLLLEKEKLQARQQ 452
```

>[sp|Q02363|ID2 HUMAN](#)  DNA-binding protein inhibitor ID-2 (Inhibitor of DNA binding 2)
Length=134

Score = 35.4 bits (80), Expect = 0.025, Method: Composition-based stats.
Identities = 19/47 (40%), Positives = 29/47 (61%), Gaps = 0/47 (0%)

```
Query 129 VNLGFATLREHVPNGAANKKMSKVETLRSAVQYIRALQQLLDEHDAV 175
          +N ++ L+E VP+   NKK+SK+E L+ + YI  LQ  LD H  +
Sbjct 39  MNDCYSKLELVPSIPQNKKVSKMEILQHVIDYILDQLALDSHPTI 85
```

>[sp|P12980|LYL1 HUMAN](#)  Protein lyl-1 (Lymphoblastic leukemia-derived sequence 1)
Length=267

Score = 35.4 bits (80), Expect = 0.025, Method: Composition-based stats.
Identities = 22/50 (44%), Positives = 31/50 (62%), Gaps = 0/50 (0%)

```
Query 129 VNLGFATLREHVPNGAANKKMSKVETLRSAVQYIRALQQLLDEHDAVSAA 178
          VN  FA LR+ +P      ++K+SK E LR A++YI  L +LL +  A  AA
Sbjct 153 VNGAFaelrKLLPThPPDRKLSKNEVLRlAMKYIGFLVRLLRDQAAALAA 202
```

- **Similarity**

The extent to which nucleotide or protein sequences are related. The extent of similarity between two sequences can be based on percent sequence identity and/or conservation. In BLAST similarity refers to a positive matrix score.

- **Identity**

The extent to which two (nucleotide or amino acid) sequences are invariant.

- **Homology**

Similarity attributed to descent from a common ancestor.

It is your responsibility as an informed bioinformatician to use these terms correctly: A sequence is either homologous or not. Don't use % with this term!

BLAST statistics to record in your bioinformatics labbook

It can be helpful to record the statistics that are found at bottom of your BLAST results

```
Database: Non-redundant SwissProt sequences
Posted date: Jun 14, 2007 5:55 PM
Number of letters in database: 9,119,588
Number of sequences in database: 16,602
Lambda      K      H
0.309      0.125    0.352
Gapped
Lambda      K      H
0.267      0.0410   0.140
Matrix: BLOSUM62
Gap Penalties: Existence: 11, Extension: 1
Number of Sequences: 16602
Number of Hits to DB: 1063550
Number of extensions: 39000
Number of successful extensions: 121
Number of sequences better than 10: 19
Number of HSP's better than 10 without gapping: 0
Number of HSP's gapped: 121
Number of HSP's successfully gapped: 19
Length of query: 231
Length of database: 9119588
Length adjustment: 94
Effective length of query: 137
Effective length of database: 7559000
Effective search space: 1035583000
Effective search space used: 1035583000
T: 11
A: 40
X1: 16 (7.1 bits)
X2: 38 (14.6 bits)
X3: 64 (24.7 bits)
S1: 42 (20.8 bits)
S2: 58 (26.9 bits)
```

```
25: 28 (58.8 bits)
27: 45 (50.8 bits)
X3: 64 (24.7 bits)
X5: 38 (14.6 bits)
X7: 18 (1.7 bits)
```

Sorting BLAST by Taxonomy

BLAST Basic Local Alignment Search Tool My NCBI 2
Home Recent Results Saved Strategies Help
Welcome joannealisonfox. [Sign Out]

► NCBI/BLAST/blastp/Formatting Results - 75AMH5J9015 [Reformat these Results] [Edit and Resubmit] [Save Search Strategy]

Job Title: gj|231571 (231 letters) ► Show Conserved Domains

▢ Your search is limited to records matching entrez query: txid9606 [ORGN].

BLASTP 2.2.16 (Mar-25-2007)

Reference:
Altschul, Stephen F., Thomas L. Madden, Alejandro A. Schäffer, Jinghui Zhang, Zheng Zhang, Webb Miller, and David J. Lipman (1997), "Gapped BLAST and PSI-BLAST: a new generation of protein database search programs", Nucleic Acids Res. 25:3389-3402.

Reference:
Schäffer, Alejandro A., L. Aravind, Thomas L. Madden, Sergei Shavirin, John L. Spouge, Yuri I. Wolf, Eugene V. Koonin, and Stephen F. Altschul (2001), "Improving the accuracy of PSI-BLAST protein database searches with composition-based statistics and other refinements", Nucleic Acids Res. 29:2994-3005.

RID: 75AMH5J9015

Database: Non-redundant SwissProt sequences
245,584 sequences; 92,640,715 total letters

If you have any problems or questions with the results of this search please refer to the [BLAST FAQs](#)
[Taxonomy reports](#)

Query= gi|231571|sp|Q02067|ASCL1_MOUSE Achaete-scute homolog 1 (Mash-1)
Length=231



BLAST

Basic Local Alignment Search Tool

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Job Title: gi|231571 (231 letters)

[Show Conserved Domains](#)

Tax BLAST Report

Index

- [Lineage Report](#)
- [Organism Report](#)
- [Taxonomy Report](#)
- [Help](#)


Lineage Report

Bilateria	[animals]				
Coelomata	[animals]				
Euteleostomi	[vertebrates]				
Tetrapoda	[vertebrates]				
Amniota	[vertebrates]				
Eutheria	[placentals]				
Euarchontoglires	[placentals]				
Glires	[placentals]				
Muroidea	[rodents]				
Murinae	[rodents]				
Mus musculus	(mouse)	466	22 hits	[rodents]	Achaete-scute homolog 1 (Mash-1)
Rattus norvegicus	(brown rat)	347	10 hits	[rodents]	Achaete-scute homolog 1
Mesocricetus auratus	(Syrian hamster)	50	2 hits	[rodents]	Neurogenic differentiation factor 1 (NeuroD1)
Oryctolagus cuniculus	(domestic rabbit)	49	1 hit	[rabbits & hares]	Heart- and neural crest derivatives-expressed
Homo sapiens	(man)	332	25 hits	[primates]	Achaete-scute homolog 1 (HASH1)
Macaca fascicularis	(cynomolgus monkey)	48	1 hit	[primates]	Neurogenic differentiation factor 6 (NeuroD6)
Bos taurus	(cow)	135	4 hits	[even-toed ungulates]	Achaete-scute homolog 2 (Mash2)
Ovis aries	(domestic sheep)	50	1 hit	[even-toed ungulates]	Heart- and neural crest derivatives-expressed
Gallus gallus	(bantam)	60	8 hits	[birds]	Heart- and neural crest derivatives-expressed
Coturnix japonica		50	1 hit	[birds]	Myogenic factor 5 (Myf-5) (Myogenic factor 3)
Xenopus laevis	(common platanna)	289	10 hits	[frogs & toads]	Achaete-scute homolog 1
Notophthalmus viridescens	(red-spotted newt)	49	1 hit	[salamanders]	Myogenic factor 5 (Myf-5)
Danio rerio	(leopard danio)	298	8 hits	[bony fishes]	Achaete-scute homolog 1a (Zash-1a) (Pituitary)
Drosophila melanogaster		74	5 hits	[flies]	Achaete-scute complex protein T5 (Achaete)
Caenorhabditis elegans	(nematode)	64	4 hits	[nematodes]	Helix-loop-helix protein 6

Organism Report

Mus musculus	(mouse)	[rodents]	taxid 10090		
sp Q02067 ASCL1_MOUSE	Achaete-scute homolog 1 (Mash-1)	466	4e-131		
sp Q35885 ASCL2_MOUSE	Achaete-scute homolog 2 (Mash-2)	103	9e-22		
sp Q9JJR7 ASCL3_MOUSE	Achaete-scute homolog 3 (bHLH transc...	75	2e-13		
sp Q61039 HAND2_MOUSE	Heart- and neural crest derivatives...	6360	7e-09		
sp P27792 LYL1_MOUSE	Protein lyl-1 (Lymphoblastic leukemia...	53	8e-07		

Nucleotide BLAST

**BLAST**
Basic Local Alignment Search Tool

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NCBI/ BLAST Home
BLAST finds regions of similarity between biological sequences. [more...](#)
[Learn more](#) about how to use the new BLAST design**BLAST Assembled Genomes**
Choose a species genome to search, or [list all genomic BLAST databases](#).

- [Human](#)
- [Mouse](#)
- [Rat](#)
- [Arabidopsis thaliana](#)
- [Oryza sativa](#)
- [Bos taurus](#)
- [Danio rerio](#)
- [Drosophila melanogaster](#)
- [Gallus gallus](#)
- [Pan troglodytes](#)
- [Microbes](#)
- [Apis mellifera](#)

Basic BLAST
Choose a BLAST program to run.

nucleotide blast	Search a nucleotide database using a nucleotide query <i>Algorithms:</i> blastn, megablast, discontinuous megablast
protein blast	Search protein database using a protein query <i>Algorithms:</i> blastp, psi-blast, phi-blast
blastx	Search protein database using a translated nucleotide query
tblastn	Search translated nucleotide database using a protein query
tblastx	Search translated nucleotide database using a translated nucleotide query

News
[New Human and Mouse pre-indexed databases](#)
Human and mouse genomic + transcript megablast searches now use a faster, indexed algorithm that typically reduces run time by two thirds, as compared with standard megablast.
2007-09-04 10:55:00
[More BLAST news...](#)**Tip of the Day**
Using Genomic BLAST
Genomic BLAST pages are helpful because they allow the genomic context of a BLAST search to be displayed in the Map Viewer. For example, discontinuous (cross-species) MegaBLAST against the human RefSeq transcript for albumin (NM_000477) can be used to identify the homolog in the rat genome.
[More tips...](#)

Algorithm parameters: Nucleotide

Algorithm parameters blastn

General Parameters

Max target sequences:
Select the maximum number of aligned sequences to display

Word size

Scoring Parameters

Match/Mismatch Scores

Gap Costs

Filters and Masking

Filter

Mask

Low complexity regions

Species-specific repeats for:

☒ Mask for lookup table only

☐ Mask lower case letters

- Prevents starting alignment in masked region
- Allows extensions through masked regions

Masks LC sequence (simple repeats)

- Masks species-specific interspersed repeats
- Essential for genomic query sequences

Human

Human

Rodents

Arabidopsis

Rice

Mammals

Fungi

C. elegans

A. gambiae

Zebrafish

Fruit fly

nt BLAST: New Output

► [NCBI/BLAST/blastn suite](#): BLASTN programs search nucleotide databases using a nucleotide query. [more...](#)

[Reset page](#)

[Bookmark](#)

Enter Query Sequence

Enter accession number, gi, or FASTA

ABI68636

[Clear](#)

Query subrange ⓘ

From

To

```
>Crab eating macaque CDC20 mRNA
AGCGGAGAGTTTAAAGAGGCGTAAGCGAGGCGTGTTAAACCCGGTCGGAAGTGCAACTTGCTC
ACGGGCTCCGCAGGCACCAACTGCAAGGACCCCTCCCGCTGCGGGCGTTCCCATGGCACAAT
GAGAGTGACCTGCACTCGCTGCTTCAGCTGGATGCACCCATCCCCAATGCACCCCTGCGCG
GCAAAGCCAAGGAAGCCTCAGGCCCGCCCCCTCACCCATGCGGGCCGCCAACCGATCCCAC
```

Or, upload file

Browse...

Job Title

Crab eating macaque CDC20 mRNA

Enter a descriptive title for your BLAST search ⓘ

Choose Search Set

Database

☒ Human genomic + transcript ☐ Mouse genomic + transcript ☐ Others (nr etc.):

Human genomic plus transcript ⓘ

Entrez Query
Optional

Enter an Entrez query to limit search ⓘ

Sortable Results

Legend for links to other resources: [U](#) UniGene [E](#) GEO [G](#) Gene [S](#) Structure

Sequences producing significant alignments:
(Click headers to sort columns)

Accession	Description	Max score	Tot score	Query coverage	E value	Max ident	Links
Transcripts							
NM_001255.1	Homo sapiens CDC20 cell division cycle 20 ho	2876	2876	95%	0.0	97%	U E G M
Genomic sequences [show first]							
NT_023935.17	Homo sapiens chromosome 9 genomic contig	2629	2629	94%	0.0	95%	
NW_924484.1	Homo sapiens chromosome 9 genomic contig	2601	2601	94%	0.0	95%	
NT_032977.8	Homo sapiens chromosome 1 genomic contig	428	3002	95%	9e-117	100%	
NW_921351.1	Homo sapiens chromosome 1 genomic contig	428	3010	95%	9e-117	100%	

Separate Sections for Transcript and Genome

Pseudogene on Chromosome 9

Functional Gene on Chromosome I

Total Score: All Segments

Legend for links to other resources: [U](#) UniGene [E](#) GEO [G](#) Gene [S](#) Structure [M](#) Map Viewer

Sequences producing significant alignments:

(Click headers to sort columns)

Accession	Description	Max score	Tot score	Query coverage	E value	Max ident	Links
Transcripts							
NM_001255.1	Homo sapiens CDC20 cell division cycle 20 hc	2876	2876	95%	0.0	97%	U E G M
Genomic sequences [show first]							
NW_921351.1	Homo sapiens chromosome 1 genomic contig	428	3010	95%	9e-117	100%	
NT_032977.8	Homo sapiens chromosome 1 genomic contig	428	3002	95%	9e-117	100%	
NT_023935.17	Homo sapiens chromosome 9 genomic contig	2629	2629	94%	0.0	95%	
NW_924484.1	Homo sapiens chromosome 9 genomic contig	2601	2601	94%	0.0	95%	

Functional Gene
Now First

Sorting in Exon Order

> ☐ [ref|NT_032977.8|Hs1_33153](#) ☒ Homo sapiens chromosome 1 genomic contig, reference assembly
Length=73835825

Sort alignments for this subject sequence by:

E value Score Percent identity
Query start position Subject start position

Features flanking this part of subject sequence:
Features in [6169 bp at 5' side: myeloproliferative leukemia virus oncogene](#)
[cell division cycle 20](#)
[223 bp at 3' side: cell division cycle 20](#)

Score = 42.0 bits (100), Expect = 1e-14
Identities = 51/53 (96%), Gaps = 0/53 (0%)
Strand=Plus/Plus

Query	965	Query	1	AGCGGAGAGTTTAAGAGGCGTAAGCGAGGCGTGTTAAACCCGGTCGGAAGTGC	53
Sbjct	13796530	Sbjct	13796530	AGCGGAGAGTTTAAGAGGCGTAAGCGAGGCGTGTTAAACCCGGTCGGAAGTGC	13796582

Query 1025
Sbjct 13796582
Features in this part of subject sequence:
[cell division cycle 20](#)

Score = 412 bits (208), Expect = 5e-112
Identities = 226/232 (97%), Gaps = 0/232 (0%)
Strand=Plus/Plus

Default
Long

Query	73	GGGCTCCGCAGGCACCAACTGCAAGGACCCCTCCCGCTGCGGGCGTTCCCATGGCACAAT	132
Sbjct	13796755	GGGCTCCGTAGGCACCAACTGCAAGGACCCCTCCCGCTGCGGGCGTTCCCATGGCACAAT	13796814
Query	133	TCGCGTTCGAGAGTGACCTGCACTCGCTGCTTCAGCTGGATGCACCCATCCCCAATGCAC	192
Sbjct	13796815	TCGCGTTCGAGAGTGACCTGCACTCGCTGCTTCAGCTGGATGCACCCATCCCCAATGCAC	13796874

Query start
position
Exon order

Genome View

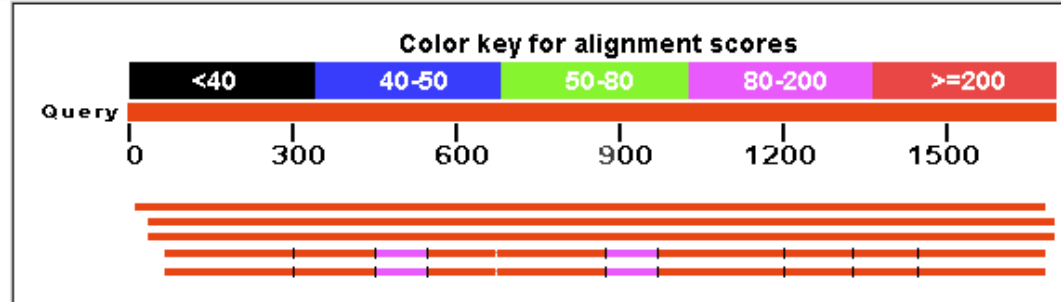
Show positions of the BLAST hits in the human genome using the Entrez Genomes MapViewer



Query= gi|67968779|dbj|AB168636.1| Macaca fascicularis testis cDNA clone: QtsA-13692,
similar to human CDC20 cell division cycle 20 homolog (S. cerevisiae)(CDC20),
mRNA, RefSeq: NM_001255.1.
Length=1696

Distribution of 23 Blast Hits on the Query Sequence

Mouse over to see the define, click to show alignments



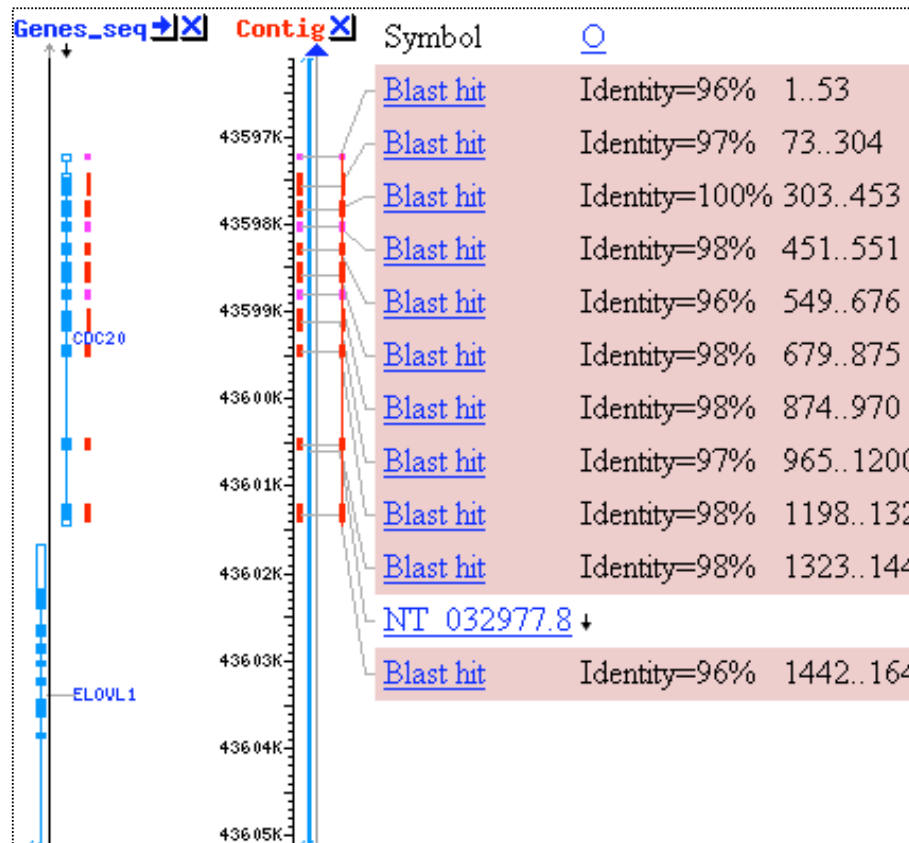
[Distance tree of results](#) **NEW**

Legend for links to other resources: **U** UniGene **E** GEO **G** Gene **S** Structure **M** Map Viewer

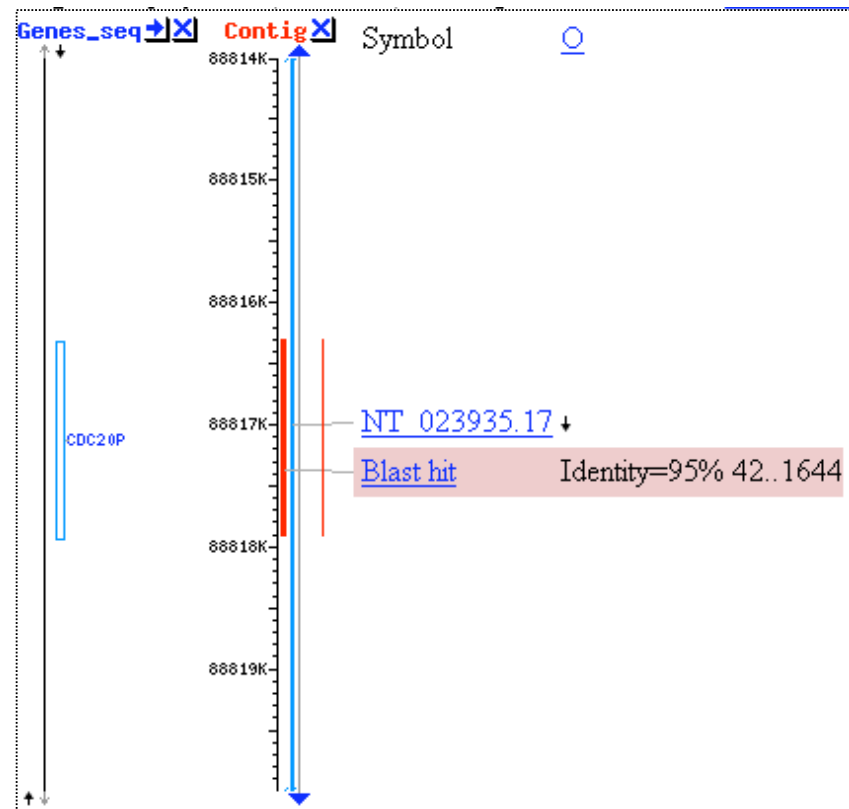
Sequences producing significant alignments:
(Click headers to sort columns)

Accession	Description	Max score	Total score	Query coverage	E value	Max ident
Transcripts						
gi 4557436 NM_001255.1	Homo sapiens CDC20 cell division cycle 20 homolog (S. cerevisiae) (C	2830	2830	98%	0.0	97%
Genomic sequences [show first]						
gi 51467245 NT_023935.17	Homo sapiens chromosome 9 genomic contig, reference assembly	2673	2673	97%	0.0	95%
gi 89030471 NW_924484.1	Homo sapiens chromosome 9 genomic contig, alternate assembly (bas	2654	2654	97%	0.0	95%
gi 88950243 NW_921351.1	Homo sapiens chromosome 1 genomic contig, alternate assembly (bas	411	2858	94%	2e-111	100%
gi 88942921 NT_032977.8	Homo sapiens chromosome 1 genomic contig, reference assembly	411	2853	94%	2e-111	100%

Links to Map Viewer

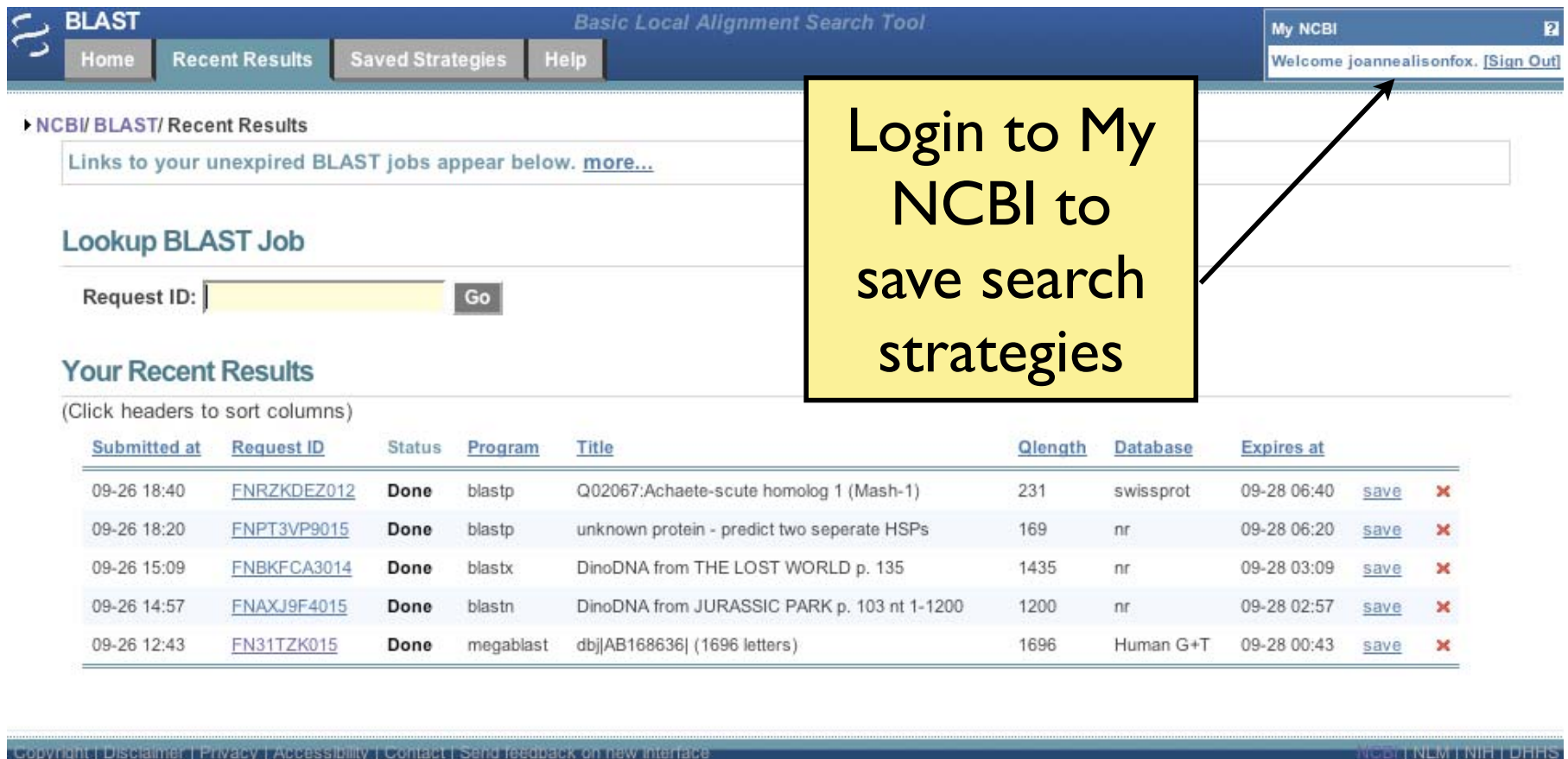


Chromosome 1



Chromosome 9

Recent and Saved Strategies



BLAST Basic Local Alignment Search Tool

Home Recent Results Saved Strategies Help

My NCBI
Welcome joannealisonfox. [Sign Out]

► NCBI/BLAST/ Recent Results

Links to your unexpired BLAST jobs appear below. [more...](#)

Lookup BLAST Job

Request ID: Go

Your Recent Results
(Click headers to sort columns)

Submitted at	Request ID	Status	Program	Title	Qlength	Database	Expires at		
09-26 18:40	FNRZKDEZ012	Done	blastp	Q02067:Achaete-scute homolog 1 (Mash-1)	231	swissprot	09-28 06:40	save	✗
09-26 18:20	FNPT3VP9015	Done	blastp	unknown protein - predict two seperate HSPs	169	nr	09-28 06:20	save	✗
09-26 15:09	FNBKFCA3014	Done	blastx	DinoDNA from THE LOST WORLD p. 135	1435	nr	09-28 03:09	save	✗
09-26 14:57	FNAXJ9F4015	Done	blastn	DinoDNA from JURASSIC PARK p. 103 nt 1-1200	1200	nr	09-28 02:57	save	✗
09-26 12:43	FN31TZK015	Done	megablast	dbj AB168636 (1696 letters)	1696	Human G+T	09-28 00:43	save	✗

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Genomic and Specialized BLAST pages

BLAST Assembled Genomes

Choose a species genome to search, or [list all genomic BLAST databases](#).

- ▣ [Human](#)
- ▣ [Mouse](#)
- ▣ [Rat](#)
- ▣ [Arabidopsis thaliana](#)
- ▣ [Oryza sativa](#)
- ▣ [Bos taurus](#)
- ▣ [Danio rerio](#)
- ▣ [Drosophila melanogaster](#)
- ▣ [Gallus gallus](#)
- ▣ [Pan troglodytes](#)
- ▣ [Microbes](#)
- ▣ [Apis mellifera](#)

Specialized BLAST

Choose a type of specialized search (or database name in parentheses.)

- ▣ Search [trace archives](#)
- ▣ Find [conserved domains](#) in your sequence (cds)
- ▣ Find sequences with similar [conserved domain architecture](#) (cdart)
- ▣ Search sequences that have [gene expression profiles](#) (GEO)
- ▣ Search [immunoglobulins](#) (IgBLAST)
- ▣ Search for [SNPs](#) (snp)
- ▣ Screen sequence for [vector contamination](#) (vecscreen)
- ▣ [Align](#) two sequences using BLAST (bl2seq)

Service Addresses

- ***General Help*** `info@ncbi.nlm.nih.gov`
- ***BLAST*** `blast-help@ncbi.nlm.nih.gov`

Telephone support: 301- 496- 2475

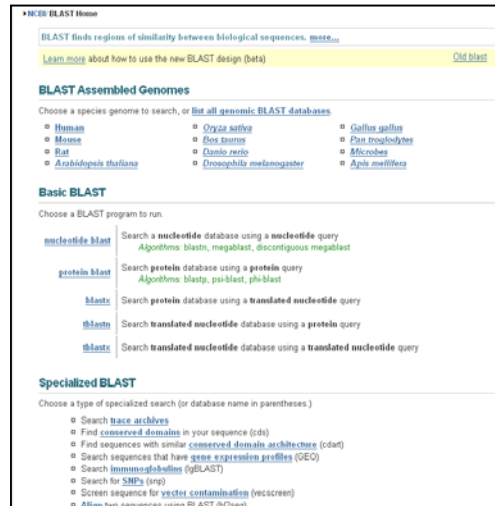
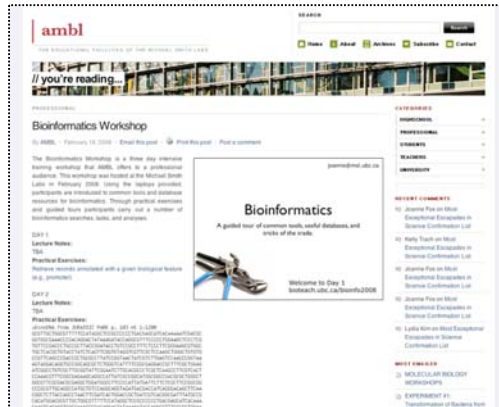


BLAST

PRACTICAL EXERCISE: The Jurassic Park Detective Story



navigate to:
bioteach.ubc.ca/bioinfo2008



Let's compare
our results

Get the sequences from the
webpage and carry out BLAST
searches



Can you identify the Dinosaur sequences?

Search #1:
Jurassic Park
sequence
use blastn

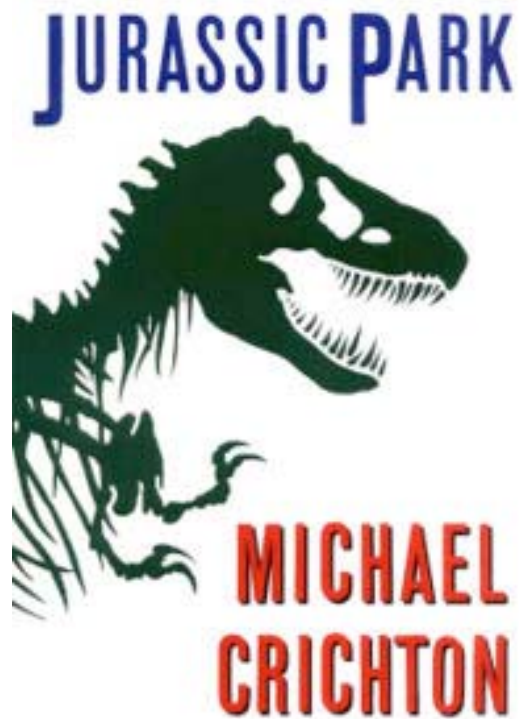
Search #2:
The Lost World
sequence
use blastx

Try some BLAST searches with
your own sequence of interest...



Explore what happens when you
change advanced parameters...

Search #1 - blastn against nr



- Most common use of blastn
 - ✓ Sequence identification
 - ✓ Establish whether an exact match for a sequence is already present in the database

>|gi|157064989|gb|EU118176.1| Cloning vector pCM433, complete sequence
Length=8081

Sort alignments for this subject sequence by:
E value Score Percent identity
Query start position Subject start position

Score = 437 bits (484), Expect = 4e-119
Identities = 297/340 (87%), Gaps = 40/340 (11%)
Strand=Plus/Plus

```
Query 1 GCGTTGCTGGCGTTTTTCCATAGGCTCCGCCCCCTGACGAGCATCACAAAAATCGACGC 60
      |||
Sbjct 7309 GCGTTGCTGGCGTTTTTCCATAGGCTCCGCCCCCTGACGAGCATCACAAAAATCGACGC 7368

Query 61 -----GGTGGCGAAACCCGACAGGACTATAAAGATACCAGGCGTTTCCCCCTGGA 110
      |||
Sbjct 7369 TCAAGTCAGAGGTGGCGAAACCCGACAGGACTATAAAGATACCAGGCGTTTCCCCCTGGA 7428

Query 111 AGCTCCCTCG-----TGTTCCGACCCTGCCGCTTACCGGATACCTGTCCGCCTTT 160
      |||
Sbjct 7429 AGCTCCCTCGTGCCTCTCCTGTTCCGACCCTGCCGCTTACCGGATACCTGTCCGCCTTT 7488

Query 161 CTCCCTTCGGGAAGCGTGGC-----TGCTCACGCTGTACCTATCTCAGTTCGGTG 210
      |||
Sbjct 7489 CTCCCTTCGGGAAGCGTGGCGCTTCTCATAGCTCACGCTGTAGGTATCTCAGTTCGGTG 7548

Query 211 TAGGTCGTTTCGCTCCAAGCTGGGCTGTGTG-----CCGTTACGCCGACCGCTGC 260
      |||
Sbjct 7549 TAGGTCGTTTCGCTCCAAGCTGGGCTGTGTGCACGAACCCCGTTACGCCGACCGCTGC 7608

Query 261 GCCTTATCCGGTAACTATCGTCTTGAGTCCAACCCGGTAA 300
      |||
Sbjct 7609 GCCTTATCCGGTAACTATCGTCTTGAGTCCAACCCGGTAA 7648
```

Score = 536 bits (594), Expect = 6e-149
Identities = 360/410 (87%), Gaps = 50/410 (12%)
Strand=Plus/Plus

```
Query 302 GTAGGACAGGTGCCGGCAGCGCTCTGGGTCAATTTTCGGCGAGGACCGCTTTCGCTGGAG- 360
      |||
Sbjct 3591 GTAGGACAGGTGCCGGCAGCGCTCTGGGTCAATTTTCGGCGAGGACCGCTTTCGCTGGAGC 3650

Query 361 -----ATCGGCCTGTCGCTTGCGGTATTTCGGAATCTTGCACGCCCTCGCTCAAGCC 411
      |||
Sbjct 3651 GCGACGATGATCGGCCTGTCGCTTGCGGTATTTCGGAATCTTGCACGCCCTCGCTCAAGCC 3710

Query 412 TTCGTCACCT-----CCAAACGTTTCGGCGAGAAGCAGGCCATTATCGCCGGCATG 461
      |||
Sbjct 3711 TTCGTCACCTGGTCCCGCCACCAAACGTTTCGGCGAGAAGCAGGCCATTATCGCCGGCATG 3770

Query 462 GCGGCCGACGCGCTGGGCT-----GGCGTTCGCGACGCGAGGCTGGATGGCCTTC 511
      |||
Sbjct 3771 GCGGCCGACGCGCTGGGCTACGTCTTGCTGGCGTTCGCGACGCGAGGCTGGATGGCCTTC 3830

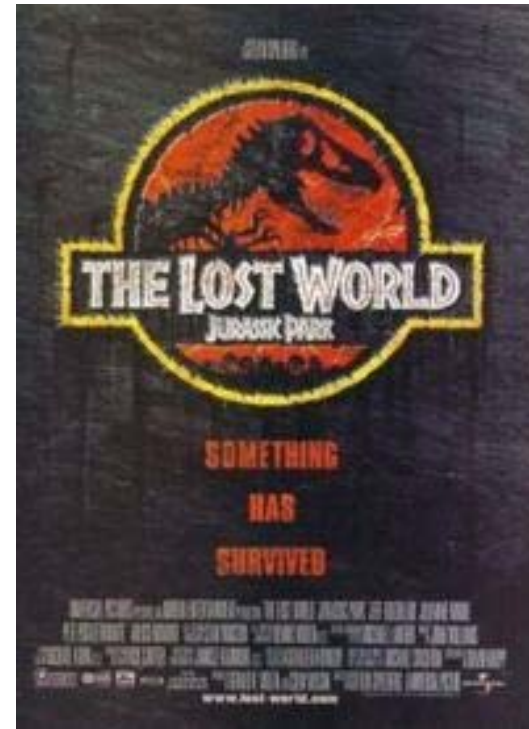
Query 512 CCCATTATGATTCTTCTCGCTTCCGGCG-----GCCCGCGTTGCAGGCCATGCTG 561
      |||
Sbjct 3831 CCCATTATGATTCTTCTCGCTTCCGGCGGCGATCGGGATGCCCGCGTTGCAGGCCATGCTG 3890

Query 562 TCCAGGCAGGTAGATGACGACCATCAGGGACAGCTTCAA-----CGGCTCTTACC 611
      |||
Sbjct 3891 TCCAGGCAGGTAGATGACGACCATCAGGGACAGCTTCAAGGATCGCTCGCGGCTCTTACC 3950

Query 612 AGCCTAACTTCGATCACTGGACCGCTGATCGTCACGGCGATTATGCCGC 661
      |||
Sbjct 3951 AGCCTAACTTCGATCACTGGACCGCTGATCGTCACGGCGATTATGCCGC 4000
```


Search #2 - blastx against nr

- Translating BLAST programs (blastx, tblastn, tblastx)
 - ✓ Look for similar proteins
 - ✓ Identify potential homologs in other species



```

>|gi|45382623|ref|NP_990795.1|UG erythroid-specific transcription factor eryf1 [Gallus gallus]
|gi|120955|sp|P17678|GATA1_CHICKG Erythroid transcription factor (GATA-binding factor 1) (GATA-1)
(Eryf1) (NF-E1 DNA-binding protein) (NF-E1A)
|gi|212629|gb|AAA49055.1|UG Eryf1 protein
Length=304

Score = 366 bits (940), Expect = 2e-99
Identities = 304/318 (95%), Positives = 304/318 (95%), Gaps = 14/318 (4%)
Frame = +1

Query 121 MEFVALGGPDAGSPTPPFDeagafllgllgggerteaggllaSYPPSGRVSLVPWADTGTLG 300
MEFVALGGPDAGSPTPPFDEAGAFLLGLGGGERTEAGLLASYPPSGRVSLVPWADTGTLG
Sbjct 1 MEFVALGGPDAGSPTPPFDEAGAFLLGLGGGERTEAGLLASYPPSGRVSLVPWADTGTLG 60

Query 301 TPQWVPPATQMEPPHYLEllqpprgspphpssgpllpssgpppCEARECVMARKNCGAT 480
TPQWVPPATQMEPPHYLELLQPPRGSPHPSSGPLLPLSSGPPPCEARECV NCGAT
Sbjct 61 TPQWVPPATQMEPPHYLELLQPPRGSPHPSSGPLLPLSSGPPPCEARECV----NCGAT 116

Query 481 ATPLWRRDGTGHYLCN WASACGLYHRLNGQNRPLIRPKRLLVSKRAGTVCSHERENCQT 660
ATPLWRRDGTGHYLCN ACGLYHRLNGQNRPLIRPKRLLVSKRAGTVCS NCQT
Sbjct 117 ATPLWRRDGTGHYLCN---ACGLYHRLNGQNRPLIRPKRLLVSKRAGTVCS----NCQT 169

Query 661 STTTLWRRSPMGDPVCNNIHACGLYYKLHQVNRPLTMRKDGITRNRKVsskgkkrppg 840
STTTLWRRSPMGDPVCN ACGLYYKLHQVNRPLTMRKDGITRNRKVSSKGKKRRPPG
Sbjct 170 STTTLWRRSPMGDPVCN ACGLYYKLHQVNRPLTMRKDGITRNRKVSSKGKKRRPPG 226

Query 841 ggnpsatagggapmggggdpsmppppppppaaappQSDALYALGPVVLSGHFLPfgnsggf 1020
GGNPSATAGGGAPMGGGGDPSMPPPPPPPPAAAPPQSDALYALGPVVLSGHFLPPGNSGGF
Sbjct 227 GGNPSATAGGGAPMGGGGDPSMPPPPPPPPAAAPPQSDALYALGPVVLSGHFLPPGNSGGF 286

Query 1021 fgggaggYTAPPGLSPQI 1074
FGGGAGGYTAPPGLSPQI
Sbjct 287 FGGGAGGYTAPPGLSPQI 304

```

Mark was here, NIH

Credits

- Materials for this presentation have been adapted with permission from the following NCBI HelpDesk course materials:

Field Guide Course Materials

Advanced Workshop for Bioinformatics Information Specialists

- NCBI BLAST

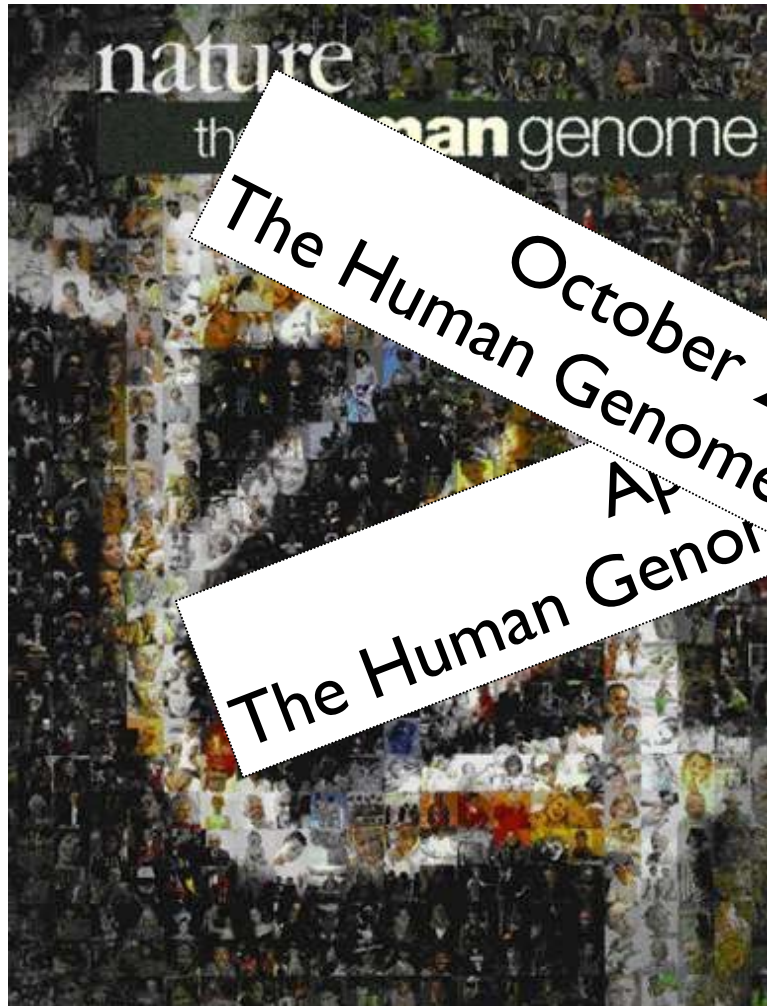
<http://www.ncbi.nlm.nih.gov/blast/Blast.cgi>

Genome Browsers

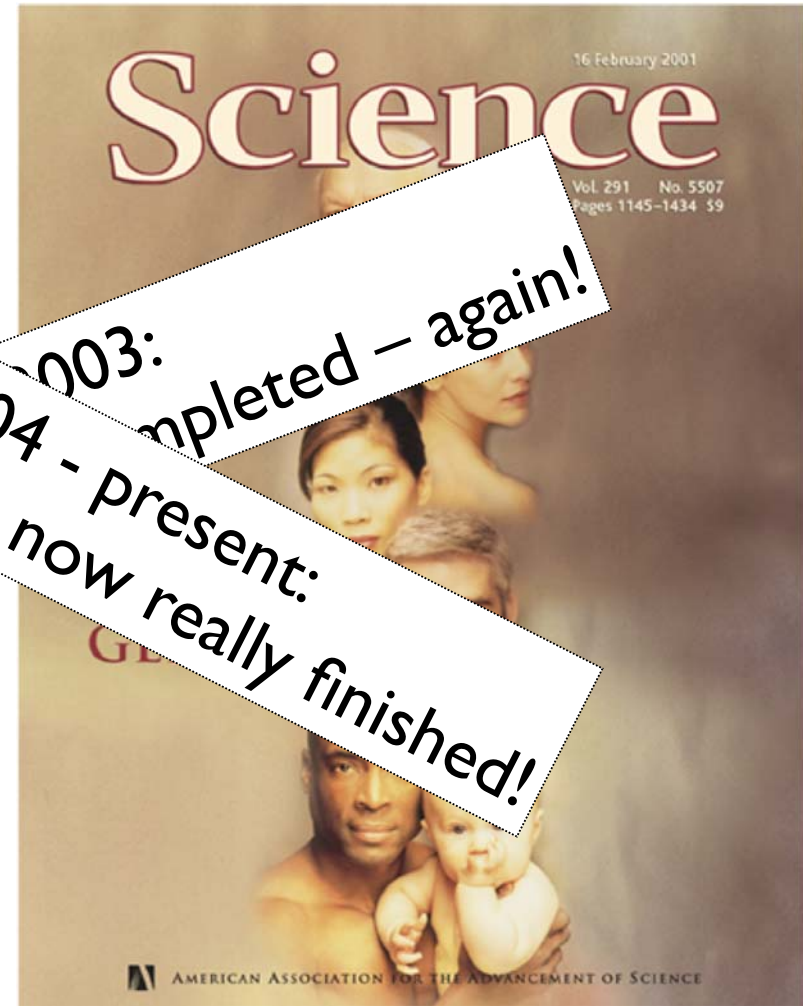
Accessing Genome Annotations &
PRACTICAL EXERCISE: Three Different
Views of the BRCA1 Gene



The Human Genome Project



October 2004 - present:
The Human Genome is now really finished!

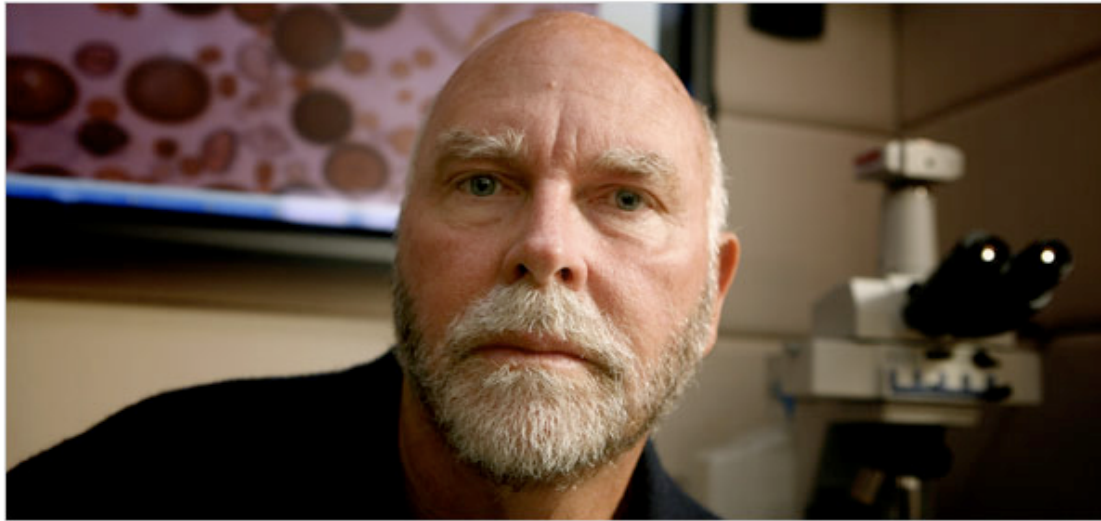


Public HGP

Celera Genomics

February 2001: Completion of the Draft Human Genome

In the Genome Race, the Sequel Is Personal



Thor Swift for The New York Times

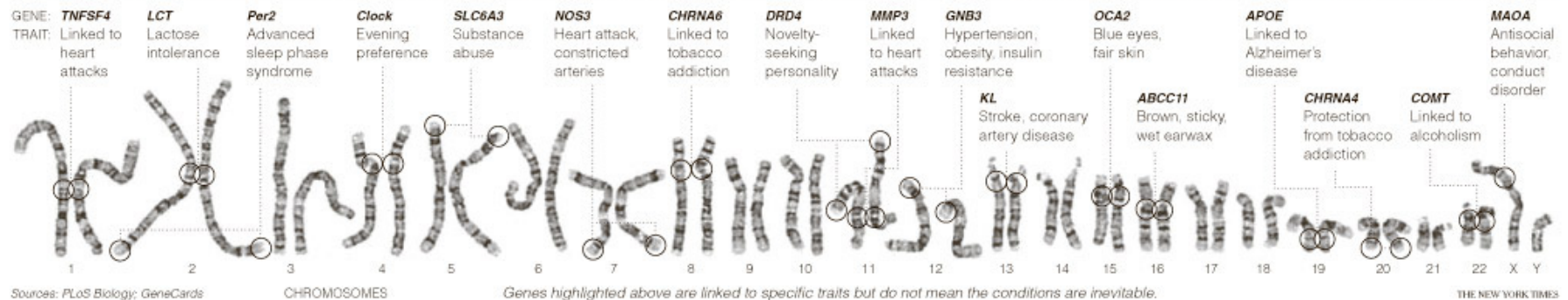
A team led by J. Craig Venter, above, has finished the first mapping of a full, or diploid, genome, made up of DNA inherited from both parents. The genome is Dr. Venter's own.

PHOTOGRAPH BY

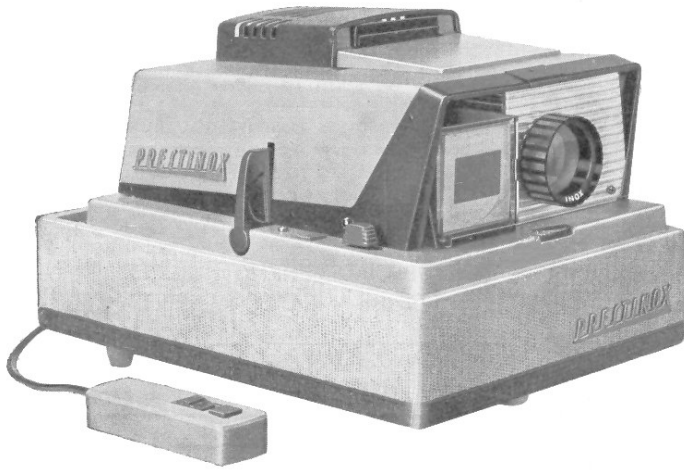
The New York Times

September 3, 2007

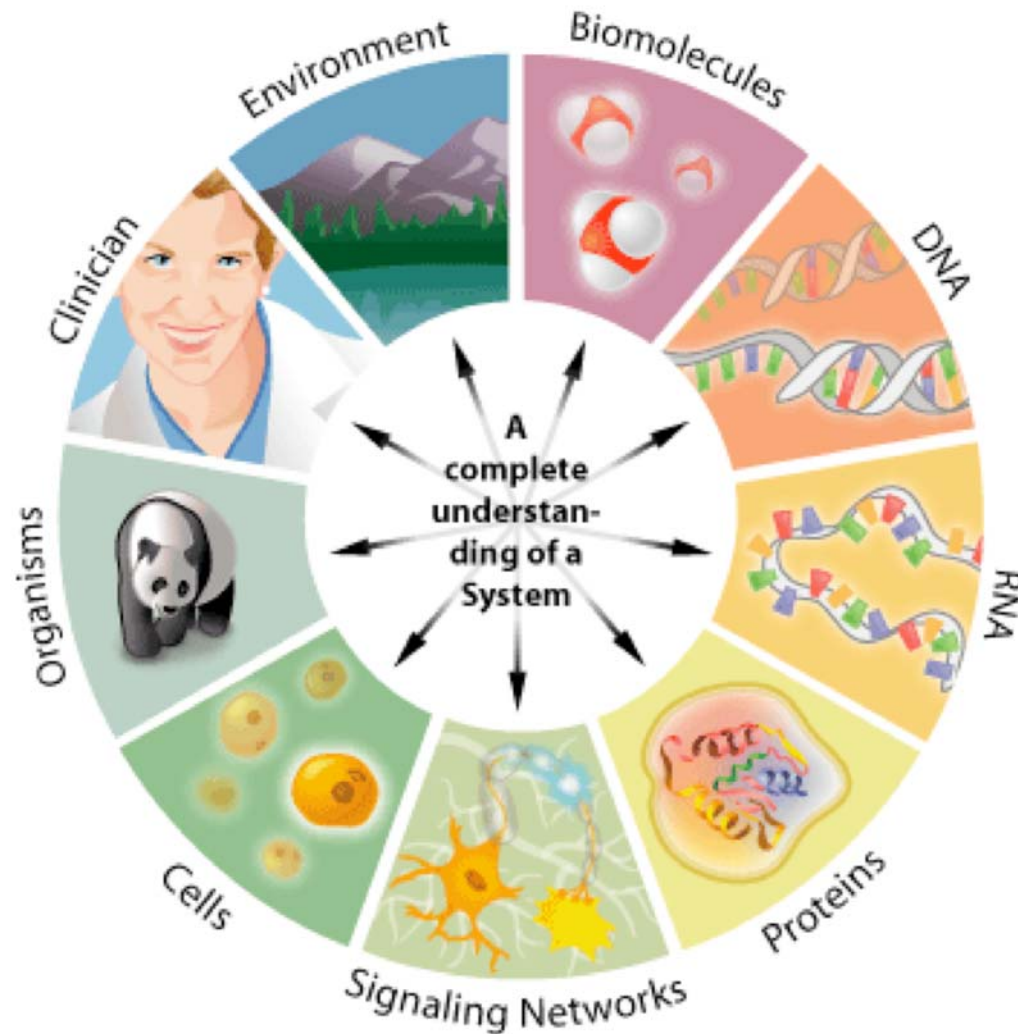
DECODING HIMSELF A team led by J. Craig Venter, above, has finished the first mapping of a full, or diploid, genome, made up of DNA inherited from both parents. The genome is Dr. Venter's own.

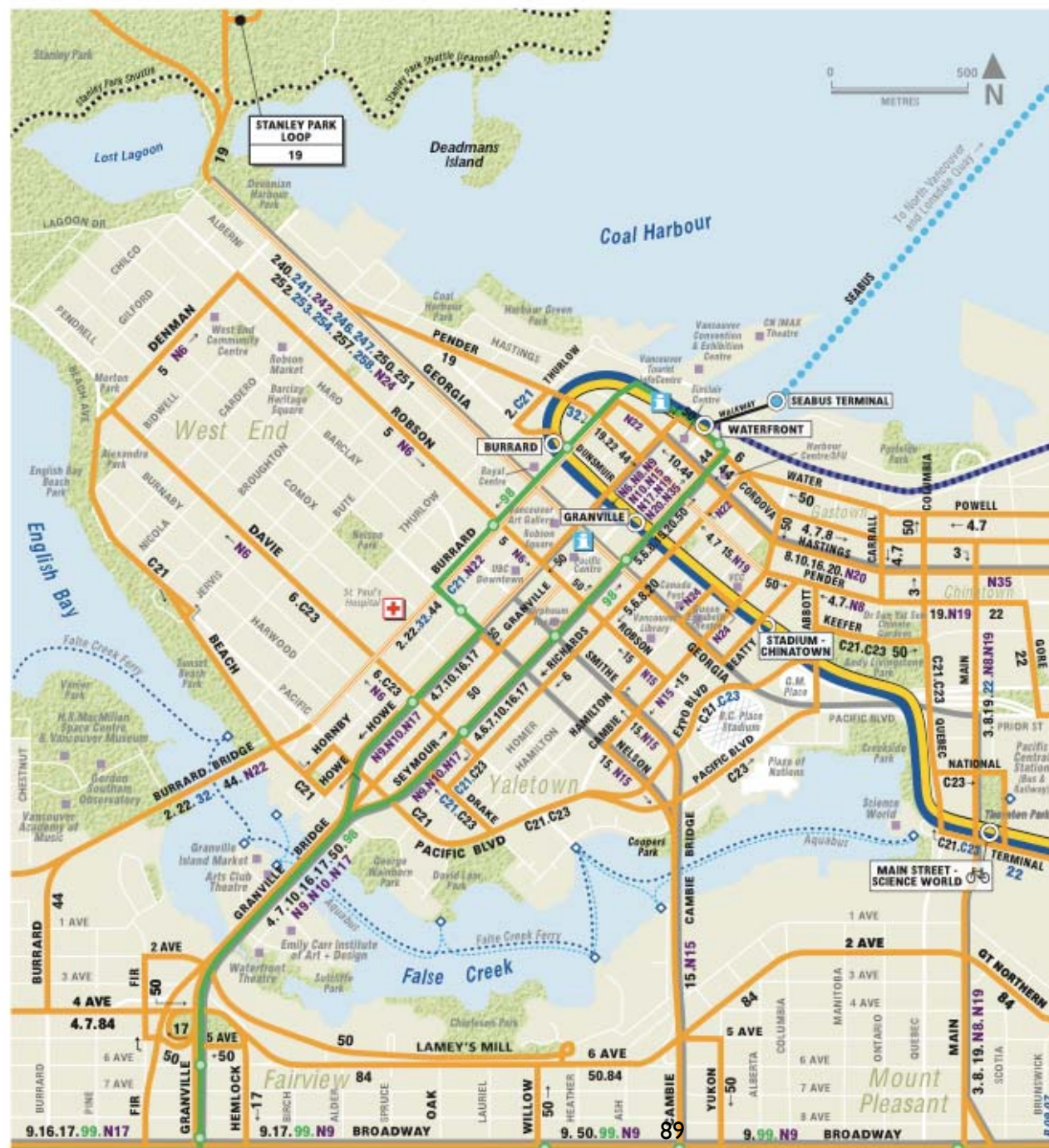


Technology



What is Bioinformatics?





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Google [Web](#) [Images](#) [Groups](#) [News](#) [Maps](#) [Scholar](#) [more »](#)

coffee 2185 East Mall, Vancouver, BC Search Businesses

[Search the map](#) [Find businesses](#) [Get directions](#)

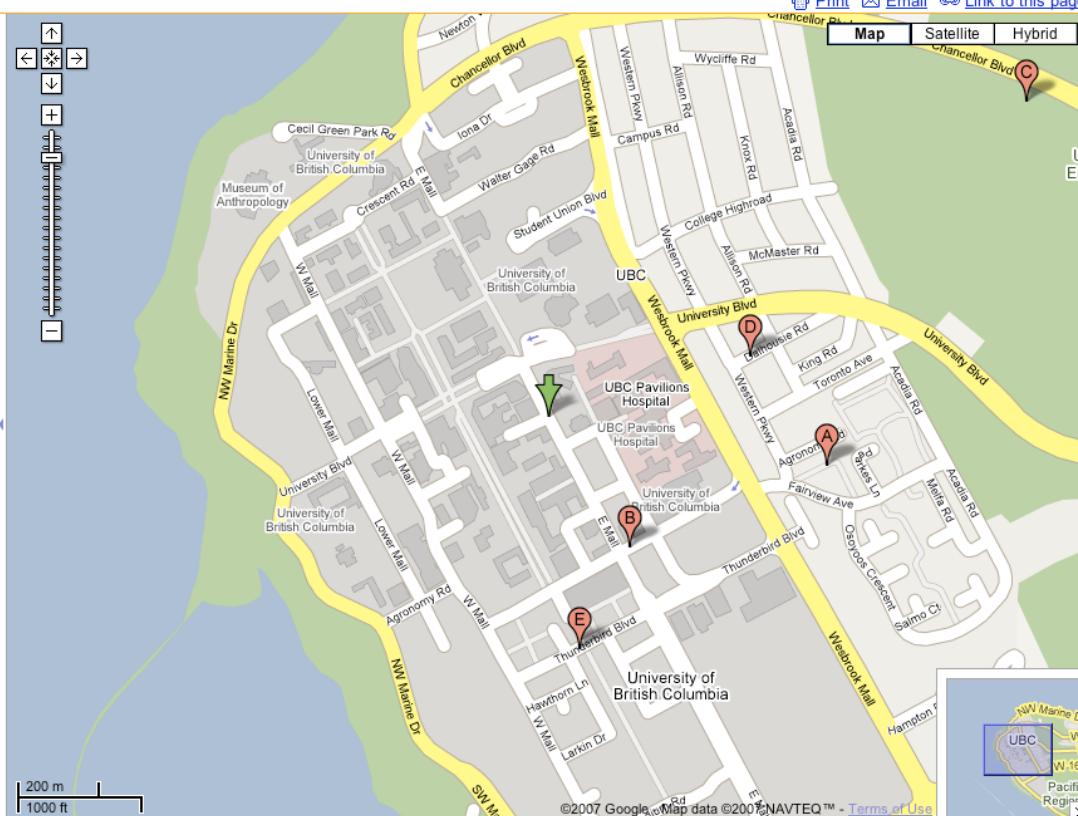
Search Results My Maps

[Text View](#) [Map View](#)

Results 1-10 of about 6,156 for **coffee** near **2185 E Mall, UBC, BC V6T** - [Modify search](#)

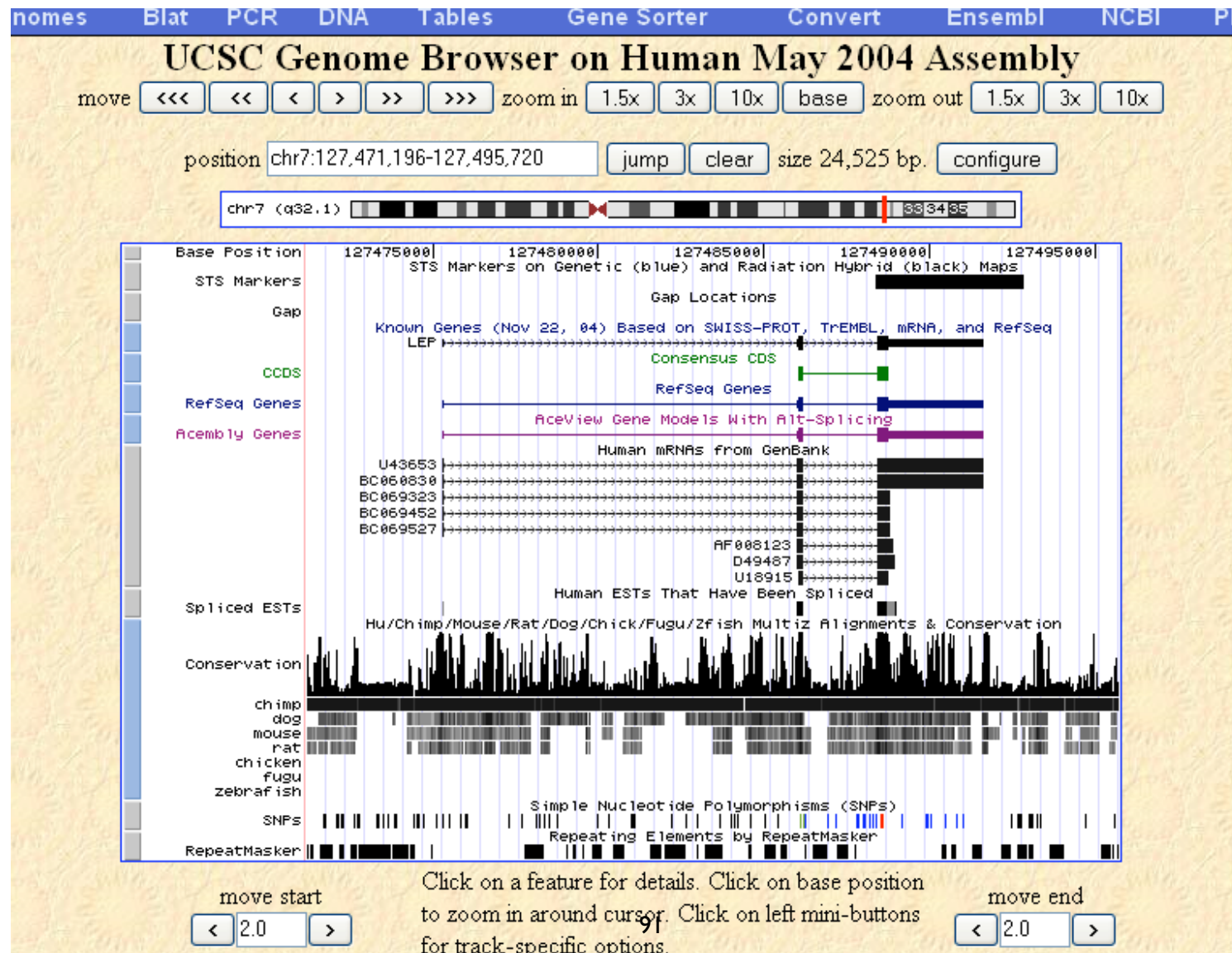
Categories: [Coffee Retail](#), [Coffee Houses](#)

- A** [Original Beanery The](#) - [more info »](#)
2706 Fairview Crescent, Vancouver, BC V6T 2B9
(604) 224-2326 - 0.7 km E
Category: Coffee Houses
- B** [Starbucks Coffee Co](#) - [more info »](#)
6190 Agronomy Road, Vancouver, BC V6T 1Z3
(604) 221-6434 - 0.4 km SE
- C** [Blue Chip Cookies](#) - [more info »](#)
6138 Sub Boulevard, Vancouver, BC V6T 2A5
(604) 822-6999 - 1.4 km NE
Category: Coffee Houses
- D** [Starbucks Coffee Co](#) - [more info »](#)
5761 Dalhousie Road, Vancouver, BC V6T 2H9
(604) 221-0200 - 0.5 km E
- E** [Salt Spring Coffee](#) - [more info »](#)
6308 Thunderbird Blvd, Vancouver, BC V6T 1Z4
(604) 221-6400 - 0.6 km S
- F** [Starbucks Coffee Co](#) - [more info »](#)
4580 10th Avenue West, Vancouver, BC V6R 2J1
(604) 222-1456 - 2.8 km E
- G** [European Crepe Cafe](#) - [more info »](#)
4544 West 10th Avenue, Vancouver, BC V6R 2J1
(604) 221-4340 - 2.8 km E



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Let's Look at the Human Genome...



Objectives

- By the end of this module:
 - ✓ You will be able to describe the following concepts: genome annotation, genome builds, and genome browsers.
 - ✓ You will view the genomic location that contains the BRCA1 gene in the human genome using three different genome browsers.
 - ✓ You will be able to compare and contrast the UCSC, Ensembl and MapViewer systems for visualizing genome information.

Genome Browsers

- What is a Genome Browser?
 - System for displaying, viewing, and accessing genome annotation data
- Genome annotations = knowledge attached to raw genome sequence.
 - Annotation information comes from many different sources
 - ✓ Computational pipelines
 - ✓ Research groups
 - ✓ Databases

The “Neopolitan Ice Cream” World of Genome Browsing:

- UCSC Genome Browser
<http://genome.cse.ucsc.edu/>

- Ensembl
<http://www.ensembl.org/>

- NCBI Map Viewer
<http://www.ncbi.nlm.nih.gov/mapview/>



**The underlying data is
common for all three
“flavors” of Genome
Browsers.**

- NCBI, UCSC and Ensembl use the same human genome assembly that is generated by NCBI
 - ─ release timing is different between sites.
- Note the version of genome assembly to which you are referring
 - ─ available precomputed info and locations of features will be different between different assemblies.

**Let's compare the view of
the BRCA1 gene in all
three genome browsers.**

Viewing the genomic region containing BRCA1

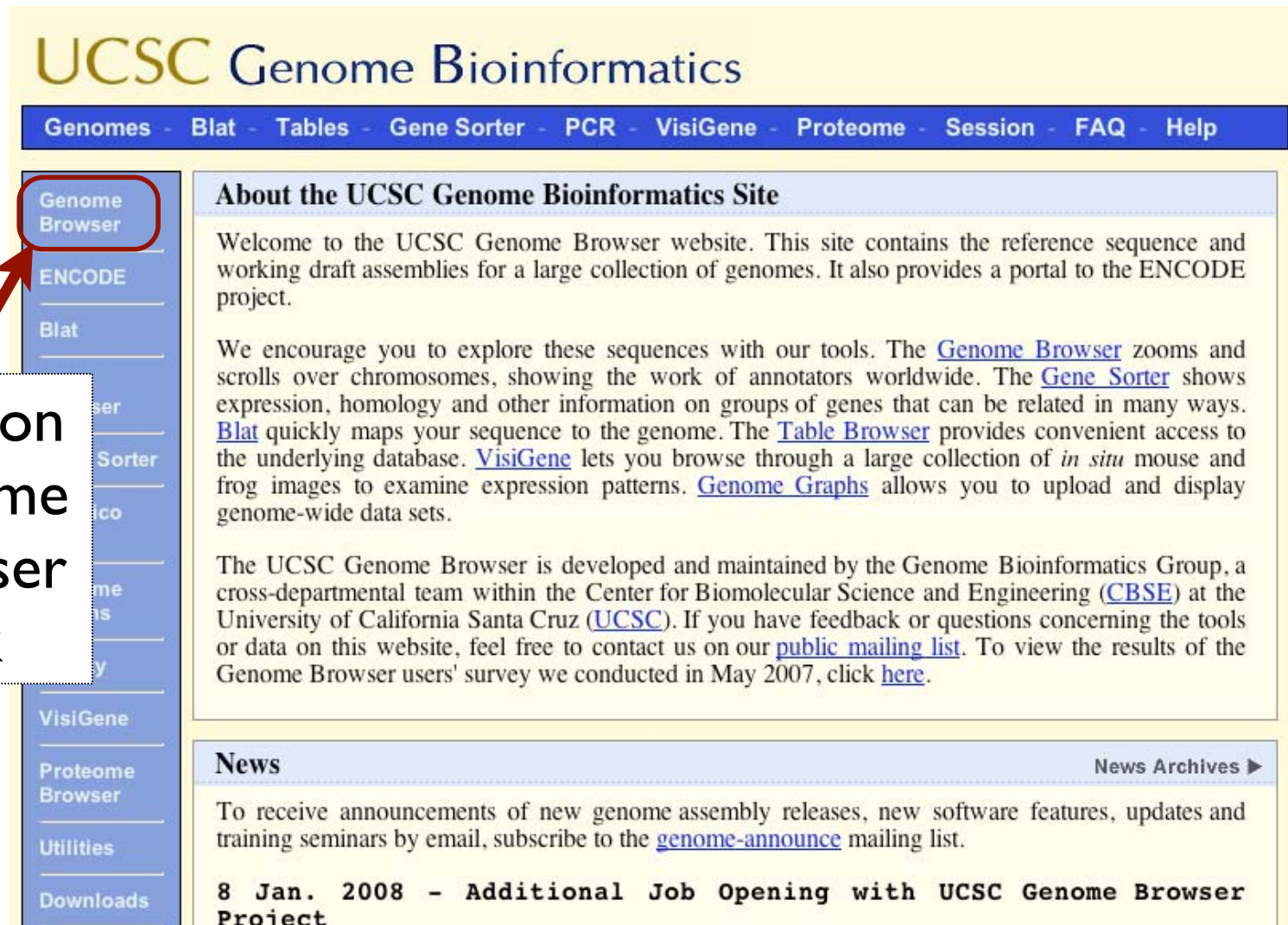
- Common features:

- ✓ Coordinate system is based on the build
- ✓ Zoom in and out
- ✓ Annotations displayed – ie. Gene features

- Major Differences:

- ✓ Each Browser has a very different look and feel
- ✓ Annotation information displayed differently
- ✓ Different ways to navigate through the information

<http://genome.cse.ucsc.edu/>



UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Proteome - Session - FAQ - Help

Genome Browser

ENCODE

Blat

ser

Sorter

co

me

is

y

VisiGene

Proteome Browser

Utilities

Downloads

About the UCSC Genome Bioinformatics Site

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides a portal to the ENCODE project.

We encourage you to explore these sequences with our tools. The [Genome Browser](#) zooms and scrolls over chromosomes, showing the work of annotators worldwide. The [Gene Sorter](#) shows expression, homology and other information on groups of genes that can be related in many ways. [Blat](#) quickly maps your sequence to the genome. The [Table Browser](#) provides convenient access to the underlying database. [VisiGene](#) lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns. [Genome Graphs](#) allows you to upload and display genome-wide data sets.

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the Center for Biomolecular Science and Engineering ([CBSE](#)) at the University of California Santa Cruz ([UCSC](#)). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our [public mailing list](#). To view the results of the Genome Browser users' survey we conducted in May 2007, click [here](#).

News

News Archives ►

To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the [genome-announce](#) mailing list.

8 Jan. 2008 - Additional Job Opening with UCSC Genome Browser Project

Click on
Genome
Browser
link

[Home](#)
[Genomes](#)
[Blat](#)
[Tables](#)
[Gene Sorter](#)
[PCR](#)
[FAQ](#)
[Help](#)

Human (*Homo sapiens*) Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#).
Software Copyright (c) The Regents of the University of California. All rights reserved.

clade
Vertebrate

genome
Human

assembly
May 2004

position or search term
BRCA1

image width
620

submit

[Click here to reset](#) the browser user interface settings to their defaults.
[add your own custom tracks](#)
[configure tracks and display](#)
[clear position](#)

About the Human May 2004 (hg17) assembly ([sequences](#))

The May 2004 human reference sequence is based on NCBI Build 35 and was produced by the International Human Genome Sequencing Consortium.

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS number, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of queries to the human genome. See the [User's Guide](#) for more information.

Request:	Genome Browser Response:
chr7	Displays all of chromosome 7
20p13	Displays region for band p13 on chr 20
chr3:1-1000000	Displays first million bases of chr 3, counting from p arm telomere
D16S3046	Displays region around STS marker D16S3046 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well.
RH18061;RH80175	Displays region between STS markers RH18061;RH80175. Includes 100,000 bases on each side as well.
AA205474	Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17
AC008101	Displays region of clone with GenBank accession AC008101
AF083811	Displays region of mRNA with GenBank accession number AF083811
PRNP	Displays region of genome with HUGO Gene Nomenclature Committee identifier PRNP
NM_017414	Displays the region of genome with RefSeq identifier NM_017414
NP_059110	Displays the region of genome with protein accession number NP_059110
pseudogene mRNA	Lists transcribed pseudogenes, but not cDNAs
human-mouse	Lists cDNAs from mouse that have human homologs

Search for
BRCA1;
Note sample
queries

Known Genes

BRCA1 (NM 007305)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007304)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007303)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007302)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007301)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007300)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007299)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007298)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007297)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform
BRCA1 (NM 007295)	at chr17:38449840-38530994	- breast cancer 1, early onset isoform

✓ All located on chr 17

✓ same chr coordinates

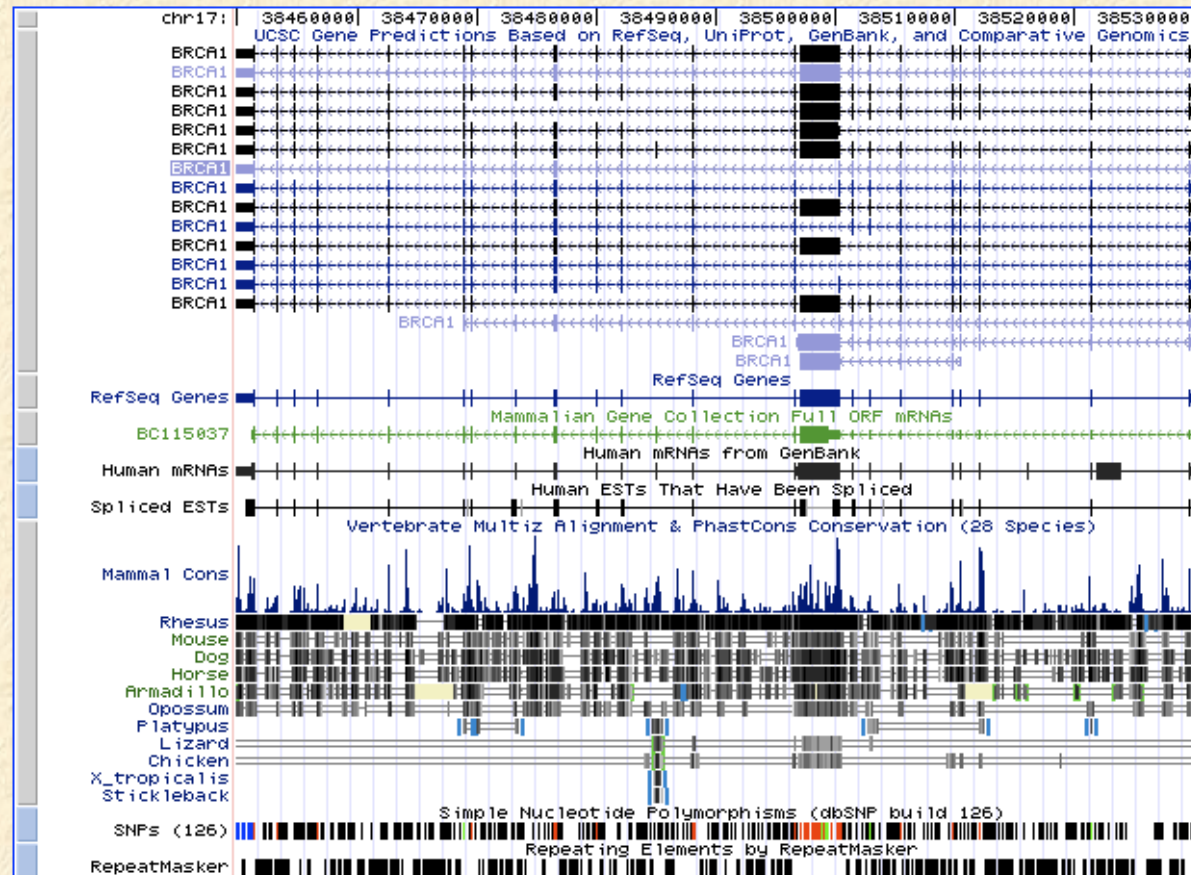
✓ different gene structures

UCSC Genome Browser on Human Mar. 2006 Assembly

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

position/search chr17:38,449,840-38,530,994 jump clear size 81,155 bp. configure

chr17 (q21.31) p12 p11.2 11.2 17q12 17q22 q25.3



move start

< 2.0 >

Click on a feature for details. Click on base position to zoom in around cursor. Click gray/blue bars on left for track options and descriptions.

move end

< 2.0 >

default tracks

hide all

add custom tracks

configure

refresh

Use drop-down controls below and press refresh to alter tracks displayed.

Two tasks

- What genes are on either side of BRCA1 on chr 17?
- Can you figure out how to download the genomic sequence for the BRCA1 region?

The screenshot shows the UCSC Genome Browser interface. At the top is a navigation bar with links: Home, Genomes, Blat, Tables, Gene Sorter, PCR, **DNA**, Convert, Ensembl, NCBI, PDF/PS, and Help. The main title is "UCSC Genome Browser on Human May 2004 Assembly". Below the title is a navigation area with "move" buttons (<<<, <<, <, >, >>, >>>) and a "zoom in" button circled in red. To the right of "zoom in" are buttons for "1.5x", "3x", "10x", "base", "zoom out", "1.5x", "3x", and "10x". Below this is a search bar with the text "position/search chr17:38,423,783-38,543,782" and buttons for "jump", "clear", "size 120,000 bp.", and "configure". Below the search bar is a chromosome map showing chromosomes 17, 18, 19, 20, 21, 22, 23, and 24. A red arrow points from the "zoom in" button to the "DNA" tab. Another red arrow points from the "DNA" tab to the "BRCA1" gene track. The "BRCA1" gene track is highlighted in blue. Below the gene track is a table of "UCSC Known Genes (June, 05) Based on UniProt, RefSeq, and GenBank mRNA". The table has columns for "Gene Name", "Transcript", "Exon", "Intron", "Start", "End", "Score", "RefSeq", "UniProt", and "GenBank". The "BRCA1" gene is listed with its transcripts and exons. The "BRCA1" gene track is highlighted in blue. Below the gene track is a table of "UCSC Known Genes (June, 05) Based on UniProt, RefSeq, and GenBank mRNA". The table has columns for "Gene Name", "Transcript", "Exon", "Intron", "Start", "End", "Score", "RefSeq", "UniProt", and "GenBank". The "BRCA1" gene is listed with its transcripts and exons. The "BRCA1" gene track is highlighted in blue. Below the gene track is a table of "UCSC Known Genes (June, 05) Based on UniProt, RefSeq, and GenBank mRNA". The table has columns for "Gene Name", "Transcript", "Exon", "Intron", "Start", "End", "Score", "RefSeq", "UniProt", and "GenBank". The "BRCA1" gene is listed with its transcripts and exons. The "BRCA1" gene track is highlighted in blue.

[Home](#) [Genomes](#) [Genome Browser](#) [Blat](#) [Tables](#) [Gene Sorter](#) [PCR](#) [FAQ](#) [Help](#)

Get DNA in Window

Get DNA for

Position

Note: if you would prefer to get DNA for features of a particular track or table, try the [Table Browser](#) using the output format sequence.

Sequence Retrieval Region Options:

Add extra bases upstream (5') and extra downstream (3')

Note: if a feature is close to the beginning or end of a chromosome and upstream/downstream bases are added, they may be truncated in order to avoid extending past the edge of the chromosome.

Sequence Formatting Options:

☒ All upper case.
☐ All lower case.
☐ Mask repeats: ☒ to lower case ☐ to N
☐ Reverse complement (get '-' strand sequence)

Note: The "Mask repeats" option applies only to "get DNA", not to "extended case/color options".

<http://www.ensembl.org/>



Ensembl release 48 - Dec 2007

[HOME](#) • [BLAST](#) • [BIOMART](#) • [SITEMAP](#) [HELP](#)

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- [About User Accounts](#)

Help & Documentation

- [Setting up an Ensembl Website](#)
- [Data Downloads](#)
- [About Ensembl](#)
- [Using Ensembl](#)

Ensembl Archive

[View previous releases](#)



Mouse Lemur
Microcebus murinus

Search Ensembl

Search: for [Go](#)
e.g. mouse chromosome 2 or rat X:10000..20000 or human gene BRCA2

Ensembl tools



Start a sequence search →

Search Ensembl for nucleotide and peptide sequences with BLAST and SSAHA.



Mine Ensembl with BioMart →

Extract information from the Ensembl database and export sequences or tables in text, html, or Excel format with BioMart



Customise Your Ensembl →

Register with Ensembl to bookmark your favourite pages, customise your home page and much more!



Fetch data with the Ensembl API →

Learn how to extract data from the public Ensembl database with this tutorial.

Ensembl 48

[Pre! species](#)

Popular genomes • [Log in to customize](#)



Human

NCBI 36 | Vega



Mouse

NCBI m37 | Vega



Zebrafish

Zv7 | Vega

All genomes

-- Select a species --

Other pre-build species are available in [Ensembl Pre!](#)

**Ensembl headlines: Release 48
(December 2007)**

[About Ensembl](#)

Click on Human

Your Ensembl

- Login or Register
- About User Accounts

Help & Documentation

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- Setting up an Ensembl Website
- About Ensembl
- Using Ensembl

Ensembl Archive

- View previous release of page in Archive!
- Stable Archive! link for this page



Mouse Lemur
Microcebus murinus



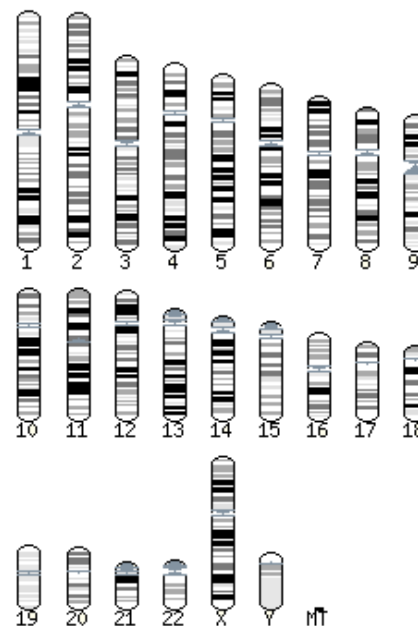
Explore the *Homo sapiens* genome

Search Ensembl *Homo sapiens*

Search: **Go**
e.g. chromosome X or 12:10000..200000 or BRCA2

Karyotype

Click on a chromosome for a closer view



Jump directly to sequence position

Chromosome:

About the Human genome

Assembly



This release is based on the NCBI 36 assembly of the [human genome](#) [November 2005]. The data consists of a reference assembly of the complete genome plus the Celera WGS and a number of alternative assemblies of individual haplotypic chromosomes or regions.

[Full list of assemblies](#)

The International Human Genome Sequencing Consortium have published their scientific analysis of the finished human genome.

- [Nature 431, 931 - 945 \(21 October 2004\)](#)
- [WT Sanger Institute Press Release](#)

Annotation

Since release 38 (April 2006) the gene annotation presented has been a combined Ensembl-Havana geneset, which incorporates more than 18,000 full-length protein-coding transcripts annotated by the Havana team with the Ensembl automatic gene build. The human genome sequence is now considered sufficiently stable that since 2004 the major genome

Ensembl release 40 - Aug 2006

Feature type

☒ **Homo sapiens (13)**

Species

☒ **Gene (13)**

Exalead Help

to exclude a category click on the "[-]".
to restrict to a category click on the name of the category.
to reset a category click on the "[R]" or its name.

Use Ensembl to...

☒ **Run a BLAST search**

☒ **Search Ensembl**

Docs and downloads

☒ **Information**

☒ **What's New**

☒ **About Ensembl**

☒ **Ensembl data**

☒ **Software**

Ensembl text search

Your query matched 13 entries in the search database. Viewing hits 1-10

1 2

Ensembl Gene: ENSG0000012048 [\[ContigView\]](#)

Ensembl gene ENSG0000012048 has 8 transcripts: ENST00000309486, ENST00000346315, ENST00000351666, ENST00000353540, ENST00000354971, ENST00000357654, ENST00000359270 and associated peptides: ENSP0000013772, ENSP000001246907, ENSP0000010938, ENSP0000012236, ENSP000001326002, ENSP000001338007, ENSP000001350283, Breast cancer type 1 susceptibility protein (RING finger protein 53). [Source:Uniprot/SWISSPROT;Acc:P38398]

The gene has the following external identifiers mapped to it:

Affymetrix Microarray Focus: 204531_s_at

Affymetrix Microarray HCG110: 1993_s_at, 604_at

Affymetrix Microarray HuGeneFL: L78833_cds1_at, U64805_s_at

Affymetrix Microarray U133: 211851_x_at, g2218153_3p_a_at, g6552300_3p_a_at, 204531_s_at

Affymetrix Microarray U95: 1993_s_at, 604_at, 33724_at

Agilent CGH: A_14_P133777, A_14_P139703, A_14_P135846

Agilent Probe: A_32_P180603, A_32_P405851, A_23_P207400

CCDS: CCDS11458, RNF53, CCDS11457.1, CCDS11456, CCDS11460.1, CCDS11457, CCDS11454.1, CCDS11460, CCDS1145

CCDS11453, CCDS11458.1, CCDS11453.1, CCDS11456.1, CCDS11459, CCDS11455

DQ190457, AF507077, DQ190456, Y08757, DQ299331, DQ299329, DQ190452, DQ299328, DQ2993

5037, AY093493, DQ190454, AF507078, AF005068, DQ299324, AY093487, AY093489, DQ299305

99320, DQ299316, U37574, DQ299325, DQ145822, DQ299308, L78833, DQ145824, AY093492, DQ

41, DQ299319, AF284812, U61268, DQ116737, DQ299315, AY751490, DQ190451, AY093490, AY0

52429, DQ145823, AY093485, U14680, DQ190450, DQ299306, DQ299326, DQ299322, AY304547,

BCT06746, DQ145826, DQ145825, DQ299307, DQ299314, AY706911, DQ299311, DQ299313, AF507076, DQ299323

EntrezGene: RNF53, 672, BRCC1

GO: GO:0007098, GO:0045786, RNF53, GO:0006260, GO:0042127, GO:0005737, GO:0050681, GO:0005515, GO:0045739, GO:

GO:0000793, GO:0006359, GO:0005622, GO:0000067, GO:0000075, GO:0016481, GO:0003684, GO:0000151, GO:0046600, GO:

GO:0051298, GO:0016567, GO:0030521, GO:0008372, GO:0045893, GO:0015631, GO:0004553, GO:0006357, GO:0005975, GO:

GO:0008630, GO:0005634, GO:0006281, GO:0005813, BRCC1, GO:0009048, GO:0004842, GO:0046872, GO:0008274, GO:0003

GO:0042981, GO:0007049, GO:0006978

HGNC Symbol: **BRCA1**, 1100

Illumina: GI_6552306

IPI: IPI00037550, RNF53, IPI0002727, IPI0002723, IPI00185298, IPI00027268, IPI00027273, IPI002189, IPI0037550

Click on
ENSG0000012048

e!Ensembl Human GeneView

Search e!Human: e.g. ENSG00000193451, ENSG00000190111

Ensembl v34 - Oct 2005

ENSG00000012048

- Gene information
- Gene splice site image
- Gene regulation info.
- Gene variation info.
- Genomic sequence
- Export data
- Transcript information
- Exon information
- Peptide information

Chromosome 17
38,449,844 - 38,530,934

- View of Chromosome 17
- Graphical view

Ensembl Gene Report for ENSG00000012048

Gene	BRCA1 (HGNC Symbol ID) (to view all Ensembl genes linked to the name click here) This gene is a member of the human CCDS set: CCDS11453 , CCDS11454 , CCDS11455 , CCDS11456 , CCDS11457 , CCDS11458 , CCDS11459 , CCDS11460
Ensembl Gene ID	ENSG00000012048
Genomic Location	This gene can be found on Chromosome 17 at location: 38,449,844-38,530,934 This start of this gene is located in Contig AC135721.1 1,150472 .
Description	Breast cancer type 1 susceptibility protein (RING finger protein 53). Source: UniProt/SwissProt P38398
Prediction Method	Genes were annotated by the Ensembl automatic analysis pipeline using either a GeneWise model from a human/vertebrate protein, a set of aligned human cDNAs followed by GenomeWise for ORF prediction or from Genscan exons supported by protein, cDNA and EST evidence. GeneWise models are further combined with available aligned cDNAs to annotate UTRs.

ENST00000309486	ENSP00000310938	NP_009228.1	[Transcript info]
ENST00000337272	ENSP00000337814	BRCA1	[Transcript info]
ENST00000346315	ENSP00000246907	NP_009230.1	[Transcript info]
ENST00000351666	ENSP00000338007	NP_009229.1	[Transcript info]
ENST00000352993	ENSP00000312236	NP_009234.1	[Transcript info]
ENST00000353540	ENSP00000013772	NP_009233.1	[Transcript info]
ENST00000354071	ENSP00000326002	NP_009231.1	[Transcript info]
ENST00000357654	ENSP00000350283	BRCA1	[Transcript info]
ENST00000359270	ENSP00000352215	NP_009237.1	[Transcript info]
ENST00000360294	ENSP00000353439	NP_009233.1	[Transcript info]

Features

Chr. 17
Length 101.09 Kb

DNA(contigs)

Ensembl trans.

click here to view genomic location

GeneView shows you information about the gene

- Run a BLAST search
- Search Ensembl
- Data mining [BioMart]
- Upload your own data
- Export data
- Download data

Docs and downloads

- Information
- What's New
- About Ensembl

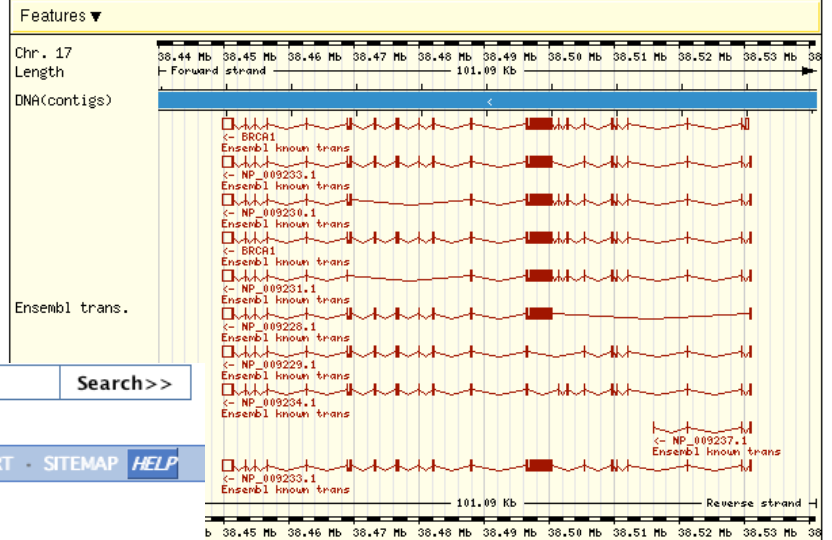
Two tasks

- Using GeneView, can you figure out how many different alternatively spliced isoforms exist for BRCA1?
- Using ContigView, can you figure out how to download the genomic sequence for the BRCA1 region?

GeneView shows
you information
about the
transcripts

Transcripts

ENST00000309486	ENSP00000310938	NP_009228.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000337272	ENSP00000337814	BRCA1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000346315	ENSP00000246907	NP_009230.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000351666	ENSP00000338007	NP_009229.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000352993	ENSP00000312236	NP_009234.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000353540	ENSP00000013772	NP_009233.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000354071	ENSP00000326002	NP_009231.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000357654	ENSP00000350283	BRCA1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000359270	ENSP00000352215	NP_009237.1	[Transcript info]	[Exon info]	[Peptide info]
ENST00000360294	ENSP00000353439	NP_009233.1	[Transcript info]	[Exon info]	[Peptide info]



e!Ensembl Human ExportView

Ensembl release 48 - Dec 2007

HOME · BLAST · BIOMART · SITEMAP [HELP](#)

Your Ensembl

- Login or Register
- About User Accounts

Chromosome 17
38,449,840 - 38,530,994

- View of Chromosome 17
- Graphical view
- Graphical overview
- Resequencing alignment
- View alignment with ...
- View alongside ...
- View Syntenic regions ...
- View region at UCSC
- View region at NCBI

Export data

- Export from region...
- Export Gene info in region
- Export SNP info in region
- Export Vega info in region

Select region/feature to Export

Choose at least one feature to export. Features must map to the current Ensembl Golden tile path.
Please note we will not export more than 5Mb.

Region

Chromosome name/fragment 17

From (type): Base pair 38449840 *

To (type): Base pair 38530994

Context

Bp upstream (to the left)

Bp downstream (to the right)

Output

Output FASTA sequence

Continue >>

Fields marked with * are required

ExportView gives
you access to
sequence data

<http://www.ncbi.nlm.nih.gov/mapview/>



NCBI Map Viewer

Map Viewer Home

Help

The Map Viewer provides a wide variety of genome mapping and sequencing data. [More..](#)

Search

Search:

for:

Go

Tools Legend

- Search or Browse the Genome
- BLAST
- Genome Resources page

News

Annotation update plans for Human Genome build 36 Sep 30, 2007
NCBI is planning to update its annotation of the human genom... [more](#)

[Show all](#)

Related Resources



- [NCBI Home](#)
- [NCBI Web Search](#)
- [NCBI Site map](#)
- [Genome Biology](#)
- [Taxonomy](#)
- [Entrez \(Global Query\)](#)
- [BLAST](#)
- [Map Viewer FTP](#)

Small Genomes

- [Bacteria](#)
- [Organelles](#)

▼ Vertebrates (15)			
▼ Mammals (13)			
▼ Primates (3)			
Scientific name	Common name	Build	Tools
<i>Homo sapiens</i>	human	Build 36.2	
		Build 35.1	
<i>Macaca mulatta</i>	rhesus macaque	Build 1.1	
<i>Pan troglodytes</i>	chimpanzee	Build 2.1	
▼ Rodents (2)			
Scientific name	Common name	Build	Tools
<i>Mus musculus</i>	laboratory mouse	Build 37.1	
		Build 36.1	
<i>Rattus norvegicus</i>	rat	RGSC v3.4	
▶ Monotremes (1)			
▶ Marsupials (1)			
▶ Other Mammals (6)			
▶ Other Vertebrates (2)			
▶ Invertebrates (8)			
▶ Protozoa (7)			
▶ Plants (42)			
▼ Fungi (17)			
Scientific name			
<i>Aspergillus clavatus</i>			
<i>Aspergillus fumigatus</i>		Build 2.1	
<i>Aspergillus niger</i>		Build 1.1	
<i>Candida glabrata</i>		Build 1.1	
<i>Cryptococcus neoformans</i>		Build 2.1	
<i>Debaryomyces hansenii</i>		Build 1.1	

Two builds of human;
Note many genomes
available



NCBI Map Viewer

PubMed
Nucleotide
Protein
Genome
Gene
Structure
PopSet
Taxonomy
Help

Map Viewer

Map Viewer Home

Map Viewer Help

Human Maps Help

Release Notes

NCBI Resources

Genome Project

TaxPlot

Consensus CoDing Sequence (CCDS)

Human Genome Resources

NCBI Handbook

RefSeq

Whole Genome Association (WGA)

Organism Data in GenBank

EST

Genomic

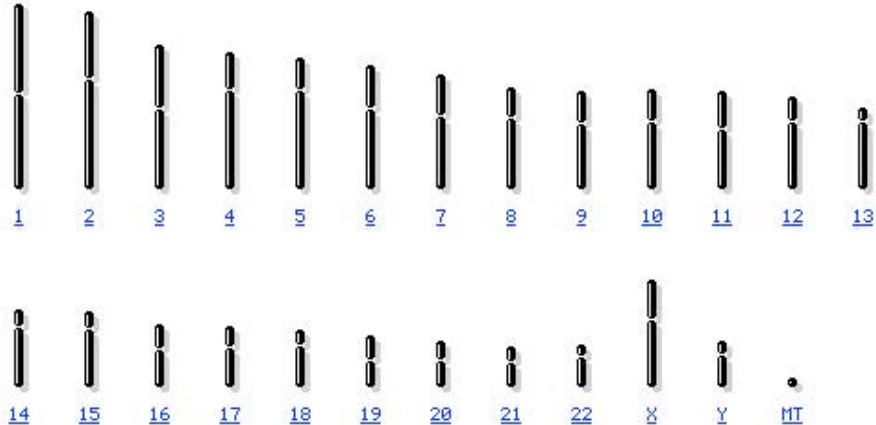
mRNA

Protein

Homo sapiens (human) genome view

[Build 36.2 statistics](#) [Switch to previous build](#)

[BLAST search the human genome](#)



Lineage: [Eukaryota](#); [Metazoa](#); [Chordata](#); [Craniata](#); [Vertebrata](#); [Euteleostomi](#); [Mammalia](#); [Eutheria](#); [Euarchontoglires](#); [Primates](#); [Haplorrhini](#); [Catarrhini](#); [Hominidae](#); [Homo](#); [Homo sapiens](#)

September 2006: NCBI released an annotation update for the human genome (NCBI Build 36.2); this update does not change the genome assembly. The previous version of the genome assembly, [NCBI Build 35.1](#), can still be accessed for Map Viewer display and for BLAST. For additional information about changes, statistics, and the status of the CCDS project please refer to:

- [Release Notes](#)
- [Statistics](#)
- [CCDS Project](#)

The NCBI Map Viewer provides graphical displays of features on the human genome sequence assembly as well as



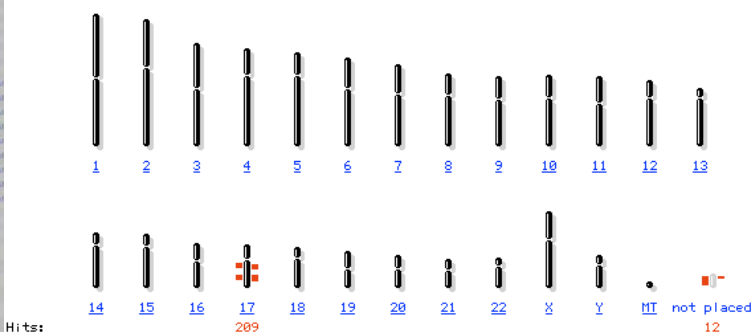
NCBI Map Viewer

PubMed Nucleotide Protein Genome Gene Structure PopSet Taxonomy Help
 Search for on chromosome(s) assembly Find

Homo sapiens (human) genome view

Build 36.2 statistics [Switch to previous build](#)

[BLAST search the human genome](#)



Hits:

Search results for query "BRCA1": 221 hits

Hits shown: 1 - 100 [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) [11](#) [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#) [MT](#) [not placed](#)

Chr Assembly Match

17 reference

[all matches](#)

Neighbor of **Brca1** gene 1
BRCA1 interacting protein C-terminal helicase 1
 Neighbor of **Brca1** gene 1
 Neighbor of **BRCA1** gene 2 (9 hits)
 Neighbor of **BRCA1** gene 1 (2 hits)
BRCA1 interacting protein C-terminal helicase 1
 Neighbor of **BRCA1** gene 1
 Neighbor of **BRCA1** gene 1 (2 hits)
BRCA1 interacting protein C-terminal helicase 1

Map element

Type

Maps

[Rn.94975](#) Rn_EST_C1
[Mm.186143](#) Mm_EST_C1
[Mm.784](#) Mm_EST_C1
[Hs.559259](#) Hs_EST_C1
[Hs.546264](#) Hs_EST_C1
[Hs.532799](#) Hs_EST_C1
[Hs.373818](#) Hs_EST_C1
[Hs.277721](#) Hs_EST_C1
[Gga.17801](#) Gga_EST_C1

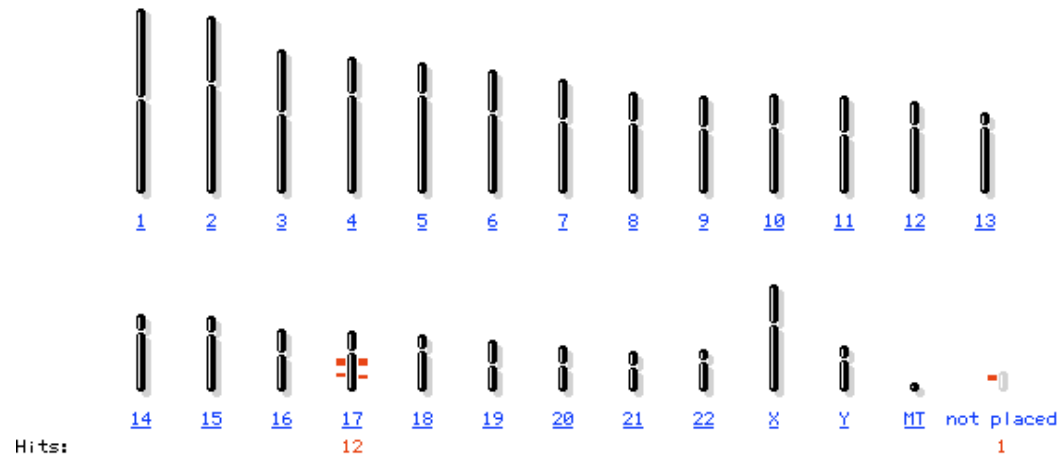
Quick Filter

☒ Gene
☐ Transcript :
☐ all
☐ RefSeq
☐ STS
☐ Unigene

Quick Filter
✓ Gene

Homo sapiens (human) genome view

Build 36.2 statistics [Switch to previous build](#)



Search results for query "BRCA1 AND gene[obj_type]": 13 hits

Chr	Assembly	Match	Map element	Type	Maps
17	reference	all matches			
		similar to neighbor of BRCA1 gene 1	LOC728560	Gene	Genes cyto Genes seq
		BRCA1P1 : like BRCA1	BRCA1P1	Gene	Genes cyto Genes seq
		BRCA1 -interacting protein 1	BRIP1	Gene	Genes cyto Genes seq
		neighbor of BRCA1 gene 2	NBR2	Gene	Genes cyto Genes seq
		neighbor of BRCA1 gene 1	NBR1	Gene	Genes cyto Genes seq
		BRCA1 : breast cancer 1, early onset	BRCA1	Gene	Genes cyto Genes seq
		BRCA1 : ENSG00000012048	BRCA1	GENE	ensGenes
17	Celera	all matches			
		BRCA1P1 : like BRCA1	BRCA1P1	GENE	Genes seq
		BRCA1 -interacting protein 1	BRIP1	GENE	Genes seq
		neighbor of BRCA1 gene 2	NBR2	GENE	Genes seq
		neighbor of BRCA1 gene 1	NBR1	GENE	Genes seq
		BRCA1 : breast cancer 1, early onset	BRCA1	GENE	Genes seq
17: not placed	reference	similar to neighbor of BRCA1 gene 1	LOC727732	GENE	Genes seq

Human genome
overview page (Build
36.2)
Human genome
overview page (Build
35.1)

[Map Viewer Home](#)

[Map Viewer Help](#)
[Human Maps Help](#)
[FTP](#)

[Data As Table View](#)

[Maps & Options](#)

[Compress Map](#) ☐

Region Shown:

38,389K

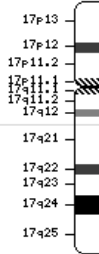
38,592K

[Go](#)

[out](#)
[zoom](#)
[in](#)

You are here:

[Ideogram](#)



☒ default
☐ master

Master Map: Genes On Sequence

[Summary of Maps](#)

[Maps & Options](#)

Region Displayed: 38,389K-38,592K bp

[Download/View Sequence/Evidence](#)

Hs Uni6	Genes_seq	Symbol	Links	E	Cyto	Description
Hs.317403 Unknown Unknown Unknown Hs.634952 Unknown Unknown Hs.175437 Unknown Unknown Unknown Hs.514193 Hs.632255 Hs.514196 Unknown Hs.603111	38390K 38400K 38410K 38420K 38430K 38440K 38450K 38460K 38470K 38480K 38490K 38500K 38510K 38520K 38530K 38540K 38550K 38560K 38570K 38580K 38590K	RUNDC1 RPL27 IFI35 VAT1 RND2 RPL21P4 BRCA1 NBR2 BRCA1P1 NBR1	HGNC sv pr dl ev mm hm sts OMIM HGNC sv pr dl ev mm hm sts OMIM HGNC sv pr dl ev mm hm sts OMIM HGNC sv pr dl ev mm hm sts OMIM HGNC sv pr dl ev mm hm sts HGNC sv dl ev mm OMIM HGNC sv pr dl ev mm hm sts HGNC sv pr dl ev mm hm sts OMIM HGNC sv pr dl ev mm hm sts HGNC sv dl ev mm OMIM HGNC sv pr dl ev mm hm sts	best RefSeq best RefSeq best RefSeq best RefSeq best RefSeq best RefSeq best RefSeq best RefSeq best RefSeq best RefSeq	17q21.31 17q21.1-q21.2 17q21 17q21 17q21 17q21 17q21 17q21 17q21 17q21.31	RUN domain containing 1 ribosomal protein L27 interferon-induced protein 35 vesicle amine transport protein 1 homolog (T call Rho family GTPase 2 ribosomal protein L21 pseudogene 4 breast cancer 1, early onset neighbor of BRCA1 gene 2 BRCA1 pseudogene 1 neighbor of BRCA1 gene 1

Two tasks

- Can you figure out how to LinkOut to the OMIM and/or Homologene entries for BRCA1?
- Can you figure out how to download the genomic sequence for the BRCA1 region?

Credits

- UCSC Genome Browser

<http://genome.cse.ucsc.edu/>

- Ensembl Genome Browser

<http://www.ensembl.org/index.html>

- NCBI MapViewer

<http://www.ncbi.nlm.nih.gov/mapview/index.html>