





MICB 405 Bioinformatics Mini-Lab #3 – Genome Browsers

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Download Artemis to Desktop

http://www.sanger.ac.uk/Software/Artemis/v9/

Google: "artemis v9"

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Feedback / Help		WWW	: Artemis V9 f	or Windows				

Objectives

- By the end of today's lecture:
 You will practice using four different genome browsers.
 - You will navigate the genomic region for BRCAI using three different genome browsers.
 - ✓ You will download genomic data for BRCAI
 - You will learn about the photosynthetic gene cluster of Rhodobacter sphaeroides using Artemis

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

UCSC Genome Bioinformatics

Genome Browser

ENCODE

Blat

Click on

Browser

link

Genome

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



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 <u>Genome Browser</u> zooms and scrolls over chromosomes, showing the work of annotators worldwide. The <u>Gene Sorter</u> shows expression, homology and other information on groups of genes that can be related in many ways. <u>Blat</u> quickly maps your sequence to the genome. The <u>Table Browser</u> provides convenient access to the underlying database. <u>VisiGene</u> lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns. <u>Genome Graphs</u> 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 (<u>CBSE</u>) at the University of California Santa Cruz (<u>UCSC</u>). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our <u>public mailing list</u>. To view the results of the Genome Browser users' survey we conducted in May 2007, click <u>here</u>.

Proteome	News Archives ►
Browser	To receive announcements of new genome assembly releases, new software features, updates and
Utilities	training seminars by email, subscribe to the genome-announce mailing list.
Downloads	8 Jan. 2008 - Additional Job Opening with UCSC Genome Browser Project

Gene Sorter Home Genomes Blat Tables 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 assembly position or search term image width genome BRCA1 620 Human May 2004 🔽 Vertebrate ~ 4 submit Click here to reset the browser user interface settings to their del configure tracks and display add your own custom tracks clear About the Human May 2004 (hg17) assembly (sequences) Search for The May 2004 human reference sequence is based on NCBI Build 35 and was produced by the International Human Genome Sec Sample position queries BRCAI; A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS m chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples Note sample human genome. See the User's Guide for more information. queries 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
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The Search Results

Known Genes

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BRCA1	(NM	007305)	at	chr17	:3844	9840-	-38530	994	-	breast	cancer	1,	early	onset	isoform	
BRCA1	(NM	007304)	at	chr17	:3844	9840-	-38530	994	-	breast	cancer	1,	early	onset	isoform	3
BRCA1	(NM	007303)	at	chr17	:3844	9840-	-38530	994	-	breast	cancer	1,	early	onset	isoform	<
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BRCA1	(NM	007301)	at	chr17	:3844	9840-	-38530	994	-	breast	cancer	1,	early	onset	isoform	E
BRCA1	(NM	007300)	at	chr17	:3844	9840-	-38530	994	-	breast	cancer	1,	early	onset	isoform	5
BRCA1	(NM	007299)	at	chr17	:3844	9840-	-38530	994	-	breast	cancer	1,	early	onset	isoform	1
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- Many BRCA1 isoforms
 - \checkmark All located on chr 17
 - \checkmark same chr coordinates
 - \checkmark different gene structures

Non-Human RefSeq Genes



Alias of STS Marker

SECAl at chr7:34489430-34689690 - (AFR067XA9 I DNA Court Double

IIuman	Augueo	micina	Dearen	results

0002999	-	Nono	sapiens BRCA1/BRCA2-containing complex, subunit 3, mFNA (cINA clone MGC:3961 IMAGE:2621917), complete cds.
C006540	- 1	Homo	sapiens BRCk1/BRCk2-containing complex, subunit 3, mRNA (cDNA clone MGC:2329 IMAGE:2621917), complete cds.
76638 -	E	menan	BRC&1-associated RING domain protein (B&RD1) mRNA, complete cds.
64805 -	Re	000 8	apiens Brcal-deltallb (Brcal) mRNA, complete cds.
	-	Homo	sapiens SRCA1 interacting protein C-terminal helicase 1, mRNA (cDNA clone MGC:126521 IMAGE:8068978), complete c
C101474	-	Homo	sapiens BRCA1 interacting protein C-terminal helicase 1, mRNA (cINA clone MGC:126523 IMAGE:8068980), complete c
		Homo	sapiens breast cancer 1, early onset, mRNA (cDNA clone IMAGE:3686198), partial cds.
	-	Homo	sapiens breast cancer 1, early onset, mRNA (cDNA clone IMAGE:6181860), complete cds.
	-	Homo	sapiens breast cancer 1, early onset, mRNA (cDNA clone IHAGE:6042052), partial cds.
	-	Homo	sapiens breast cancer 1, early onset, mRNA (cDNA clone NGC:131629 INAGE:7961446), complete cds.
1597810	1	Homo	sapiens chromosome 17 neighbor of SFCA1 gene 2 (NSF2) mFNA, complete cds, alternatively spliced.
Y438031	1	Homo	sapiens BRC&1/BRC&2-containing complex subunit 45 (BRE) mBNA, complete cds.
Y436030	-	Homo	sapiens BRCA1/BRCA2-containing complex subunit 36 (BRCC36) mRMA, complete cds.
X223405	-	Homo	sapiens mRNA for SRCA1 associated RING domain 1 variant, clone: FCC109A11.
F464935		Homo	sapiens cofactor of BRC&1 (COBR&1) HRN&, complete cds.
F360549	-	Homo	sapiens BRCA1-binding helicase-like protein BACH1 mRNA, complete ods.
	-	Homo	sapiens putative SRCA1-interacting protein (SRIP1) mRNA, partial cds.
2045583	-	Homo	sapiens SFCA1 associated protein 1 (BAP1) mFNA, complete cds.
1035620	1	HORO	Sapiens skial-associated protein 2 (skar2) mkma, complete cds.
		nono	Saplens maa for packi associated protein variant protein.
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	-	Moreo	saminas cofactor of BECAL MENA (CDNA clone NGC:19805 INAGE:1991719), complete cds.
	2	Homo	sablens meichhor of SPCA1 cene 1, mPNA (CDNA clone BCC:1377 INACE:2089212), complete cds.
0000924	12	Homo	sapiens meighbor of BRCA1 gene 2, mBNA (cDNA clone MCC15031 INAGE13496931), complete cds.
2094499		Horeo	samiens dNA FL/37180 fis, close BRAL72001838, highly similar to Mono samiens BECAL associated protein 1 (BAPL)
		Homo	sapiens cDNA FLJ35406 fis. clone SENSH2007429, highly similar to Homo sapiens SECA1 associated protein 1 (BAP1)
F274503	-	Hoteo	sapiens breast and ovarian cancer susceptibility (BRCA1) useudomene, partial mRNA sequence.
	-	Homo	capiens meighbor of BRCA1 gene 2, mRNA (CDNA clone INAGE:4026050], with apparent retained intron.
	12	Homo	sapiens cofactor of BRCA1, mENA (CDNA cione IMAGE:4554322).
C012847	-	Homo	sapiens meighbor of BRCA1 gene 1, mBNA (cINA clone INAGE:3858519), with apparent retained intron.



Tasks

- What genes are on either side of BRCA1 on chr 17?
- Can you figure out how to download the genomic sequence for the BRCAI region?
- Can you figure the display to add/remove tracks that are (or are not) of interest to you?



Home Genomes Genome Browser Blat Tables Gene Sorter PCR FAQ Help

Get DNA in Window

Get DNA for

Position chr17:38,451,220-38,530,831

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 0 extra bases upstream (5') and 0 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)

get DNA extended case/color options

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

DNA link Download Sequence

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•	Variation and Repeats	refresh
+	Pilot ENCODE Regions and Gene	refresh

http://www.ensembl.org/

CEnsembl

Login / Register | BLAST/BLAT | BioMart | Docs & FAQs

2] -







Annotation

Since release 38 (April 2006) the gene annotation presented has been a combined Ensembl-<u>Havana</u>, 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 browsers have come together to produce a common set of identifiers where CDS annotations of transcripts can be agreed and these identifiers are also shown.

More information about the <u>CCDS project</u>.

The ENCODE (ENCyclopedia Of DNA Elements) project aims to find functional elements in the human genome.

• More information about the ENCODE resources at Ensembl.

Vega* Additional manual annotation of this genome can be found in Vega

Ensembl release 52 - Dec 2008 © WTSI / EBI

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<i>C</i> Ensembl	
Home > Human Genome	Login / Register BLAST/BLAT BioMart Docs & FAQs
Search Ensembl	Ensembl text search
 Feature type (22) Domain (3) Homo sapiens (3) Gene (18) Homo sapiens (18) Marker (1) Homo sapiens (1) Species (1) Homo sapiens (22) Domain (3) Gene (18) Marker (1) 	brca1 corporate/tree:"Top/Species/Homo sapiens" corp Search Your query matched 22 entries in the search database. Viewing hits 1-10 1 2 3 Ensembl Marker: BRCA1 A marker with 2 synonyms (262743 BRCA1) Source: e52; Feature type: Marker; Homo sapiens; Species: Homo sapiens; Marker; Ensembl protein_coding Gene: ENSG0000012048 (HGNC (automatic): BRCA1) [Region in detail] Ensembl protein_coding gene ENS G0000012048 has 10 transcripts: ENST00000309486, ENST00000346315, ENST00 10.51666, ENST000003522093, ENST00000353540, ENST00000354071, ENST00000346315, ENST00 10.51666, ENST00000322682, ENST000003283691, preprinted pontider:
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Tasks

- Explore the information presented in the Gene Summary views.
 - Can you figure out how to visualize the alternatively spliced isoforms for BRCAI?
 - What can you find out about known variations in this gene?
- Using the Location Based Displays, can you figure out how to download the genomic sequence for the BRCA1 region?





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Home > Human Location: 17:38,449,84	10-38,530,994 Gene: BRC	Login / Register BLAST/BLAT BioMart Docs & FAQs
Location-based	Chromosome 17: 38,4	49,840-38,530,994
- Whole genome	chromosome 17	p12 p11.2 q12 q22 q25.3
- Chromosome summary		Export image
- Region in detail	« Region overview	Region in detail he!p Genomic alignments »
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On your own....

Follow the next few slides to explore the genomic region for BRCA1 using the

NCBI MapViewer system.

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

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



<u>Homo sapiens (human)</u> genome view Build 36.2 statistics <u>Switch to previous build</u>



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

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17	reference	all matches			
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		BRCA1-interacting protein 1	BRIP1	Gene	Genes cyto Genes seq
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		neighbor of BRCA1 gene 1	NBR1	GENE	Genes seq
		BRCA1 : breast cancer 1, early onset	BRCA1	GENE	Genes seq
17:not plz_ed	reference	similar to neighbor of BRCA1 gene 1	LOC727732	GENE	Genes seq



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?



Artemis genome browser for prokaryotic genomes

Download Artemis to Desktop

http://www.sanger.ac.uk/Software/Artemis/v9/

Google: "artemis v9"

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Rhodobacter sphaeroides

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The photosynthetic co-complex



Co-complex Structure



The photosynthetic gene cluster



Download and Extract Sequence File for Chromosome I

I. Go to <u>www.rhodobacter.org</u> 2. Click 'Finished Genome + GenBank files'



Download and Extract Sequence File for Chromosome I

3. Find 'rsph_chrl.zip' file.

4. Right click on file and then choose 'Extract to rsph_chrl\'



Start Artemis and Load Chromosome I File

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Top Pane: Overview



3 Reverse strand reading frames

Middle Pane: Sequence Detail



Four Tasks

- Find the *pufM* and *pufL* genes of the reaction center (part of the *puf* operon)
- How many nucleotides overlap between the coding sequence of these genes?
- Are the genes in the same frame?
- Export a picture of the *puf* operon (*pufQBALMX*)

Bottom Pane: Feature List

CDS	1976931 1978313	COGO665: Glycine/D-amino acid oxidases (DadA).
CDS	1978340 1980286 c	Experimental evidence in Rhodobacter capsulatu
CDS	1980460 1980708 c	Citation: McGlynn P, Hunter CN, Jones MR. (199
CDS	1980721 1981647 c	pfam: Photosynthetic reaction center protein,
CDS	1981640 1982488 c	pfam: Photosynthetic reaction center protein,
CDS	1982612 1982788 c	pfam: Antenna complex alpha/beta subunit,
CDS	1982802 1982951 c	pfam: Antenna complex, alpha/beta subunit
CDS	1982953 1983015 c	Citation: Gong L, Kaplan S. (1996) Microbiolog
CDS	1083082 1083315 -	Citation: Rever CF Marre RI (1088) Droc Matl

Links

• 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

Artemis

http://www.sanger.ac.uk/Software/Artemis/