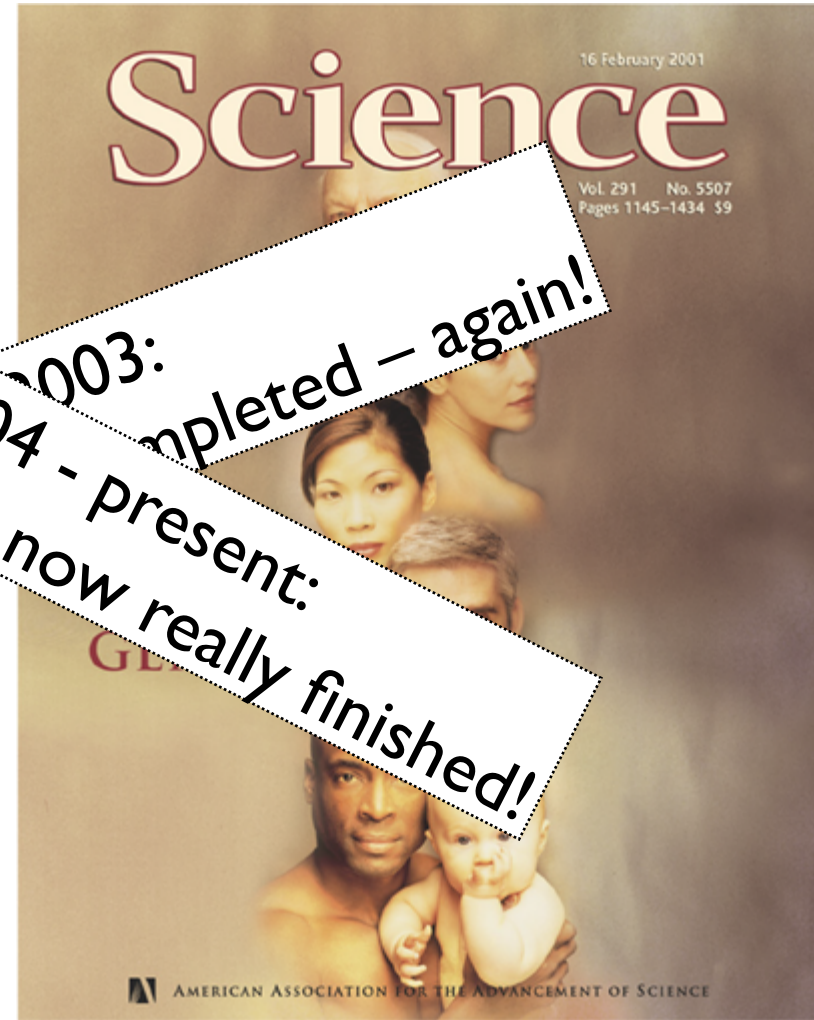
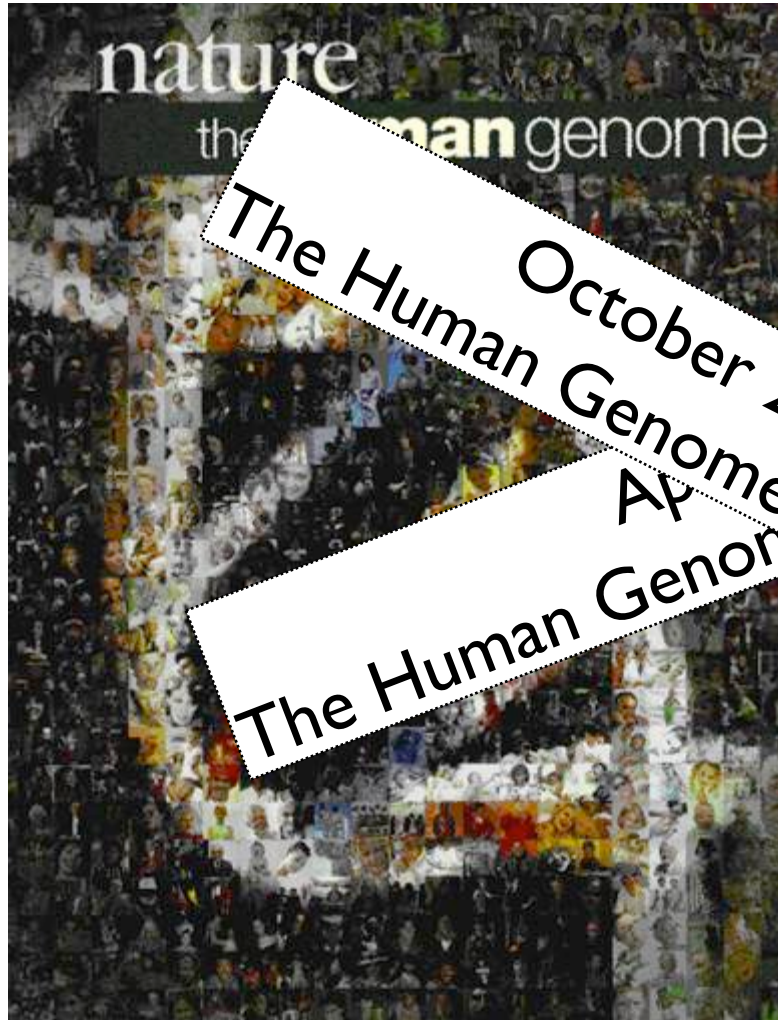


Genome Browsers

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



The Human Genome Project

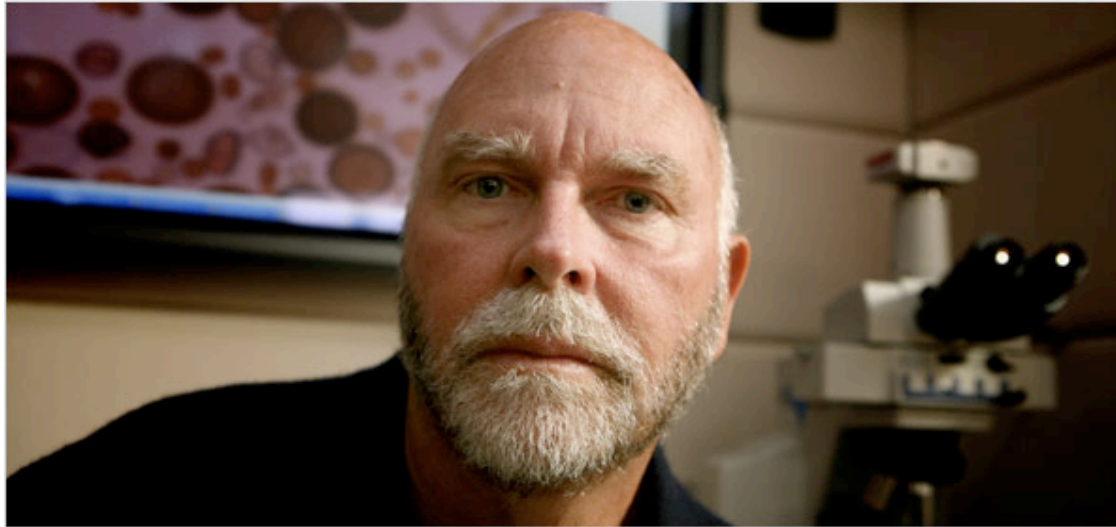


Public HGP

Celera Genomics

February 2001: Completion₂ of the Draft Human Genome

In the Genome Race, the Sequel Is Personal



Thor Swift for The New York Times

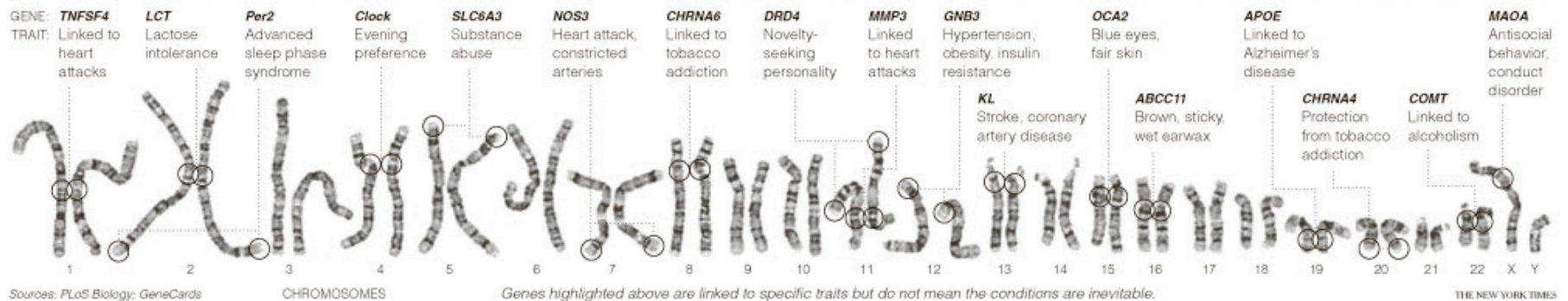
A team led by J. Craig Venter, above, has finished the first mapping of a full, or diploid, genome, made up of DNA inherited from both parents. The genome is Dr. Venter's own.

© MICHAEL MARR

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.





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Maps Canada

[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 Beany 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) 221-4240 - 2.8 km E

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Let's Look at the Human Genome...

UCSC Genome Browser on Human May 2004 Assembly

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

position chr7:127,471,196-127,495,720 jump clear size 24,525 bp. configure

chr7 (q32.1)

Base Position 127475000 127480000 127485000 127490000 127495000

STS Markers on Genetic (blue) and Radiation Hybrid (black) Maps

Gap Locations

Known Genes (Nov 22, 04) Based on SWISS-PROT, TrEMBL, mRNA, and RefSeq

LEP

Consensus CDS

RefSeq Genes

AceView Gene Models With Alt-Splicing

Human mRNAs from GenBank

U43653

BC060838

BC069323

BC069452

BC069527

AF008123

D49487

U18915

Spliced ESTs

Human ESTs That Have Been Spliced

Hu/Chimp/Mouse/Rat/Dog/Chick/Fugu/Zfish Multiz Alignments & Conservation

Conservation

chimp

dog

mouse

rat

chicken

fugu

zebrafish

SNPs

Simple Nucleotide Polymorphisms (SNPs)

Repeating Elements by RepeatMasker

RepeatMasker

move start < 2.0 >

Click on a feature for details. Click on base position to zoom in around cursor. Click on left mini-buttons for track-specific options.

move end < 2.0 >

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

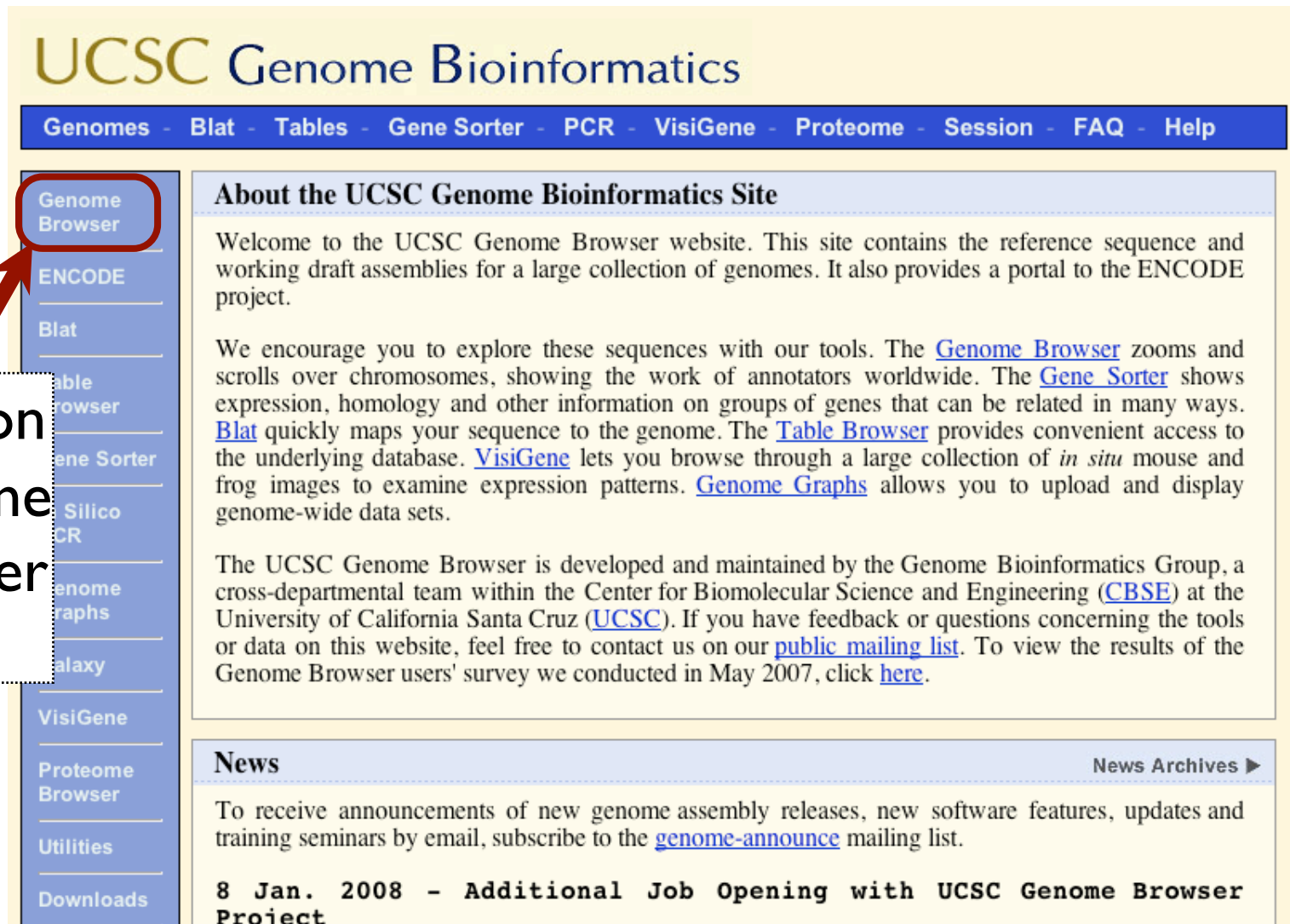
- Common features:

- ✓ 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

Table Browser

Gene Sorter

Silico CR

Genome Graphs

Galaxy

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

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 | genome | assembly | position or search term | image width | |
| Vertebrate | Human | May 2004 | BRCA1 | 620 | submit |
| <p>Click here to reset the browser user interface settings to their defaults.</p> <p> <input type="button" value="add your own custom tracks"/> <input type="button" value="configure tracks and display"/> <input type="button" value="clear position"/> </p> | | | | | |

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 marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of queries for 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 |
| homologous pseudogene | Lists mRNAs for pseudohomologous genes |

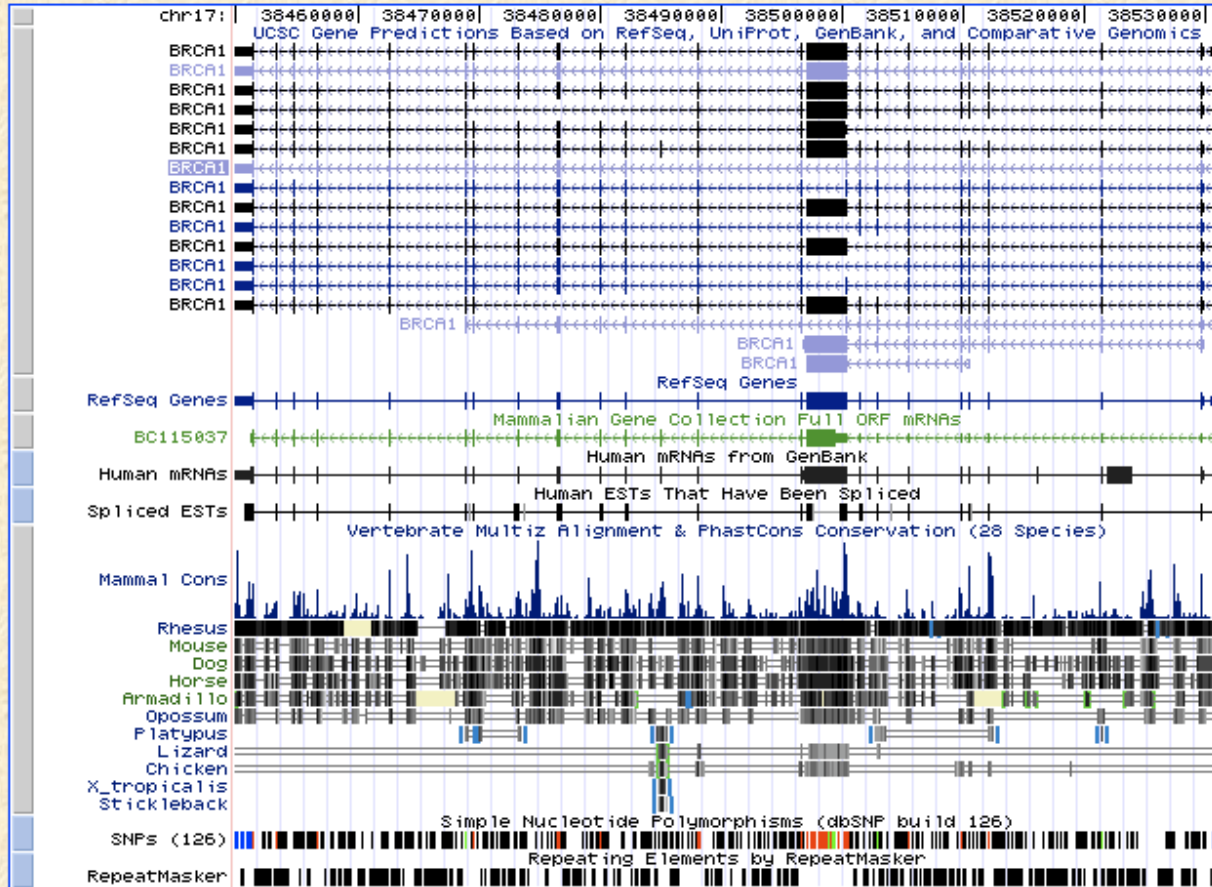
Search for
BRCA1;
Note sample
queries

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

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?
- Can you figure the display to add/remove tracks that are (or are not) of interest to you?

Home Genomes Blat Tables Gene Sorter PCR **DNA** Convert Ensembl NCBI PDF/PS Help

UCSC Genome Browser on Human May 2004 Assembly

move <<< << < > >> >>> zoom in 1x 3x 10x base zoom out 1.5x 3x 10x

position/search chr17:38,423,783-38,543,782 jump clear size 120,000 bp. configure

chr17 (q21.31) p12 p11.2 q11.21 q12 22 q23.2 q25.3

Click on a feature for details. Click on base position

Zoom in
Zoom out

DNA link
Download
Sequence

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".

collapse all Use drop-down controls below and press refresh to alter tracks displayed. expand all
 Tracks with lots of items will automatically be displayed in more compact modes.

- Mapping and Sequencing Tracks refresh

| | | | | | |
|--|---|---------------------------------------|---|--|---------------------------------------|
| Base Position dense ▾ | Chromosome Band hide ▾ | STS Markers hide ▾ | FISH Clones hide ▾ | Recomb Rate hide ▾ | Map Contigs hide ▾ |
| Assembly hide ▾ | Gap hide ▾ | Coverage hide ▾ | BAC End Pairs hide ▾ | Fosmid End Pairs hide ▾ | GC Percent hide ▾ |
| Short Match hide ▾ | Restr Enzymes hide ▾ | | | | |

+ Phenotype and Disease Associations refresh

- Genes and Gene Prediction Tracks refresh

| | | | | | |
|---|--|---------------------------------------|--------------------------------------|---|---|
| UCSC Genes pack ▾ | Old UCSC Genes hide ▾ | Alt Events hide ▾ | CCDS hide ▾ | RefSeq Genes dense ▾ | Other RefSeq hide ▾ |
| MGC Genes pack ▾ | ORFeome Clones hide ▾ | TransMap... hide ▾ | Vega Genes hide ▾ | Ensembl Genes hide ▾ | AceView Genes hide ▾ |
| SIB Genes hide ▾ | N-SCAN hide ▾ | CONTRAST hide ▾ | SGP Genes hide ▾ | | |
| Exoniphy hide ▾ | Augustus hide ▾ | RNA Genes hide ▾ | ACEScan hide ▾ | | |
| Pos Sel Genes hide ▾ | | | | | |

Drop down controls
configure the data shown
in the image above

+ mRNA and EST Tracks refresh

+ Expression refresh

+ Regulation refresh

+ Comparative Genomics refresh

+ Variation and Repeats refresh

+ Pilot ENCODE Regions and Genes refresh

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

Search Ensembl

Search: for




e.g. human gene BRCA2 or rat X:100000..200000 or insulin

Browse a Genome

The Ensembl project produces genome databases for vertebrates and other eukaryotic species, and makes this information freely available online.

Click on a link below to go to the species' home page.

Popular genomes [\(Log in to customize this list\)](#)







-  **Human**
NCBI36
-  **Mouse**
NCBIM37
-  **Zebrafish**
ZFISH7

All genomes

-- Select a species --

New to Ensembl?

Did you know you can:

-  [Add custom tracks](#) using our new Control Panel
-  [Upload your own data](#) and save it to your Ensembl account
-  [Search for a DNA or protein sequence](#) using BLAST or BLAT
-  [Fetch only the data you want](#) from our public databases
-  [Download our data](#) in FASTA, MySQL and other formats
-  [Mine Ensembl with BioMart](#) and export sequences or tables in text, html, or Excel format

Still got questions? [Try our FAQs](#)

NEW! **The new Ensembl website**
We've made some changes to our site, to make it faster and easier to use. [Find out more about what we've changed and why!](#)

Click on Human

e!Ensembl
Home > Human

Location: 6:131,533,782-131,677,240 | Gene: AKAP7 | Transcript: AKAP7-001

Search Ensembl, EBI or Sanger Institute

Jump from gene to location using tabs

Location-based displays

- Whole genome
- Chromosome summary
- Region overview
- Region in detail**
- Comparative Genomics
 - Genomic alignments (35)
 - Multi-species comp. (39)
 - Synteny (10)
- Genetic Variation
 - Resequencing (6)
 - Markers
 - Export location data

• Bookmark this page
• Configure this page
• Add custom data to page

Chromosome 6: 131,533,782-131,677,240

Assembly exceptions
chromosome 6

Assembly exceptions
COX
QBL

Click and drag the mouse to recentre the display

« Region overview | Region in detail | »

Cortigs

Ensembl/Havana gene

1.00 Mb Forward strand

131.20 Mb 131.50 Mb 131.80 Mb

EPB41L2 AKAP7 ARG1 CRSP3 ENPP3

About this species

- Description
 - Genome Statistics
 - Assembly and Genebuild
 - Top 40 InterPro hits
 - Top 500 InterPro hits
 - What's New
- Sample entry points
 - Karyotype
 - Location (AL032821.2)
 - Gene (BRCA2)
 - Transcript (FOXP2-203)

- Configure this page
- Add custom data to page
- Export data
- Bookmark this page

Search Ensembl Human

Search for:
 e.g. **gene BRCA2** or **AL032821.2.1.143563** or **muscular dystrophy**

Description

[Assembly and Genebuild >](#)

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 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 [CCDS project](#).

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

- More information about the [ENCODE resources](#) at Ensembl.



Additional manual annotation of this genome can be found in [Vega](#)

Search Ensembl

- 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)

- [Configure this page](#)
- [Add custom data to page](#)
- [Export data](#)
- [Bookmark this page](#)

Ensembl text 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; **Species:** Homo sapiens; Marker;**Ensembl protein_coding Gene:** **ENSG00000012048 (HGNC (automatic): BRCA1)** [\[Region in detail\]](#)

Ensembl protein_coding gene ENSG00000012048 has 10 transcripts: ENST00000309486, ENST00000346315, ENST00000346316, ENST00000352493, ENST00000353540, ENST00000354071, ENST00000357654, ENST00000393680, ENST00000393683, ENST00000393691, associated peptides: ENSP00000013772, ENSP000000246907, ENSP000000310938, ENSP000000312236, ENSP000000326002, ENSP000000338577, ENSP000000350283, ENSP000000377285, ENSP000000377288, ENSP000000377294 and 35 exons: ENSE000000371140, ENSE000000729436, ENSE000000865492, ENSE000000865496, ENSE000000865503, ENSE000000865520, ENSE000000865521, ENSE000000865524, ENSE000000865528, ENSE000000865546, ENSE000000865551, ENSE000000865553, ENSE000000865557, ENSE000000865565, ENSE000001297284, ENSE000001312675, ENSE000001360157, ENSE000001360198, ENSE000001360203, ENSE000001360204, ENSE000001368002, ENSE000001383775, ENSE000001383927, ENSE000001473234, ENSE000001473237, ENSE000001473240, ENSE000001473241, ENSE000001473245, ENSE000001516235, ENSE000001516237, ENSE000001516259, ENSE000001577499,

Click on
ENSG00000012048

septibility protein (RING finger protein 53) [Source:UniProtKB/Swiss-Prot;Acc:P38398]

external identifiers mapped to it:

ArrayExpress microarray focus: 204531_s_at
Affymx Microarray HCG110: 1993_s_at, 604_at
Affymx Microarray HuGeneFL: L78833_cds1_at, U64805_s_at
Affymx Microarray Human Exon 1.0 ST v2: 3722383, 3482826, 3800710, 2324530, 3722373, 3722386, 3722372, 3722385, 3722425, 3679671, 3282866
Affymx Microarray U133: 211851_x_at, g6552300_3p_a_at, g2218153_3p_a_at, 204531_s_at
Affymx Microarray U95: 1993_s_at, 604_at, 33724_at
Agilent CGH: A_14_P133777, A_14_P135846, A_14_P139703
Agilent Probe: A_32_P180603, A_32_P405851, A_23_P207400
CCDS: CCDS11458, CCDS11454, CCDS11457.1, CCDS11455.1, CCDS11459.1, CCDS11453, CCDS11458.1,

Location: 17:38,449,840-38,530,994 Gene: BRCA1

Gene: BRCA1

Gene: BRCA1 (ENSG0000012048)

Best cancer type 1 susceptibility protein (RING finger protein 53) Source: UniProtKB/Swiss-Prot P38398

Gene summary

Chromosome 17: 38,449,840-38,530,994 reverse strand.

Location

Transcripts

There are 10 transcripts in this gene: [hide transcripts](#)

| | | | |
|-----------|---------------------------------|---------------------------------|----------------|
| BRCA1-201 | ENST00000309486 | ENSP00000310938 | protein_coding |
| BRCA1-202 | ENST00000346315 | ENSP00000246907 | protein_coding |
| BRCA1-203 | ENST00000351666 | ENSP00000338007 | protein_coding |
| BRCA1-204 | ENST00000352993 | ENSP00000312236 | protein_coding |
| BRCA1-205 | ENST00000353540 | ENSP00000313772 | protein_coding |
| BRCA1-206 | ENST00000354071 | ENSP0000026002 | protein_coding |
| BRCA1-207 | ENST00000357654 | ENSP0000030283 | protein_coding |
| BRCA1-208 | ENST00000358680 | ENSP000003 | |
| BRCA1-209 | ENST0000039368 | ENSP000003 | |
| BRCA1-210 | ENST00000393691 | ENSP000003 | |

Gene Summary shows you information about the gene

click here to view genomic location

- page
- Export data
- Bookmark this page

Gene type
Prediction Method

[BRCA1](#) (HGNC (automatic))

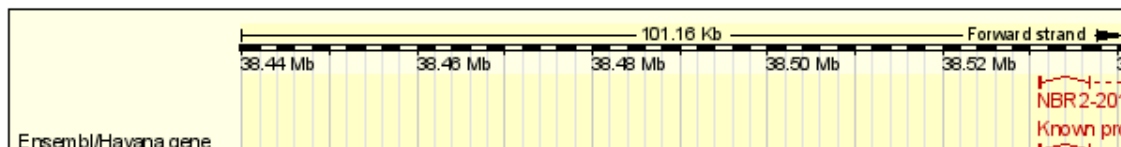
BRCC1, RNF53 [To view all Ensembl genes linked to the name [click here](#).]

This gene is a member of the Human CCDS set: [CCDS11453](#), [CCDS11454](#), [CCDS11455](#), [CCDS11456](#), [CCDS11457](#), [CCDS11458](#), [CCDS11459](#)

Known protein coding

Transcripts were annotated by the Ensembl [genebuild](#).

Transcripts



Tasks

- Explore the information presented in the Gene Summary views.
 - Can you figure out how to visualize the alternatively spliced isoforms for BRCA1?
 - 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?

- Gene: BRCA1
- Gene summary
 - Splice variants (10)**
 - Supporting evidence
 - Sequence
 - External references (15)
 - Regulation
 - Comparative Genomics
 - Genomic alignments (3)
 - Gene Tree
 - Gene Tree (text)
 - Gene Tree (alignment)
 - Orthologues (28)
 - Paralogues (0)
 - Protein families (1)
 - Genetic Variation
 - Variation Table
 - Variation Image
 - External Data
 - ID History
 - Gene history

- Configure this page
- Add custom data to page
- Export data
- Bookmark this page

Gene: BRCA1 (ENSG0000012048)

Breast cancer type 1 susceptibility protein (RING finger protein 53) [Source: UniProtKB/Swiss-Prot P](#)

Location [chr17:41,210,840-28,530,994 reverse strand.](#)

Transcripts There are 10 transcripts in this gene: [hide transcripts](#)

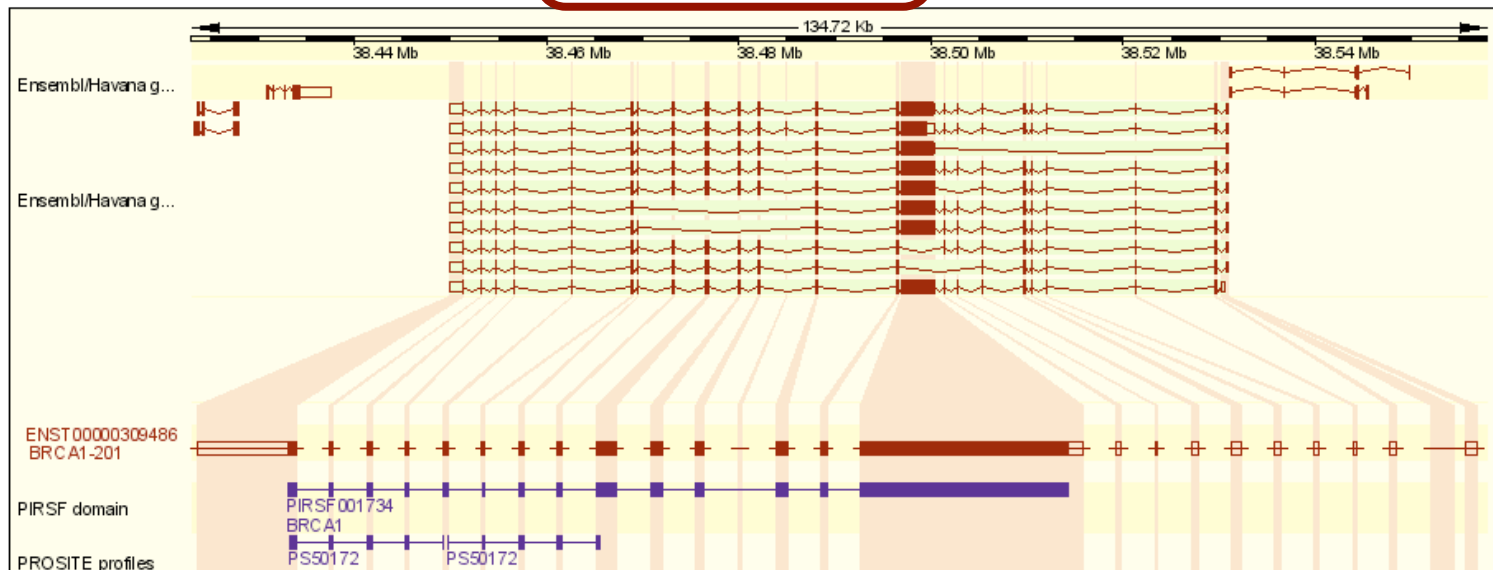
| | | | |
|---------------------------|---------------------------------|---------------------------------|----------------|
| BRCA1-201 | ENST00000309486 | ENSP00000310938 | |
| BRCA1-202 | ENST00000346315 | ENSP00000246907 | |
| BRCA1-203 | ENST00000351666 | ENSP00000338007 | |
| BRCA1-204 | ENST00000352993 | ENSP00000312236 | |
| BRCA1-205 | ENST00000353540 | ENSP00000013772 | |
| BRCA1-206 | ENST00000354071 | ENSP00000326002 | |
| BRCA1-207 | ENST00000357654 | ENSP00000350283 | protein_coding |
| BRCA1-208 | ENST00000393680 | ENSP00000377288 | protein_coding |
| BRCA1-209 | ENST00000393683 | ENSP00000377288 | protein_coding |
| BRCA1-210 | ENST00000393691 | ENSP00000377294 | protein_coding |

The Splice Variants page shows you information about the transcripts

[← Gene summary](#)

Splice variants [help](#)

[Supporting evidence →](#)



Gene: BRCA1

- Gene summary
- Splice variants (10)
- Supporting evidence
- Sequence
- External references (15)
- Regulation
- Comparative Genomics
 - Genomic alignments (3)
 - Gene Tree
 - Gene Tree (text)
 - Gene Tree (alignment)
 - Orthologues (28)
 - Paralogues (0)
 - Protein families (1)
- Genetic Variation
 - Variation Table
 - Variation Image**
- External Data
- ID History
 - Gene history

- [Configure this page](#)
- [Add custom data to page](#)
- [Export data](#)
- [Bookmark this page](#)

Gene: BRCA1 (ENSG0000012048)

Breast cancer type 1 susceptibility protein (RING finger protein 53) [Source: UniProtKB/Swiss-Prot P38398](#)

Location [Chromosome 17: 38,449,840-38,530,994 reverse strand.](#)

Transcripts There are 10 transcripts in this gene: [show transcripts](#)

« Variation Table

Variation Image [help](#)

External Data



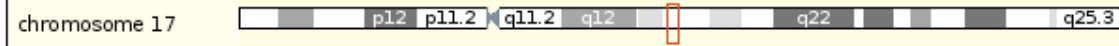
Variation Data;
configure display to
show more/less
details

Location-based displays

- Whole genome
- Chromosome summary
- Region overview
- Region in detail**
- Comparative Genomics
 - Genomic alignments
 - Synteny (10)
- Genetic Variation
 - Resequencing (6)
 - Linkage Data
- Markers

- Configure this page
- Add custom data to page
- Export data
- Bookmark this page

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

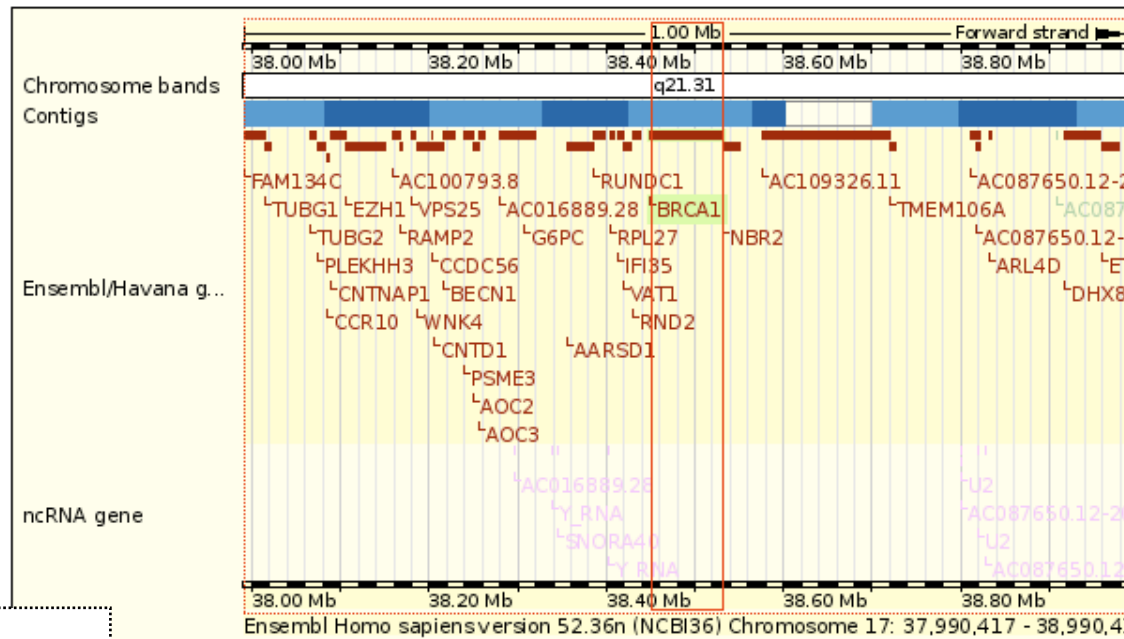


Export image

< Region overview

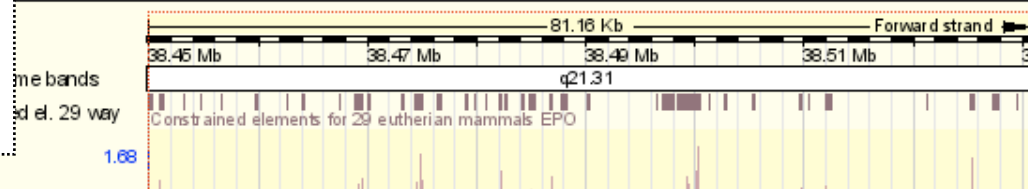
Region in detail help

Genomic alignments >



Export image

Chromosome: 17 : 38449840 - 38530994 Go>



Export options available on all pages

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

NCBI Home GenBank BLAST

Map Viewer Home > Help

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

Search

Search:

for:

Tools Legend

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

| Scientific name | Common name | Build | Tools |
|---------------------------------|------------------|----------------------------|---|
| Vertebrates (16) | | | |
| Mammals (14) | | | |
| Primates (3) | | | |
| <i>Homo sapiens</i> | human | Build 36.3 | <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> |
| | | Build 35.1 | <input type="radio"/> <input type="radio"/> <input type="radio"/> |
| <i>Macaca mulatta</i> | rhesus macaque | Build 1.1 | <input type="radio"/> <input type="radio"/> <input type="radio"/> |
| <i>Pan troglodytes</i> | chimpanzee | Build 2.1 | <input type="radio"/> <input type="radio"/> <input type="radio"/> |
| Rodents (2) | | | |
| Mus musculus | | | |
| | laboratory mouse | Build 37.1 | <input type="radio"/> <input type="radio"/> <input type="radio"/> <input type="radio"/> |
| | | Build 36.1 | <input type="radio"/> <input type="radio"/> |
| <i>Rattus norvegicus</i> | rat | RGSC v3.4 | <input type="radio"/> <input type="radio"/> <input type="radio"/> |
| Monotremes (1) | | | |
| Marsupials (1) | | | |
| Other Mammals (7) | | | |
| Other Vertebrates (2) | | | |
| Invertebrates (12) | | | |
| Protozoa (18) | | | |
| Plants (46) | | | |
| Fungi (17) | | | |
| Scientific name | | | |
| <i>Aspergillus clavatus</i> | | | |
| <i>Aspergillus fumigatus</i> | | | |
| <i>Aspergillus niger</i> | | | |
| <i>Candida glabrata</i> | | Build 1.1 | <input type="radio"/> <input type="radio"/> |
| <i>Cryptococcus neoformans</i> | | Build 2.1 | <input type="radio"/> <input type="radio"/> |
| <i>Debaryomyces hansenii</i> | | Build 1.1 | <input type="radio"/> <input type="radio"/> |
| <i>Encephalitozoon cuniculi</i> | | Build 1.1 | <input type="radio"/> <input type="radio"/> |
| <i>Eremothecium oosporii</i> | | Build 3.1 | <input type="radio"/> <input type="radio"/> |

Two builds of human; Note many genomes available

News

Annotation update released for human genome build 36 Mar 23, 2010

An annotation update for the human genome (NCBI Build 36.3) ... [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

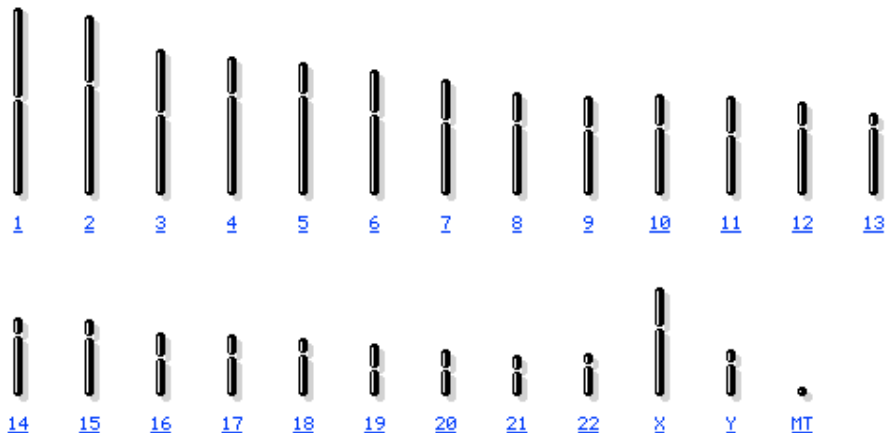
- Bacteria

[Search for](#) [on chromosome\(s\)](#)

[BLAST search the human genome](#)

- 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](#)

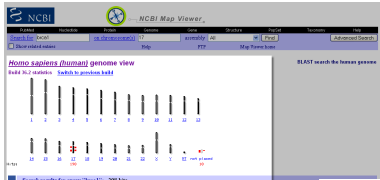


Lineage: [Eukaryota](#); [Metazoa](#); [Chordata](#); [Craniata](#); [Vertebrata](#); [Euteleostomi](#); [Mammalia](#); [Eutheria](#); [Euarchontoglires](#); [Primates](#); [Haplorrhini](#); [Catarrhini](#); [Hominidae](#); [Homo](#); [Homo sapiens](#)

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

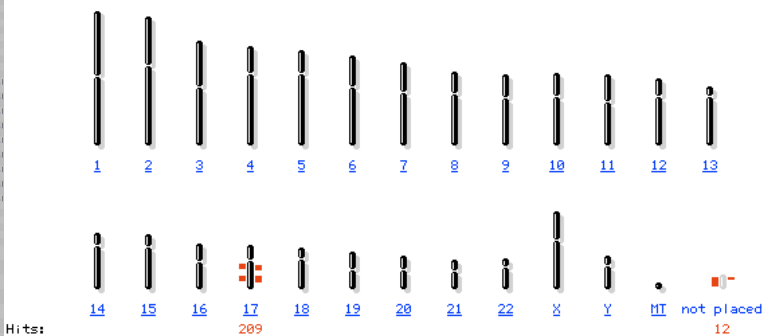


PubMed Nucleotide Protein Genome Gene Structure PopSet Taxonomy Help

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



Search results for query "BRCA1": 221 hits
Hits shown: 1 - 100

| Chr | Assembly | Match | Map element | Type | Maps |
|-----|-----------|--|---------------------------|------------|--------------------------|
| 17 | reference | all matches | | | |
| | | Neighbor of Brcal1 gene 1 | Rn.94975 | Rn_EST_C1 | Rn Un G |
| | | BRCA1 interacting protein C-terminal helicase 1 | Mm.186143 | Mm_EST_C1 | Mm Un G |
| | | Neighbor of Brcal1 gene 1 | Mm.784 | Mm_EST_C1 | Mm Un G |
| | | Neighbor of BRCA1 gene 2 (9 hits) | Hs.559259 | Hs_EST_C1 | Hs Un G |
| | | Neighbor of BRCA1 gene 1 (2 hits) | Hs.546264 | Hs_EST_C1 | Hs Un G |
| | | BRCA1 interacting protein C-terminal helicase 1 | Hs.532799 | Hs_EST_C1 | Hs Un G |
| | | Neighbor of BRCA1 gene 1 | Hs.373818 | Hs_EST_C1 | Hs Un G |
| | | Neighbor of BRCA1 gene 1 (2 hits) | Hs.277721 | Hs_EST_C1 | Hs Un G |
| | | BRCA1 interacting protein C-terminal helicase 1 | Gga.17801 | Gga_EST_C1 | Gga Un G |

Quick Filter

Gene

Transcript

all

RefSeq

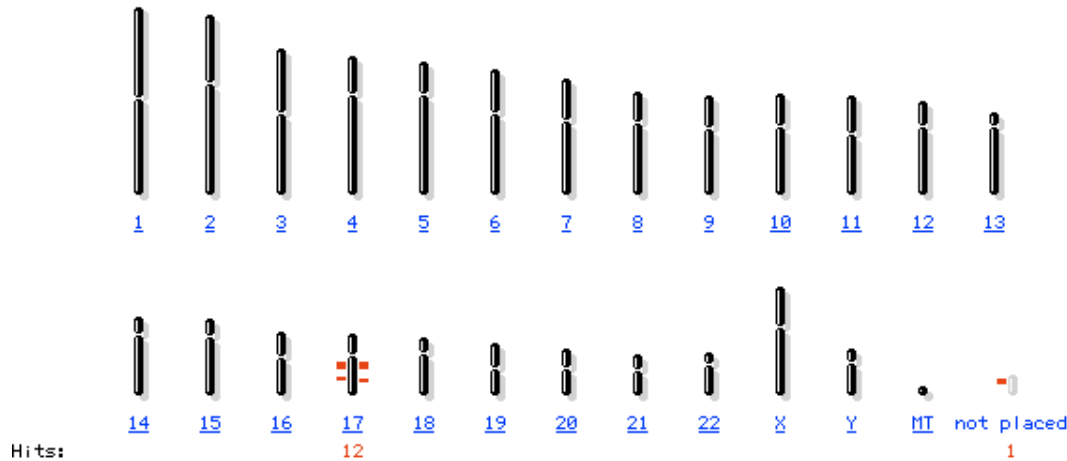
STS

Unigene

Quick Filter
✓ Gene

Homo sapiens (human) genome view

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



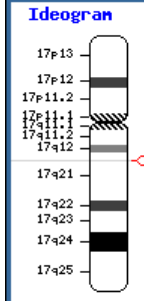
Search results for query "BRCA1 AND gene[obj_type]": 13 hits

| 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](#)
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 Human Maps Help
 FTP
 Data As Table View
[Maps & Options](#)
 Compress Map
 Region Shown:
 38,389K
 38,592K

out
 zoon
 in

You are here:



- default
- master

Master Map: Genes On Sequence

[Summary of Maps](#)

[Maps & Options](#)

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

[Download/View Sequence/Evidence](#)

| Hs UniG | Genes_seq | Symbol | Links | E | Cyto | Description |
|-----------|-----------|---------|---|-------------|---------------|--|
| Hs.317403 | | RUNDC1 | HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21.31 | RUN domain containing 1 |
| Unknown | | RPL27 | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21.1-q21.2 | ribosomal protein L27 |
| Unknown | | IFI35 | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | interferon-induced protein 35 |
| Hs.634952 | | VAT1 | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | vesicle amine transport protein 1 homolog (T cell) |
| Unknown | | RND2 | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | Rho family GTPase 2 |
| Unknown | | RPL21P4 | HGNC sv dl ev mm sts | best RefSeq | 17q21 | ribosomal protein L21 pseudogene 4 |
| Hs.194143 | | BRCA1 | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | breast cancer 1, early onset |
| Unknown | | NBR2 | HGNC sv pr dl ev mm sts | best RefSeq | 17q21 | neighbor of BRCA1 gene 2 |
| Unknown | | BRCA1P1 | HGNC sv dl ev mm sts | best RefSeq | 17q21 | BRCA1 pseudogene 1 |
| Unknown | | NBR1 | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21.31 | 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?

Human genome overview page (Build 36.2)
 Human genome overview page (Build 35.1)
[Map Viewer Home](#)
 Map Viewer Help
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 FTP
 Data As Table View
Maps & Options
 Compress Map
 Region Shown:
 38,389K
 38,592K
 out
 zoom
 in
 You are here:
Ideogram

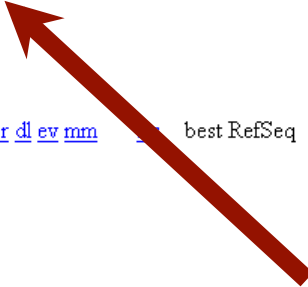
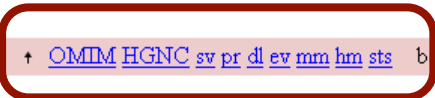
 default
 master

Master Map: Genes On Sequence

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

[Summary of Maps](#) [Maps & Options](#) [Download/View Sequence/Evidence](#)

| Hs UniG | Genes_seq | Symbol | Links | E | Cyto | Description |
|-----------|-----------|---------------------------|---|-------------|---------------|--|
| Hs.317403 | Unknown | RUNDC1 ↓ | HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21.31 | RUN domain containing 1 |
| Unknown | Unknown | RPL27 ↓ | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21.1-q21.2 | ribosomal protein L27 |
| Unknown | Unknown | IFI35 ↓ | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | interferon-induced protein 35 |
| Hs.634952 | Unknown | VAT1 ↑ | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | vesicle amine transport protein 1 homolog (T cell) |
| Unknown | Unknown | RND2 ↓ | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | Rho family GTPase 2 |
| Unknown | Unknown | | | | | |
| Hs.194143 | Unknown | | | | | |
| Unknown | Unknown | | | | | |
| Unknown | Unknown | RPL21P4 ↓ | HGNC sv dl ev mm | best RefSeq | 17q21 | ribosomal protein L21 pseudogene 4 |
| Unknown | Unknown | | | | | |
| Unknown | Unknown | BRCA1 ↑ | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | 17q21 | breast cancer 1, early onset |
| Unknown | Unknown | | | | | |
| Unknown | Unknown | | | | | |
| Hs.373818 | Unknown | NBR2 ↓ | HGNC sv pr dl ev mm | best RefSeq | | |
| Unknown | Unknown | | | | | |
| Hs.601045 | Unknown | | | | | |
| Hs.601912 | Unknown | | | | | |
| Unknown | Unknown | | | | | |
| Unknown | Unknown | BRCA1P1 ↑ | HGNC sv dl ev mm | best RefSeq | | |
| Unknown | Unknown | | | | | |
| Hs.626603 | Unknown | NBR1 ↓ | OMIM HGNC sv pr dl ev mm hm sts | best RefSeq | | |
| Unknown | Unknown | | | | | |
| Hs.277721 | Unknown | | | | | |
| Hs.546264 | Unknown | | | | | |



LinkOut

OMIM = disease
 sv = sequence view
 pr = protein record
 dl = download
 hm = Homologene