

UCSC Genome Browser: Latest resources for variant interpretation

Luis R Nassar¹, Galt P Barber¹, Benet-Pagès A², Jonathan Casper¹, Hiram Clawson¹, Mark Diekhans¹, Clay Fischer¹, Jairo Navarro Gonzalez¹, Angie S Hinrichs¹, Christopher M Lee¹, Gerardo Perez¹, Brian J Raney¹, Maximilian Haeussler¹

¹University of California Santa Cruz, Genomics Institute, Santa Cruz, CA ²Medical Genetics Center (Medizinisch Genetisches Zentrum), Munich Germany

Introduction

The UCSC Genome Browser¹ is a free resource that contains much of the information required for variant effect prediction. The use of this information is also facilitated by our Recommended Track Set (RTS) feature. Here we summarize our improvements for clinical users:

- **Data:** New Exon Relevance RTS and a brief list of most relevant datasets recently released, such as **SpliceAI**
- **Features:** Some of the latest features that facilitate variant interpretation and education, including a new **clinical tutorial**

Determine Exon Relevance RTS

UCSC Genome Browser on Human (GRCh38/hg38)

Move <<< << < > >> >>> Zoom in 1.5x 3x 10x Base Zoom out 1.5x 3x 10x 100x

region chr17:29,625,938-29,930,228 304,291 bp. gene, chromosome range, search terms, help pages, see exa

Search box

Menu

Configure c f
Track Search t s
Short Exact DNA Match
Recommended Track Sets
Reset All User Settings c r

Scale chr17: 29,650,000

Track set selection menu

Click gray bar to description page and configure

Recommended Track Sets

These links provide track sets selected and pre-configured for specific user scenarios. They are designed to be useful at different genomic loci. Clicking a link below will create a browser window with these tracks visible, without changing the locus.

- [Clinical SNVs](#): Assess potential disease contributions of single nucleotide variants in coding regions
- [Clinical CNVs](#): Assess potential disease contributions of structural variants in coding regions
- [Non-coding SNVs](#): Investigate functional aspects of non-coding variants
- **Determine Exon Relevance**: Examine if variants are present in an exon required for the function of the expressed gene product
- [Problematic Regions](#): Evaluate if annotations are on potential low confidence regions due to high homology or other reported concerns
- [ENIGMA BRCA1/BRCA2 VCFP](#): Asses potential disease contribution of variants on BRCA1 and BRCA2 according to the ENIGMA VCFP guidelines

Return to [Default](#) browser tracks.

Exit recommended track sets

This tool is for research use only. For personal medical or genetic advising, consult a qualified physician.

Recommended Track Sets are pre-selected track groupings for specific user scenarios. The **Determine Exon Relevance** track set examines if variants are present in an exon required for the function of gene product.

New Clinically Relevant Data

- **AbSplice Prediction Scores** (Splicing impact prediction)
- **AlphaMissense scores** (Missense variant pathogenicity)
- **Bionano DLE-1 CTTAAG sites** (SV detection)
- **CADD v1.7** (Variant deleteriousness)
- **ClinGen CSpec** (Loci with expert panel criteria)
- **Clinical Interpretation of Variants in Cancer** (CIViC)
- **COSMIC** (Curation cancer somatic mutations)
- **DECIPHER Population CNVs** (CNV pop variance)
- **Denovo-db** (Germine de novo variants)
- **Developmental Disorders Genotype-to-Phenotype** (DDG2P)
- **gnomAD pext** (Proportion expressed transcripts)
- **Illumina SpliceAI** (Splicing impact prediction)
- **M-CAP** (Rare missense pathogenicity predictor)
- **MaveDB Experiment Heatmaps** (Multiplexed assays var effects)
- **MITOMAP** (chrMT variation)
- **MutScore** (SNV pathogenicity predictor)
- **PanelApp Australia** (Diagnostic disease panels)
- **SpliceVarDB** (Experimentally assayed splice data)

Are we missing any data you would like to see?
Email us and let us know!

New Features

Clinical Tutorial

We have a new **interactive tutorial** showcasing the resources we offer that could be useful in variant interpretation.

It covers topics such as searching for variants and data, recommended track sets, and how to save and share browser configurations.

Help About Us

FAQs & Search
Browser Documentation
Training
Keyboard Shortcuts ?
Contact Us
Interactive Tutorials

Interactive Tutorials

These interactive tutorials will provide step-by-step guides to help navigate through various tools and pages on the UCSC Genome Browser.

- [Basic tutorial](#): An introductory tutorial to help users navigate the UCSC Genome Browser tracks display. Learn how to configure display settings, search for tracks, and view the negative strand (3' to 5').
- Advanced tutorial for clinicians** (only available on hg19 & hg38): A tutorial focused on clinical genetics to showcase resources that may be useful in variant interpretation. Learn how to search for variants, view recommended track sets, and save your configuration settings to share with others.
- [Table Browser tutorial](#): An introductory tutorial to navigate and configure the UCSC Table Browser. Learn how to switch between assemblies, select primary or related tables for a track, and select your output format.

Download Data in Current Region

You can now download all visible data in the current browser region from the tracks display. This improves reproducibility when writing variant reports or publications as data can update over time. You can select from visible tracks, and export them in various formats, including TSV/CSV, that can be opened in Excel.

Download track data in view

Use this selection window to download track data for the current region (chr7:155,799,529-155,812,871). Please note that large regions may be slow to download.

Check All Clear All

- ☒ ClinVar CNVs
- ☒ ClinVar SNVs
- ☒ ClinVar Interp
- ☒ MANE
- ☒ OMIM Alleles
- ☒ OMIM Genes

Enter an output file name hg38.tracks

Choose an output format CSV

Download Cancel

Downloads My Data

Genome Data
Source Code
Genome Browser Store
Utilities
FTP
MySQL Access
Cloud Access
REST API
Download Current Track Data

To access this feature hover over **Downloads** and click **Download Current Track Data**

MANE Transcripts More Prominent

Matches to the MANE transcripts track now appear at the top of the search results, and the MANE transcript in our default genes display is now colored to match the MANE track for easy identification.

Scale chr13: 32,330,000 32,340,000 32,350,000 32,360,000 32,370,000 32,380,000 32,390,000 32,400,000

GENCODE V48 (14 items filtered out)

BRCA2

MANE Select Plus Clinical: Representative transcript from RefSeq & GENCODE

More Information

UNIVERSITY OF CALIFORNIA
SANTA CRUZ Genomics
Institute



For any questions or comments, email us at genome@soe.ucsc.edu

Funding provided by **NHGRI [U41HG002371]**

References

The UCSC Genome Browser database: 2024 update. Raney BJ, Barber GP, Benet-Pagès A, Casper J, Clawson H, Cline MS, Diekhans M, Fischer C, Navarro Gonzalez J, Hickey G *et al.* *Nucleic Acids Res.* 2024 Jan 5;52(D1):D1082-D1088. PMID: [37953330](#).

Variant interpretation: UCSC Genome Browser Recommended Track Sets. Benet-Pagès A, Rosenbloom KR, Nassar LR, Lee CM, Raney BJ, Clawson H, Schmelter D, Casper J, Gonzalez JN, Perez G, Lee BT, Zweig AS, Kent WJ, Haeussler M, Kuhn RM. *Hum Mutat.* 2022 Jan 28; PMID: [35088925](#)