UCSC Genome Browser: Latest resources for variant interpretation

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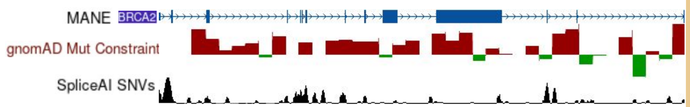
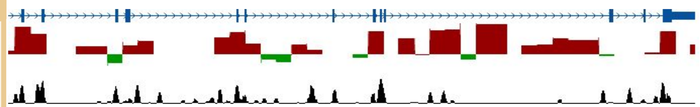


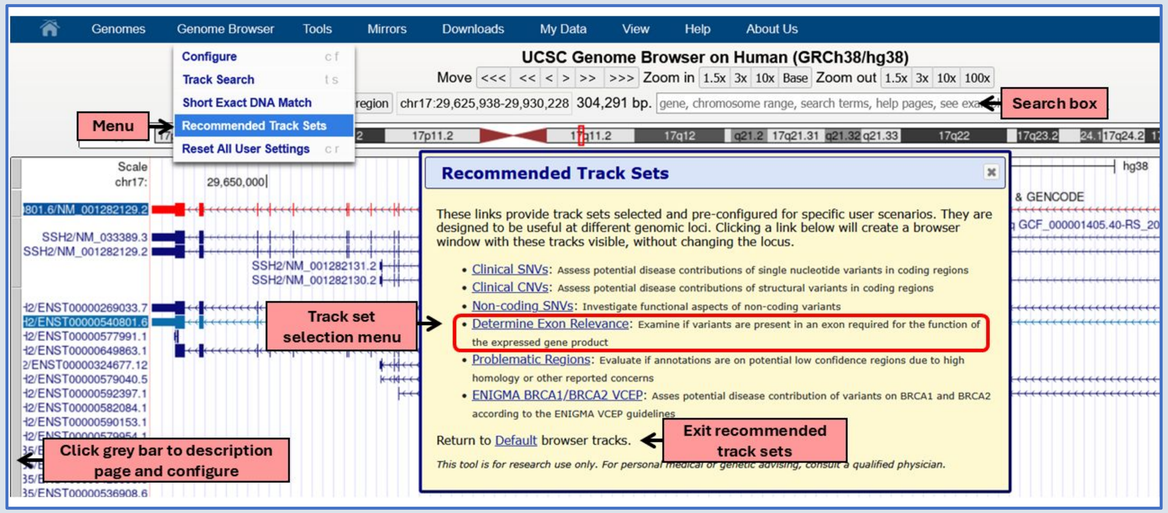
Introduction

The UCSC Genome Browser[1] 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



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)

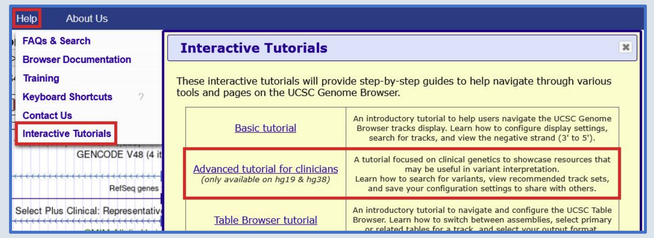
Are we missing any data you would like to see?
Email us and let us know!

New Features

Clinical Tutorial

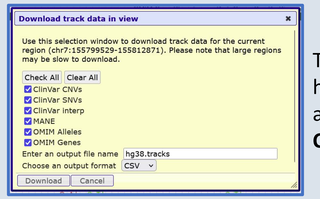
We have a new **interactive tutorial** showcasing the resources we offer that could be useful in variant interpretation.

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

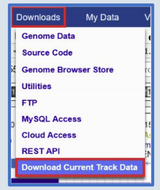


Download Data in Current Region

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.

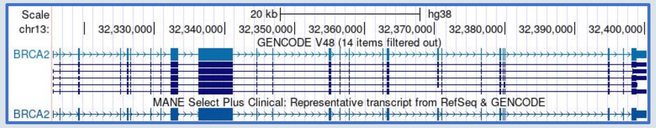


To access this feature hover over **Downloads** and click **Download Current Track Data**



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.



More Information

UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute

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

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