

UCSC Genome Browser: Latest developments for variant interpretation

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Introduction

The UCSC Genome Browser¹ is a free resource that contains much of the information required for variant effect prediction. This information is presented in the following two categories:

- **Data:** Brief list of most relevant datasets recently released, notably **gnomAD4.1** and **SpliceAI**
- **Features:** Some of the latest features that facilitate variant interpretation and education, including a new **clinical tutorial**

New Data

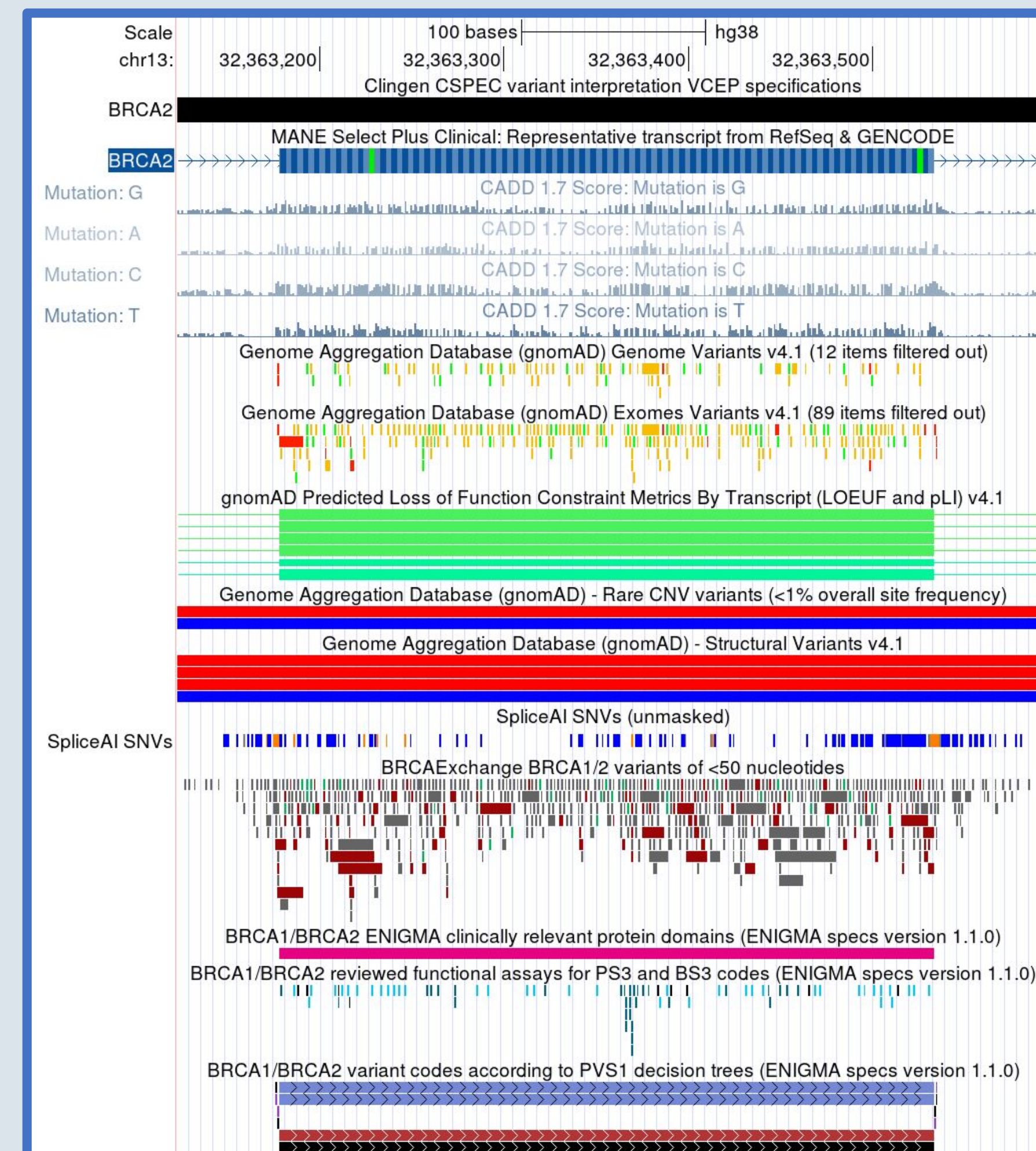
We strive to keep our annotations up to date. Below are recent data releases relevant to variant interpretation:

- **gnomAD4.1:** gnomADv4.1 April, 19 2024 release
 - **Genome and exome variants:** 943M variants
 - **Structural variants:** 2.1M variants >=50 nucleotides
 - **Predicted constraints:** LOEUF/pLI and z-score constraint metrics
 - **Rare CNV variants** (<1% overall site frequency)
- **SpliceAI:** Splicing prediction scores from Illumina
- **ClinGen CSPEC:** Locations with a ClinGen CSPEC
- **CADD 1.7:** Variant deleteriousness score
- **BRCA Exchange public hub:** Hub with BRCA Exchange data
- **ENIGMA public hub:** Hub assisting with ENIGMA VCEP interpretation

We also continue to automatically update the following relevant data:

- **DECIPHER** variants
- **Orphadata:** Aggregated Data From Orphanet
- **ClinGen** Curation (Dosage Sensitivity and Gene-Disease Validity)
- **OMIM** Genes and Phenotypes
- **Gene Curation Coalition** (GenCC) annotations
- **NCBI RefSeq** gene annotations
- **MANE Select Plus** Clinical gene annotations

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

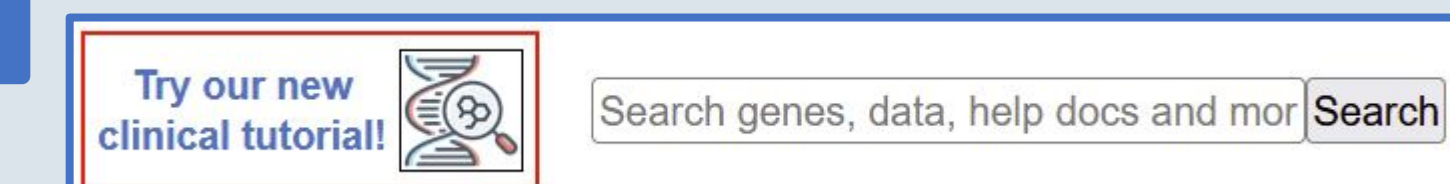


The region above shows exon 18 of the BRCA2 MANE transcript. The displayed tracks include many of our new releases listed.

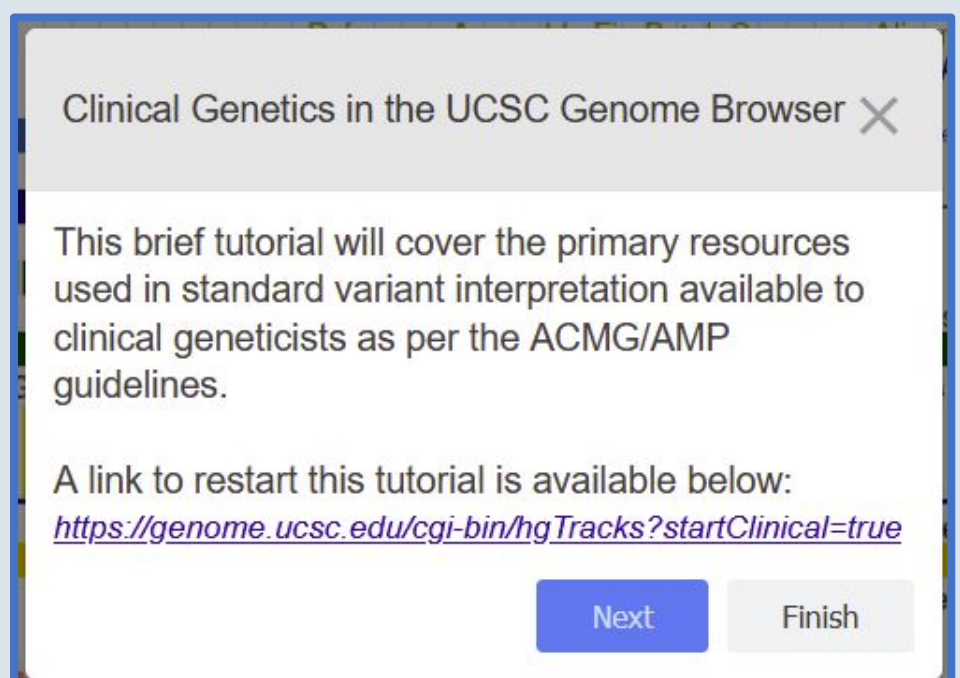
New Features

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.

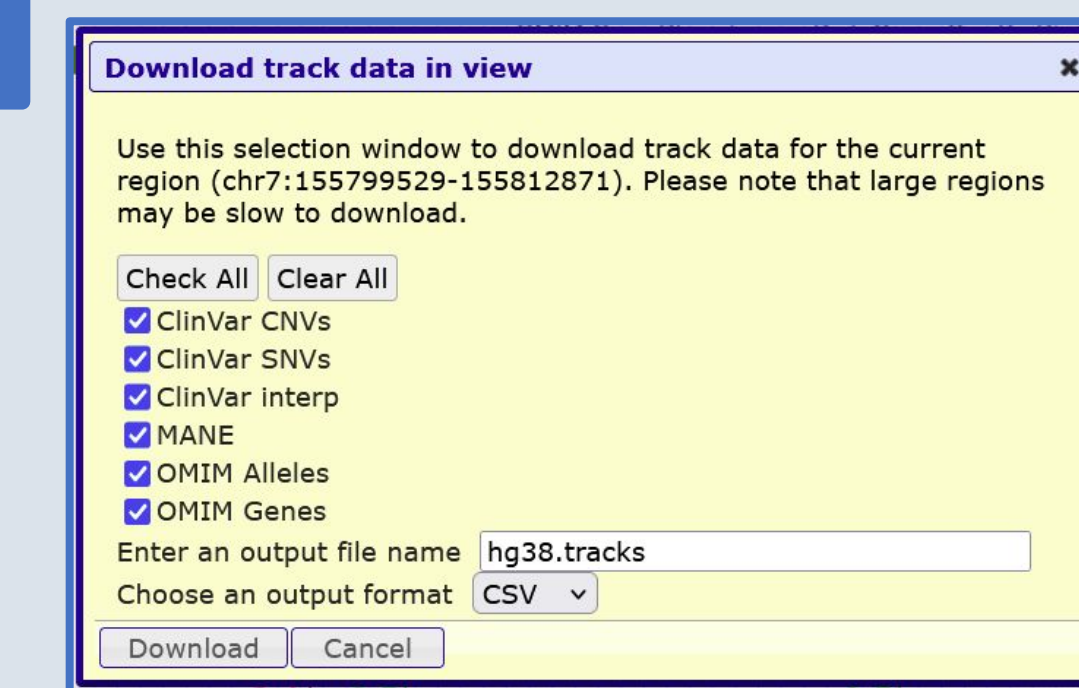


Look for the icon on our homepage (<https://genome.ucsc.edu>) to initiate the tutorial on hg38.

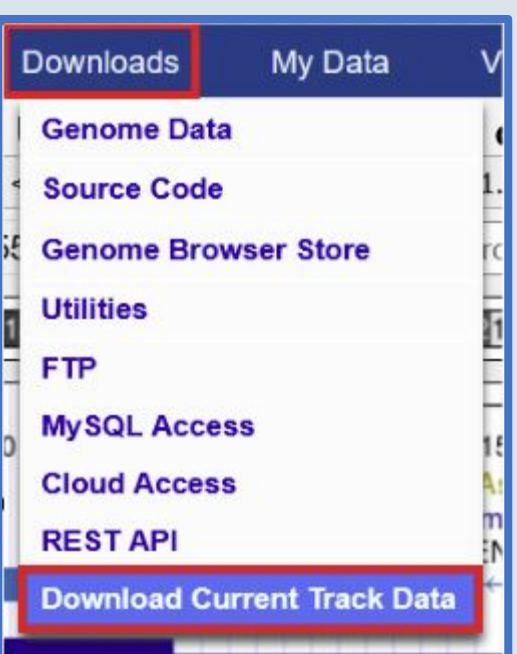


Download data in current region

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 load into Excel.



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



Search historical RefSeq NMs

Searching for historical RefSeq (NM_) transcripts is now available on hg38. This issue was most notable when searching variants in papers or workflow examples. This search can be used to look up **entire NM accessions** as well as when using **HGVS terms**.

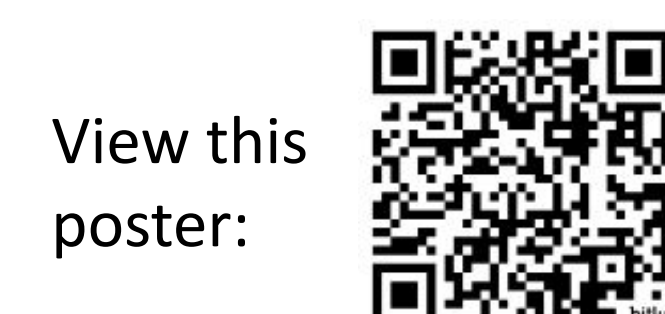
Example 1:

NM accession search: NM_198056.2
Now matches the entire length of transcript (.4 is latest)

Example 2:

HGVS term: NM_198576.2(AGRN):c.1057C>T
Now matches proper C>T position in historical transcript

More Information

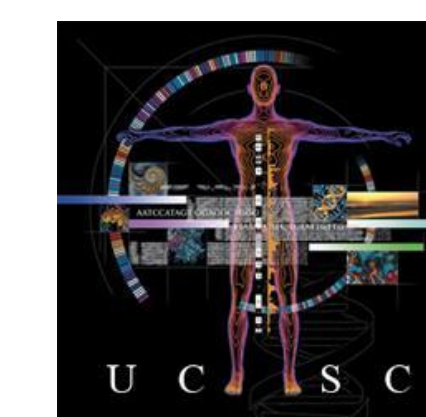


View this poster:



For any questions or comments, email us at genome@soe.ucsc.edu or scan the QR code.

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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](#)