

# UCSC Genome Browser 2026: Latest resources for variant analysis

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## Introduction

The UCSC Genome Browser<sup>1</sup> is a free resource that contains much of the information required for variant analysis and interpretation. Below is a summary of the latest resources and features available:

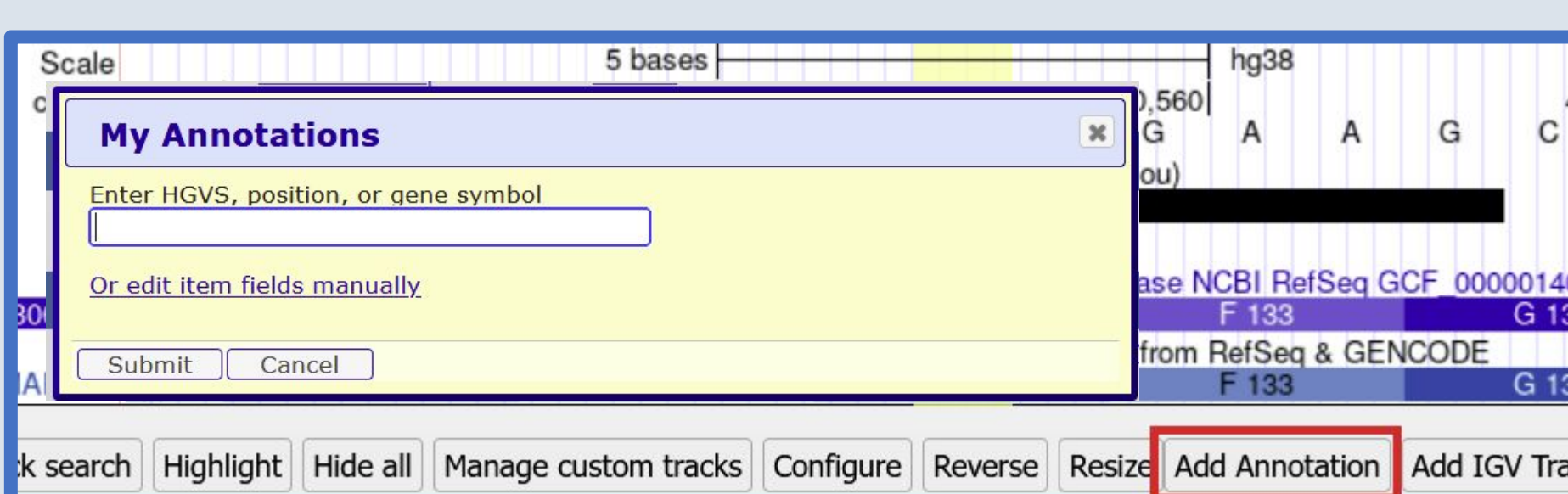
- **My Annotations:** A new feature that allows you to create and share annotations (e.g. variants) directly from the tracks display
- **Recommended Track Sets (RTS):** This feature simplifies the selection process by loading a preconfigured set of annotations
- **ClinGen InSiGHT Hub:** A new RTS with data hubs aimed at interpretation of lynch syndrome variants on MLH1, MSH2, MSH6, and PMS2
- **Latest annotations:** Some of the latest data released in the last year that can be used in variant analysis

## My Annotations (Add your own variants)

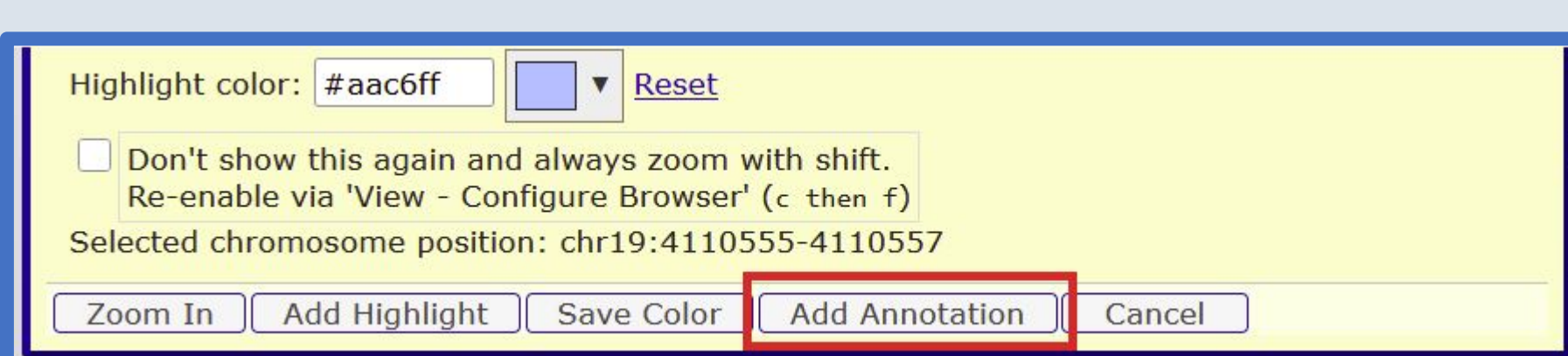
We have a new feature in development that allows you to **create, configure, and share annotations**, such as variants, directly from the tracks display.

### Adding annotations

Annotations can be added by HGVS via button:

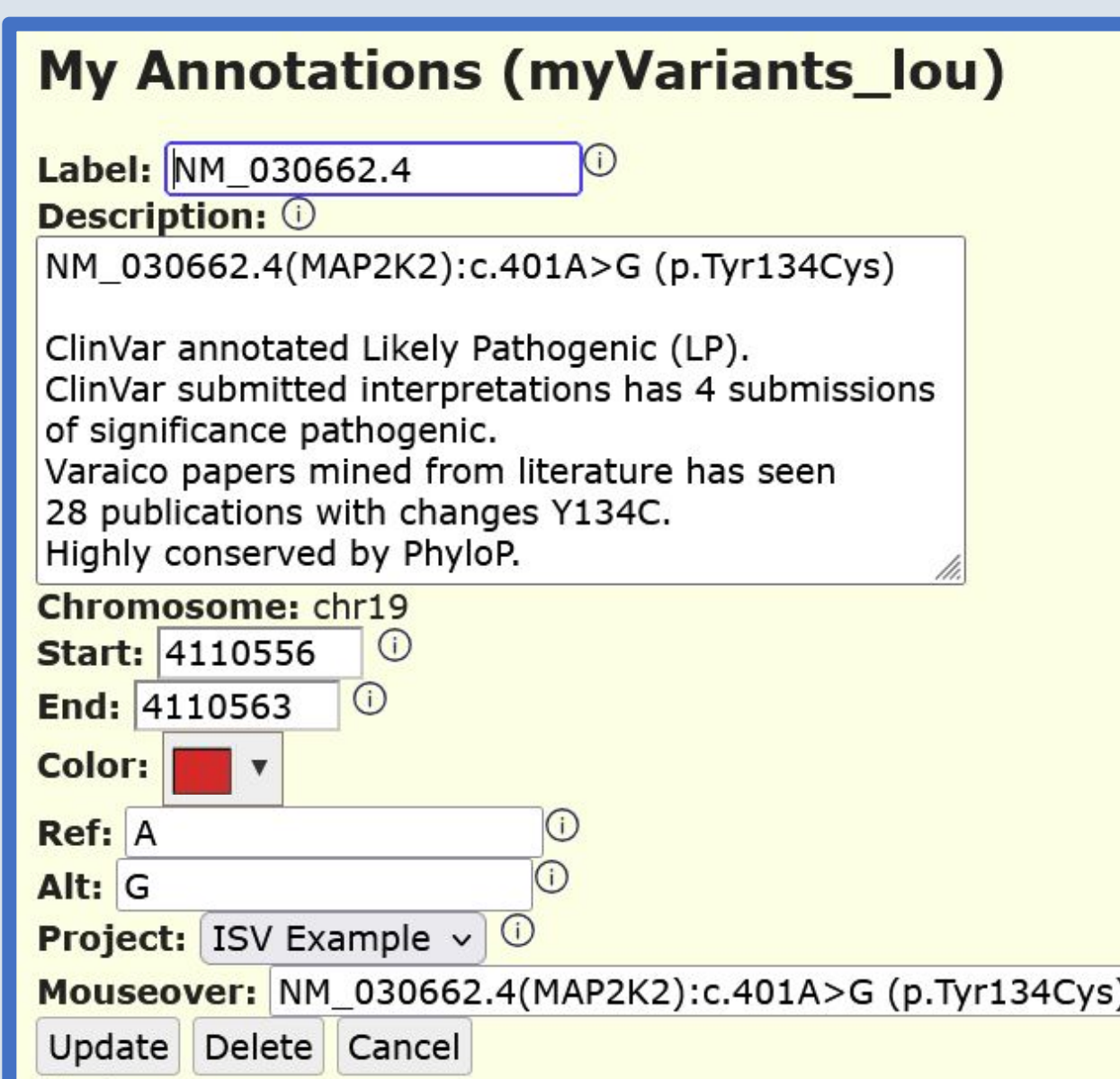


Or by **shift + drag** to draw directly on the image:

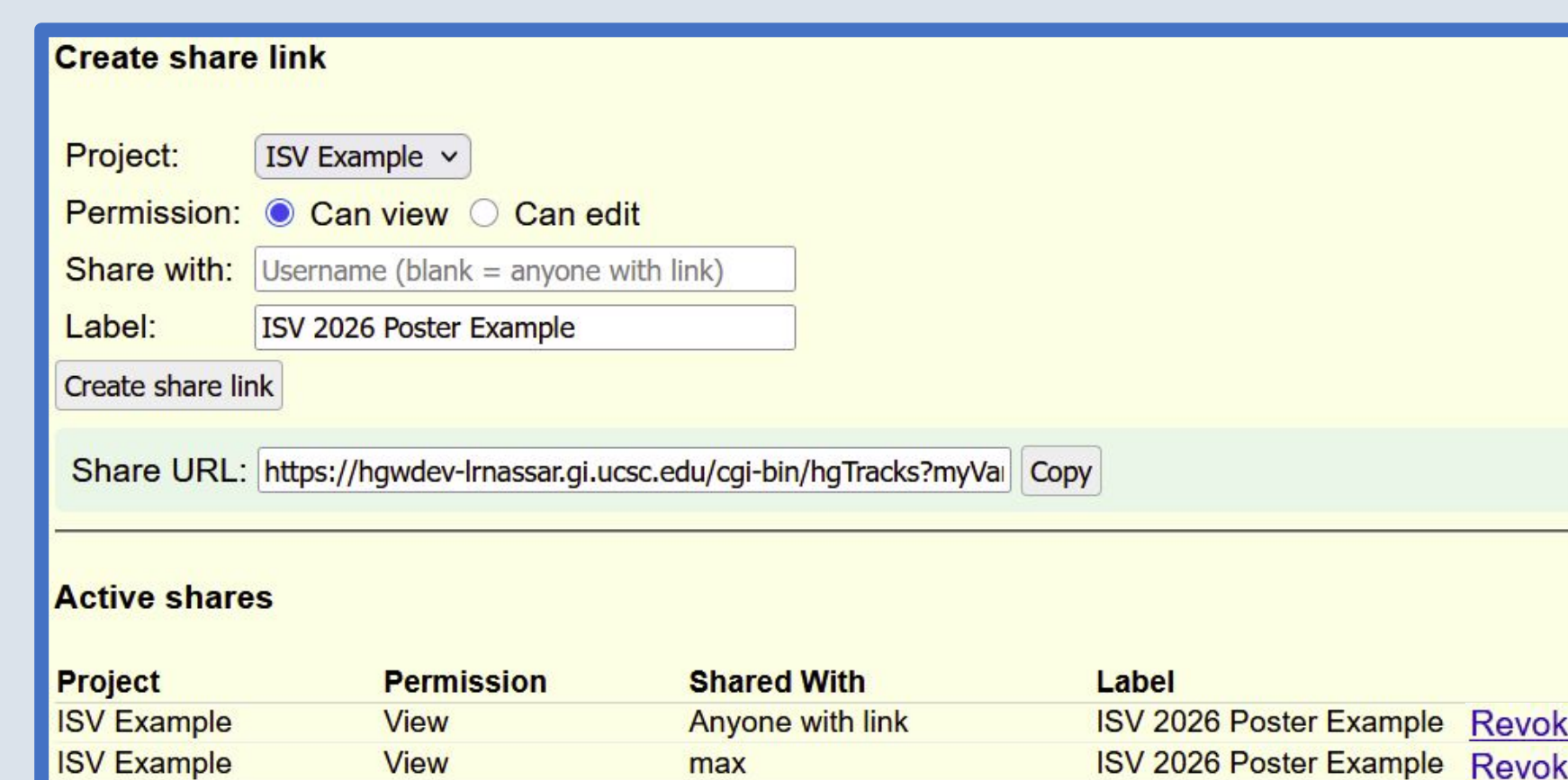


### Configuring annotations

Most item details can be configured:

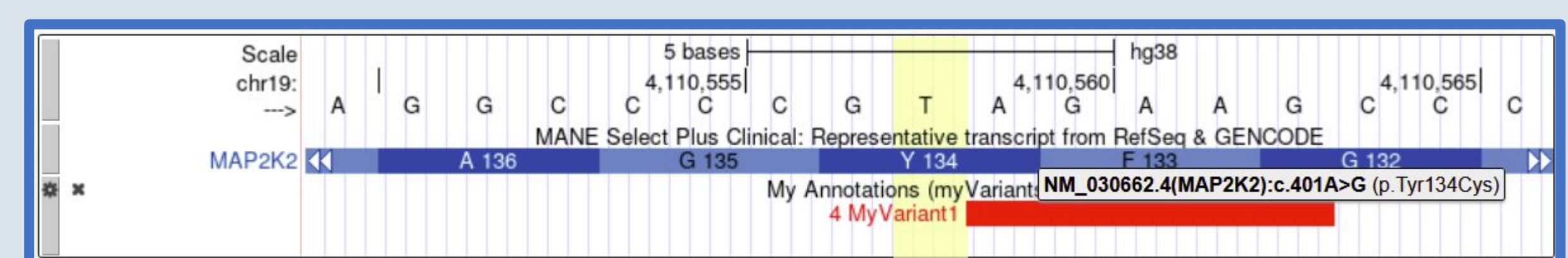


### Sharing annotations



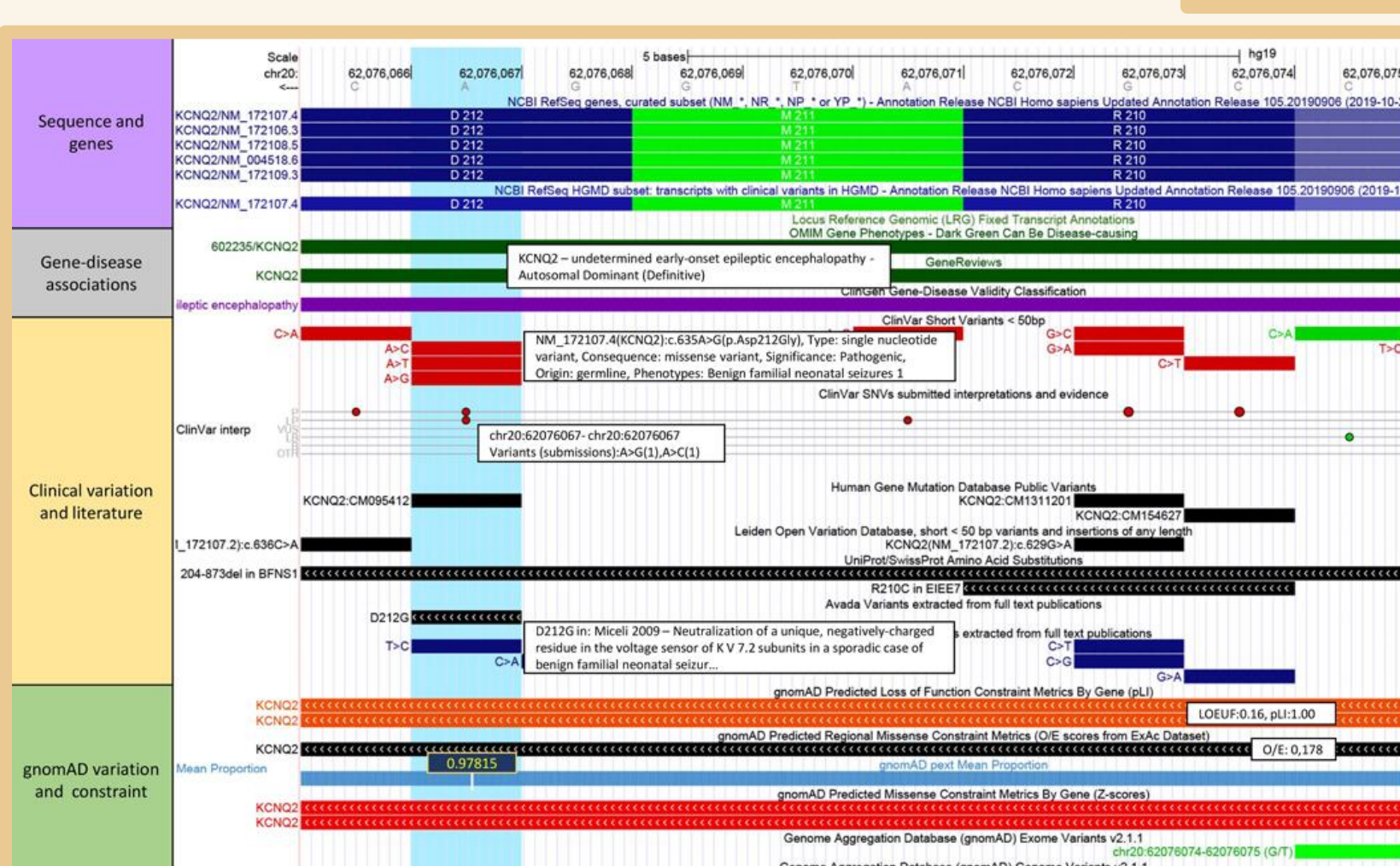
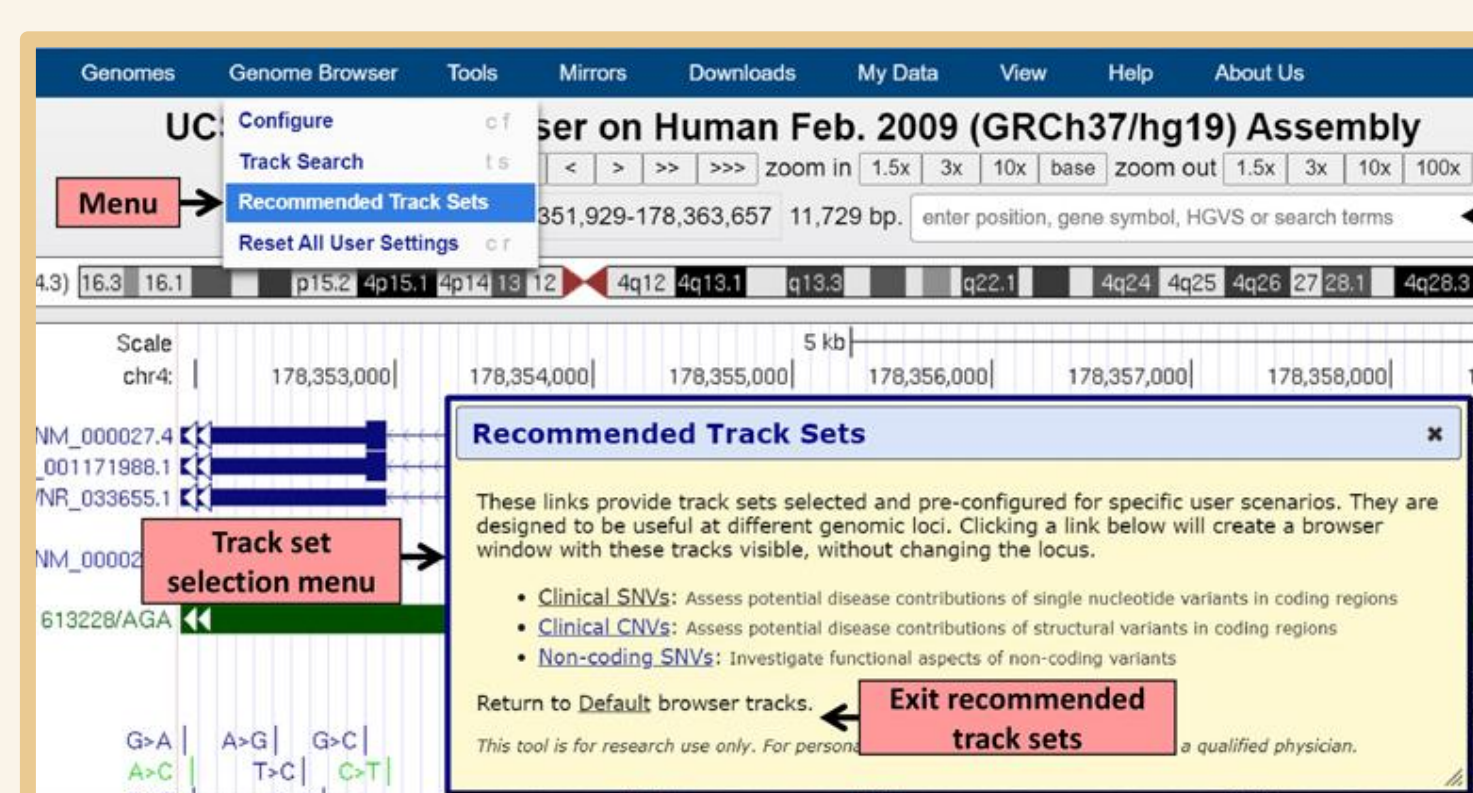
Sharing can be customized for specific users, with **view** or **edit** permissions.

Have **feedback?**  
Let us know!



## Recommended Track Sets

Recommended track sets (RTS) are **curated track groupings** designed with specific themes. They facilitate clinical variant interpretation by displaying the most relevant tracks in a single selection. They can be accessed via the **Genome Browser blue bar menu** (right).

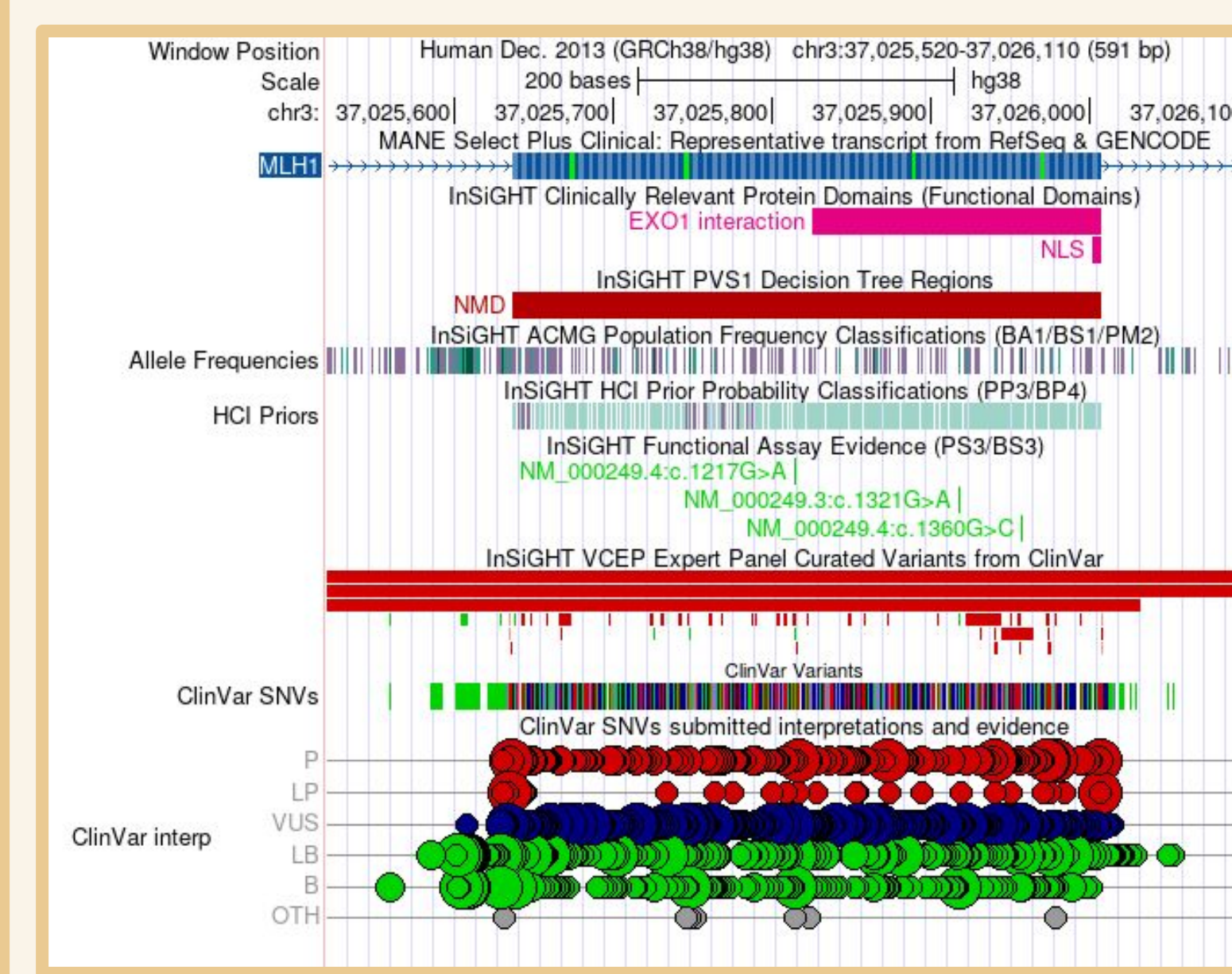


RTSs are available on both **hg19** and **hg38**. The current track sets available are:

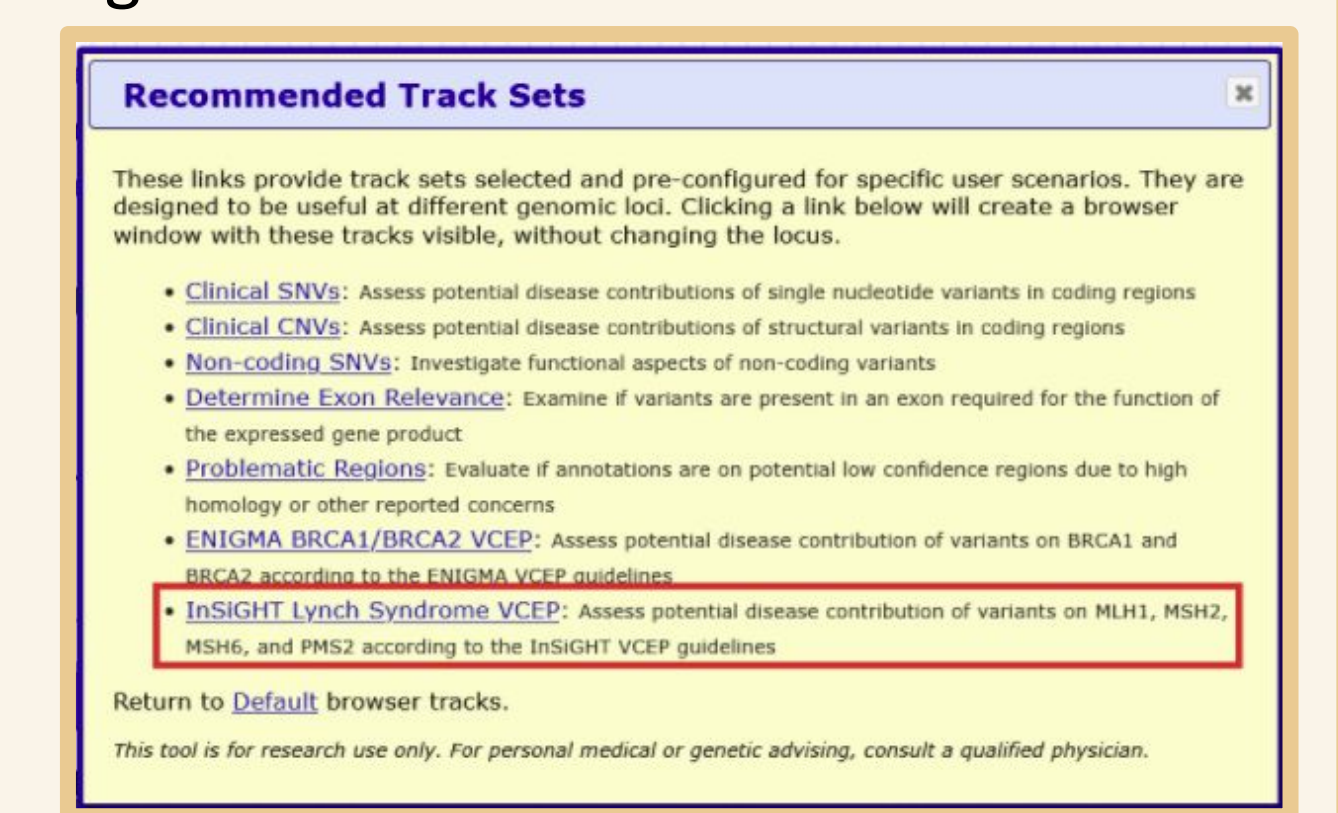
- Clinical SNVs (shown left)
- Clinical CNVs
- Non-coding SNVs
- Determine Exon Relevance
- Problematic Regions
- ENIGMA BRCA1/BRCA2 VCEP
- InSiGHT Lynch Syndrome VCEP

## InSiGHT Hereditary Colon Cancer/Polyposis VCEP

Hubs allow anyone to display **custom annotations** with all the configuration options available to native Browser tracks. Here, we collaborated to create a hub implementing the **InSiGHT Hereditary Colorectal Cancer/Polyposis VCEP** specifications (CSpec v2.0.0) for the Lynch syndrome mismatch repair genes MLH1, MSH2, MSH6, and PMS2. This hub includes all data and speeds up the process of bioaction while enhancing the accuracy of genomic variant interpretation. Hubs like these can be created by anyone.



See left for an image of the RTS on hg38/MLH1. See below to find how to enable the InSiGHT RTS on hg19 and hg38 via the RTS menu:



## New Annotation Tracks

### Non-coding + phased variants

**ENCODE4 cCREs:** Candidate cis-Regulatory Elements (2.3M)  
**Human Methylation Atlas:** WGBS DNA methylation across 39 human cell types  
**Phased variants:** Population phased genotypes from 4 projects (HGDP, SGDP, gnomAD HGDP+1KG, Mexico Biobank)

### Pathogenicity predictions

**NMD Escape:** Regions escaping nonsense-mediated mRNA decay  
**PrimateAI-3D:** Missense pathogenicity from 3D structure + primate variation  
**PromoterAI:** Non-coding promoter variant pathogenicity scores  
**MutScore:** Missense pathogenicity from variant clustering  
**M-CAP:** Rare missense pathogenicity classifier for clinical use  
**AlphaMissense:** AlphaFold-based missense pathogenicity predictions  
**gnomAD MPC:** Missense-depleted regional constraint score

### Databases

**Varaico Variants:** 5.5M+ variants mined from clinical literature  
**denovo-db:** Curated germline de novo variants  
**SpliceVarDB:** 50K+ variants assayed for splicing effects  
**MITOMAP:** Mitochondrial DNA variation and disease mutations  
**MaveDB Experiment Heatmaps and Alignment:** Multiplexed assays of variant effects  
**CoLoRSdb:** Long-read small and structural variants

### Phenotype + gnomAD + STRs

**PanelApp Australia:** Crowdsourced diagnostic disease gene panels  
**DDG2P:** Genes linked to severe developmental disorders  
**gnomAD v4 Exome / v3 Genome coverage:** Per-base sequencing coverage  
**gnomAD pext:** Isoform expression across 50 GTEx tissues  
**Short Tandem Repeats (STR):** Population-level tandem repeat variation

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

## More Information

View this poster:



Make a suggestion:



For any questions or comments, email us at [genome@soe.ucsc.edu](mailto:genome@soe.ucsc.edu) or scan the QR code.



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**SANTA CRUZ** Genomics Institute



## References

The UCSC Genome Browser database: 2025 update, Casper J, Barber GP, Benet-Pagès A, 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](https://pubmed.ncbi.nlm.nih.gov/37953330/).

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