

## UCSC GENOME BROWSER

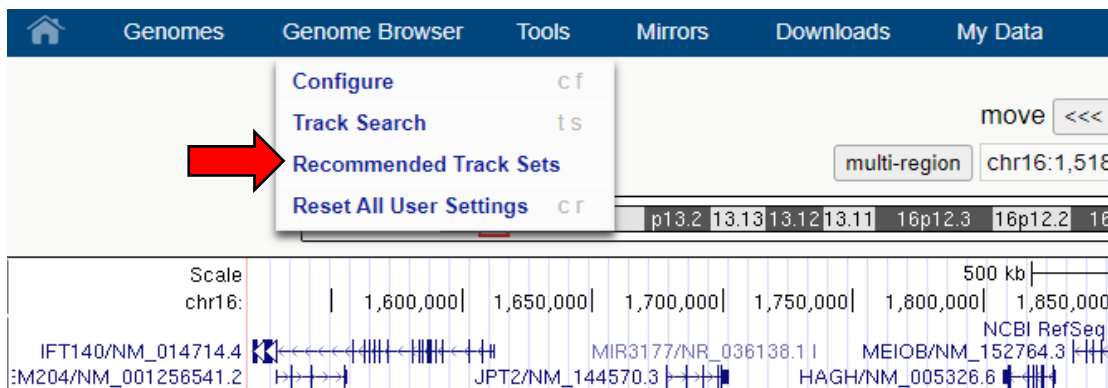
- The UCSC genome browser is an exceptional genomic resource
- Tracks are fully customizable, allowing data from a variety of databases to be displayed
- Due to the plethora of track choices, it can seem overwhelming to set your view
- Fortunately, UCSC provides Recommended Track Sets – if you select one of these options, you can instantly optimize your view for microarray analysis (“Clinical CNVs” track set) or for sequencing analysis (“Clinical SNVs” track set)
- *Follow the instructions below to quickly optimize your browser view for the analysis of constitutional copy number variants!*

Using **Google Chrome**, go to->

<http://www.genome.ucsc.edu/cgi-bin/hgGateway> ->

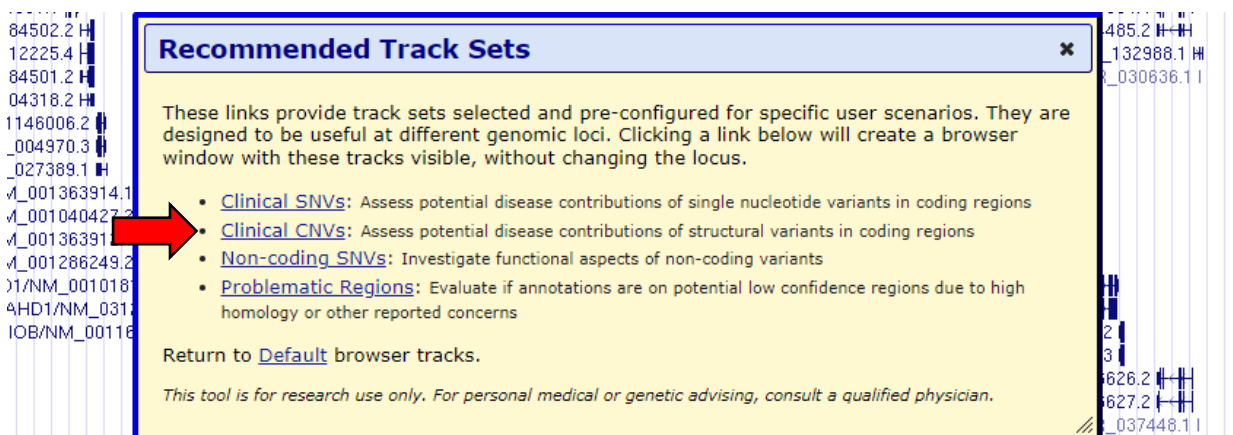
Click “Genome Browser” ->

Click “Genome Browser” again and select “Recommended Track Sets” ->



The screenshot shows the UCSC Genome Browser interface. At the top, there is a navigation bar with options: Genomes, Genome Browser, Tools, Mirrors, Downloads, and My Data. A dropdown menu is open under 'Genome Browser', with a red arrow pointing to 'Recommended Track Sets'. Other options in the menu include 'Configure', 'Track Search', and 'Reset All User Settings'. Below the menu, the browser displays a genomic track for chromosome 16, showing various annotations and a scale bar.

Select “Clinical CNVs” ->



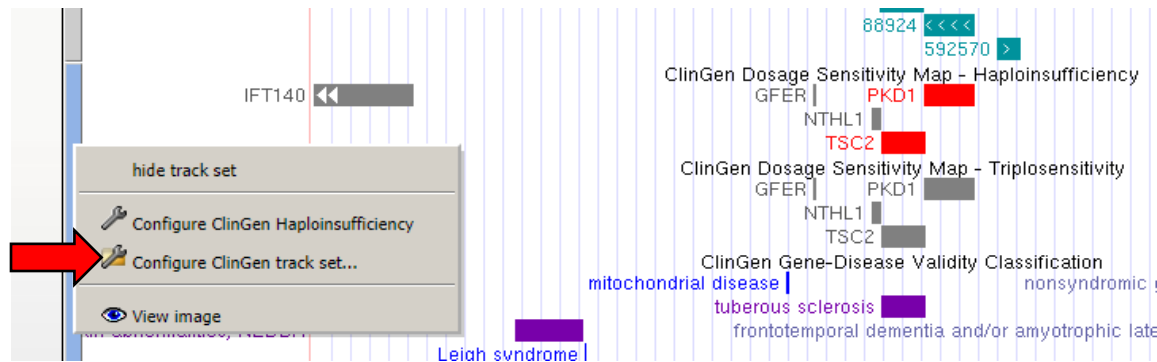
The screenshot shows the 'Recommended Track Sets' dialog box. The dialog contains the following text: 'These links provide track sets selected and pre-configured for specific user scenarios. They are designed to be useful at different genomic loci. Clicking a link below will create a browser window with these tracks visible, without changing the locus.' Below this text is a list of track sets:

- [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
- [Problematic Regions](#): Evaluate if annotations are on potential low confidence regions due to high homology or other reported concerns

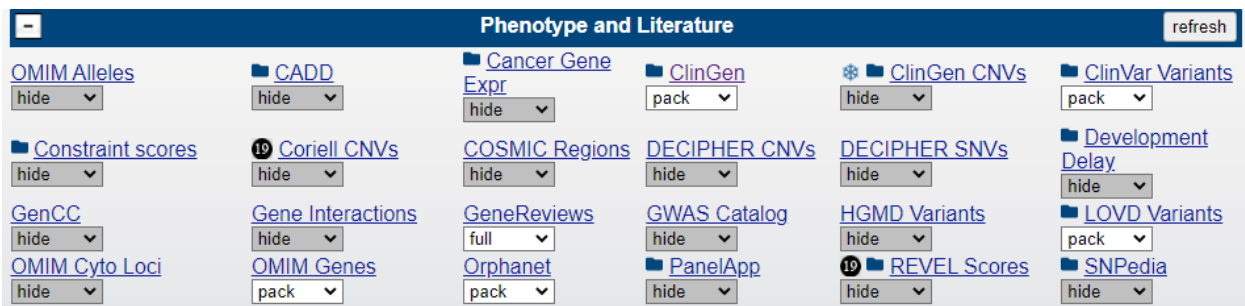
A red arrow points to the 'Clinical CNVs' link. Below the list, there is a link to 'Return to Default browser tracks.' and a disclaimer: 'This tool is for research use only. For personal medical or genetic advising, consult a qualified physician.'

- This will load all the main tracks needed for assessing disease contributions of CNVs, including:
  - NCBI transcripts with the HGMD transcript subset,
  - OMIM gene phenotypes,
  - ClinGen Gene-Disease Validity (e.g. HI and TS scores), and
  - Data from case and control databases (ClinVar, DECIPHER, DGV/Gold Standard, gnomad)

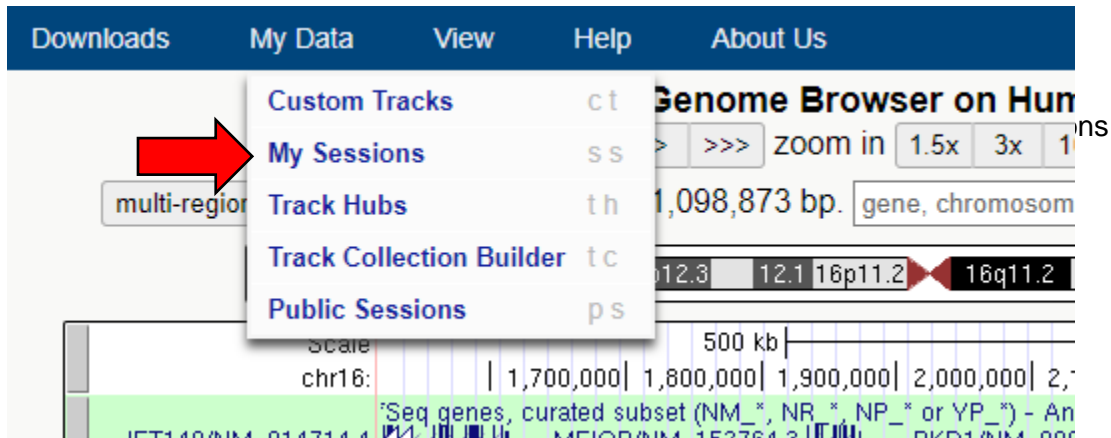
OPTIONAL: Tracks can be further customized by right-clicking on the grey bar bordering the far right of the track. Note, after you right click on the grey bar it will become blue as in the screenshot below. You can then choose to include more or less data from the dropdown menu (e.g. “hide track set”). Details about interpreting the data (e.g. the data’s color code key) can often be found by clicking on “Configure” (red arrow below).



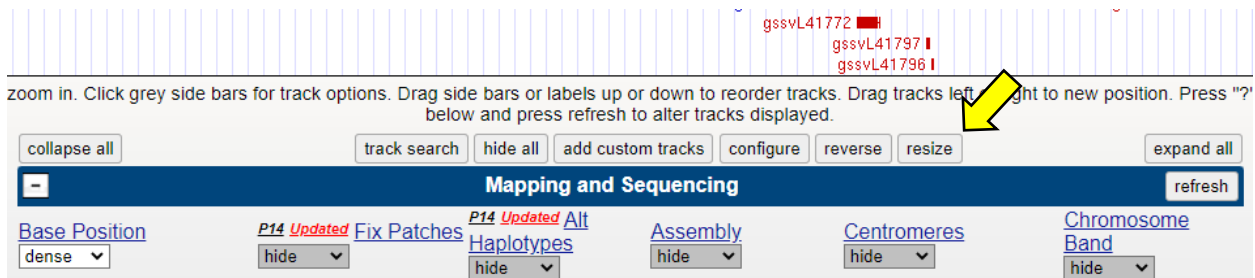
OPTIONAL: Tracks can be added or removed from view at the bottom of the screen using the drop-down boxes.



OPTIONAL: Custom track layouts can then be saved as “sessions’ in your free UCSC account. These sessions can be shared with colleagues, which is especially helpful for collaborations.



NOTE: The browser can be expanded to automatically fill your screen by clicking the “**Resize**” button.



Convert coordinates between different genome builds using the UCSC **LiftOver** tool (found in the “Tools” tab at the top of the screen, in the blue header)

**Contact the UCSC Support team if you have questions or suggestions. They are responsive and helpful, and they are open to suggestions about improvements or additions to the Genome Brower: <https://genome.ucsc.edu/contacts.html>**

### Author Information and Affiliations:

Casey Brewer, PhD, FACMG  
Cincinnati Children’s Hospital  
Division of Human Genetics

Date Created: 4/10/2024

---

