❍Demo search from GDV home page: NM_001302688 or take user suggestions

❍Example #1: TUBA1A-associated neurological disorders

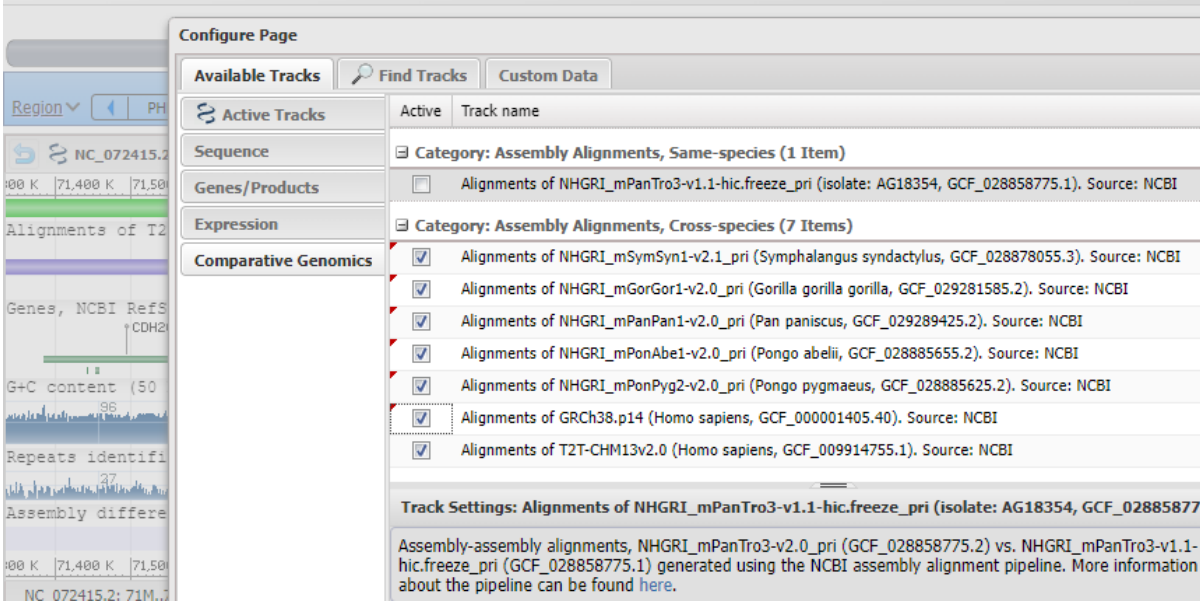- •Article: https://www.ncbi.nlm.nih.gov/labs/pmc/articles/PMC6371496/
- •GDV home page: https://www.ncbi.nlm.nih.gov/genome/gdv/
- •Goal/DIY #1: GDV search term: NM_006009.3:c.1307G>A – make sure you are looking at the right assembly
- •Add ClinVar Track w/ Precise Endpoints track to confirm
  - •Result: "Share this page" link – good for 90 days: https://www.ncbi.nlm.nih.gov/gdv/browser/genome/?cfg=NCID_1_29439921_130.14.22.10_9146_1722988474_1653042897
- •Goal/DIY #2: Variants in TUBA1A: What do we know about their allele frequencies?
- •Remote tracks, ALFA: Configure Hub -> Track Hub -> ALFA (see all track hubs)
- •ALFA home page: https://www.ncbi.nlm.nih.gov/snp/docs/gsr/alfa/
  - •Check out: https://trackhubregistry.org/
  - •Populations legend: https://www.ncbi.nlm.nih.gov/snp/docs/gsr/data_inclusion/#population

Compare chimp and human assemblies

1. Go to https://www.ncbi.nlm.nih.gov/cgv/
   - Search for human in box 1 – select Homo sapiens
   - Search for chimp in box 2 – select Pan troglodytes
   - Select T2T assemblies in boxes 3 and 4 for human and chimpanzee
   - Press View comparison
2. Note changes between genome structure between human and chimp genomes
   - Purple for reversed alignments, green for forward alignments
   - Chromosome 1 entirely reversed – maybe the genome creators submitted in reverse by accident?
   - Some other chromosomes are rearranged – part of the chimp chromosome aligns to one human chromosome, while part of it aligns to another human chromosome
   - Select chromosomes to explore further
   - Use mouse scroll to zoom in and out.
   - Click on alignments to get details
   - Undo arrow or reset to genome view to go back
   - Try searching for your favorite gene
3. Navigate to GDV using the eye icon for either the top or bottom assembly
   - Notice the alignment is displayed as a track in GDV (the top track)
   - Go to the Tracks configure wheel and the Comparative Genomics tab

- Add alignments for more species/assemblies

*Zoom into a gene in GDV to explore differences among these assemblies.*

Topics:

- Set up an alignment
- What are we looking at in the viewer?
- Navigation and zoom
- Gene search
- Link to a view of an alignment in GDV, our Genome Data Viewer

Goal: *Analyze an expansion within the GPI gene, glucose 6 phosphate isomerase, in T2T-CHM13v2.0. This region was incorrectly collapsed in GRCh38.p14. GPI deficiency is a rare cause of chronic hemolytic anemia.*

Here is the workflow exploring the GPI expansion in T2T-CHM13:

5. Set up an alignment between human T2T-CHM13v2.0 and human GRCh38.p14.
6. Use "Find a gene in this alignment" to search for glucose 6 phosphate isomerase.
7. Click on the gene name under "Description". In this case, either assembly will work.
8. Notice the unaligned region in the long intron in the 3-prime half of GPI.
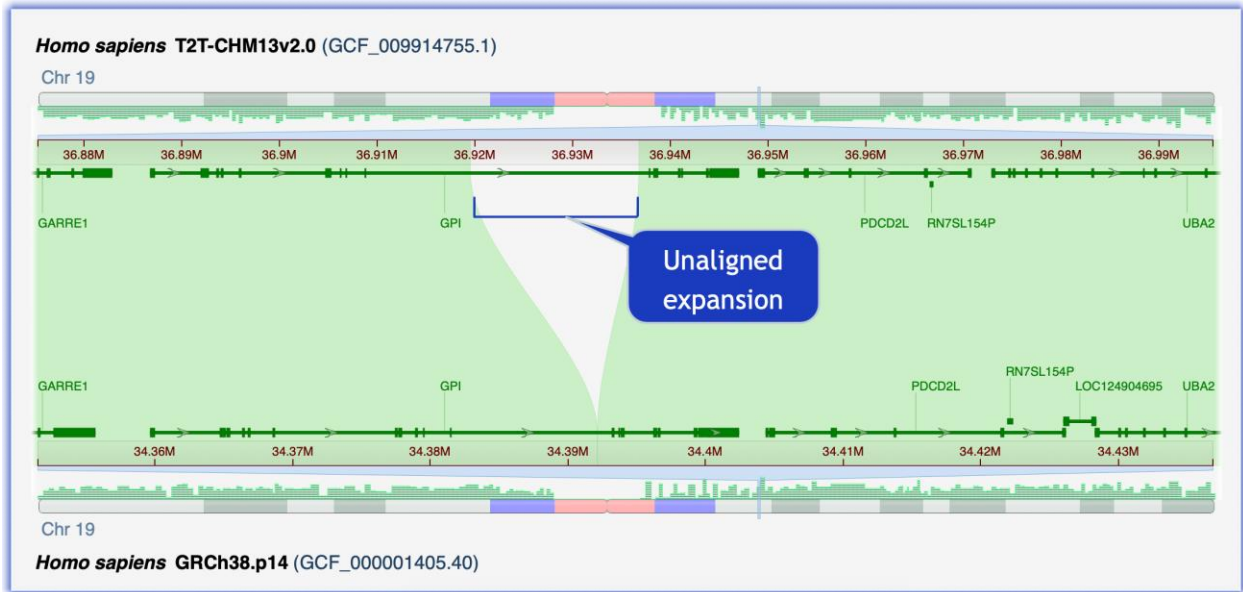
9. You can analyze the unaligned region by linking to the Genome Data Viewer, GDV. Right-click on either of the alignments to see the pop-up below. I right-clicked on the upstream alignment, the one on the left. Then select, 'View the alignment on T2T-CHM13v.20 in GDV'
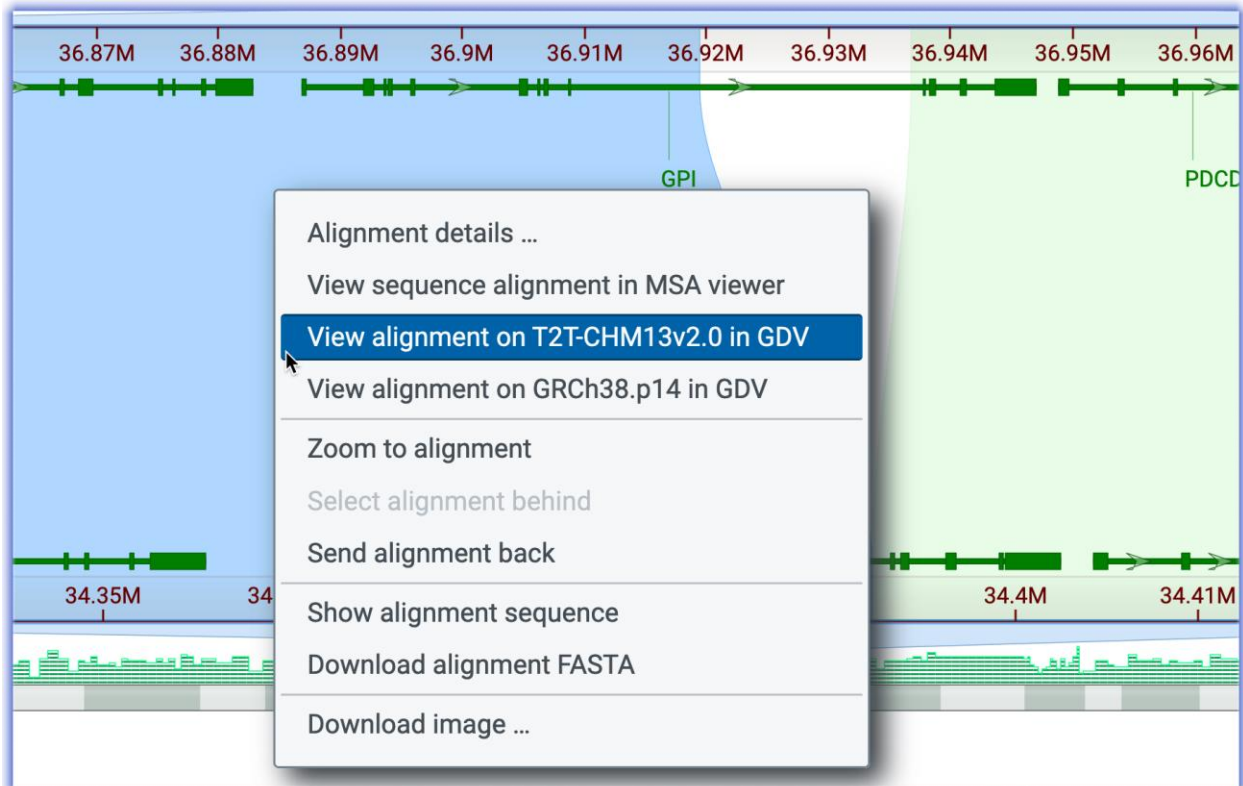
In GDV, if necesary, add the track 'T2T/HPRC Assembly-assembly alignments (T2T aligned to h38)' and the track for 'RepeatMasker Repeats.'