ODemo search from GDV home page: NM\_001302688 or take user suggestions

## OExample #1: TUBA1A-associated neurological disorders

- •Article: <u>https://www.ncbi.nlm.nih.gov/labs/pmc/articles/PMC6371496/</u>
- •GDV home page: <u>https://www.ncbi.nlm.nih.gov/genome/gdv/</u>
- •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: <u>https://www.ncbi.nlm.nih.gov/gdv/browser/genome/?cfg=NCID\_1\_29439921\_13</u> <u>0.14.22.10\_9146\_1722988474\_1653042897</u>

- •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: <u>https://www.ncbi.nlm.nih.gov/snp/docs/gsr/alfa/</u>
  - •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

Configure Page		
	Available Tracks	Find Tracks Custom Data
Region 🗸 🚺 PH	S Active Tracks	Active Track name
S NC_072415.2	Sequence	□ Category: Assembly Alignments, Same-species (1 Item)
3 К  71,400 К  71,50	Genes/Products	Alignments of NHGRI_mPanTro3-v1.1-hic.freeze_pri (isolate: AG18354, GCF_028858775.1). Source: NCBI
Alignments of T2	Expression	∃ Category: Assembly Alignments, Cross-species (7 Items)
	Comparative Genomics	Alignments of NHGRI_mSymSyn1-v2.1_pri (Symphalangus syndactylus, GCF_028878055.3). Source: NCBI
Genes, NCBI RefS		Alignments of NHGRI_mGorGor1-v2.0_pri (Gorilla gorilla, GCF_029281585.2). Source: NCBI
		Alignments of NHGRI_mPanPan1-v2.0_pri (Pan paniscus, GCF_029289425.2). Source: NCBI
		Alignments of NHGRI_mPonAbe1-v2.0_pri (Pongo abelii, GCF_028885655.2). Source: NCBI
+C content (50		Alignments of NHGRI_mPonPyg2-v2.0_pri (Pongo pygmaeus, GCF_028885625.2). Source: NCBI
		Alignments of GRCh38.p14 (Homo sapiens, GCF_000001405.40). Source: NCBI
epeats identifi		Alignments of T2T-CHM13v2.0 (Homo sapiens, GCF_009914755.1). Source: NCBI
- Janadan 370adaa		
ssembly differe		Track Settings: Alignments of NHGRI_mPanTro3-v1.1-hic.freeze_pri (isolate: AG18354, GCF_0288587
0 К  71,400 К  71,50 NC 072415.2: 71М.,7		Assembly-assembly alignments, NHGRI_mPanTro3-v2.0_pri (GCF_028858775.2) vs. NHGRI_mPanTro3-v1.1 hic.freeze_pri (GCF_028658775.1) generated using the NCBI assembly alignment pipeline. More information about the pipeline can be found here.

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

Topics:

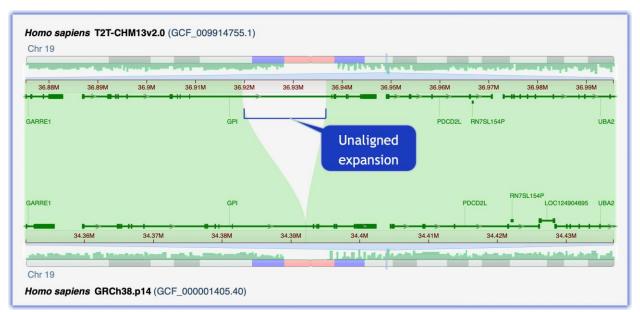
4.

- 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

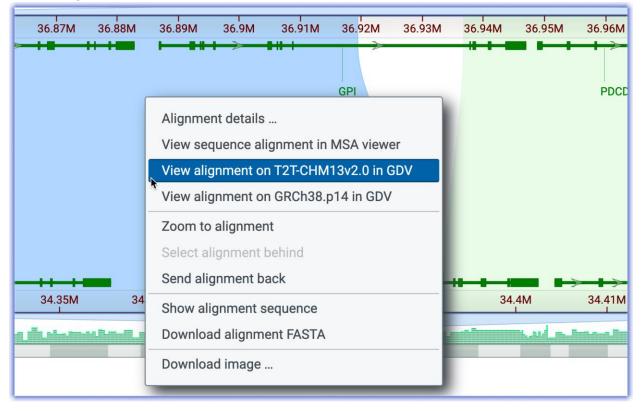
**Goal:** Analyze an expansion within the GPI gene, glucose 6 phosphate isomerase, in T2T-CHM13v2.0. This region was incorrectly collapsed in GRCh38.p14. GPI deficiency is a rare cause of chronic hemolytic anemia.

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.



 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 rightclicked 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.'