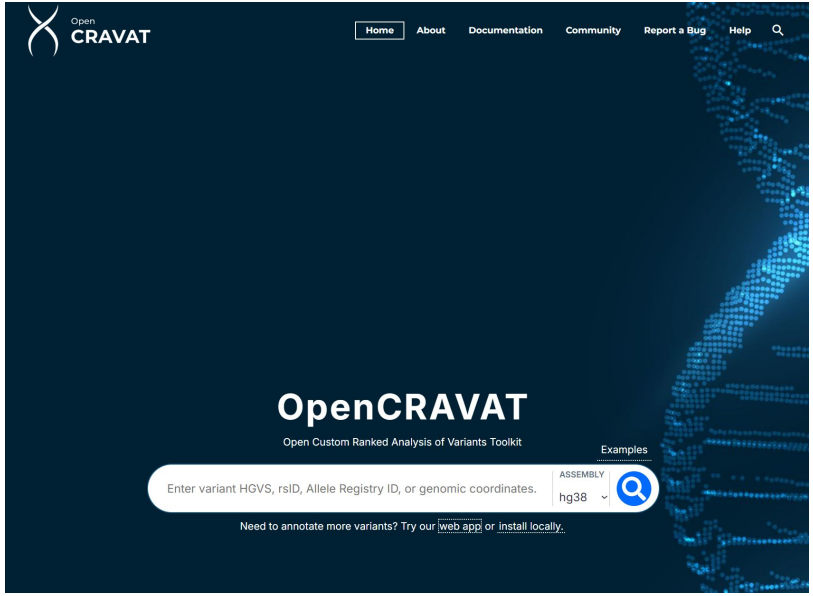
Tutorial to analyze and interactively visualize a VCF file using free web-based tools

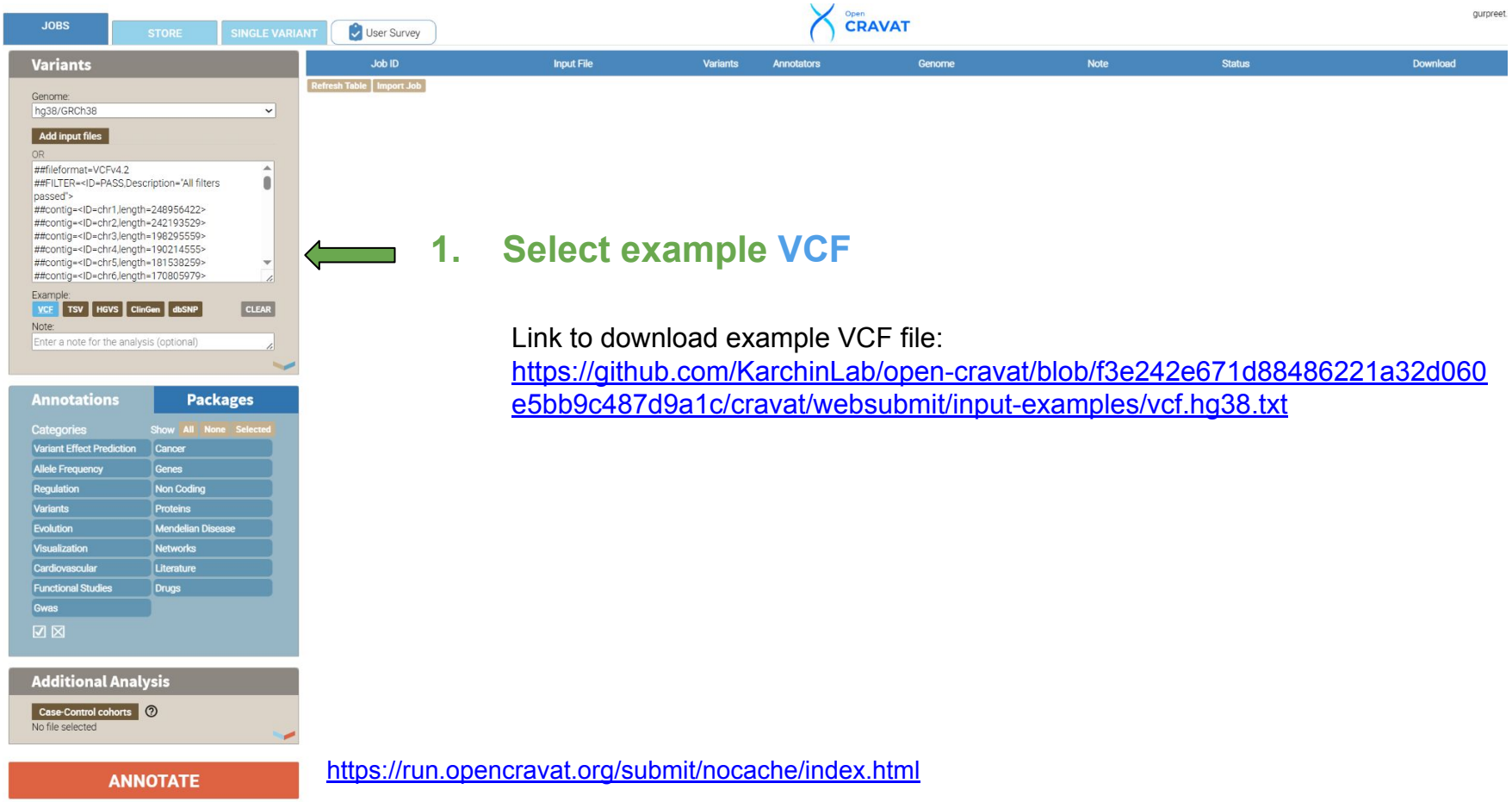
Gurpreet Kaur, Ph.D.

https://www.linkedin.com/in/gurpreet-bioin4/

1. OpenCRAVAT
Web-app



https://www.opencravat.org/
https://github.com/KarchinLab/open-cravat

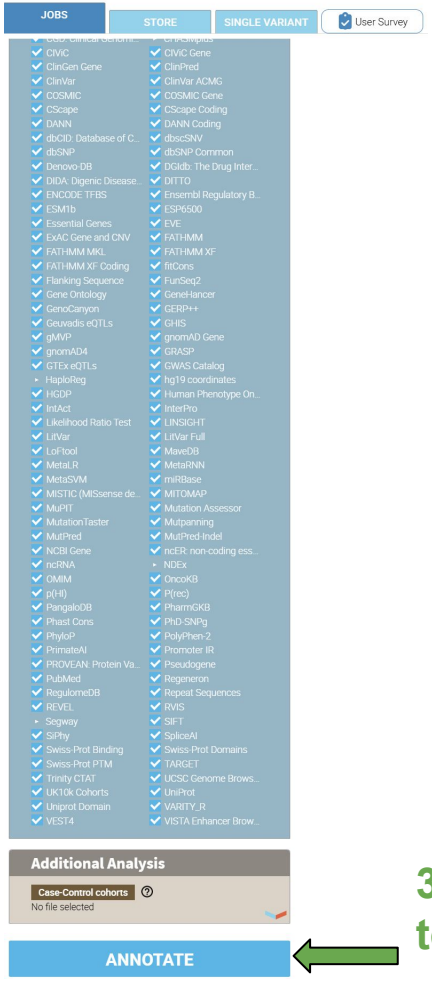


1. Select example VCF

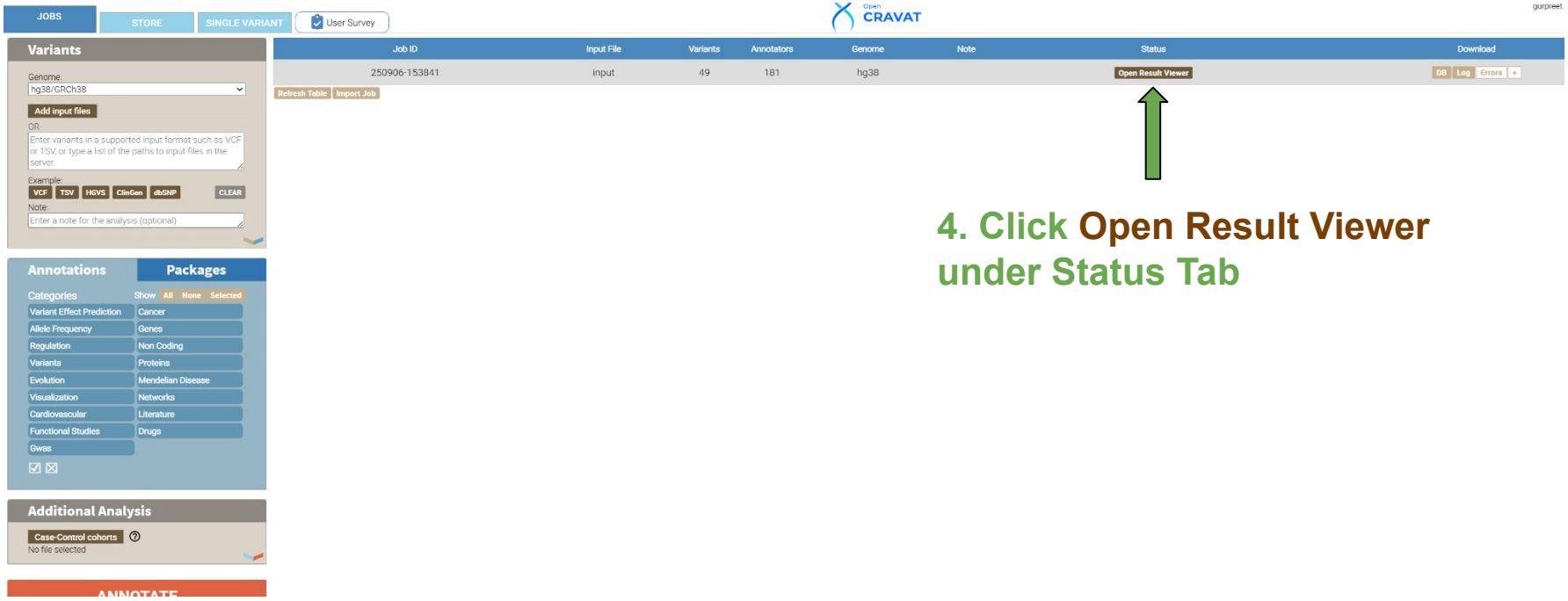
Link to download example VCF file:
https://github.com/KarchinLab/open-cravat/blob/f3e242e671d88486221a32d060e5bb9c487d9a1c/cravat/websubmit/input-examples/vcf.hg38.txt

https://run.opencravat.org/submit/nocache/index.html

2. Select Annotator (I selected all)

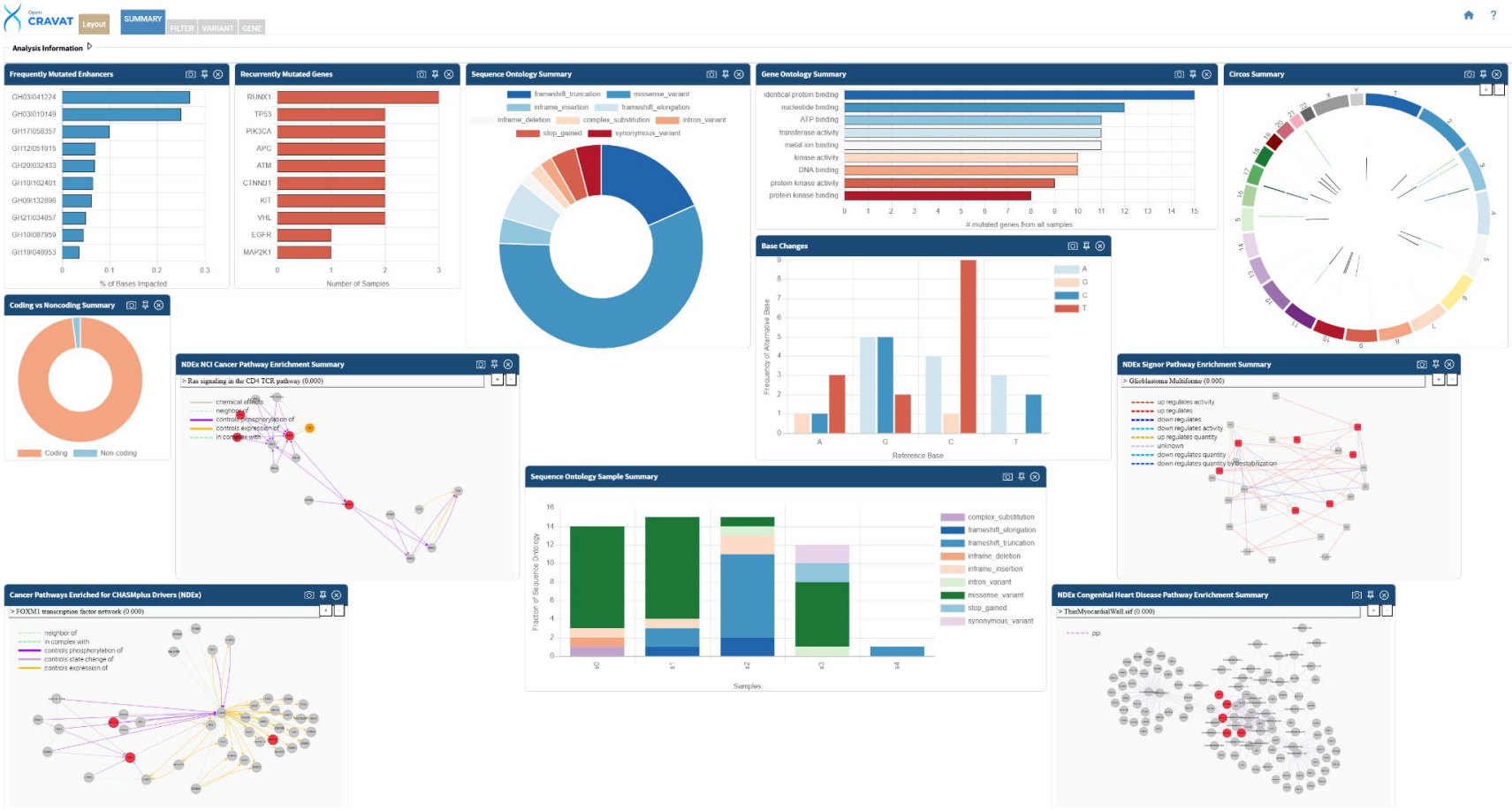


3. Click ANNOTATE to submit job



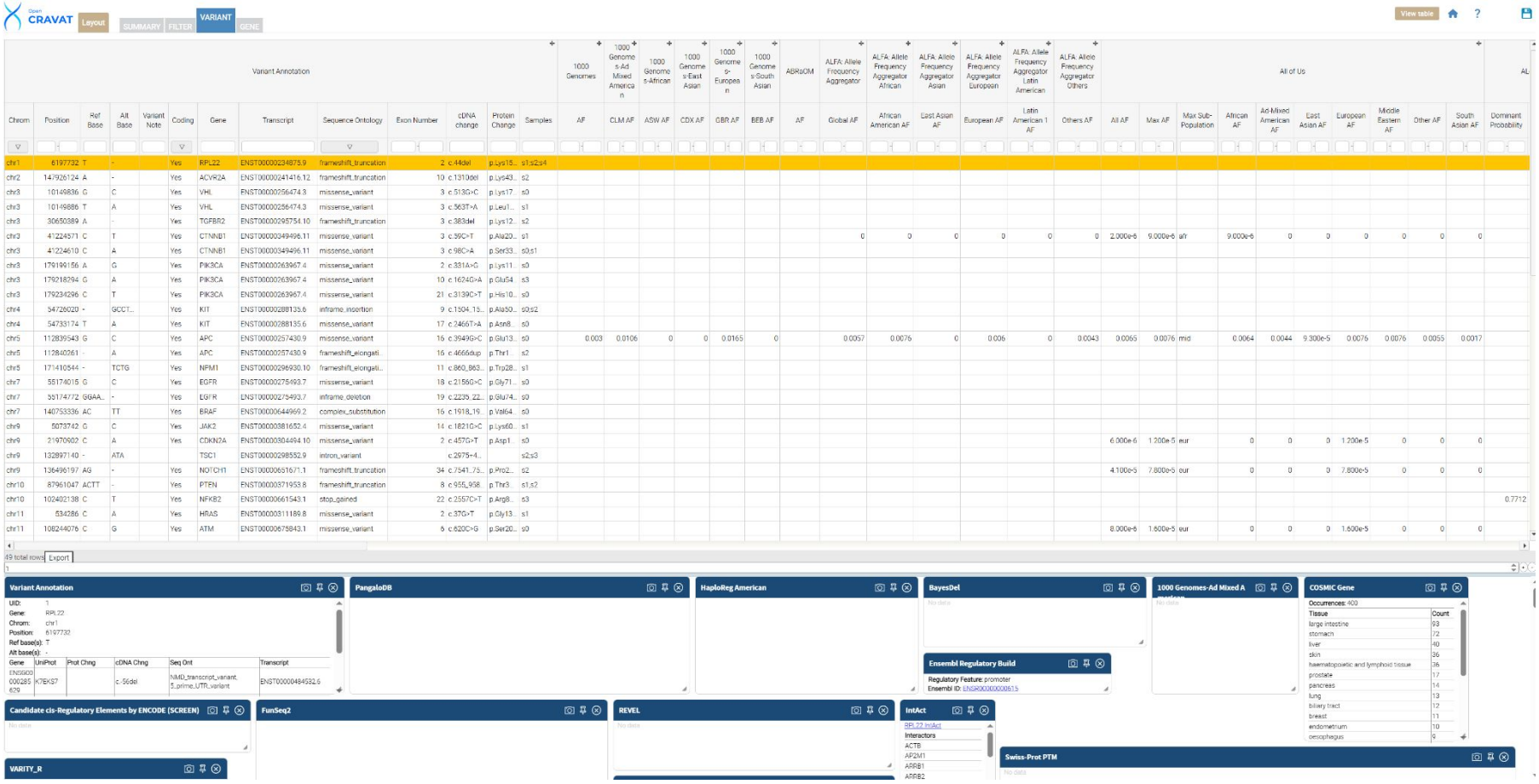
4. Click Open Result Viewer under Status Tab

5. Result Viewer: (i) Explore SUMMARY tab to interactively visualize annotated variants

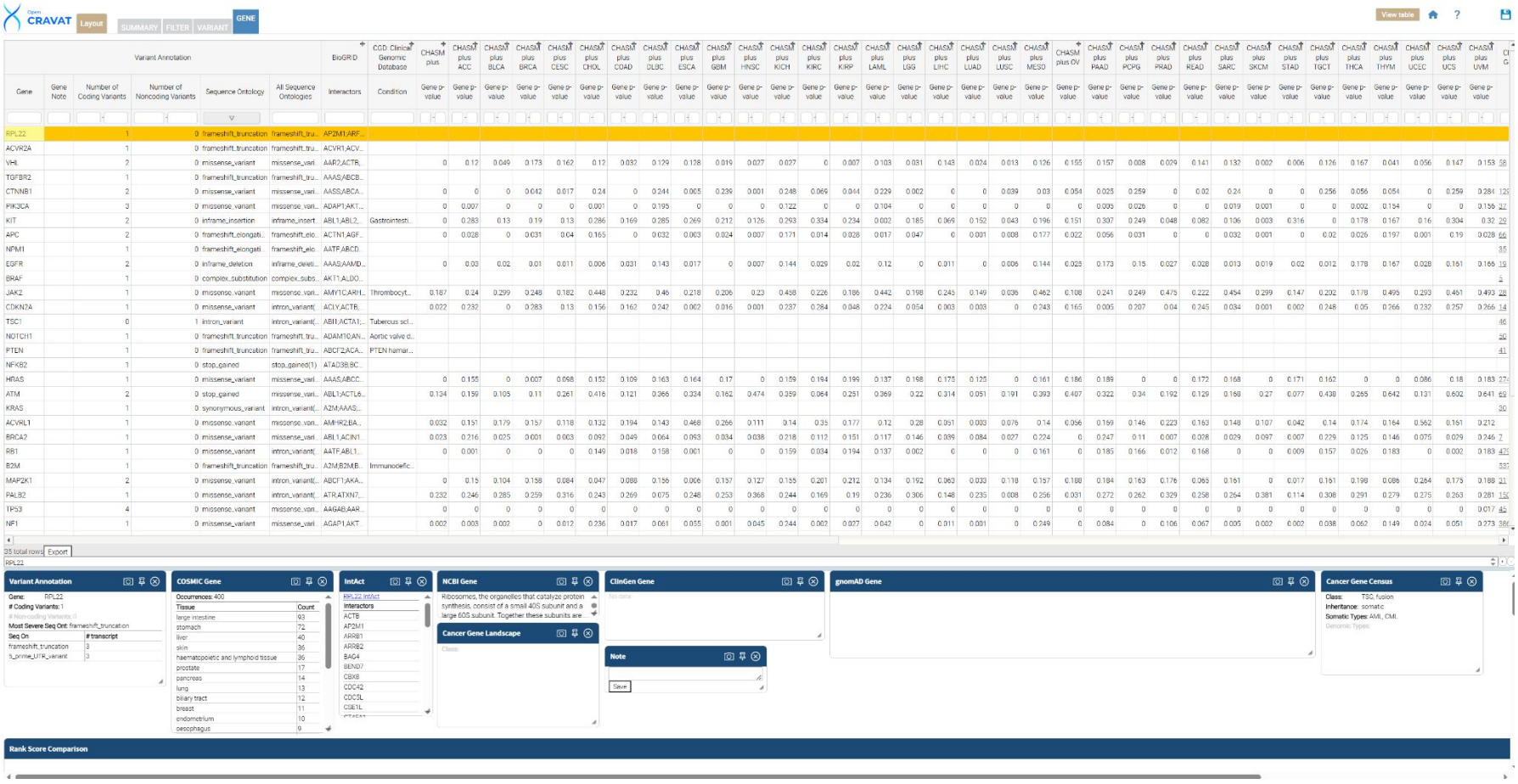


5. Result Viewer: (ii) Explore FILTER tab for various filtering options

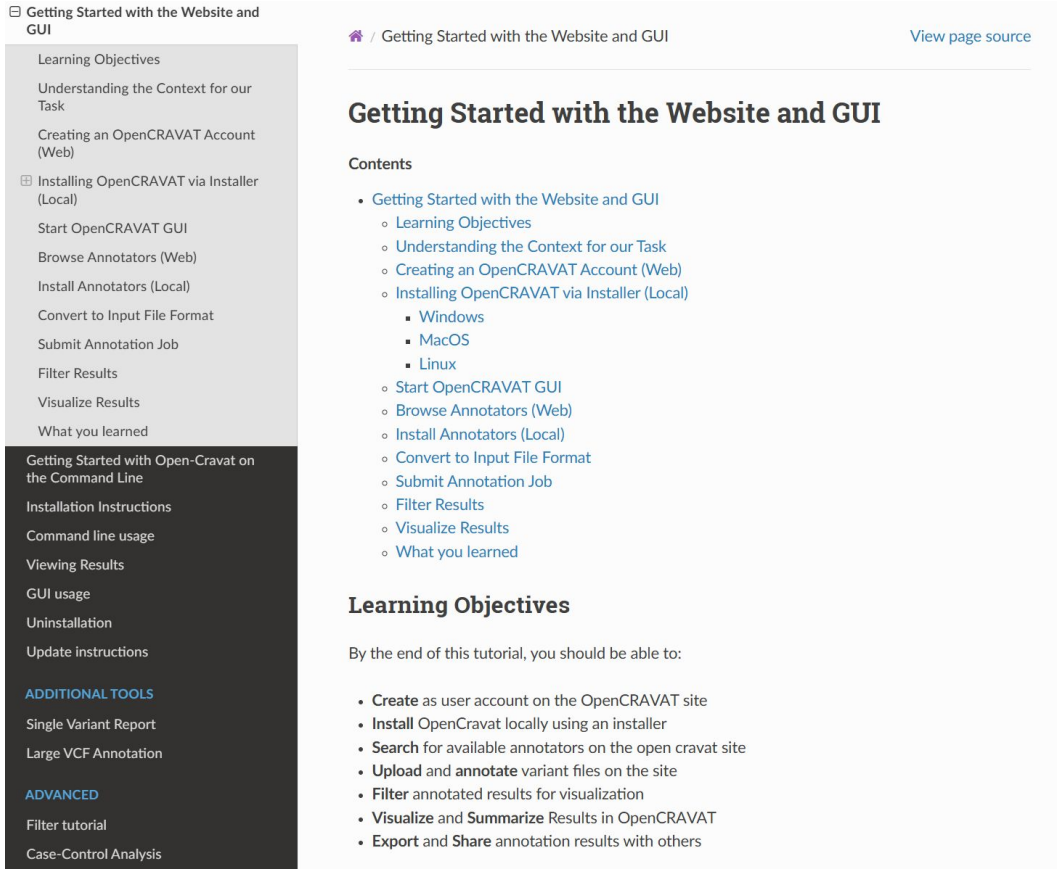
5. Result Viewer: (iii) Explore VARIANT tab for detailed variant table and export results



5. Result Viewer: (iv) Explore GENE tab for gene-based table and export results



6. For more details: https://docs.opencravat.org/en/latest/getting_started_web.html



Getting Started with the Website and GUI

- Learning Objectives
- Understanding the Context for our Task
- Creating an OpenCRAVAT Account (Web)
- Installing OpenCRAVAT via Installer (Local)
- Start OpenCRAVAT GUI
- Browse Annotators (Web)
- Install Annotators (Local)
- Convert to Input File Format
- Submit Annotation Job
- Filter Results
- Visualize Results
- What you learned
- Getting Started with Open-Cravat on the Command Line
- Installation Instructions
- Command line usage
- Viewing Results
- GUI usage
- Uninstallation
- Update instructions

ADDITIONAL TOOLS

- Single Variant Report
- Large VCF Annotation

ADVANCED

- Filter tutorial
- Case-Control Analysis

/ Getting Started with the Website and GUI — View page source

Getting Started with the Website and GUI

Contents

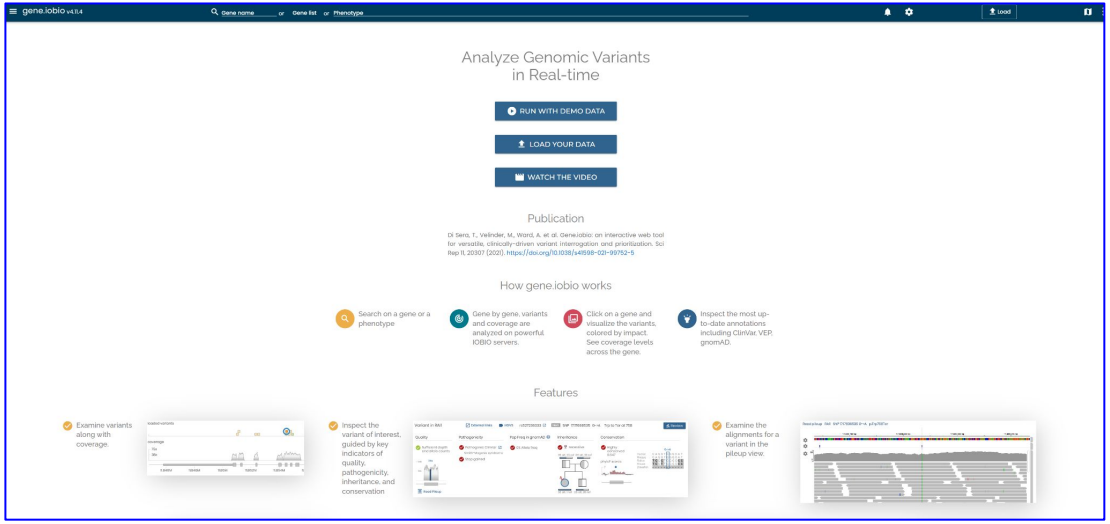
- Getting Started with the Website and GUI
  - Learning Objectives
  - Understanding the Context for our Task
  - Creating an OpenCRAVAT Account (Web)
  - Installing OpenCRAVAT via Installer (Local)
    - Windows
    - MacOS
    - Linux
  - Start OpenCRAVAT GUI
  - Browse Annotators (Web)
  - Install Annotators (Local)
  - Convert to Input File Format
  - Submit Annotation Job
  - Filter Results
  - Visualize Results
  - What you learned

Learning Objectives

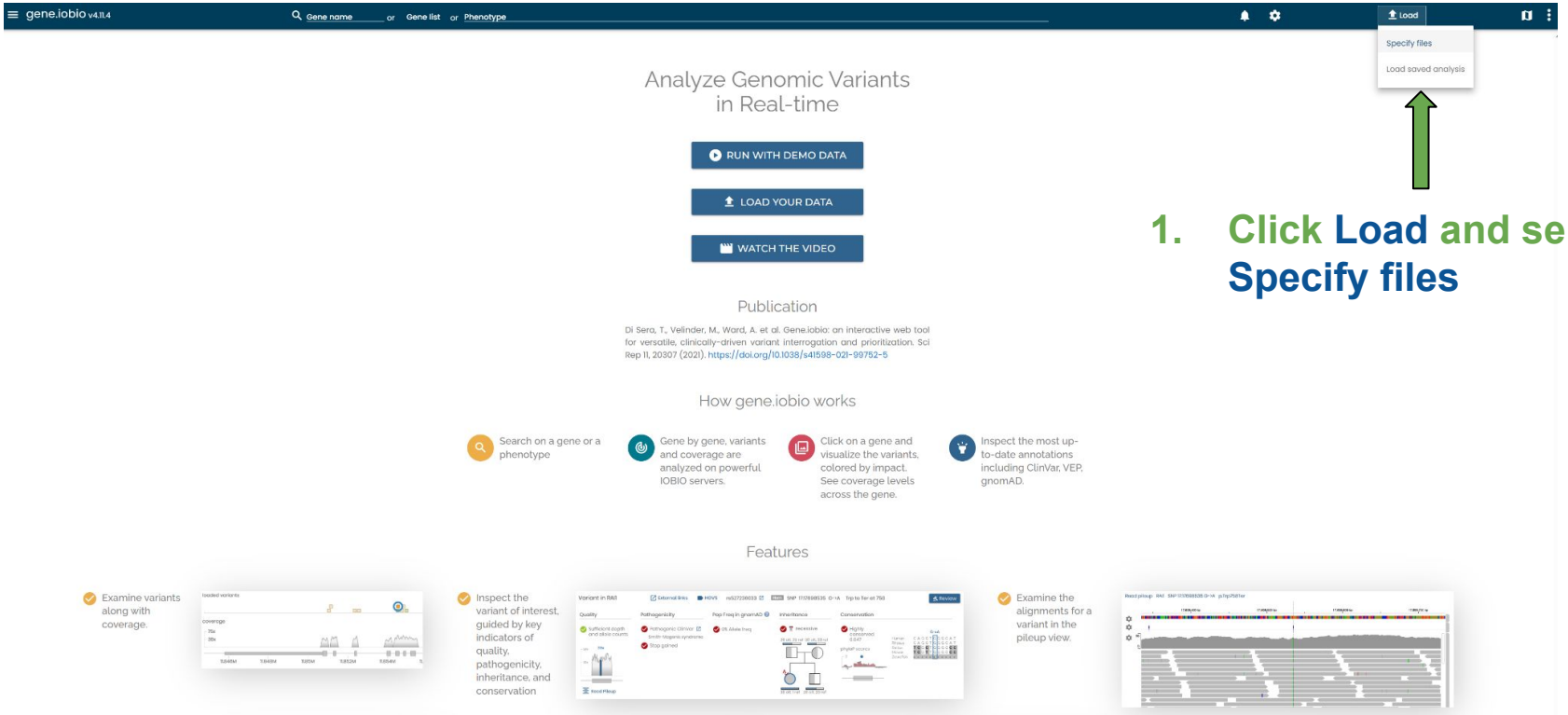
By the end of this tutorial, you should be able to:

- **Create** as user account on the OpenCRAVAT site
- **Install** OpenCravat locally using an installer
- **Search** for available annotators on the open cravat site
- **Upload** and **annotate** variant files on the site
- **Filter** annotated results for visualization
- **Visualize** and **Summarize** Results in OpenCRAVAT
- **Export** and **Share** annotation results with others

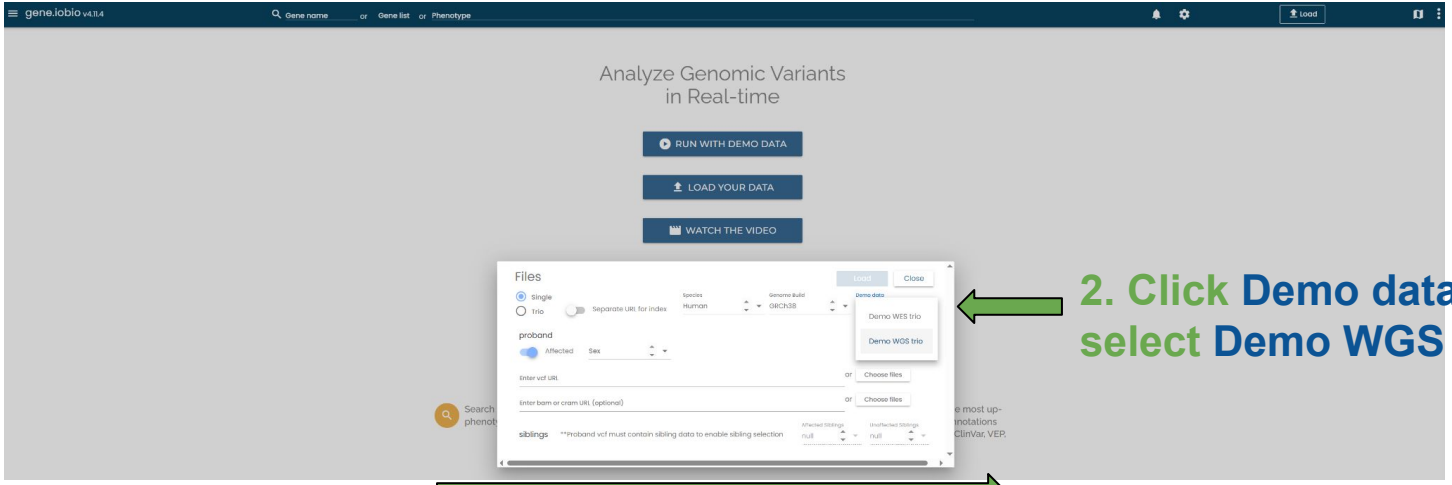
2. Gene.iobio
Web-app



https://gene.iobio.io/
https://github.com/iobio/gene.iobio

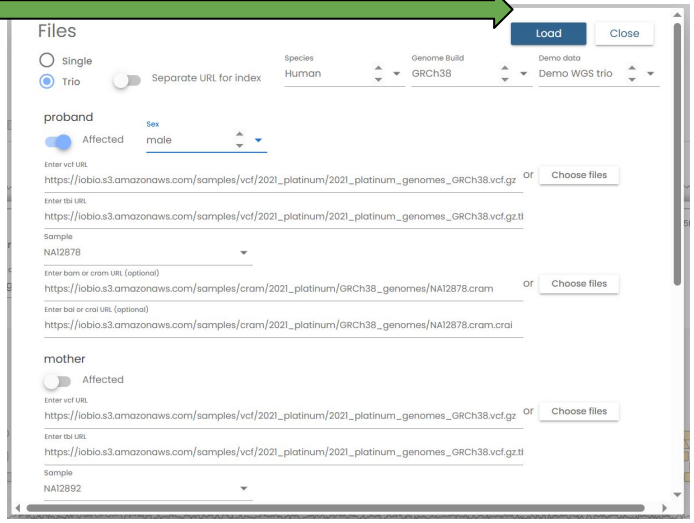


1. Click **Load** and select **Specify files**

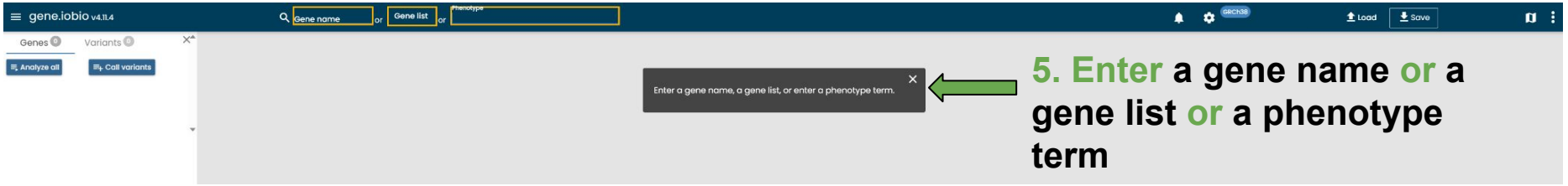


2. Click **Demo data** and select **Demo WGS trio**

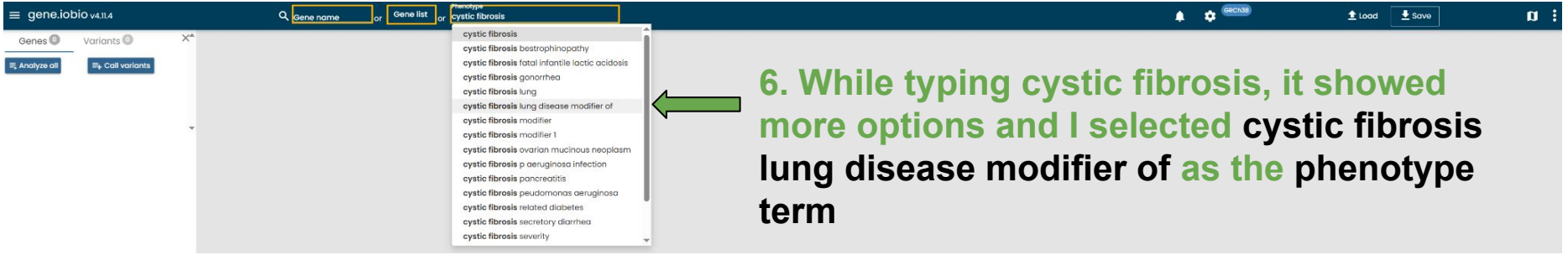
4. Click **Load** to load **Demo data**



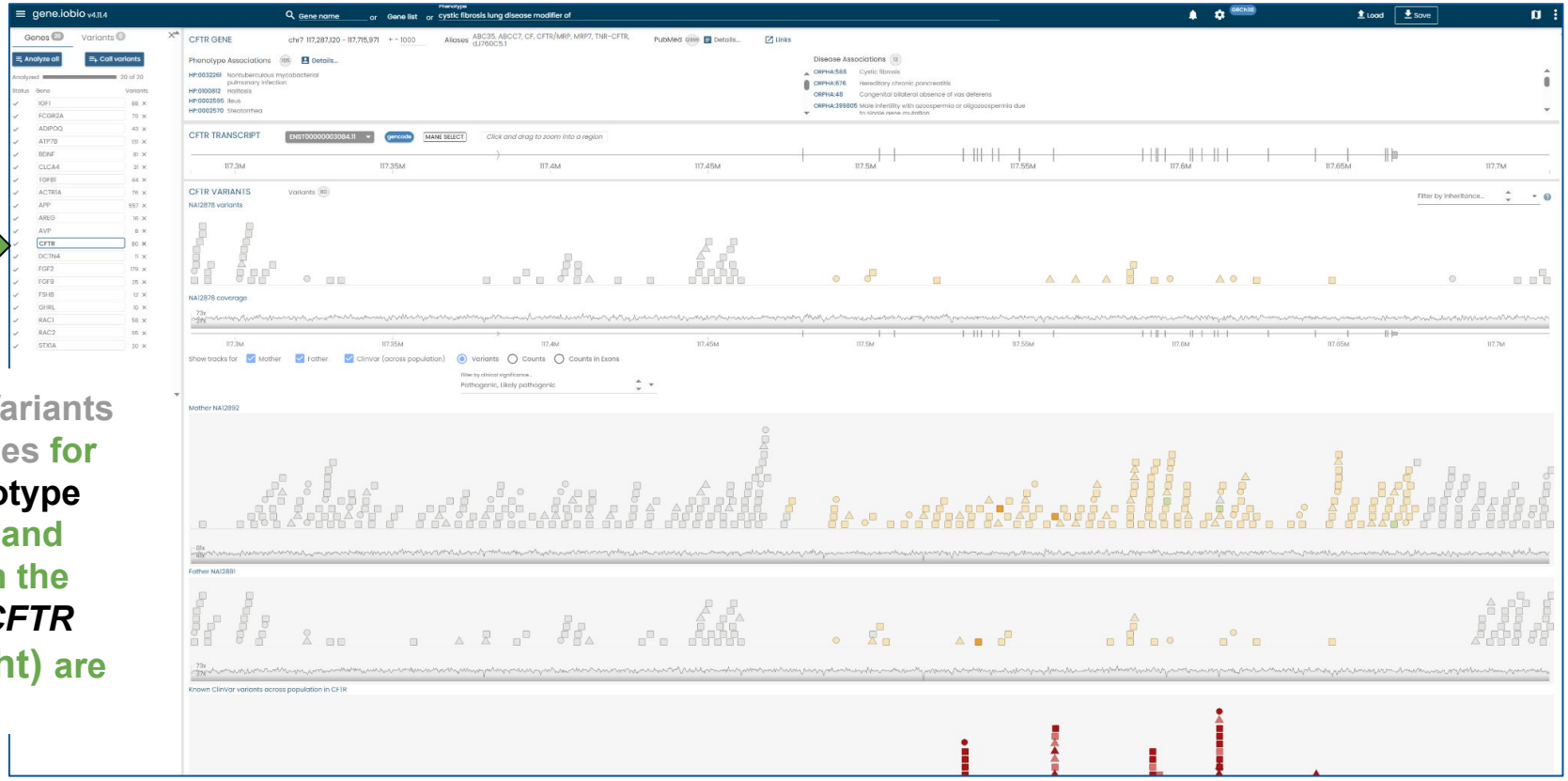
3. It will open **WGS trio** samples information: proband, mother and father for exploration



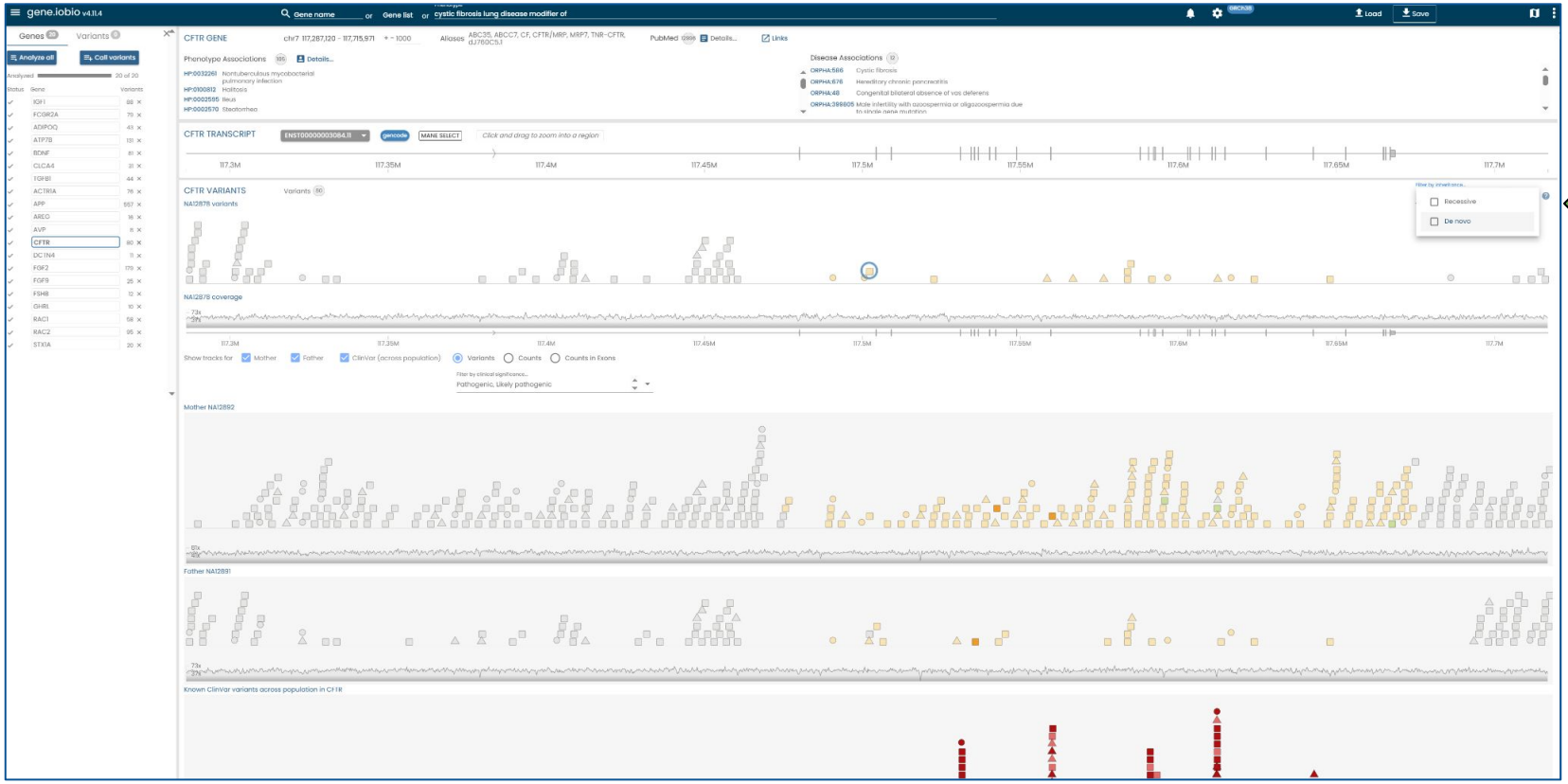
5. Enter a gene name or a gene list or a phenotype term



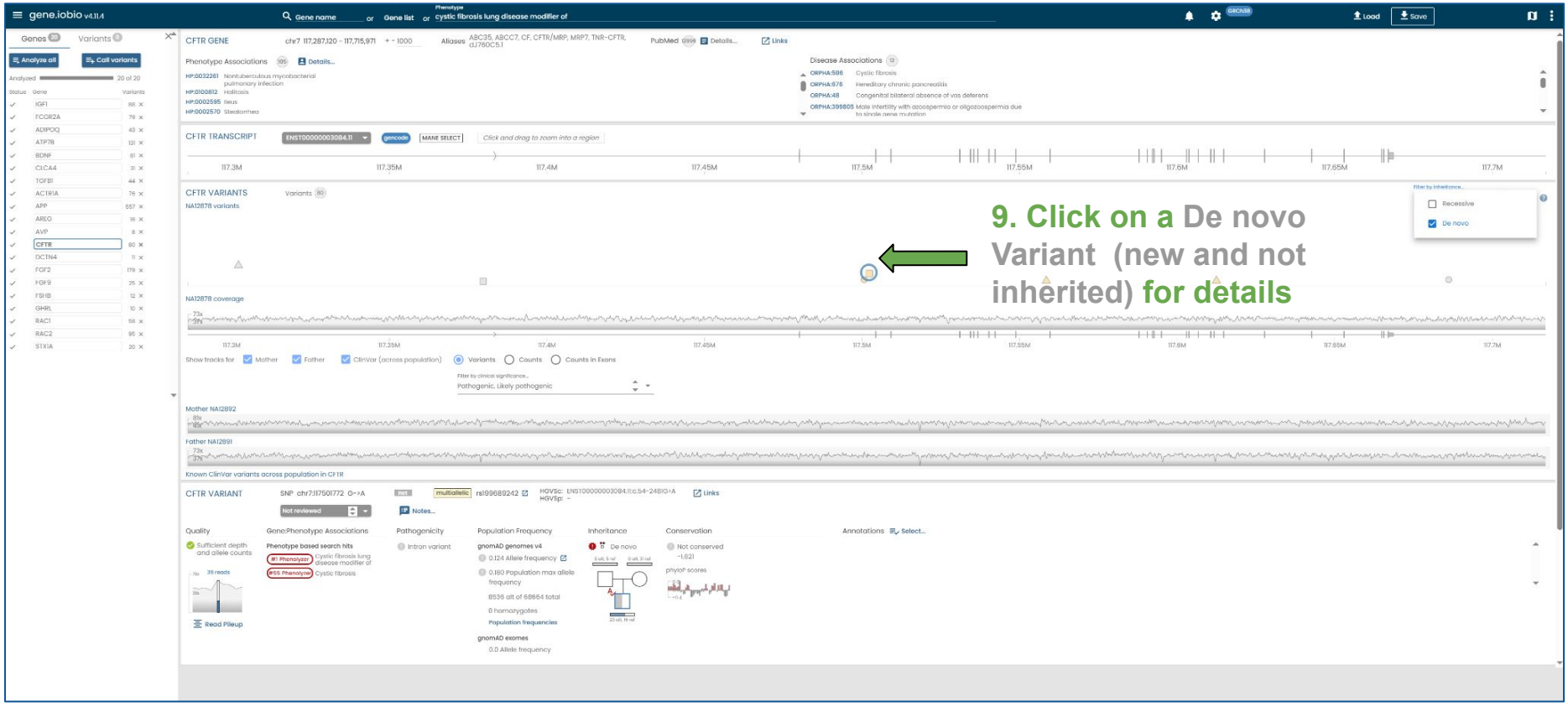
6. While typing cystic fibrosis, it showed more options and I selected cystic fibrosis lung disease modifier of as the phenotype term



7. No. of Variants in the Genes for that phenotype term (left) and Variants in the selected *CFTR* Gene (right) are shown



8. Filter by inheritance:
De novo



9. Click on a De novo Variant (new and not inherited) for details

10. For more details and options: https://gene.iobio.io/tutorial