

Genomic Public Resource Workshop: Integrative Genomics Viewer (IGV)

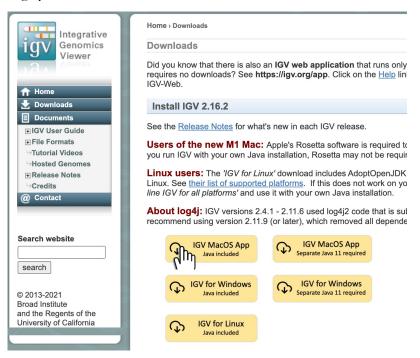
Genomic researchers use genomic visualization browsers such as IGV to explore the genome.

Common
Terminology
&
Supporting
Resources

- Gene: A gene is the basic physical and functional unit of heredity (link).
- Genetic variant: A gene variant is a permanent change in the DNA sequence that makes up a gene (link).
- Variant nomenclature: Genetic variants (and sometimes mutations or polymorphisms), are described using
 a specific language. A standard has been developed for this language, the HGVS nomenclature standard (link).
- rsID: Single Nucleotide Polymorphism (SNP) is a location in the genome that is known to differ between individuals. The rs number is used by researchers and databases to refer to specific SNPs (link).
- **Genome build:** Successive "versions" of the human genome reference, commonly called assemblies or builds, have been published since the original draft Human Genome Project publication (link).
- Tracks: Genome Browser annotation tracks are based on files in line-oriented format. Each line in the file defines a display characteristic for the track or defines a data item within the track. Annotation files contain three types of lines: browser lines, track lines, and data lines (link).

To download IGV (desktop version):

Go to https://software.broadinstitute.org/software/igv/download, and select the version based on your operating system.



Note: We recommend you use the version that includes Java.

- Once downloaded to your local computer, unzip file (for Mac), then begin install by opening IGV_2.16.2 (Mac) or IGV_Win_2.16.2-WithJava-installer.exe (Windows).
- 3 You have now downloaded IGV and are on your way to becoming a genomicist.

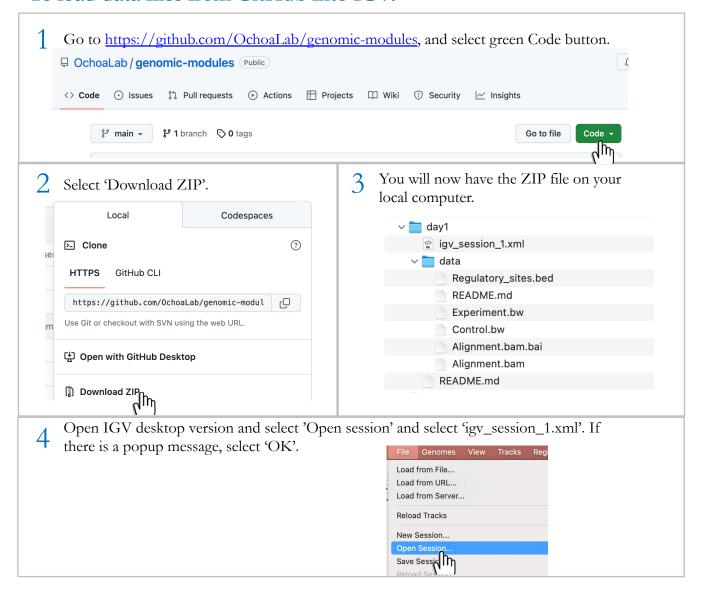


Rev. 9/7/23 1 of 2



Genomic Public Resource Workshop: Integrative Genomics Viewer (IGV)

To load data files from GitHub into IGV:



Rev. 9/7/23 2 of 2