African Genome Variation Database

Tutorial

Searching

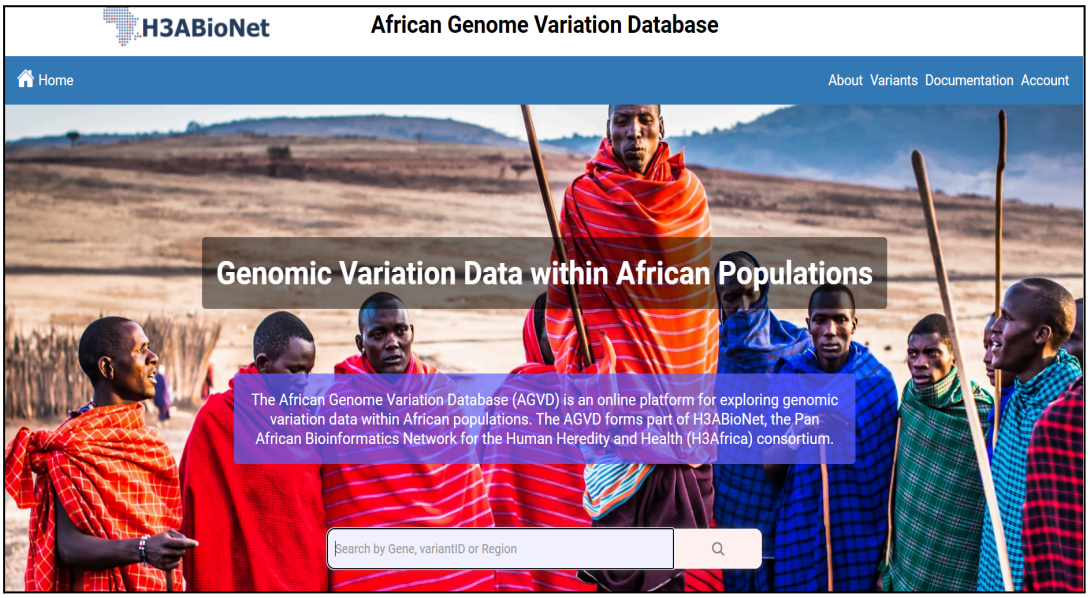


Figure 1: AGVD main search page

1. On the homepage, users can explore a world map view presenting the number of samples per country along with their corresponding projects. This feature utilizes data to visually represent the sample counts and their associated projects, all displayed based on their geographical locations.

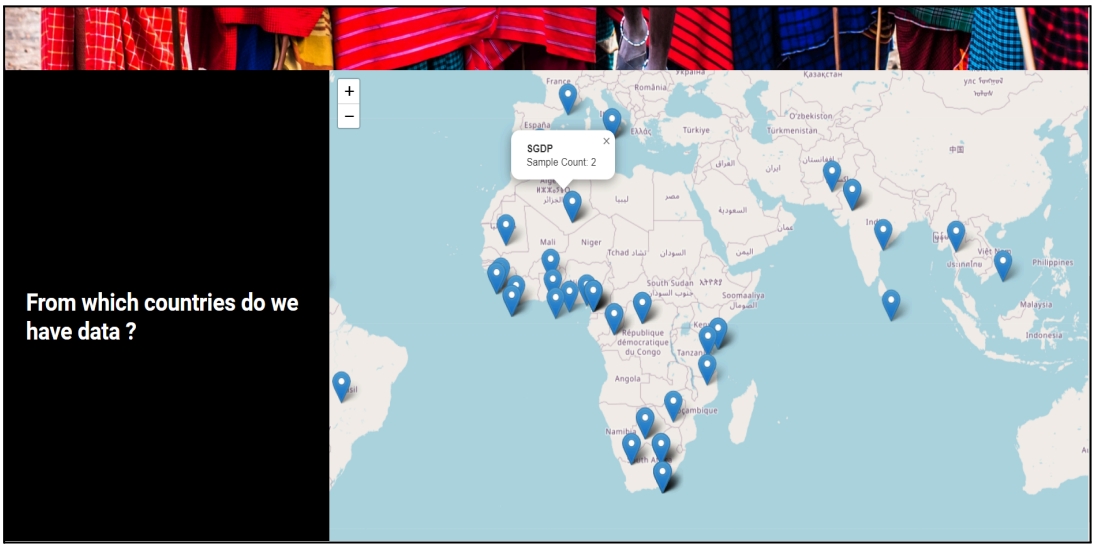


Figure 2: Interactive World Map Displaying Sample Counts and Project Associations

2. **Search by Data category:**

4.1 Search by variant: The search bar enables users to search for variants using various formats, including Reference Sequence (RefSeq) identifiers (e.g., rs116600158), Ensembl gene or transcript identifiers (e.g., ENSG00000100342, ENST00000397278), or genomic locations or ranges (e.g., 19:7177679:C:T)

4.2 Search by Gene name: The search bar enables the users to search from the list of Genes names, e.g WASH2P

4.3 Search by specific genomic region: The search bar enable users to search for region in the format chr:start-end, e.g. 1:1000-40000

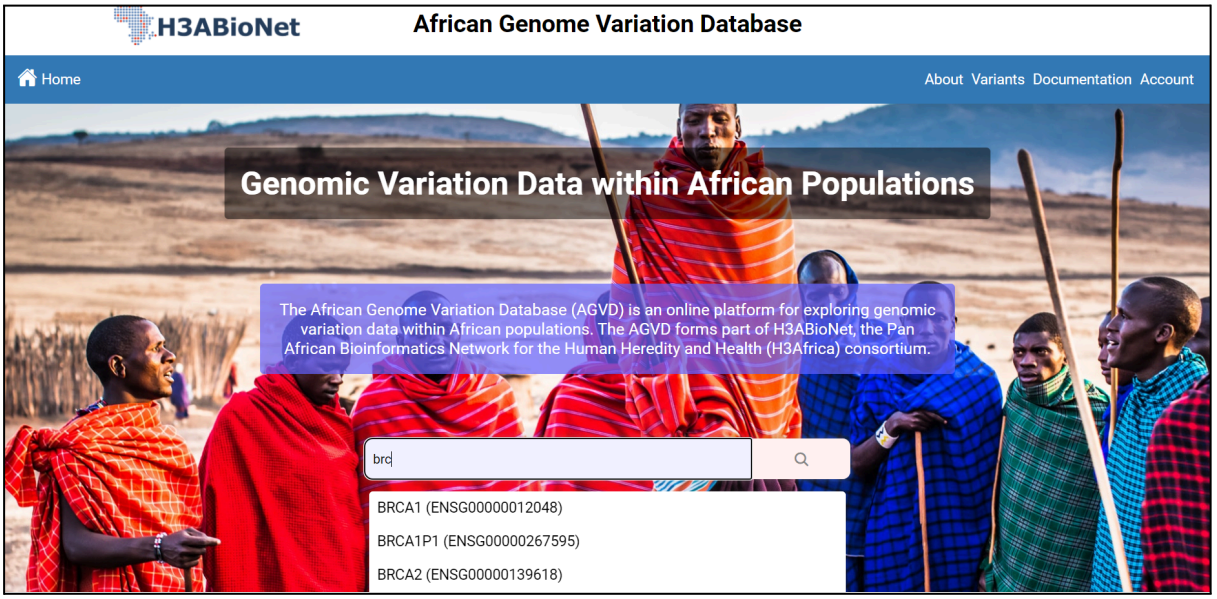


Figure 3: AGVD search by Variant. The search bar also includes a “type-hint” feature with the ability to auto-complete a search

Results

The following section discusses the results pages in relation to results obtained from WASH2P, 1:1000-40000, and 2-113601195-G-A searches.

1. **Gene:** When searching by gene, a list of relevant genes will appear below the search box. Simply click on your desired gene and press enter to access the gene results page

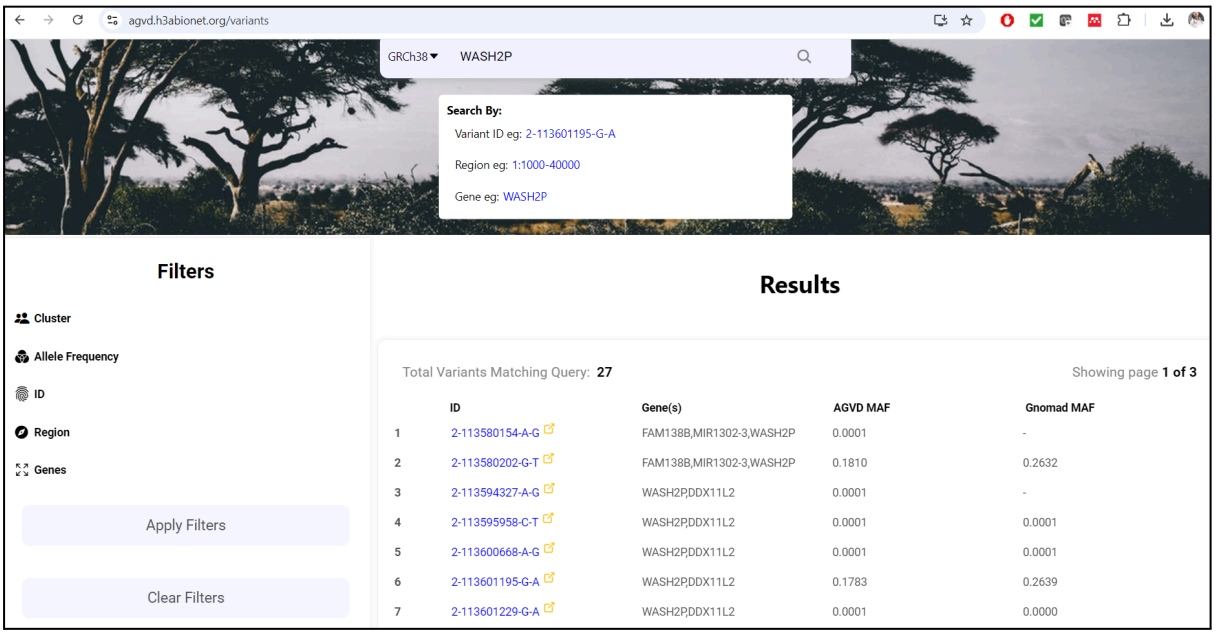
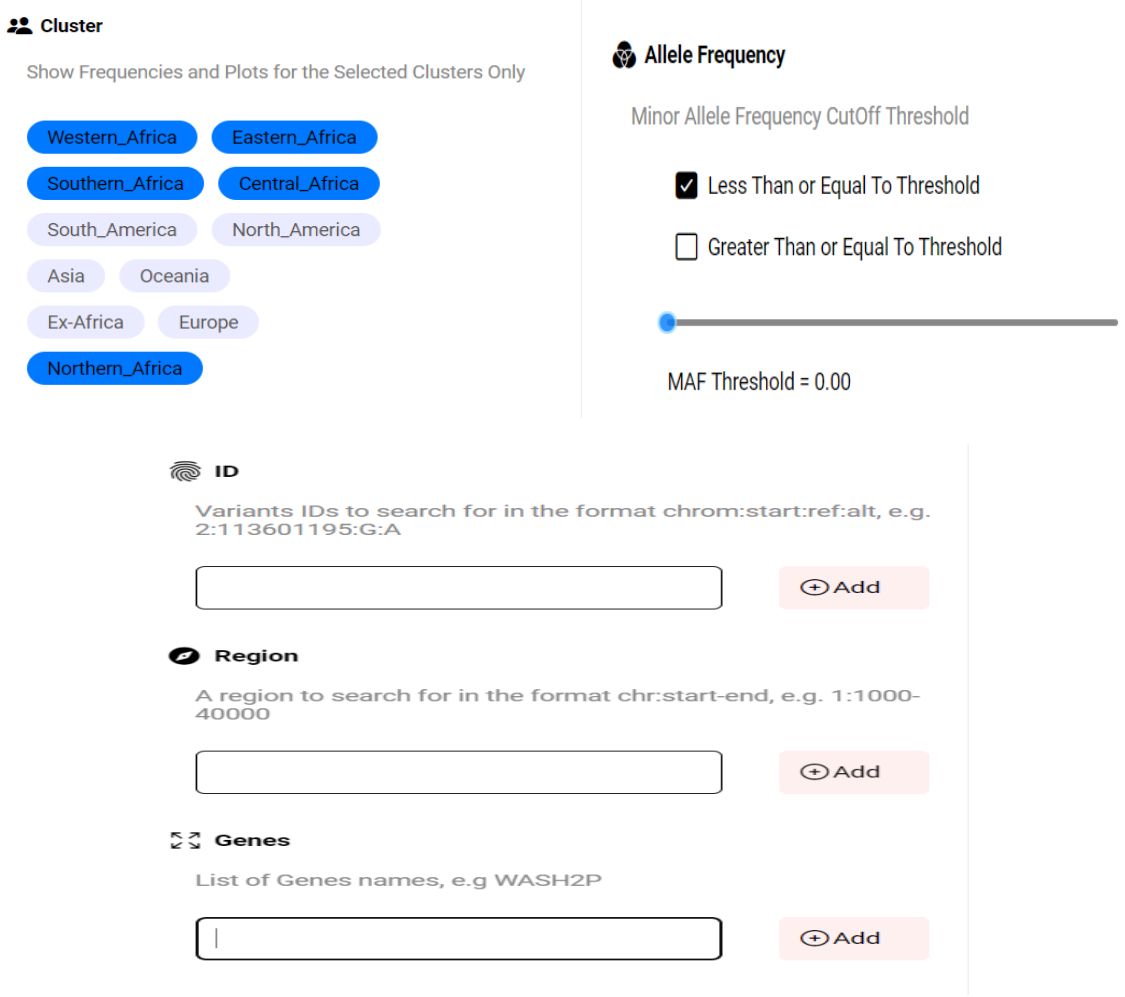


Fig 4: *WASH2P* gene results page

The gene results page displays a list of variants matching the selected gene, along with the AGVD MAF and GenomAD MAF for each variant.

❖ Filters

On the results page, users are presented with a list of filter parameters. Users can access these filters by interacting with the accordion menu in the filter panel. The available filter menus in the pilot release include “Cluster,” “Allele Frequency,” “VariantIDs,” , “Region,” and “Genes.” Each filter menu, when expanded, provides explanatory text to assist users in understanding the acceptable types of parameters, the correct format for inputting them, and examples of valid search terms for each type of filter parameter.



2. **Variant ID:** Type your variantID on the search box. Then, click on the search button to access the variant results page

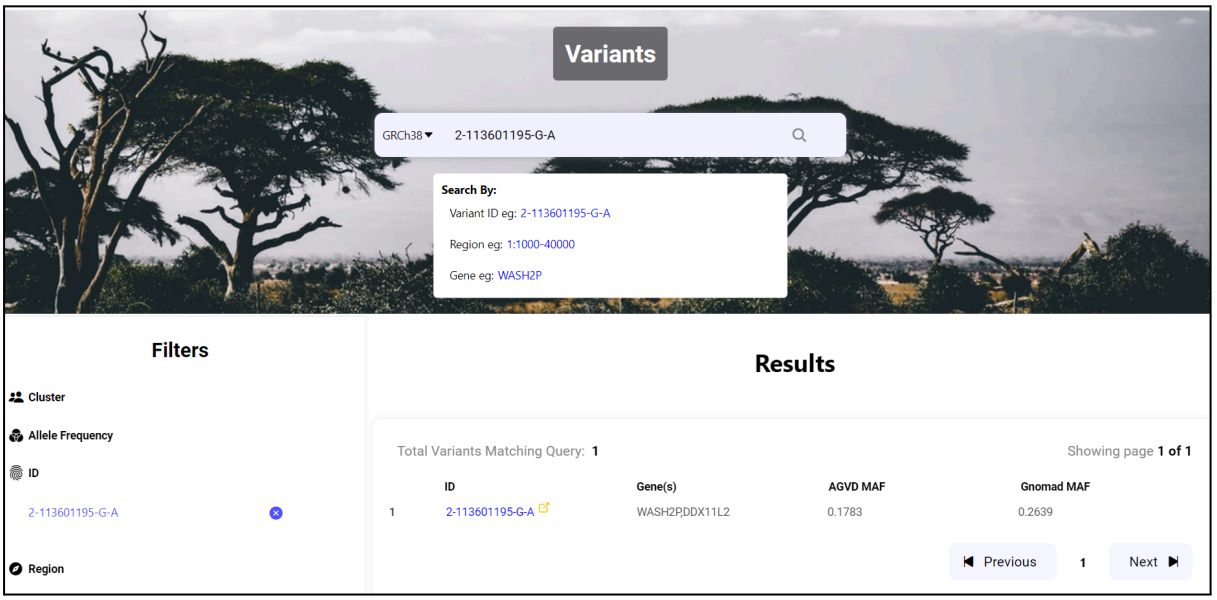


Fig: *2-113601195-G-A* variant results page

The variant results page displays the variant and the related gene, along with the AGVD MAF and GenomAD MAF for this variant.

3. **Region:** Type the rgeion you are searching for on the search box. Then, click on the search button to access the region results page

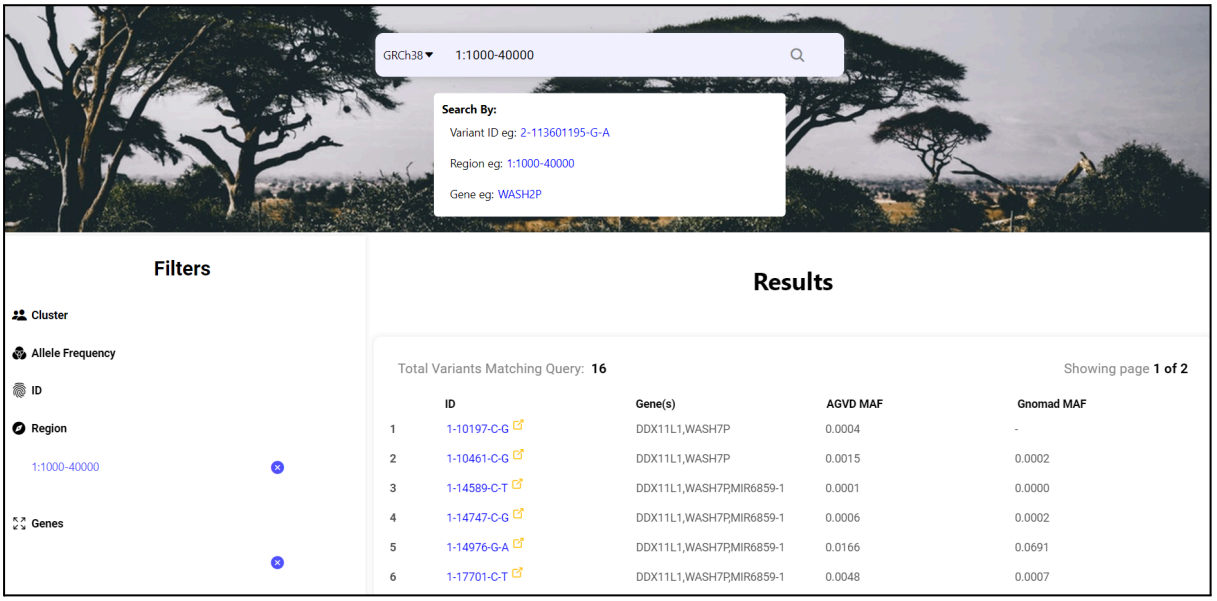


Fig: *2-113601195-G-A* region results page

The region results page displays a list of variants and their related genes matching the selected region, along with the AGVD MAF and GenomAD MAF for each variant.

Filter file

If you have a list of genes, rsids or positions (and alleles), this can be used as a filter byt using the File Upload feature...