

African Genomic Medicine Portal

Tutorial



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Introduction

The African Genomic Medicine Portal (AGMP) functions as a curated resource for researchers around the world who are conducting genomics research on African and African-ancestry populations. The portal may also be useful for individuals working in the health sector, such as healthcare workers, pharmacists, and policymakers, though it was designed as a research tool and should not be used for clinical decisions. The portal functions as a gateway to data relevant for African genomic medicine research, including pharmacogenomics and clinical/disease research, accessing, and providing African-specific data from existing resources in an easily accessible manner.

AGMP retrieves and curates data from various resources. The current release contains data from PharmGKB and DisGeNET.

This tutorial provides a step by step guide to searching data in the Portal.

Search:

1. Access AGMP using the following link: <https://agmp.h3abionet.org/>. On this page, **four main data categories** will be displayed: **Disease**, **Drug**, **Variant**, and **Gene**, as illustrated **Figure 1**.

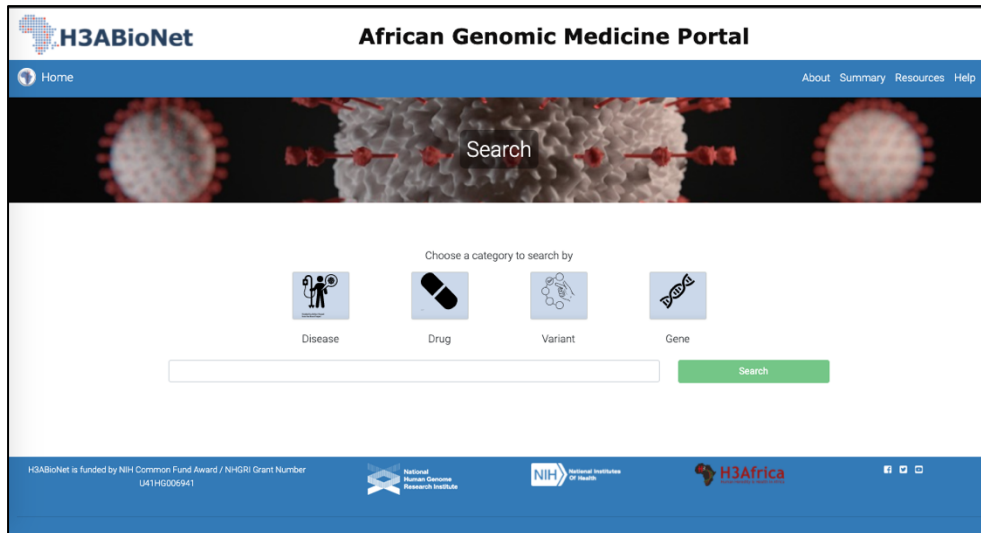


Figure 1. AGMP search page.

2. A user may search based on their preferred data category by selecting the *corresponding data category logo* (Disease, Drug, Variant or Gene), as illustrated in **Figure 2A-D**. **Results are customized according to the data category selected.**

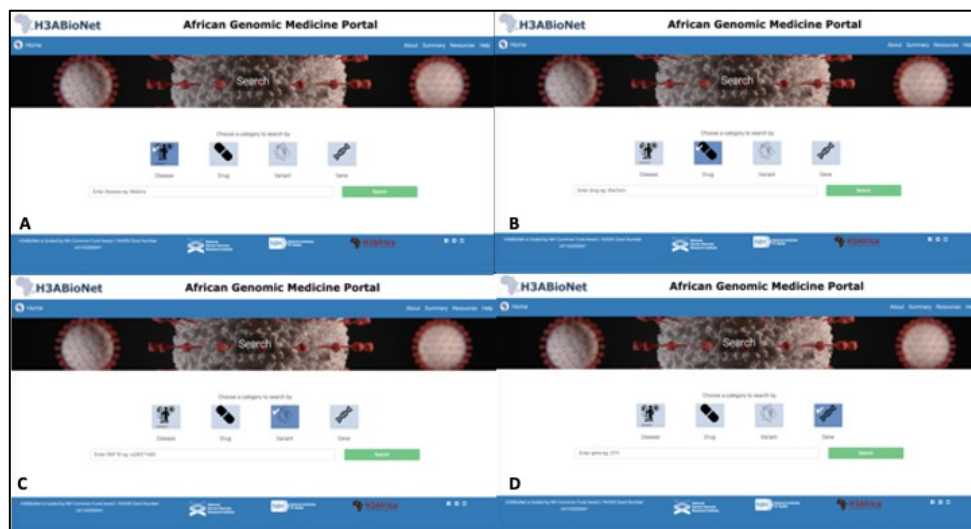


Figure 2. A) Disease category selected; B) Drug category selected; C) Variant category selected; D) Gene category selected.

3. Enter a search term into the search box and select *Search*. Matching results will appear below the text box, as illustrated in **Figure 3A-D**.

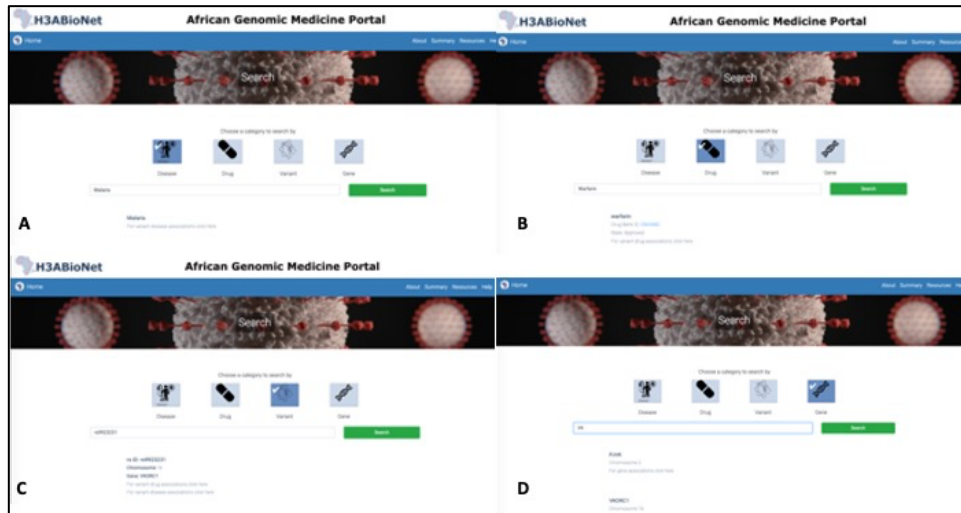


Figure 3. A) Disease result; B) Drug result; C) Variant result; D) Gene result.

Results:

*NB: Results are discussed by the data search category selected during **Search**.*

1. Disease Results:

- 1.1. When searching by a disease, a list of relevant results appears below the search box. To proceed to complete results, select the “For variant-disease associations click here” button, as illustrated in **Figure 3A**.
- 1.2. As illustrated in **Figure 4**, the results page will contain a list of variant-disease associations. Table contents are described in **Table 1**.

Variant	Disease	Gene	Significance	Country	Studies
rs10192428	Malaria	SPATA3	0.00000051	The Gambia	Genome-wide and fine-resolution association analysis of malaria in West Africa.
rs1046089	Malaria	PRRC3A	< 0.0001	The Gambia	A genetic association study in the Gambia using tagging polymorphisms in the major histocompatibility complex class III region implicates a HLA-B associated transcript 2 polymorphism in severe malaria susceptibility.
rs10900585	Malaria	ATP2B4	0.000000061	Ghana	Genome-wide association study indicates two novel resistance loci for severe malaria.
rs10900585	Malaria	ATP2B4	0.0052	Ghana	Genome-wide association study indicates two novel resistance loci for severe malaria.
rs11335470	Malaria	LINC00944	0.00000904	Tanzania	Novel genetic polymorphisms associated with severe malaria and under selective pressure in North-eastern Tanzania.
rs114169033	Malaria	FRG1-01	0.000000562	Tanzania	Novel genetic polymorphisms associated with severe malaria and under selective pressure in North-eastern Tanzania.
rs12405994	Malaria	AC092813.1	0.00000082	The Gambia	Genome-wide and fine-resolution association analysis of malaria in West Africa.
rs12788102	Malaria	MMP25	< 0.001	The Gambia, Kenya, Malawi	Imputation-based meta-analysis of severe malaria in three African populations.
rs12788102	Malaria	OR51F1	< 0.001	The Gambia, Kenya, Malawi	Imputation-based meta-analysis of severe malaria in three African populations.
rs12789492	Malaria	MMP25	< 0.001	The Gambia, Kenya, Malawi	Imputation-based meta-analysis of severe malaria in three African populations.

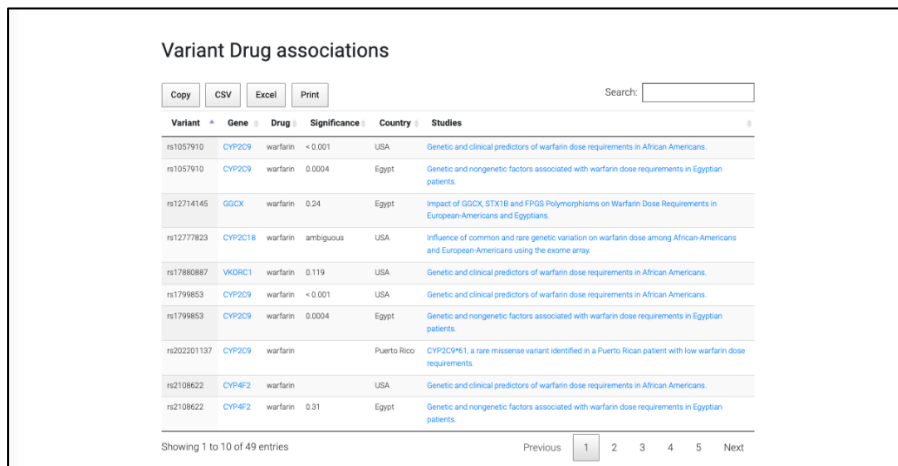
Figure 4. Tabulated Disease result.

Table 1. Descriptions of Disease results column headers.

Variant	The genetic variants which have been associated with the disease of interest.
Disease	The disease of interest entered in the search box.
Gene	The gene in which the variant is located.
Significance	The p-value observed for the association in the given study.
Country	The country of origin of the research participants.
Studies	The study associated with the result.

2. Drug Results:

- 2.1. When searching by a drug, a list of relevant results appears below the search box. To proceed to complete results, select the “For variant-drug associations click here” button, as illustrated in **Figure 3B**.
- 2.2. As illustrated in **Figure 5**, the results page will contain a list of variant-drug associations. Table contents are described in **Table 2**.



Variant	Gene	Drug	Significance	Country	Studies
rs1057910	CYP2C9	warfarin	< 0.001	USA	Genetic and clinical predictors of warfarin dose requirements in African Americans.
rs1057910	CYP2C9	warfarin	0.0004	Egypt	Genetic and nongenetic factors associated with warfarin dose requirements in Egyptian patients.
rs12714145	GGCX	warfarin	0.24	Egypt	Impact of GGCK, STX1B and FPGS Polymorphisms on Warfarin Dose Requirements in European-Americans and Egyptians.
rs12777823	CYP2C18	warfarin	ambiguous	USA	Influence of common and rare genetic variation on warfarin dose among African-Americans and European-Americans using the exonuclease array.
rs1780887	VKORC1	warfarin	0.119	USA	Genetic and clinical predictors of warfarin dose requirements in African Americans.
rs1799853	CYP2C9	warfarin	< 0.001	USA	Genetic and clinical predictors of warfarin dose requirements in African Americans.
rs1799853	CYP2C9	warfarin	0.0004	Egypt	Genetic and nongenetic factors associated with warfarin dose requirements in Egyptian patients.
rs202201137	CYP2C9	warfarin		Puerto Rico	CYP2C9*1, a rare missense variant identified in a Puerto Rican patient with low warfarin dose requirements.
rs2108622	CYP4F2	warfarin		USA	Genetic and clinical predictors of warfarin dose requirements in African Americans.
rs2108622	CYP4F2	warfarin	0.31	Egypt	Genetic and nongenetic factors associated with warfarin dose requirements in Egyptian patients.

Figure 5. Tabulated Drug result.

Table 2. Descriptions of Drug results column headers.

Variant	The genetic variants which have been associated with the drug of interest.
Drug	The drug of interest entered in the search box.
Gene	The gene in which the variant is located.
Significance	The p-value observed for the association in the given study.
Country	The country of origin of the research participants.
Studies	The study associated with the result.

3. Variant Results:

- 3.1. When searching by a variant, a list of relevant results appears below the search box. To proceed to complete results, select either the “For variant-

disease associations click here” or the “For variant-drug associations click here” button, as illustrated in **Figure 3C**.

- 3.2. Based on the selected button, the results page will contain a list of either variant-disease or variant-drug associations, as illustrated in **Figure 4** and **5**. Table contents are described in **Table 3**.

Table 3. Descriptions of Variant results column headers.

rsID	The genetic variant of interest entered in the search box.
Disease	The diseases associated with the variant of interest.
Drug	The drugs associated with the variant of interest.
Gene	The gene in which the variant of interest is located.
Significance	The p-value observed for the association in the given study.
Country	The country of origin of the research participants.
Studies	The study associated with the result.

4. Gene Results:

- 4.1. When searching by a gene, a list of relevant results appears below the search box. To proceed to complete results, select the “For gene associations click here”, as illustrated in **Figure 3D**.
- 4.2. As illustrated in **Figure 6A-B**, the results page will contain a description section (**6A**), a Pharmacogenomics Associations section (**6A**) and a Disease Associations section (**6B**). Table contents are described in **Table 4**.

COMT

Gene Name COMT

Uniprot ID P21964

Function Catalyzes the O-methylation, and thereby the inactivation, of catecholamine neurotransmitters and catechol hormones. Also shortens the biological half-lives of certain neuroactive drugs, like L-DOPA, alpha-methyl DOPA and isoproterenol.

Pharmacogenomics Associations

SNPs

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Search:

rs ID	Genotype	Drug	Description	P-value	Study	Regions	Countries
rs4680	G	morphine	Allele G is not associated with dose of morphine in people with Pain as compared to allele A.	0.2928	29259946	North African	Tunisia
rs727865	AA	bupropion	Genotype AA is associated with decreased response to bupropion in smokers as compared to genotypes AG + GG.	0.05	16876132	African American/Afro-Caribbean	USA

Showing 1 to 2 of 2 entries

Previous 1 Next

Star notation Genotype Drug Description P-value Study Regions Country of Participants

No data available in table

Showing 0 to 0 of 0 entries

Previous Next

Disease Associations

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Search:

rs ID	Disease	P-value	Study	Regions	Country of Participants
rs6265	Schizophrenia	< 0.05	22521161	North African	Egypt
rs759854365	Schizophrenia	0.008	22521161	North African	Egypt

Showing 1 to 2 of 2 entries

Previous 1 Next

Figure 6. Gene result page.

Table 4. Descriptions of Gene results column headers.

Variant	The genetic variants located in the gene which have been studied in African populations
Genotype	The genotype of a genetic variant associated with a given drug association.
Drug	The drug associated with genetic variant.
Description	A description of the drug association.
P-value	The p-value observed for a given association.
Study	The PMID associated with the associated study.
Regions	The region(s) from which the research participants originate.
Country of Participants	The Country(ies) from which the research participants originate.
Disease	The disease associated with genetic variant.

- 4.3. Using the task bars found in each table, the user can access different information on either the disease, drug, variant or gene. Table 1 provides an overview of the different types of information found.

Other Resources

- When clicking on the **Summary** tab, a summary of the Total Number of Genes, Drugs, Diseases and Variants, included in the portal, is provided. The locations from where the data is derived is also illustrated in a user-friendly map.

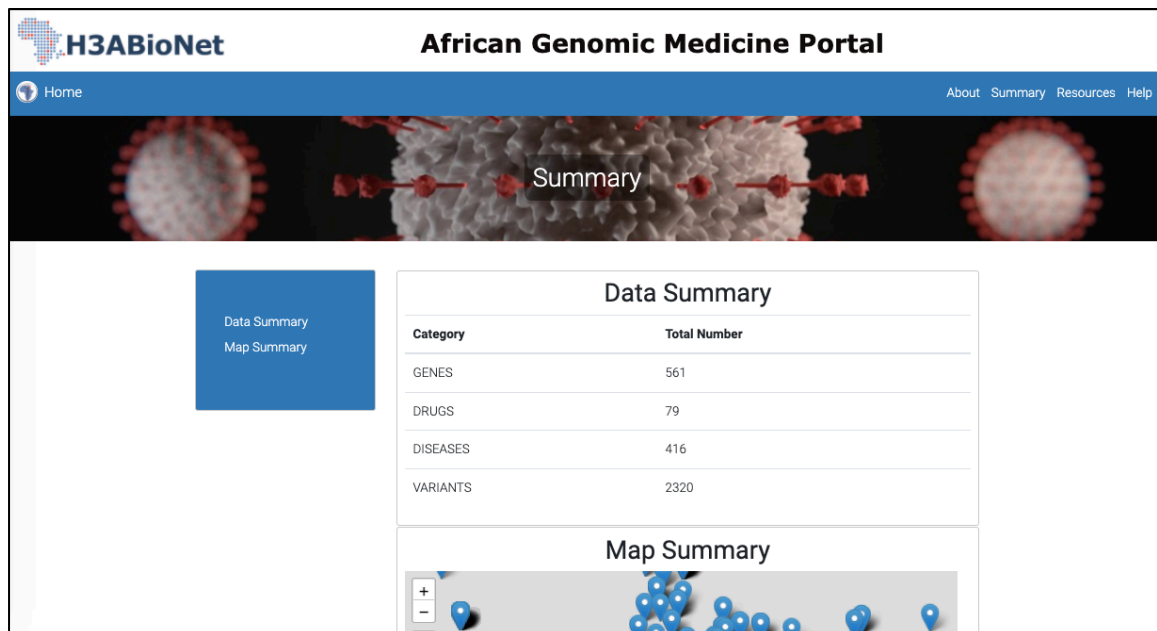
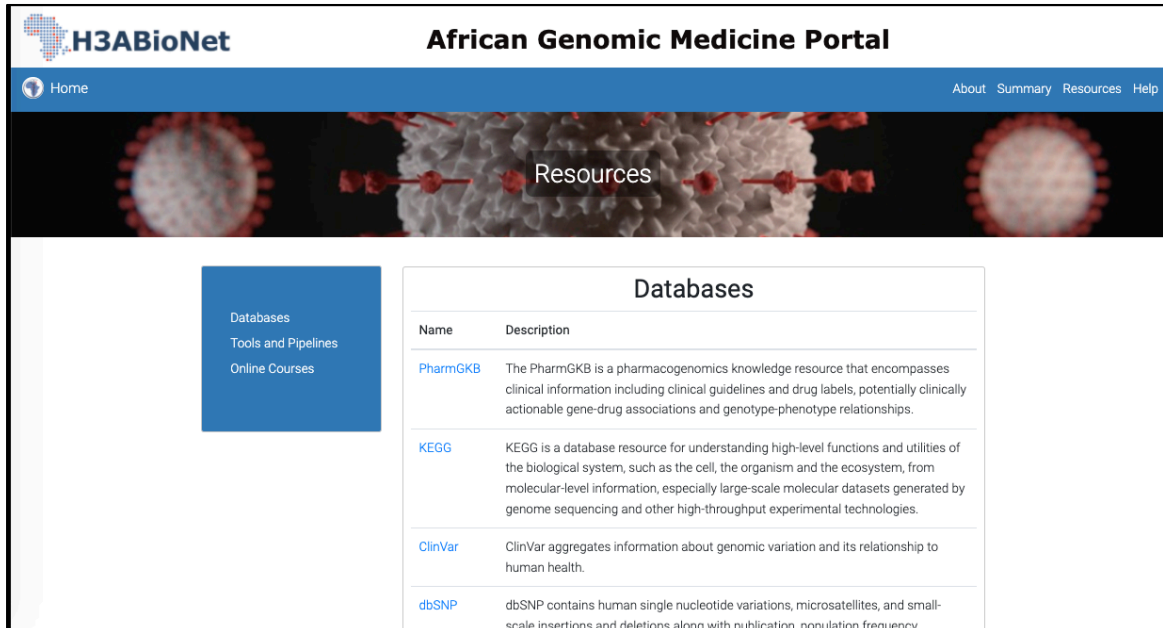


Figure 4: Summary tab.

- When clicking on the Resources tab, a list of additional H3ABioNet and relevant

external resources are provided, these include: Databases; Tools & Pipelines; and Online Courses.



The screenshot shows the H3ABioNet African Genomic Medicine Portal. The header includes the H3ABioNet logo and the title "African Genomic Medicine Portal". A navigation bar contains links for Home, About, Summary, Resources, and Help. The main content area is titled "Resources" and features a sidebar with links to Databases, Tools and Pipelines, and Online Courses. The "Databases" section is active, displaying a table of resources.

Name	Description
PharmGKB	The PharmGKB is a pharmacogenomics knowledge resource that encompasses clinical information including clinical guidelines and drug labels, potentially clinically actionable gene-drug associations and genotype-phenotype relationships.
KEGG	KEGG is a database resource for understanding high-level functions and utilities of the biological system, such as the cell, the organism and the ecosystem, from molecular-level information, especially large-scale molecular datasets generated by genome sequencing and other high-throughput experimental technologies.
ClinVar	ClinVar aggregates information about genomic variation and its relationship to human health.
dbSNP	dbSNP contains human single nucleotide variations, microsatellites, and small-scale insertions and deletions along with publication, population frequency.

Figure 5: Resources tab.