African Genomics Medicine Portal

Tutorial

Introduction

The African Genomics Medicine Portal (AGMP) functions as a one-stop resource for researchers around the world who are conducting biomedical research on African and African-related populations. The portal can also be useful for other individuals working in the health sector, such as healthcare workers, pharmacists and policymakers.

The portal functions as a gateway to African genomics medicine research, including pharmacogenomics and clinical/disease research, accessing and providing African-specific data from existing resources, and providing it in an easily accessible manner.

AGMP retrieves and curates data from various resources. The update log below highlights the resources implemented with each release.

Update Log:

Release Date:	AGMP launches. Data retrieved and curated from PharmGKB and DisGeNET.
Update Date:	TBD.

Search The Portal

1. To access AGMP, click on the following link: https://agpm.knust.edu.gh/. Once accessed, **four main data categories** will be displayed: **Disease**, **Drug**, **Variant**, and **Gene**.

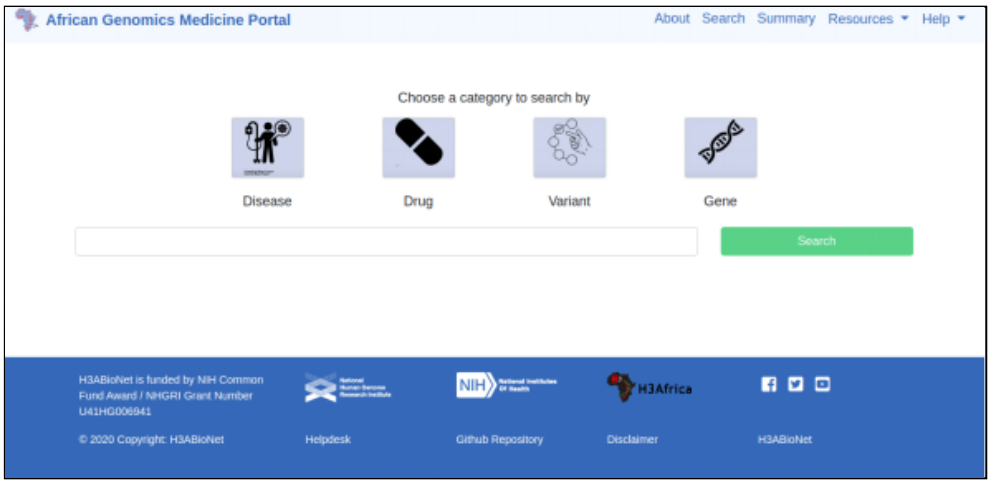


Figure 1: AGMP main search page, illustrating the main data categories: Disease, Drug, Variant and Gene

2. Search by Data category:
Click on the preferred data category logo (Disease, Drug, Variant or Gene) followed by the name of the related search field.

2.1. **Search by Disease**: Enter the name of the disease at the search field and click “search”. The resulting page, shown in Figure 2.1.1, contains a list of disease related sub-categories.

Figure 2.1: AGMP search by disease data category.

ILLUSTRATION TABLE TO BE INSERTED AS SOON AS IT'S AVAILABLE

2.2. **Search by Drug**: Enter the name of the drug at the search field and click "search". The resulting page, shown in Figure 2.2.1, contains a list of drugs.

Figure 2.2: AGMP search by drug data category.

Figure 2.2.1: AGMP results of selecting the of "Variant Drug associations" sub-category.

2.3. **Search by Variant**: Enter the name or accession number of the variant at the search field and click "search". The resulting page, shown in the Results section, contains a list of gene related sub-categories.

Figure 2.3: AGMP search by variant data category.

2.4. **Search by Gene**: Enter the name of the gene at the search field and click "search". The resulting page, shown in the Results section, contains a list of gene related sub-categories.

Figure 2.4: The AGMP database illustrating information on search by gene data category

Results

The following section discusses the results pages in relation to results obtained from BRCA and CYP2B6 searches.

3. As indicated in Section 2, when searching by a gene or variant, a list of relevant genes or variants, with their chromosome number, appear below the search box.
   3.1. Click on the “click here for gene association” button.

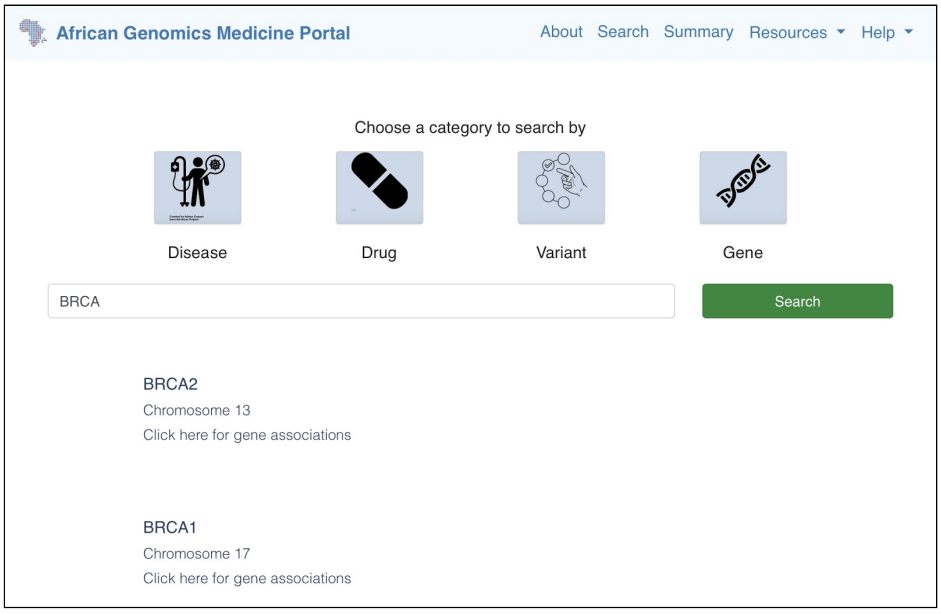


Figure 3.1: Search results following a BRCA search.

3.2. At the top of the results page, you will find:
- Gene name
- Uniprot ID hyperlinked to uniprot page
- Brief description of the protein encoded by the query gene.

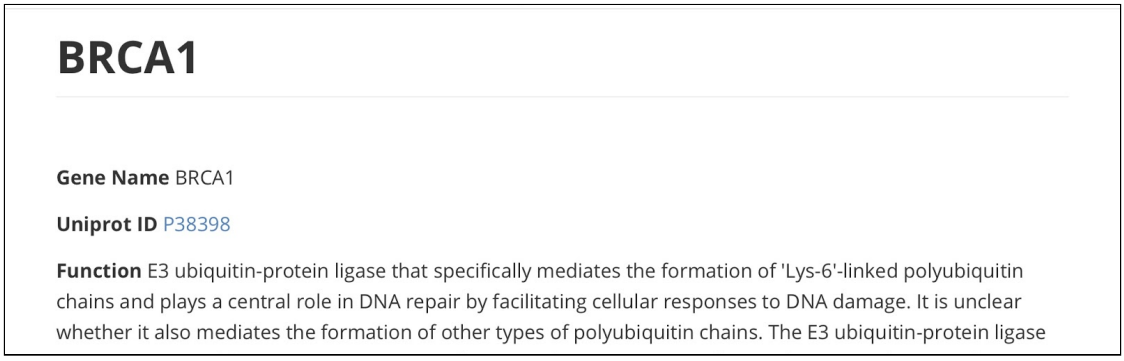
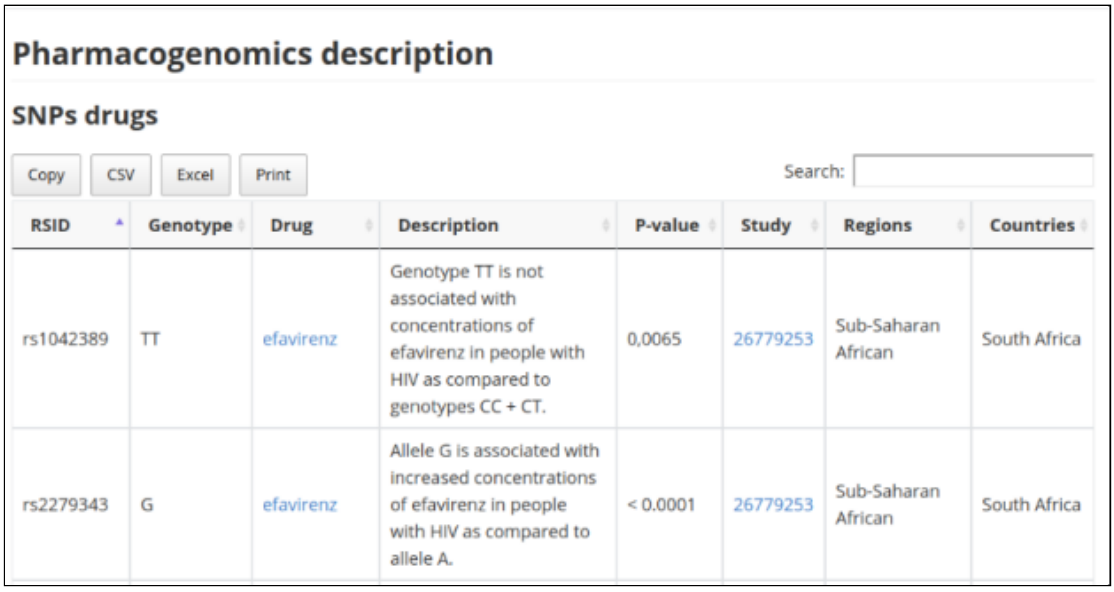


Figure 3.2: BRCA1 gene results page.

3.3. Below you will find two tables, one related to pharmacogenomics associations (CYP2B6 example) and one related to disease associations (BRCA1 example). The content of these tables are, in order:

- **Pharmacogenomics Associations**



Pharmacogenomics description

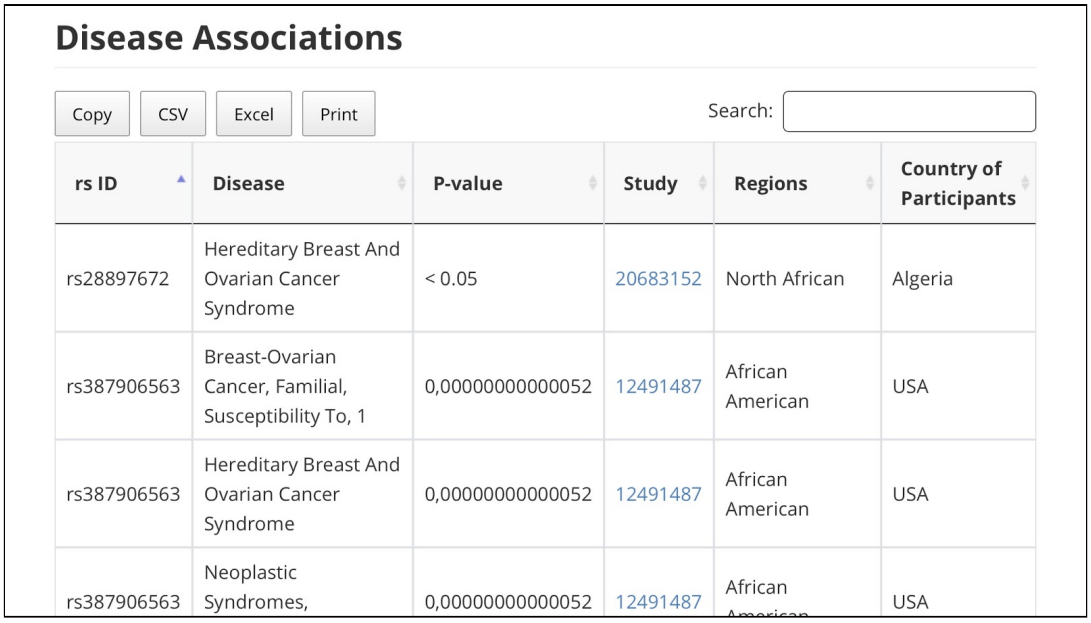
SNPs drugs

Copy CSV Excel Print Search:

RSID	Genotype	Drug	Description	P-value	Study	Regions	Countries
rs1042389	TT	efavirenz	Genotype TT is not associated with concentrations of efavirenz in people with HIV as compared to genotypes CC + CT.	0,0065	26779253	Sub-Saharan African	South Africa
rs2279343	G	efavirenz	Allele G is associated with increased concentrations of efavirenz in people with HIV as compared to allele A.	< 0.0001	26779253	Sub-Saharan African	South Africa

Figure 3.3.2: Table of Disease associations.

- **Disease Associations**:
  rs ID, Disease, P-value, Study Link to pubmed ID , Regions, Country of participants.



Disease Associations

Copy CSV Excel Print Search:

rs ID	Disease	P-value	Study	Regions	Country of Participants
rs28897672	Hereditary Breast And Ovarian Cancer Syndrome	< 0.05	20683152	North African	Algeria
rs387906563	Breast-Ovarian Cancer, Familial, Susceptibility To, 1	0,00000000000052	12491487	African American	USA
rs387906563	Hereditary Breast And Ovarian Cancer Syndrome	0,00000000000052	12491487	African American	USA
rs387906563	Neoplastic Syndromes,	0,00000000000052	12491487	African	USA

Figure 3.3.2: Table of Disease associations.

3.4. 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 and functions that can be enabled on various subsections

Table 1: Gene page functionalities.	
TASK BAR	INFORMATION PROVIDED BY SUBCATEGORIES
Gene Icon	Links to a page containing an overview of information regarding a specific gene, or the variants within the gene which influences drug processing or disease etiology. Gene pages can be accessed by clicking on the gene logo, then in the gene name in the search field.
Drug Icon	Provides a list of drugs of which their PK/PD properties have been associated with a particular variant, disease or gene along with their Pubmed IDs.
rs ID	Provides a list of variants which have been associated with disease and drug metabolism in African populations, either located within, or close to the gene
Significant Icon Study Icon Country Icon P-value	Elaborate on the study population, the country the studies were conducted in and their level of significance. The study accession number can be used to link to the Pubmed entry for the associated study.
Description icon	Provide a description of the association found in the associated study.
Drop arrows	Arrange tables in ascending and descending order given a particular column..
Copy icon Print icon CSV icon Excel	Enables the results to be either copied, downloaded (in either CSV or Excel spreadsheet format) or printed.
Search bar	Extract rows according to certain keywords within the table.

Other Resources

The following section discusses other resources provided on the Portal as well as additional resources associated with Precision Medicine initiatives.

4. 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.

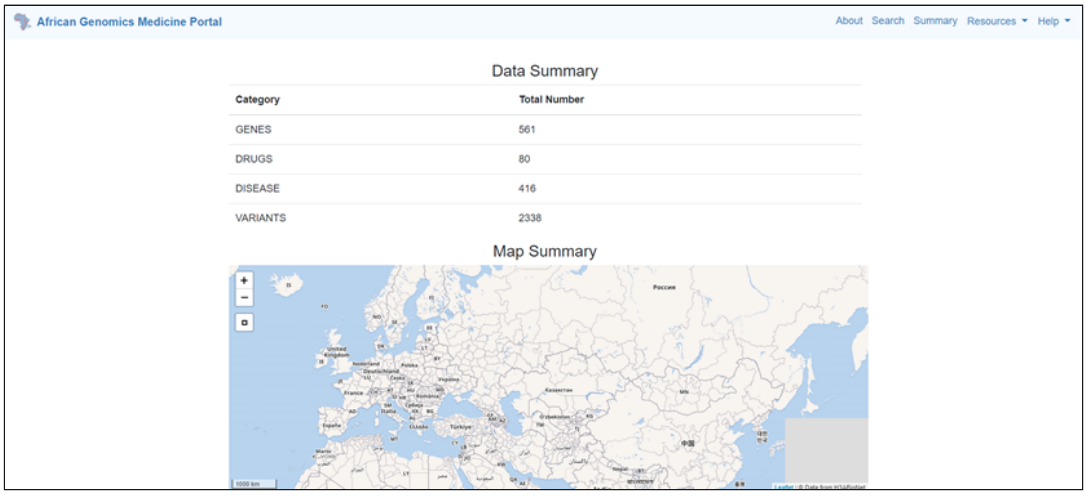


Figure 4: Summary page, illustrating location and total numbers.

5. When clicking on the resources tab, a list of external resources are provided, these include:
   - Databases
   - Tools & Pipelines
   - Online Courses

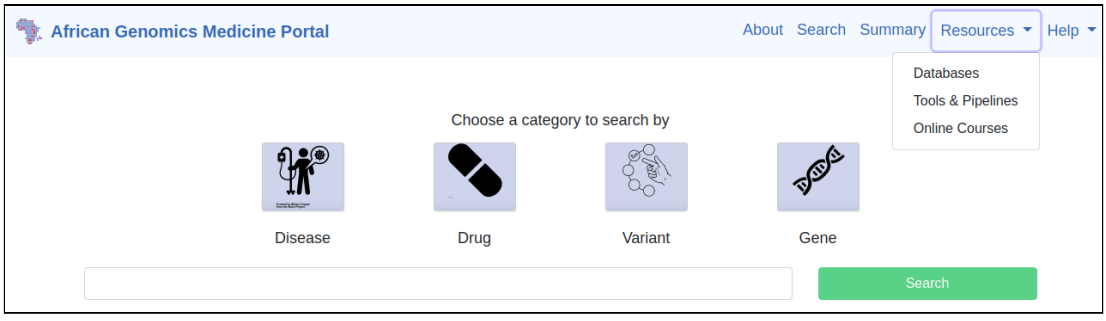


Figure 5: Resources button.

Some of these resources are tabulated in **Table 2.**

Table 2. Examples of some of the resources available online to help provide a basic understanding of genetics and bioinformatics concepts and terms.

Resource Area	Resource Name	What You Will Learn?
Bioinformatics	Pan African Bioinformatics Network for H3Africa (H3ABioNet)	Research Projects pertinent to the development of bioinformatics capacity in Africa. Various courses and hands on training.
General Genetics	Genetics Home Reference (NIH)	Terms and concepts related to genetics and what genes cause different genetic conditions.
	Human Genetics and Medical Research: A Revolution in Progress (NIH)	General genetics concepts, including what genes are, information on the HGP, and how gene therapy works.
	Human Genome Project Information (Oak Ridge National Laboratory)	The HGP
	Learn.Genetics, Genetic Science Learning Center (The University of Utah)	Genetics.
	DNA from the Beginning (Cold Spring Harbor Laboratory)	General genetics
	Gene Screen app (Cold Spring Harbor Laboratory: Dolan DNA Learning Center, Harlem DNA Lab & DNA Learning Center West)	Interactive explanations of general genetics concepts.
Genetics & Diseases	Genes and Disease (National Center for Biotechnology Information)	Genes and the genetic disorders.
	Your Genes, Your Health (Cold Spring Harbor Laboratory: Dolan DNA Learning Center)	Genetic diseases.
Gene Testing	The Genetics of Cancer (National Cancer Institute at the National Institutes of Health)	Gene testing.
Clinical Genetics	Clinical Variation	A public archive of reports of the relationships among human variations and phenotypes, with supporting evidence.

Pharmacogenetics	Pharmacogenetics Implementation Consortium (CPIC)	Freely available, peer-reviewed, evidence-based, updatable, and detailed gene/drug clinical practice guidelines
Pharmacogenetics	PharmGKB	Provides clinical information including clinical guidelines and drug labels, potentially clinically actionable gene-drug associations and genotype-phenotype relationships.
Pharmacogenetics	Pharmacogenomics Research Network (PGRN)	Research in precision medicine for the discovery and translation of genomic variation influencing therapeutic and adverse drug effects. The network has several useful resources.