

Variant Atlas

A Gen-Phen platform for the Australian Genomics Flagship Data

“Discover – Explore – Compare – Subset”

What is Variant Atlas?

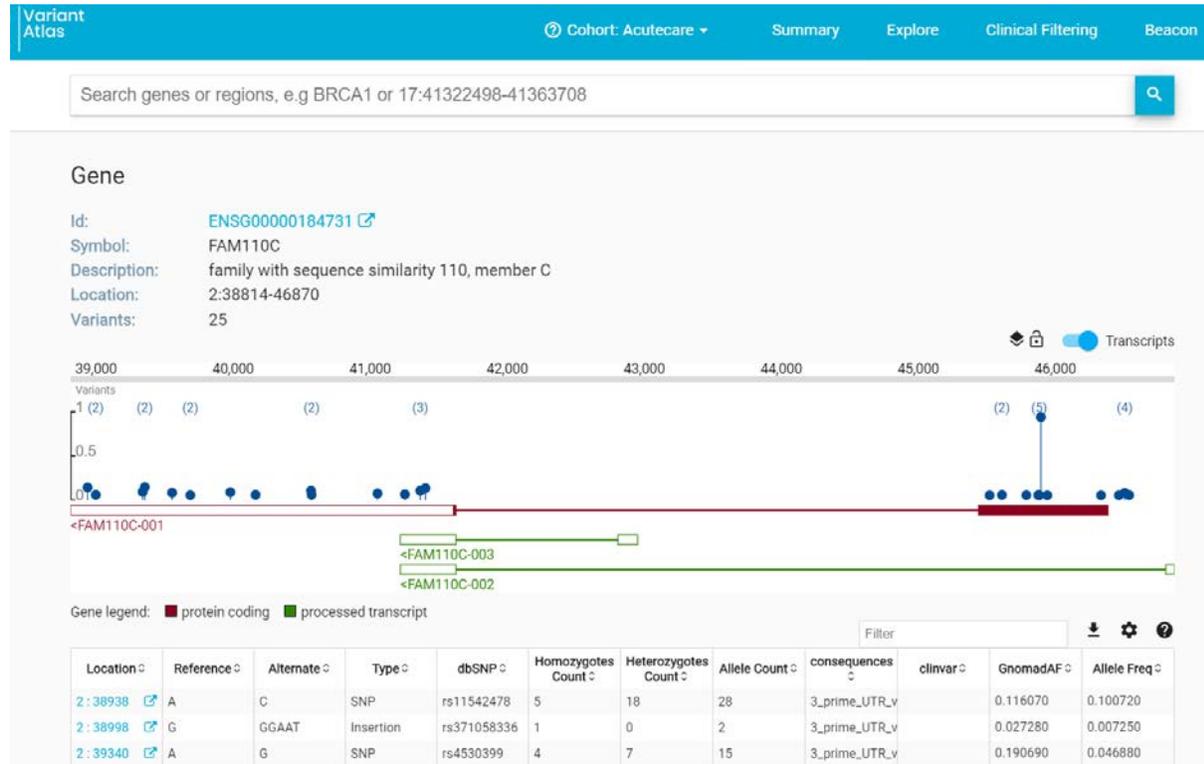
Variant Atlas is a controlled-access, web-based interface for exploring and discovering a cohort’s genomic and phenotypic data. Variant Atlas is hosted at the Garvan Institute (<https://variantatlas.org.au/>)

The platform combines Flagship participants’ genomic information, and links a participant’s variant data to a selection of clinical information collected at recruitment. With this, members and approved researchers can explore genomic characteristics at the cohort-level, query specific genes or variants of interest across the Flagship, and discover variants in patient subgroups with particular clinical features.

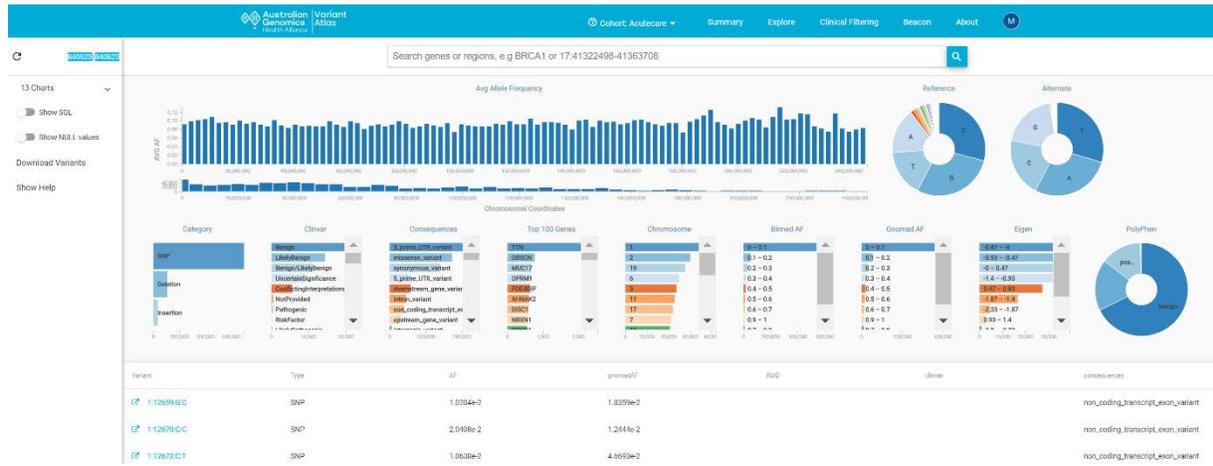
Features

Summary – Explore – Clinical Filtering – Beacon

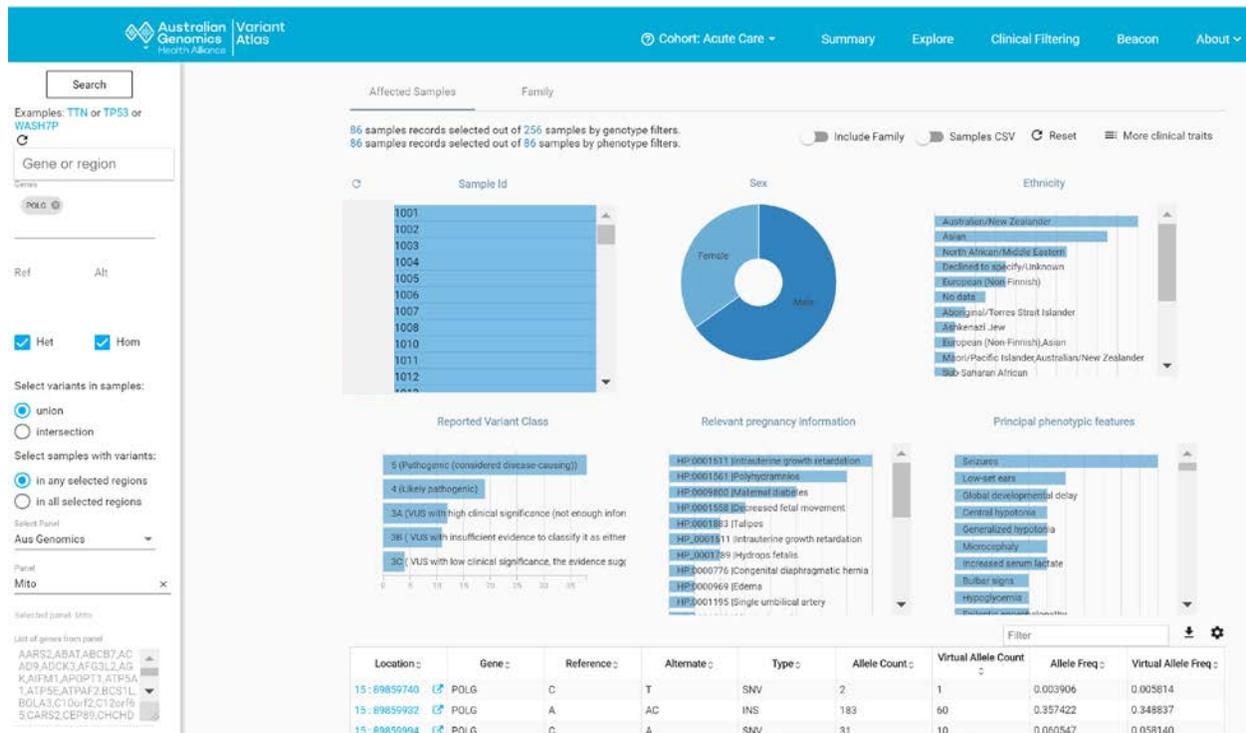
‘Summary’: Locate and compare summary statistics across variants and genes in Flagships



'Explore': Discover genomic characteristics of Flagship cohorts

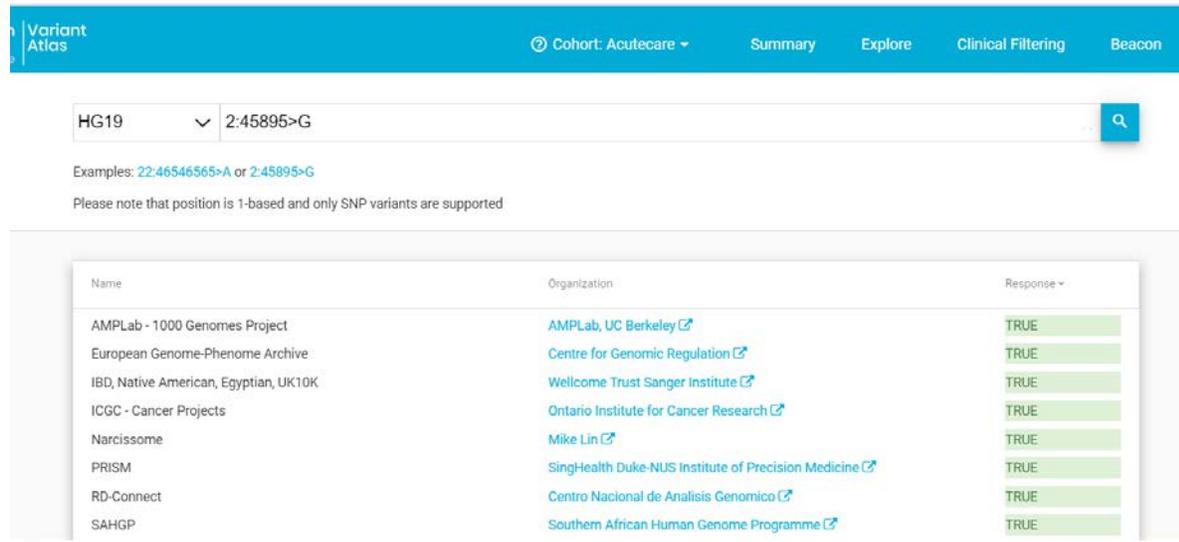


'Clinical Filtering': Filter patient data according to clinical traits **



** Restricted Access

'Beacon': Query the world's cohort datasets for specific variants



The screenshot shows the Variant Atlas Beacon interface. At the top, there is a navigation bar with 'Variant Atlas' on the left and 'Cohort: Acutecare', 'Summary', 'Explore', 'Clinical Filtering', and 'Beacon' on the right. Below the navigation bar is a search input field containing 'HG19' and '2:45895>G'. Below the search field, there are examples of variant queries: '22:46546565>A or 2:45895>G'. A note states: 'Please note that position is 1-based and only SNP variants are supported'. Below the search field is a table with three columns: 'Name', 'Organization', and 'Response'. The table lists several cohort datasets and their corresponding organizations and response status.

Name	Organization	Response
AMPLab - 1000 Genomes Project	AMPLab, UC Berkeley	TRUE
European Genome-Phenome Archive	Centre for Genomic Regulation	TRUE
IBD, Native American, Egyptian, UK10K	Wellcome Trust Sanger Institute	TRUE
ICGC - Cancer Projects	Ontario Institute for Cancer Research	TRUE
Narcissome	Mike Lin	TRUE
PRISM	SingHealth Duke-NUS Institute of Precision Medicine	TRUE
RD-Connect	Centro Nacional de Analisis Genomico	TRUE
SAHGP	Southern African Human Genome Programme	TRUE

Why have we created Variant Atlas?

We created Variant Atlas to:

- ✓ Promote discovery and support secondary analyses of Flagship data for Australian Genomics researchers and clinicians
- ✓ Enable Flagships to control access and share genomic and clinical information with collaborators
- ✓ Make summary-level variant information accessible to all researchers
- ✓ Allow our data to be discoverable through the Global Alliance Beacon for future access requests.

How do I obtain access to Flagship data on Variant Atlas?

Register for access to 'Summary' and 'Explore' Aggregated data here: [Registration Form](#)

For other access enquires please contact: datarequest@australiangenomics.org.au

Instructions and open-access demonstration instance

An instruction video on how to use the Variant Atlas platform and its features, is available: [Demo Video](#)

An open-access instance of Variant Atlas is available for anyone to begin exploring now: [Demo Dataset](#)

Questions, comments or suggestions?

We are always looking for ways to improve the interface and make it as useful as possible for our Flagships and other researchers. We would love to hear from you!

Contact: Marie-Jo Brion, *Program Two Manager* Marie-Jo.Brion@gimrberghofer.edu.au
Warren Kaplan, *Variant Atlas Lead* W.Kaplan@garvan.org.au

Variant Atlas Software Engineers
Dmitry Degrave, Principal Software Engineer
Andre Hermanto, Software Engineer