

### Workshop Report: Future Directions of the NHGRI Analysis, Visualization, and Informatics Lab-space (AnVIL)

Marylyn D. Ritchie, Ph.D.

Department of Genetics Director, Institute for Biomedical Informatics University of Pennsylvania





#### **Presentation Overview**

- AnVIL Analysis Workspaces Components
- Overview of Future Directions Of The NHGRI AnVIL Workshop
- AnVIL SWOT Analysis

Next Steps for AnVIL



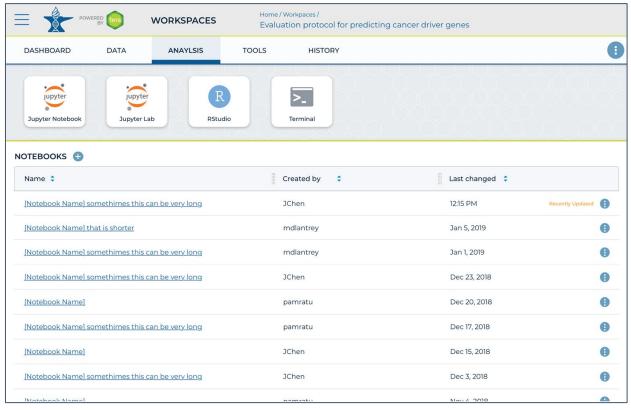
## AnVIL Analysis Workspaces Components

Faceted search

Established pipelines

**Exploratory Analysis** 

Integrated development environment





AnVIL / Dockstore: sharing containerize tools and workflows















**Genome Browser** 

# Overview of Future Directions Of The NHGRI AnVIL Workshop

Virtual Meeting Oct 29, 2021; 12pm-5pm ET

Identify gaps, challenges, and future opportunities related to NHGRI's Goal investments in the AnVIL's cloud-based infrastructure, tools, and services. Discuss The current state of AnVIL Activities needed to expand, diversify, and support the AnVIL user Identify community

## Workshop Breakout Rooms



Data submission and consortia engagement



**Analysis tools** 



Infrastructure



Outreach and training

### Cross Cutting Themes

How cloud-based platforms can better serve the needs of genomic researchers

What tools and services would better support clinical genomic researchers

How to improve interoperability with other cloud-based resources in a federated genomic data ecosystem

#### **Workshop Attendees**

- Invited participants and assigned discussants
- AnVIL key personnel
- NHGRI AnVIL staff
- AnVIL External Consultants Committee
- Other NHGRI staff as listeners
- 105 people attended

# **SWOT Analysis**Breakout Report Format



#### Strengths - Where does AnVIL excel?

Strong commitment to data security

A versatile platform for training students and consortia on the use of the cloud for genomic analyses

NHGRI's leadership of the NCPI efforts and AnVIL's extensive use of APIs and GA4GH standards facilitates interoperability

Extensive documentation, which enables users to help themselves and help each other

Ability by third party groups to build on the platform while still prioritizing security

NHGRI's robust engagement with other NIH Institutes and Centers to encourage the adoption of the GA4GH Data Use Ontology (DUO) and Data Use Oversight System (DUOS) to streamline the data access review process

#### Weaknesses: Where is AnVIL at a disadvantage?

Curation of tools and workflows to facilitate searches by users could be improved

Lack of phenotypic harmonization across programs

The relationships between AnVIL and data coordination centers of NHGRI funded consortia could be better defined

Significant hurdles required to access AnVIL just to test the platform

Lack of video documentation

**Users Cannot log in anonymously** 

AnVIL lacks embedded personnel (e.g., key personnel and developers) with clinical research priorities

#### Opportunities: Where AnVIL can grow and improve?

AnVIL could create a safe space for groups hesitant to host diverse controlled access datasets in public repositories

AnVIL could transition over to tools development and analyses for discovery, in addition to acting as a data repository

The AnVIL team could demonstrate that AnVIL can work for the clinical community

AnVIL could add additional data standards and data models to improve the interoperability model for Terra

The outreach team could conduct robust research into how AnVIL is being used

AnVIL has the opportunity to introduce cloud computing for the next generation of scientists

AnVIL could integrate a diversity of human genetic datasets and reference genomes and make them backwards compatible

#### Threats: Which factors jeopardize AnVIL?

Cloud costs are a major barrier for many users.

Challenges in making AnVIL interoperable with other platforms.

Difficulties in shifting scientific culture to the cloud.

Challenges in making tools and resources in a manner that meets users where they are

AnVIL faces data security threats, both by outsiders and by people with authorized access.

Potential institutional fear of investing in a dead-end utility if NHGRI's long term commitment to the resource is unclear.

Potential lack of users' skill transferability between AnVIL and other cloud platforms.

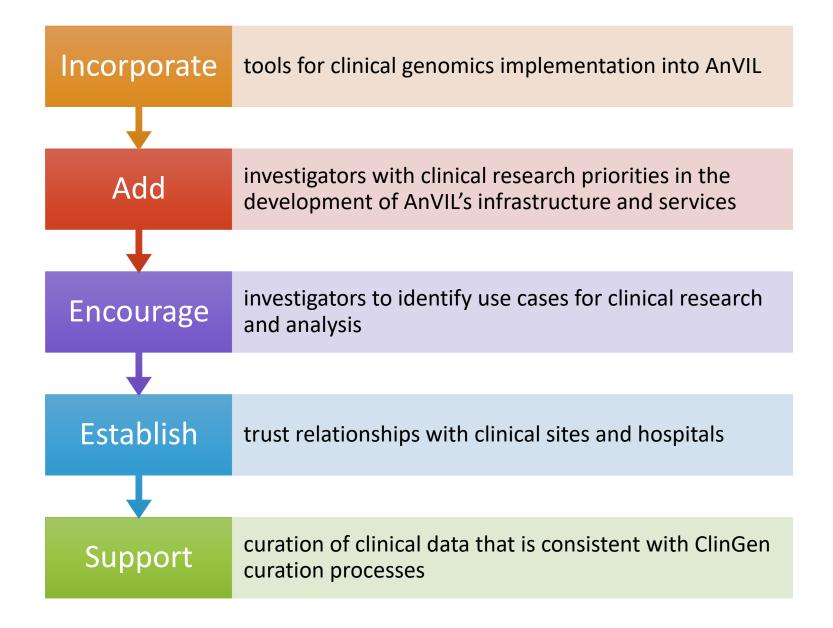
# Challenges Related To The Use Of The Cloud

Culture change for most investigators who have exclusively worked using on-premises infrastructure

Onboarding users who are inexperienced with cloud resources

Cloud costs are both a practical and psychological barrier

## Support For The Clinical Research Community



#### Make AnVIL accessible to naïve users by:

- generating and sharing popular analysis workflows
- developing and advertising an extensive video guide to its features
- providing streamlined billing and payment for cloud services
- providing easier to access data in the interactive workspaces

Make onboarding easier for all users, in particular for users from low-resource institutions

Leverage the research communities funded by NHGRI to build a sense of community around the platform

Identify use cases to demonstrate AnVIL's capabilities and promote them

# Interoperability, Analysis Tools, And Other Considerations



Continue to engage more broadly with other NIH Institutes and Centers



Increase phenotypic data harmonization efforts across NHGRIfunded consortia and initiatives



Support more data standards and common data models to improve semantic interoperability with other platforms



Support the development and availability of more analysis tools for both basic and clinical genomics researchers



Increase the search capabilities for datasets, tools, and workflows that are available on the platform



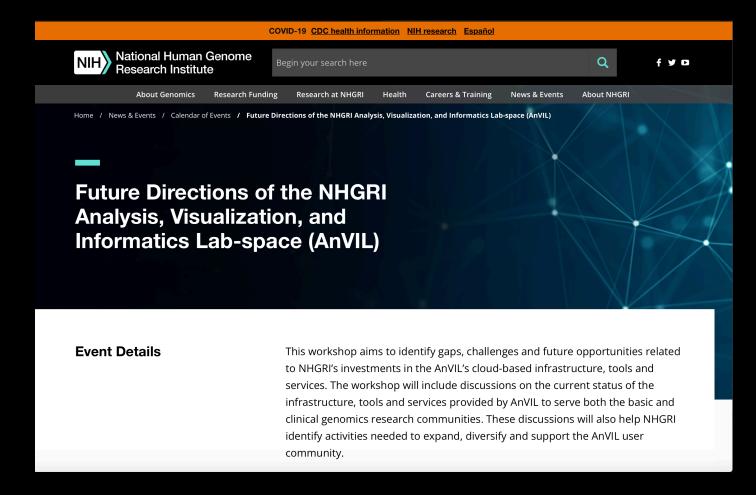
Rebrand AnVIL from a data submission site to a multi-functional discovery platform



Clarify relationship between AnVIL and data coordination centers



NHGRI to prioritize AnVIL's future activities and prepare for renewal



https://www.genome.gov/event-calendar/Future-Directions-of-the-NHGRI-Analysis-Visualization-and-Informatics-Lab-Space-AnVIL

# Thank You Workshop Participants and Organizers

Questions?