

## Workshop Report

# Future Directions of the NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space (AnVIL)

Workshop date: October 29, 2021

### Workshop Background

The [NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space](#) ('AnVIL') is a secure, cloud-based environment where researchers can store, share, and analyze unrestricted- and controlled-access genomic datasets and associated phenotypic data or metadata, particularly those generated by NHGRI consortia and initiatives. Since 2018, NHGRI has been funding and managing AnVIL through two cooperative agreements awarded to groups lead at the Broad Institute and Johns Hopkins University.

Given that AnVIL had recently completed its third year of funding, NHGRI convened a to inform future directions of the AnVIL program as it continues to mature. The goal of this workshop was to identify the gaps, challenges, and opportunities related to NHGRI's investments in AnVIL's cloud-based infrastructure, tools and services. Participants from the genomics research community (including basic genomics, clinical genomics and genomic data science) helped NHGRI identify the activities that are needed by the AnVIL resource to expand, diversify, and support genomics researchers and the AnVIL user community.

The workshop agenda included four breakout rooms, focused on the following topics: (1) Data submission and consortia engagement, (2) Analysis tools, (3) Infrastructure, and (4) Outreach and training. In each session NHGRI was particularly interested in hearing feedback on the following cross-cutting topics: (a) how cloud-based platforms can better serve the needs of genomics researchers; (b) what tools and services would better support clinical genomics researchers; and (c) how to improve interoperability with other NIH cloud-based resources in a federated genomic data ecosystem.

Detailed information about the workshop's goals, agenda, and meeting participants can be found in the workshop booklet in **Appendix 1** and on the workshop [webpage](#).

The workshop discussion in each breakout room was conducted using a Strengths, Weaknesses, Opportunities, and Threats (SWOT) analysis framework. Specifically, participants were asked to discuss the following: Strengths – Where does AnVIL excel?; Weaknesses – Where is AnVIL at a disadvantage?; Opportunities: Where can AnVIL grow and improve?; and Threats – Which factors jeopardize AnVIL?

The summary of the SWOT analyses conducted by the participants in each breakout room are combined and available in **Appendix 2**. Recurring themes, general considerations, and suggestions for improvement are summarized here.

## Summary of Workshop's Findings

### ***Strengths***

Participants highlighted the excellent progress was made during the first 3 years of the AnVIL program. There was recognition of the numerous accomplishments (see p. 13). AnVIL's commitment to facilitate NHGRI-funded consortia's data sharing and access is evident from the impressive growth rate of the data submitted to AnVIL. AnVIL is in a position to reduce the time to share and access datasets and exponentially increase the rate of discovery by consortium members as more datasets get integrated in the platform. Participants were particularly excited by NHGRI's engagement with other NIH Institutes and Centers (ICs) to encourage the adoption of the GA4GH Data Use Ontology (DUO) and the Broad's Data Use Oversight System (DUOS), which are both potential 'game changers' to speed up the data access process. Participants were impressed by AnVIL's commitment to data security and encouraged AnVIL to maintain a high security standard, as well as to ensure that users are aware of the high security standards that the platform is implementing. Workshop participants expressed their enthusiasm and satisfaction for the ease of use and extensive catalog of available tools, including hundreds of workflows and thousands of tools already available within the AnVIL platform. Participants appreciated AnVIL's extensive documentation, which enables users to help themselves and to help each other. Extremely positive comments were shared about the established outreach efforts, such as AnVIL's ongoing work with researchers at a variety of institutions that enables the expansion of genomic data science education and both synchronous and asynchronous user support of novices through power users. AnVIL was viewed as a potentially effective platform for training students and consortia on the use of the cloud for genomic analyses. Finally, participants praised NHGRI's efforts to facilitate systems interoperability and access to data residing on different NIH platforms through the NIH Cloud Platform Interoperability ([NCPI](#)) effort and AnVIL's extensive use of APIs and GA4GH standards. Overall, the workshop participants expressed great enthusiasm and support of the AnVIL project and highlighted the relevance of the achievements made in the first three years.

### ***Suggestions for Improvement***

#### **Challenges related to the use of the cloud**

Participants noted that transitioning to the cloud for data sharing, storage, and analysis is a big culture change for most investigators who have exclusively worked using on-premises infrastructure. Onboarding users who are inexperienced with cloud resources is a significant challenge. Cloud costs are a barrier for new users, both practical and psychological (e.g., paying for compute is still a new concept). AnVIL could explore ways for researchers' institutions to streamline setting up Anvil workspaces and associated billing accounts. AnVIL could also look into the feasibility of adding a "free tier," in which new users can become familiar with AnVIL's capabilities before having to pay for cloud costs. The AnVIL team could make available estimated cloud costs incurred for popular analysis workflows available on the platform to help investigators budget for future projects. Finally, NHGRI could find new ways to leverage

the NIH STRIDES cloud credits and discounts to support users, especially those from low-resource institutions.

### **Support for the clinical research community**

Participants encouraged AnVIL to add tools for clinical genomics implementation to support outreach to clinical genomics groups and help convince them that “AnVIL isn’t just for basic research.” To make AnVIL more appealing to clinical researchers, AnVIL should consider embedding investigators with clinical research priorities in the development of AnVIL’s infrastructure and services as well as to identify use cases for clinical research and analysis. Given AnVIL’s aim to adopt the FHIR standard for clinical data exchanges, it could also identify clinical use cases for FHIR beyond data exchanges. AnVIL could aim to establish trust relationships with clinical sites and hospitals and look for ways to bring translational impact of AnVIL services into the clinical domain. AnVIL could also support curation of clinical data that is consistent with ClinGen curation processes.

### **User outreach and training**

Participants suggested several ways for the AnVIL to broaden its user base, in particular students, postdocs, and other trainees, including those who are less skilled in computer science and informatics. For example, AnVIL could become more 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, and making it easier to access data in the interactive workspaces. AnVIL could make onboarding easier for all users, in particular for users from low-resource institutions. For example, the NHGRI Diversity Action Plan program could collaborate with AnVIL to offer opportunities for mentored skills development on the platform. AnVIL could invest in re-training faculty in data science and supporting curriculum development and documentation for undergraduates.

AnVIL could also leverage the research communities funded by NHGRI (and embedded in the consortia that are integrating into AnVIL) to build a sense of community around the platform. AnVIL could provide support or examples to assist users with describing their analyses in a way that supports publications. The AnVIL team could identify use cases to demonstrate AnVIL’s capabilities, promote them in a variety of venues, and provide information for how to get involved and contribute to the platform.

### **Interoperability, analysis tools, and other considerations**

Participants encouraged NHGRI to continue to engage more broadly with other NIH Institutes and Centers (ICs) to help the AnVIL program achieve its goal of improving systems interoperability across NIH cloud platforms. AnVIL could increase its phenotypic data harmonization efforts across NHGRI-funded consortia and initiatives that share datasets through the platform and could support more data standards and common data models to improve semantic interoperability with other platforms.

Supporting the development and availability of more analysis tools would make AnVIL more attractive and accessible for basic and clinical genomics researchers. AnVIL could increase the search capabilities for datasets, tools, and workflows that are available on the platform and rebrand itself from being a data submission site (i.e., repository only) to a multi-functional discovery platform for consortia as well as other genomics and clinical researchers.

NHGRI is encouraged to clarify the relationship of the AnVIL and the data coordination centers of NHGRI-funded consortia and initiatives that are expected to leverage the AnVIL services and to better delineate and communicate its long-term plans for the platform; this will help address investigators' fear of investing time and resources in a new resource that may be short-lived.

### Acknowledgements

NHGRI wishes to thank the members of the [AnVIL External Consultant Committee](#) and, in particular, the ECC's moderators of the workshop breakout rooms: Ms. Karen Davis, Dr. Siddharth Pratap, Dr. Adam Resnick, and Dr. Marylyn Ritchie as well as all the meeting participants for their valuable suggestions and feedback.

### NHGRI Workshop Report Writers

- Valentina Di Francesco
- Carolyn Hutter
- Ana Stevens
- Chris Wellington
- Ken Wiley

# *Appendix 1*

## *Workshop's Booklet*

### Purpose of the workshop

The NHGRI Genomic Data Science Analysis, Visualization, and Informatics Lab-space (AnVIL) is a secure, cloud-based environment where researchers can store, share, and analyze unrestricted- and controlled-access genomic datasets and associated phenotypic data or metadata, particularly those generated with NHGRI support. Since 2018 NHGRI has been funding and managing AnVIL through two cooperative agreements awarded to the Broad Institute and Johns Hopkins University.

The goal of this workshop is to identify gaps, challenges, and future opportunities related to NHGRI's investments in the AnVIL's cloud-based infrastructure, tools, and services.

At this workshop we will discuss the current status of AnVIL's ability to serve both the basic and clinical genomics research communities, and identify the activities that are needed to expand, diversify, and support the AnVIL user community.

The workshop agenda includes two sessions, each with two concurrent virtual breakout rooms, focused on the following topics:

1. Data submission and consortia engagement
2. Analysis tools
3. Infrastructure
4. Outreach and training

In each session NHGRI is particularly interested in discussing the following cross-cutting topics, although discussions won't be limited to these:

- (a) how cloud-based platforms can better serve the needs of genomic researchers;
- (b) what tools and services would better support clinical genomic researchers;
- (c) how to improve interoperability with other NIH cloud-based genomic resources in a federated data ecosystem.

All meeting materials and recordings will be made publicly available on the workshop website soon after the end of the workshop.

## Agenda

October 29, 2021 – 12pm-5pm ET

- 12:00-12:10 Welcome and purpose of the workshop  
*Ms. Valentina Di Francesco (NHGRI) and Dr. Ken L. Wiley Jr. (NHGRI)*
- 12:10-12:20 Data Science at the Forefront of Enhancing Diversity in Genomics  
*Mr. Vence L. Bonham Jr., J.D. (NHGRI)*
- 12:20-12:35 Introduction to AnVIL  
*Dr. Anthony A. Philippakis (Broad) and Dr. Michael C. Schatz (Johns Hopkins)*
- 12:35-1:50 Session 1: Breakout rooms

Data submission and consortia engagement		Analysis tools	
<i>Moderators: Dr. Adam Resnick (Children's Hospital of Philadelphia) and Ms. Valentina Di Francesco (NHGRI)</i>		<i>Moderators: Dr. Marylyn Ritchie (University of Pennsylvania) and Dr. Ken L. Wiley, Jr. (NHGRI)</i>	
12:35-12:40	Moderator introductions	12:35-12:40	Moderator introductions
12:40-12:55	AnVIL presentation: <i>Dr. Brian O'Connor (Broad) and Dr. Frederick Tan (Carnegie)</i>	12:40-12:55	AnVIL presentation: <i>Dr. Vincent Carey (HMS) and Dr. Anne O'Donnell-Luria (Broad)</i>
12:55-1:40	Discussion	12:55-1:40	Discussion
1:40-1:50	Prepare breakout report	1:40-1:50	Prepare breakout report

- 1:50-2:15 Report back from Session 1  
*Rapporteurs: Dr. Adam Resnick and Dr. Marylyn Ritchie*

- 2:15-2:30 15 min break

- 2:30-3:45 Session 2: Breakout rooms

Infrastructure		Outreach and training	
<i>Moderators: Ms. Karen M. Davis (RTI International) and Dr. Carolyn M. Hutter (NHGRI)</i>		<i>Moderators: Dr. Siddharth Pratap (Meharry Medical College) and Mr. Christopher Wellington (NHGRI)</i>	
2:30-2:35	Moderator introduction	2:30-2:35	Moderator introduction
2:35-2:50	AnVIL presentation: <i>Dr. Jeremy Goecks (OHSU) and Dr. Benedict Paten (UCSC)</i>	2:35-2:50	AnVIL presentation: <i>Dr. Jeffrey Leek (JHU) and Ms. Tiffany Miller (Broad)</i>
2:50-3:35	Discussion	2:50-3:35	Discussion
3:35-3:45	Prepare breakout report	3:35-3:45	Prepare breakout report

- 3:45-4:10 Report back from Session 2  
*Rapporteurs: Ms. Karen M. Davis and Dr. Siddharth Pratap*

- 4:10-5:00 Summary and closing  
*Ms. Valentina Di Francesco and Dr. Ken L. Wiley Jr.*

## Discussant assignments to breakout rooms

### Session 1

#### Breakout room: Data submission and consortia engagement

*Moderators: Ms. Valentina Di Francesco and Dr. Adam Resnick*

Dr. Elizabeth (Liz) Blue  
Dr. David Crosslin  
Dr. Iftikhar Kullo  
Dr. Tara Matise

Dr. Aleksandar Milosavljevic  
Dr. Minoli Perera  
Dr. Stephen (Steve) Rich  
Dr. Kenneth (Ken) Rice

#### Breakout room: Analysis tools

*Moderators: Dr. Marylyn Ritchie and Dr. Ken Wiley, Jr.*

Dr. Nadav Ahituv  
Dr. Joshua (Josh) Akey  
Dr. Mark Craven  
Dr. Sean Davis  
Dr. Barbara Engelhardt  
Dr. James Knight

Dr. Anshul Kundaje  
Dr. Karen Miga  
Dr. Adam Phillippy  
Dr. Timothy (Tim) Reddy  
Dr. Chunhua Weng

### Session 2

#### Breakout room: Infrastructure

*Moderators: Ms. Karen Davis and Dr. Carolyn Hutter*

Mr. Samuel (Sandy) Aronson  
Dr. Vivien Bonazzi  
Dr. Brandi Davis-Dusenbery  
Dr. Richard Gibbs  
Dr. George Hripcsak

Dr. Eimear Kenny  
Dr. Lucila Ohno-Machado  
Dr. Shannon McWeeney  
Mr. Luke Rasmussen

#### Breakout room: Outreach and training

*Moderators: Dr. Siddharth Pratap and Mr. Christopher Wellington*

Dr. Cinnamon Bloss  
Dr. C. Titus Brown  
Dr. Carol Bult  
Dr. John Kwagyan  
Dr. Andrew Lee

Dr. Robert Meller  
Dr. Peter Robinson  
Dr. Sourav Roy  
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All NHGRI extramural staff is invited to participate in this workshop as listeners only.

## **Appendix 2**

### ***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.