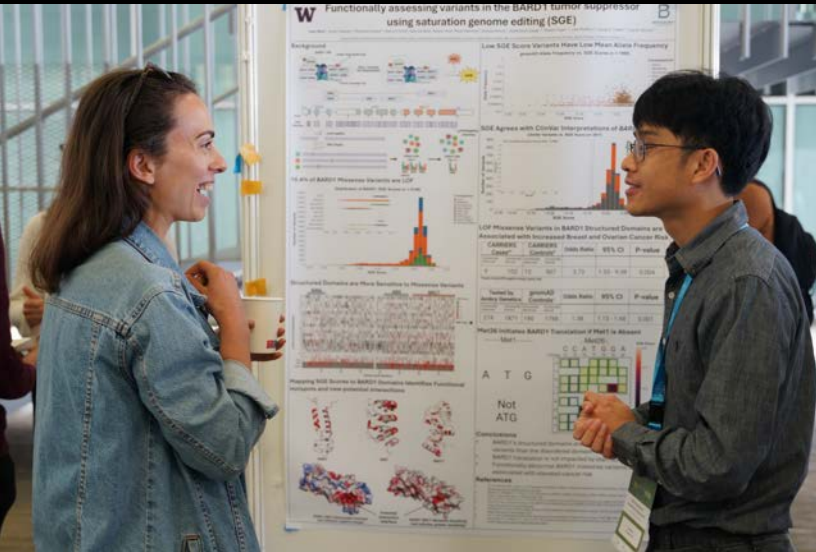


ATLAS OF VARIANT EFFECTS ALLIANCE

2025 ANNUAL REPORT



Atlas of Variant Effects Alliance

2025 - A YEAR OF MOMENTUM AND CONNECTION

Message from the Co-Chairs of the AVE Executive Committee

As we reflect on 2025, we are filled with gratitude and excitement for the remarkable strides our community has made. This year has been a testament to the power of collaboration, curiosity, and innovation in advancing variant effect research.

In 2025, our collective efforts reached new heights. Through our growing community of practice, we've now generated over 7 million variant effect measurements spanning more than 700 human and model organism genes. Much of the data has direct implications in human health.

Through dedicated workstreams and cross-institutional collaborations, AVE members have also contributed to a growing body of community resources and educational materials. There are now more than a dozen peer-reviewed publications that advance MAVE technologies, establish experimental and analytical standards, and inform clinical variant interpretation. These outputs reflect the strength of our community and its commitment to open, rigorous, and translational science.

We'd like to highlight a significant new collaboration that emerged this year. In April, our Clinical Variant Interpretation (CVI) workstream joined with ClinGen to launch the ClinGen-AVE Functional Data Working Group, convening experts from AVE, ClinGen, IGVF, GA4GH and NIH to develop guidance for incorporating functional data in clinical practice.

One of the year's most energizing moments was the Mutational Scanning Symposium in Barcelona. The event brought together researchers from across experimental biology, computational modeling, and clinical translation, creating a vibrant space for sharing ideas and forging new collaborations. The quality of the science and the depth of engagement underscored the momentum behind our field. We extend our sincere thanks to the organizers, sponsors, speakers, and attendees who made it such a meaningful gathering.

Beyond the symposium, we've witnessed tremendous growth in our community. Membership has expanded globally and we now have over 800 members from 58 countries. The AVE Alliance continues to grow—not just in numbers, but in ambition, reach, and impact. We are proud to be part of a community advancing science, as well as nurturing the next generation of researchers. Thank you for your continued dedication, insight, and passion. Together, we are building a future where variant effect research is transformational to human health.

With appreciation,



Matt Hurles and Doug Fowler
Co-Chairs, AVE Executive Committee

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ABOUT US

The Atlas of Variant Effects (AVE) Alliance, founded in 2020, is an international collaboration dedicated to developing, disseminating, and democratizing technologies for mapping variant effects. Our vision is to create a comprehensive atlas of variant effect maps for key regions of human and pathogen genomes to aid in the diagnosis, prognosis, and treatment of diseases. We unite genomics researchers, curators, health practitioners, and funders from around the world to set standards, share tools, and strategize. Our goal is to facilitate the creation of comprehensive variant effect maps and establish community-driven resources for sharing and analyzing large-scale variant effect data.

OUR VALUES AND PRINCIPLES

- Inclusivity
- Open Access Science
- Research Excellence and Integrity
- Respect, Relevance, Reciprocity, Responsibility, Relationships



BY THE NUMBERS



800+
members
58
countries



4
committees



1
interest group



4
work-streams



8
Mutational
Scanning
Symposiums



90+
Seminar Series
speakers



300+
MAVE
Registry
Projects



7M+
variant effect
measurements
in MAVEDB



2+
workshops
per year



200+
educational
videos



>50
manuscripts



>10
Online
resources
and toolkits



8
podcast
episodes



1.2K
youtube
subscribers



1.4K
social
media
followers



16K
unique visits

LEADERSHIP



David Adams



Benedetta Bolognesi



Anna Gloyn



Irene Gallego Romero



Matthew Hurles



Doug Fowler



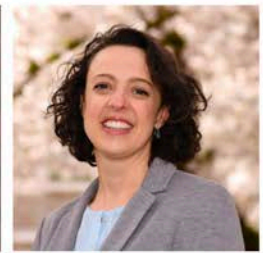
Jonathan Mill



Fritz Roth



Alan Rubin



Lea Starita



Executive Committee: David Adams, Benedetta Bolognesi, Anna Gloyn, Irene Gallego Romero, Matthew Hurles (co-chair), Doug Fowler (co-chair), Jonathan Mill, Frederick 'Fritz' Roth, Alan Rubin, Lea Starita

Executive Office Staff: Lara Muffley and Alex Hopkins-Sathe

Workstream Co-chairs:

- Lea Starita and Clare Turnbull - ClinGen AVE Functional Data Working Group (FDWG)
- Julia Foreman and Alan Rubin - Data Coordination and Dissemination (DCD)
- Alex Nguyen Ba and Sven Diederichs - Experimental Technology and Standards (ETS)
- Joseph Marsh and Vikas Pejaver - Analysis, Modelling and Prediction (AMP)

Virology Interest Group Chair: Taha Y Taha

Communications Working Group: Lara Muffley, Dean Owen, Rachael Smith

Podcast committee: Alex Nguyen, Ziyi Dai, Moez Dawood, Jerome Freudenberg, Kortni Kindree, Katie Partington, Adrine de Souza

Variant Effect Seminar Series (VESS) Organizing Committee: Priyanka Bajaj, Ohanna Bezerra, Matteo Cagiada, Ziyi Dai, Jingyou Rao, Adelaide Tovar

Code of Conduct Committee: Jeffrey Calhoun, Lara Muffley, Irene Gallego Romero, Rehan Villani

OUR MEMBERS

Membership growth

- The Alliance exceeded 800 members in 2025. These individuals include early and mid-career scientists, along with senior-level individuals in industry, government, and academia, as well as patient advocacy groups and foundations - a total of more than 380 institutions. The overwhelming majority (80 percent) serve in the academic sector.
- Membership is geographically diverse. Our members are located in the following 58 geographic regions: Algeria, Australia, Austria, Bangladesh, Belgium, Brazil, Bulgaria, Canada, Chile, China, Croatia, Cyprus, Denmark, Egypt, Estonia, Finland, France, Germany, Ghana, Greece, Hong Kong, Hungary, India, Iran, Ireland, Israel, Italy, Japan, Latvia, Lebanon, Luxembourg, Malaysia, Mexico, Nepal, Netherlands, New Zealand, Nigeria, Norway, Pakistan, Philippines, Poland, Qatar, Republic of Korea, Romania, Russia, Saudi Arabia, Singapore, South Africa, Spain, Sweden, Switzerland, Thailand, Tunisia, Turkey, UAE, Ukraine, the United Kingdom, and the United States.



Global Representation of Alliance Members

HIGH IMPACT VARIANT EFFECT MAPS

This past year, our global community made exceptional progress in particular towards realizing a [Clinical Atlas of Variant Effects](#). The completion of these variant effect maps marks a significant milestone in clinical genomics. Notable genes mapped in 2025 include:

- AIRE | Autoimmune Disease ([preprint](#))
- BARD1 | Breast and ovarian cancer ([preprint](#))
- LDLR | Cardiovascular Disease ([publication](#))
- F9 | Hemophilia B, Bleeding Disorders ([publication](#))
- FGFR kinase domain | Tumorigenesis ([publication](#))
- KCNQ1 | Arrhythmia ([preprint](#))
- MUTYH | Colorectal Cancer ([publication](#))
- MYBPC3 | Hypertrophic cardiomyopathy ([preprint](#))
- SOD1 | Neurodegenerative Disease/ familial ALS ([publication](#))

To learn more about the growing application of MAVE data in clinical settings, we recommend reading the following review articles published in 2025:

📄 Creating an atlas of variant effects to resolve variants of uncertain significance and guide cardiovascular medicine [DOI: 10.1038/s41569-025-01201-7](https://doi.org/10.1038/s41569-025-01201-7)

📄 Multiplexed assays of variant effect for clinical variant interpretation [DOI: 10.1038/s41576-025-00870-x](https://doi.org/10.1038/s41576-025-00870-x)



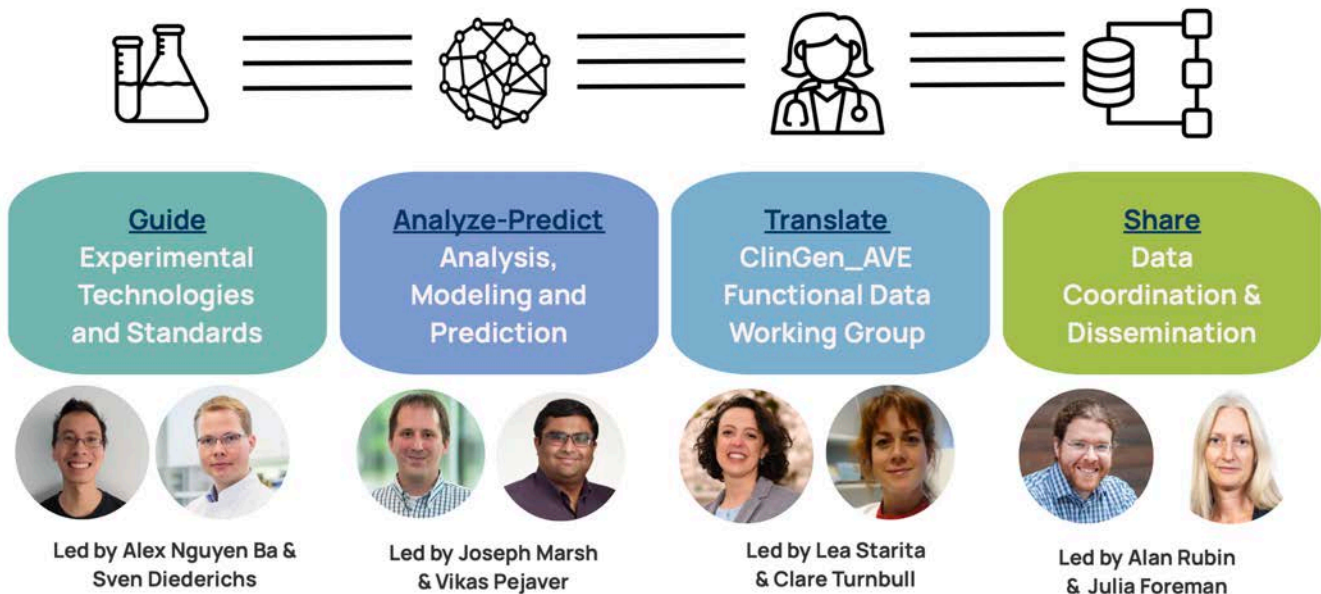
Left: Silvia Casadei - SGE team lead (Starita lab) Brotman Baty Institute, UW Seattle, USA

Right: Larissa Matsuyama, Fernanda Arriaga González, Rebeca Olvera León (Adams group) Wellcome Sanger, Hinxton, UK

WORKSTREAM UPDATES

The Alliance has four interconnected work streams. All have been making strides in setting standards, providing tools, and disseminating information to advance the field of variant effect mapping. <https://www.varianteffect.org/workstreams/>

How we work: 4 interconnected workstreams



Experimental Technology and Standards (ETS) Workstream

Co-Chairs: Alex Ngyuen Ba (University of Toronto) and Sven Diederichs (University of Freiburg & German Cancer Research Center (DKFZ), Heidelberg).
Outgoing ETS chair Andrew Glazer (Vanderbilt University)

The ETS workstream supports the MAVE community by facilitating the development, evaluation, and dissemination of experimental methods for variant effect mapping. The focus is on establishing best practices and quality standards across the experimental pipeline—from library construction through data generation.

This past year, a centralized protocols resource on protocols.io was launched, providing the community with accessible, standardized protocols for variant effect mapping experiments. Looking ahead, a comprehensive review article is being prepared that will offer technology-focused guidance and recommendations for experimental design based on different mutagenesis approaches. Finally, ETS will launch a series of educational coding notebooks for analyzing and producing variant effect maps and quality control metrics from sequencing reads.

Analysis, Modelling and Prediction (AMP) Workstream

Co-Chairs: Joseph Marsh (The University of Edinburgh) and Vikas Pejaver (Icahn School of Medicine at Mount Sinai)

The AMP workstream focuses on computational approaches for analyzing MAVE data and predicting variant effects, with the aim of improving interpretation, mechanistic discovery and downstream clinical application. The workstream emphasizes best-practice guidelines, evaluation and promotion of existing methods, and integration of experimental assays with computational analyses.

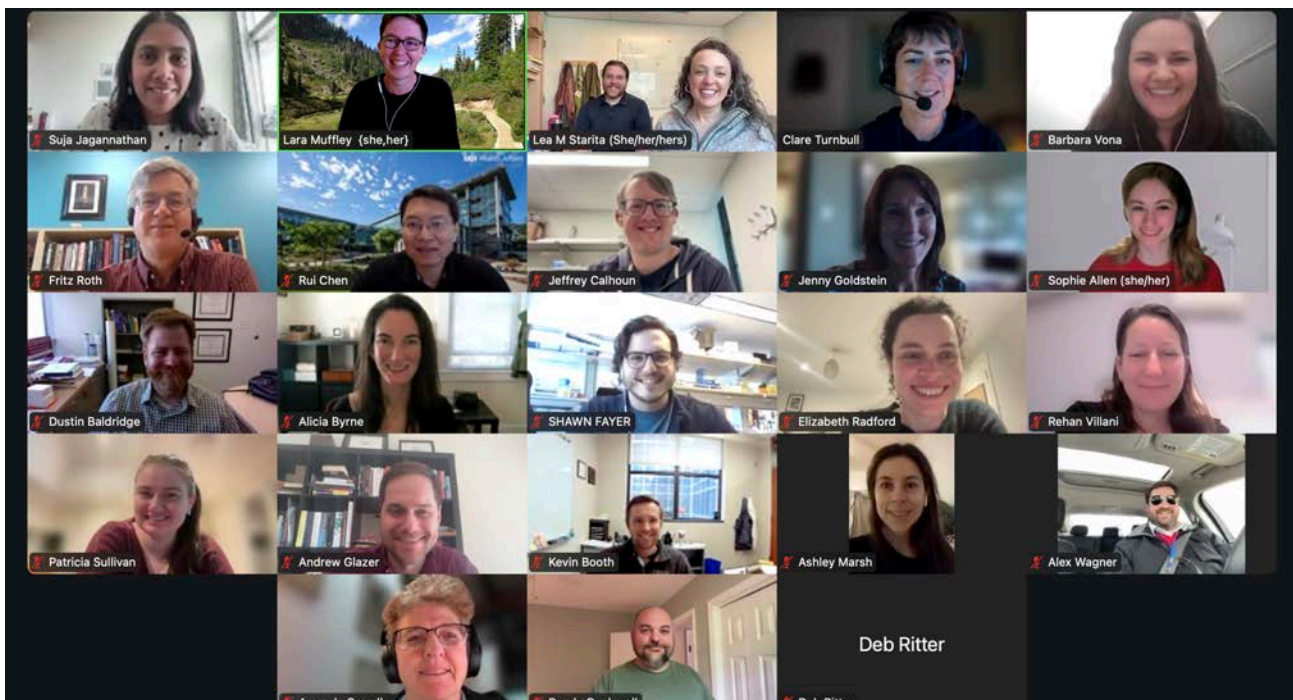
This past year, the AMP-led paper '[Guidelines for Releasing a Variant Effect Predictor](#)' was published in Genome Biology, building on our [Variant Effect Predictors resource](#). In addition, with the Data Coordination and Dissemination (DCD) workstream, we published an overview of '[Variant Scoring Tools for Deep Mutational Scanning](#)' in Molecular Systems Biology, to accompany our [MAVE Analysis Tools](#) resource. A ranked priority list is currently being developed to guide the selection of high-impact targets for future MAVE experiments, helping to strategically direct community resources toward the most valuable genes and domains. Much of this work was built on presentations and discussions with early career researchers in monthly meetings, supporting their scientific and professional development while furthering workstream objectives.

Collaborations with the Clinical Genome ([ClinGen](#)) Resource and the Critical Assessment of Genome Interpretation ([CAGI](#)) consortia have enabled the assessment of state-of-the-art variant effect predictors as evidence for clinical variant classification and in the prediction of MAVE measurements, respectively, shedding light on the relationships between assay data, computational predictions and clinical pathogenicity.

ClinGen AVE Functional Data Working Group

Co-Chairs: Lea Starita (UW, BBI) and Clare Turnbull (ICR)

The ClinGen-AVE Functional Data Working Group is a collaborative effort between members of the Atlas of Variant Effects Alliance and ClinGen, a National Institutes of Health (NIH)-funded community dedicated to building a central resource that defines the clinical relevance of genes and variants for use in precision medicine and research. The working group's purpose is to provide high-level generalized guidance for using functional data in variant classification.



ClinGen AVE Functional Data Working Group zoom kickoff meeting (April 2025)

Key activities of the group include: developing approaches for calibration of functional assays, guiding Variant Curation Expert Panel (VCEP) use of multiplexed assays of variant effects (MAVE) data, facilitating the development and testing of VCEP curated variants in MAVE and other functional assays, and informing priorities for large-scale functional data generation efforts.

Education and outreach are core activities of this working group. This past year, two in-person workshops were held: "How to release MAVE datasets that clinicians can use" at the Mutational Scanning Symposium in Barcelona, Spain (May 2025), and a Functional Evidence Workshop at Curating the Clinical Genome in Hinxton, UK (June 2025).

An accessible online educational resource was launched (<https://www.varianteffect.org/clinical-application/>) offering guidance on the clinical applications of functional evidence.

This year the group also [published a commentary](#) outlining the use case for combining multiple MAVE datasets as well as best practices to achieve outcomes such as improved pathogenicity classification.

Current efforts are focused on providing updated guidance for using functional evidence according to the new ACMG/AMP/CAP/ClinGen Sequence Variant Classification v4 recommendations.

Data Coordination and Dissemination (DCD) Workstream

Co-Chairs: Julia Foreman (DECIPHER, EMBL-EBI) and Alan Rubin (University of Melbourne)

The DCD workstream facilitates the sharing and discoverability of MAVE projects and data. Focus areas include: defining and promoting standards for data deposition and dissemination, engaging with clinical and non-clinical data resources to enable data sharing and federation, and overseeing the growth and management of [MaveDB](#) - the open access public repository for datasets from Multiplexed Assays of Variant Effect (MAVEs).

In 2025, workstream members made significant progress in making MAVE data more accessible and clinically actionable. A major publication, "[Mapping MAVE data for use in human genomics applications](#)," described efforts to map MAVE data from the MaveDB community database to human reference sequences, creating an extensive set of machine-readable homology mappings that have been incorporated into widely used human genomics applications.

This important workstream project helped enable the launch of a new product: [MaveMD](#) (MAVEs for MeDicine), a functional data interface for the MaveDB database that integrates

with external resources such as ClinVar and the ClinGen Allele Registry, displays clinical evidence calibrations, provides intuitive visualizations, and exports structured evidence compatible with ACMG/AMP variant classification guidelines.

Current efforts include developing data quality metrics in consultation with other workstreams and academic and industry partners, as well as expanding model organism data integration to improve sharing and delivery of annotations that guide cross-organism comparisons and applications.

VIROLOGY INTEREST GROUP

We established a new virology interest group this year under the leadership of Taha Y. Taha from the Gladstone Institute. Meeting monthly via Zoom, this group unites researchers who study viral genetics, evolution, and host-virus interactions using variant effect mapping approaches.

Interested in joining this group? Reach out! <https://www.varianteffect.org/contact-us/>

PUBLICATIONS

Recommended Reading List (2025) - This curated publication list includes collaborative workstream-led efforts and articles which cite the Atlas of Variant Effects Alliance.

- [The functional landscape of coding variation in the familial hypercholesterolemia gene LDLR](#)
- [An evolving understanding of multiple causal variants underlying genetic association signals](#)
- [acmgscaler: an R package and Colab for standardized gene-level variant effect score calibration within the ACMG/AMP framework](#)
- [Creating an atlas of variant effects to resolve variants of uncertain significance and guide cardiovascular medicine](#)
- [Variant scoring tools for deep mutational scanning](#)
- [Landscapes of missense variant impact for human superoxide dismutase 1](#)
- [Combining multiplexed functional data to improve variant classification](#)
- [Multiplexed assays of variant effect for clinical variant interpretation](#)

- [Mapping MAVE data for use in human genomics applications](#)
- [Consultation informs strategies for improving the use of functional evidence in variant classification](#)
- [Validating data from multiplex assays of variant effect: A CanVIG-UK national survey of NHS clinical scientists](#)
- [Insights on improving accessibility and usability of functional data to unlock their potential for variant interpretation.](#)
- [Atlas of Variant Effects 2030 Roadmap: resolving human variants of uncertain significance](#)
- [Guidelines for releasing a variant effect predictor](#)
- [MaveDB 2024: a curated community database with over seven million variant effects from multiplexed functional assays](#)
- [Site-saturation mutagenesis of 500 human protein domains](#)
- [Epitope mapping via in vitro deep mutational scanning methods and its applications](#)

MUTATIONAL SCANNING SYMPOSIUM

The symposium's 8th iteration took place in Barcelona, Spain May 21–23, 2025, establishing its first presence in continental Europe. The event included two specialized satellite workshops: "How to choose a model for variant interpretation and protein design" (May 20th) and "How to release MAVE datasets that clinicians can use" (May 23rd), providing practical training on critical translation and implementation issues.

Held at the Barcelona Biomedical Research Park (PRBB), the 2.5-day symposium attracted 310 registrants from 20 countries (250 in-person, 60 virtual), featuring 31 speakers, 2 satellite workshops with 7 instructors, and 127 poster presentations showcasing cutting-edge advances in variant interpretation and functional genomics with notable emphasis on AI integration.

Special recognition goes out to organizers: Benedetta Bolognesi (IBEC), Mafalda Dias (CRG), Jonathan Fraser (CRG), Lara Muffley (University of Washington), Lea Starita (UW), and Ben Lehner (CRG) - supported by a fantastic local events team who brought it all together.

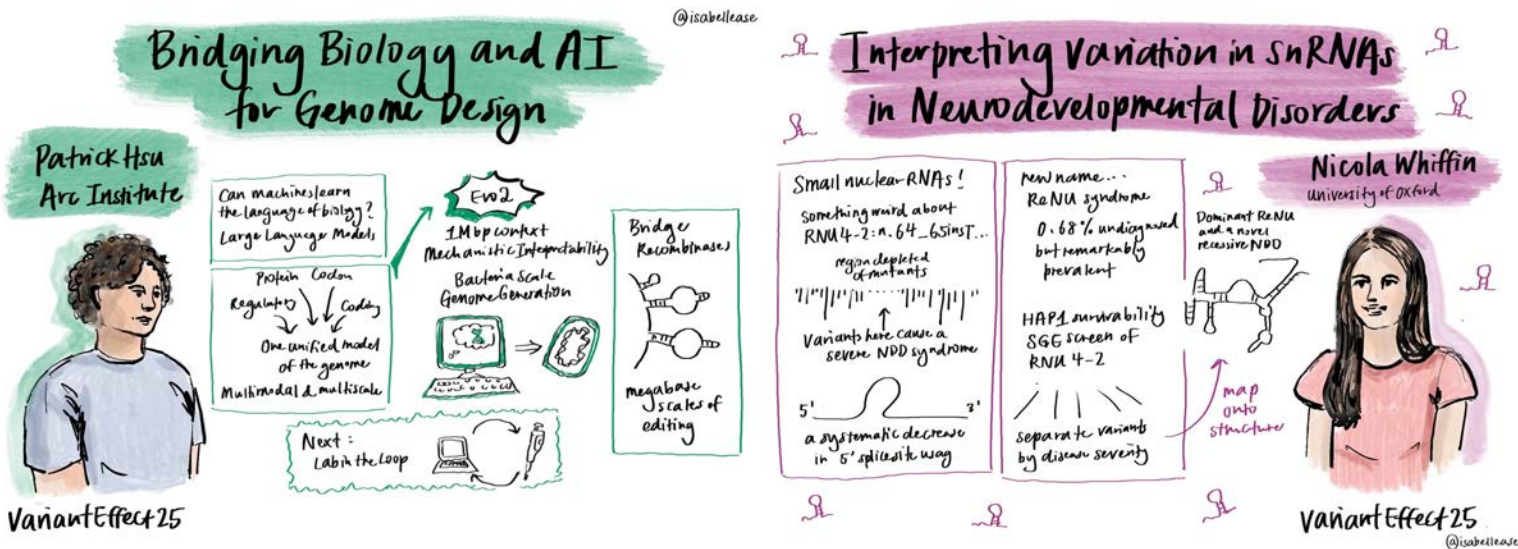


Group photo from the 8th annual Mutational Scanning Symposium (Barcelona, Spain)



Photos of speakers and poster presenters at the Mutational Scanning Symposium (Barcelona, Spain)

Our surprise live conference illustrator [Isabelle Zane](#) captured [visual summaries of each talk](#). Recordings from the event can be found on the Alliance's [YouTube](#) channel,



You can also read more about the symposium in the articles linked below:

- [MSS 2025 Debrief: 'I Ended Up Attending Every Talk and Gained Valuable Insights from the Poster Sessions'](#)
- [Spotlight on MSS 2025: Lindorff-Larsen to Address Symposium on Interface Between Variant Effects and Interpretation](#)
- [MSS 2025 Update: BioMarin Sponsorship Aims to Drive Progress in Development of Therapies for Rare Diseases](#)
- [MSS 2025 Speakers will Offer Perspectives on AI, Bioengineering, and Other Topics](#)

The event was generously supported by AstraZeneca, BioMarin, Brotman Baty Institute for Precision Medicine, Centre for Genomic Regulation (CRG), Chan Zuckerberg Initiative (CZI BioHUB), Constantiam, GCATbio, Illumina, Institute for Bioengineering of Catalunya (IBEC), La Caixa Foundation, and Twist Biosciences.



WORKSHOPS

Two workshops, held in May 2025 adjacent to the Mutational Scanning Symposium, addressed critical challenges in variant interpretation and clinical genomics. The first workshop guided participants in: selecting appropriate computational tools for variant interpretation and protein design, covering the landscape of available analysis, and modeling approaches while emphasizing how to evaluate tool performance for specific research needs. The second workshop focused on: translating MAVE (Multiplexed Assays of Variant Effect) datasets into clinically actionable formats, teaching participants how to standardize functional data according to ACMG and ClinGen guidelines, implement FAIR data-sharing principles, and utilizing resources like MaveDB and ClinVar to make their datasets directly accessible to clinicians.



Workshop attendees (May 2025 - Barcelona, Spain)

Workshop 1: May 20th “How to choose a model for variant interpretation and protein design” Instructors: Noelia Ferruz (CRG), Albert Escobedo (CRG) and Pascal Notin (Harvard University)

Workshop 2: May 23rd “How to release MAVE datasets that clinicians can use” Instructors: Lea Starita (UW), Alan Rubin (WEHI/University of Melbourne), Abbye McEwen (UW), Malvika Tejura (UW)

MAVE EDUCATIONAL COURSE

As part of our commitment to advancing genomic medicine, in partnership with Wellcome Connecting Science, we launched a new educational course focused on Multiplex Assays of Variant Effects (MAVEs). The Multiplex Assays of Variant Effects (MAVEs): Approaches, Analysis, and Interpretation was held 23–28 November 2025, in Hinxton, UK.

This inaugural week-long course brought together an international cohort of PhD students, postdoctoral scientists, clinicians, and clinical scientists engaged in genetic variant interpretation. The course equipped participants with a foundation in the principles, tools, and applications of multiplex assays of variant effects (MAVEs) for clinical and research applications.

The course was led by the following AVE Affiliated MAVE experts: David Adams (Wellcome Sanger Institute), Sophie Allen (The Institute of Cancer Research), Jorge Batista da Roche (EMBL-EBI), Matthew Coelho (Wellcome Sanger Institute), Miranda Durkie (NHS North East Yorkshire Genomic Laboratory Hub), Greg Findlay (The Francis Crick Institute), Sebastian Gerety (Wellcome Sanger Institute), Helen Firth (Newnham College, University of Cambridge), Julia Foreman (EMBL EBI), Irene Gallego Romero (St Vincent's Institute for Medical Research, Australia), Adam Hunter (Wellcome Sanger Institute), Sarah Hunt (EMBL-EBI), Daniel Jaramillo Calle (Wellcome Sanger Institute), Joseph Marsh (University of Edinburgh), Sofia Obolenski (Wellcome Sanger Institute), Victoria Offord (Wellcome Sanger Institute), Rebeca Olvera-León (Wellcome Sanger Institute), Elizabeth Radford (Cambridge University Hospitals NHS), Charlie Rowlands (The Institute of Cancer Research), Alan Rubin (University of Melbourne, Australia), Likhitha Surapaneni (EMBL-EBI), Andrew Waters (Wellcome Sanger Institute) and Clare Turnbull (The Institute of Cancer Research).

VARIANT EFFECTS SEMINAR SERIES (VESS)

This seminar series is a platform for early career scientists to share their research related to interpreting human genetic variation. In September of 2025, VESS celebrated its four-year anniversary! Seminars are open to the public and recordings are posted to our [YouTube channel](#).

The top three most viewed VESS videos in 2025 were:

- "[Bridge RNAs direct programmable recombination of target and donor DNA](#)"
Matthew Durrant & Nicholas Perry (4.3K views)
- "[Leveraging single-cell transcriptomics to study complex diseases](#)"
Annique Claringbould (1.9K views)
- "[Disease variant prediction with deep generative models of evolutionary data](#)"
Jonathan Frazer and Mafalda Dias (1.6K views)

Current members of the [VESS Organizing Committee](#) are: Priyanka Bajaj, Ohanna Bezerra, Matteo Cagiada, Ziyi Dai, Jingyou Rao, and Adelaide Tovar. The Alliance also thanks all previous VESS organizing committee members - Steven Erwood, Yann Ilboudo, Mariano Martín, and Mireia Seuma who have helped make our series so successful.

VARIANTS AND US PODCAST

Under the direction of Alex Nguyen Ba at the University of Toronto, the Variants and Us podcast (VUSPod) delivers accessible insights into state-of-the-art functional genomics research. Sponsored by the AVE Alliance and debuting in 2024, the program features interviews with experts exploring both foundational and clinical research on genetic variant impacts in disease contexts, including rare disorders.

Podcast committee members are: Alex Nguyen, Ziyi Dai, Moez Dawood, Jerome Freudenberg, Kortni Kindree, Katie Partington, and Adrine de Souza.



VUS Pod

14+ Interviews
7+ Episodes

850+ listens
170+ followers

Listeners tuning in
from 35 countries:
36.5% US,
18.8% Canada,
9% UK

VUS Pod Episodes: <https://www.varianteffect.org/podcast/>

It's in your blood

Vijay Sankaran and John Doench

Biocuration: from Evidence to Classification

Heidi Rehm & Courtney Thaxton

Deep Thought

Debora Marks & Joe Marsh

Live from the Mutational Scanning Symposium

Interviews with attendees

Your genes on drugs: context matters

Frederick Roth, Iris Cohn & Michelle Axford

Melanoma and Variant Effects: Beyond Sunscreen

Dave Adams & Jiyeon Choi

Variant Effect What

Stan Fields, Doug Fowler & Greg Costain

NEWSWORTHY MENTIONS

The Atlas of Variant Effects Alliance was mentioned in the news throughout 2025, here are a few top stories:

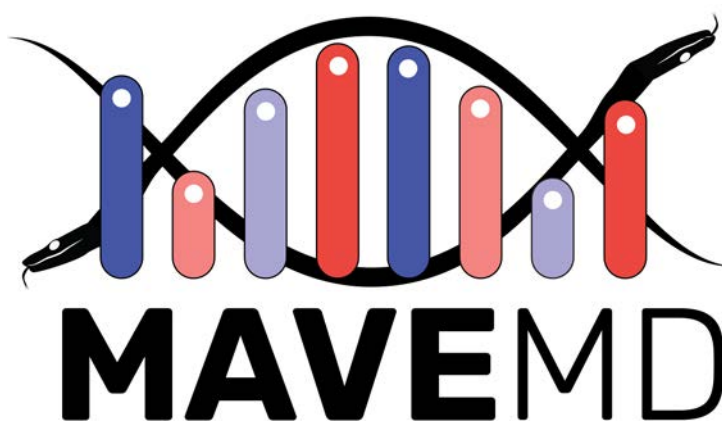
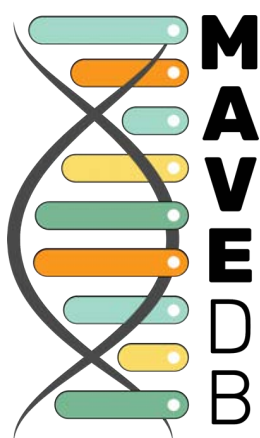
- [Two Organizers of the Variant Effects Seminars Share Observations and Insights](#)
- [Study Links Genetic Variants to Higher "Bad" Cholesterol and Heart Attack Risk](#)
- [AVE Alliance Working Group Developing International Guidelines for Genetic Variant Classification](#)
- [Researchers Offer Guidelines for Variant Effect Predictors](#)
- [New guidelines aim to improve transparency and trust in genetic prediction tools](#)

And more! Read them all here: <https://www.varianteffect.org/news>

MAVE RESOURCES AND TOOLS

Multiplexed assays of variant effects (MAVEs) allows the assessment (in a single experiment) of thousands of variants simultaneously. This technique, which harnesses the power of next-gen sequencing, yields large-scale data sets that can shed light on the functional consequences of protein variants and, ultimately, on the landscape of human genetic variation. The Alliance provides (and continually updates) a suite of resources and tools for variant effect mapping which include:

MAVEDB | MAVEMD



MaveDB is the open source platform and database of record for variant effect mapping and the deposition of MAVE datasets. MaveDB now contains over 7 Million variant effect measurements.

This year, a new interface was introduced - MaveMD (MAVEs for MeDicine) to make MAVE data more accessible and usable for clinical variant classification and interpretation. Read more about this project here: [MaveMD: A functional data resource for genomic medicine](https://www.mavedb.org/mavemd) or explore directly online <https://www.mavedb.org/mavemd>.

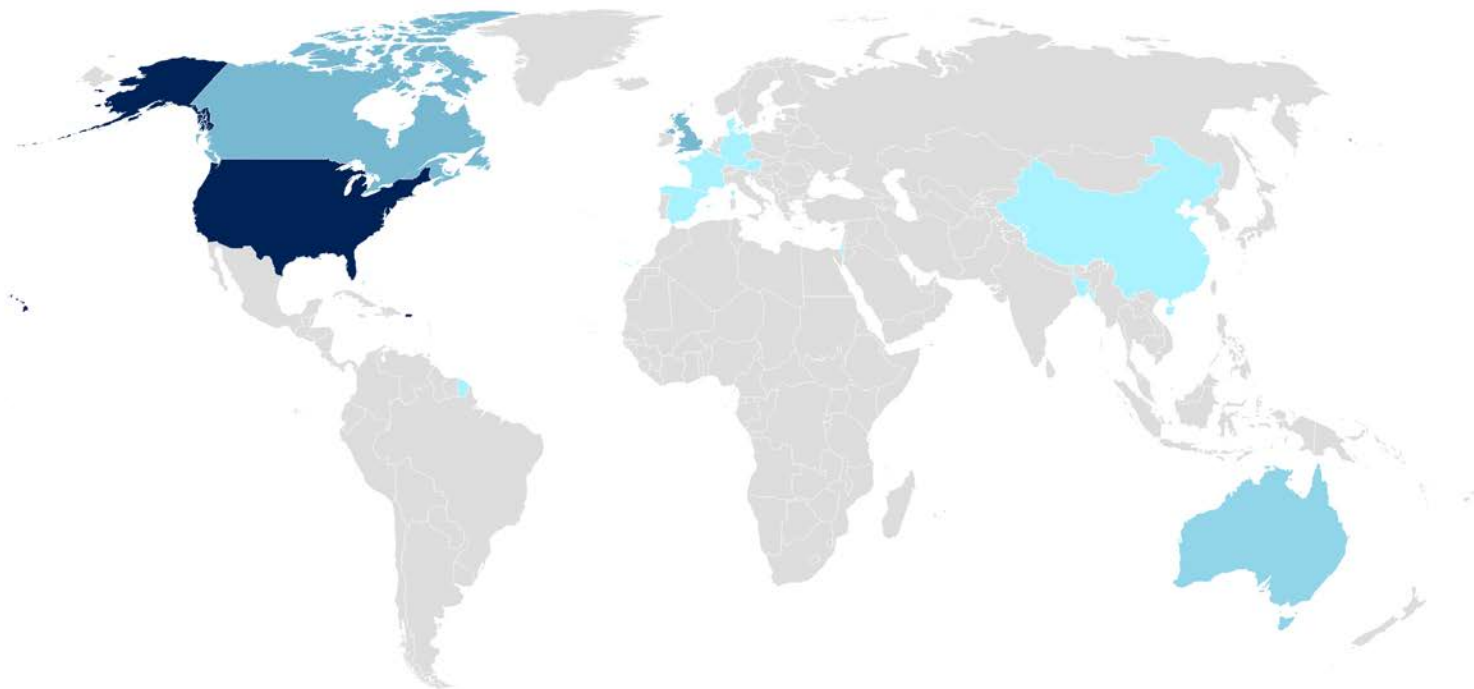
MAVE REGISTRY

[MaveRegistry](#) continues to serve as the coordination hub for the growing international community engaged in multiplexed assays of variant effect (MAVEs). The registry facilitates collaboration, enables discovery of ongoing work, and helps researchers communicate progress to key stakeholders including fellow scientists, clinicians, and funding agencies. The site allows users to receive updates about projects and teams that they follow, propose MAVE targets, and register their own projects.

As of December 2025, there are 528 users and 344 registered projects. 124 registered research teams are distributed across 13 countries and 18 U.S. states.

MaveRegistry Team Locations

Number of Teams



Created with Datawrapper

EDUCATIONAL RESOURCES

We provide numerous resources in order to provide guidance and to help make MAVEs more accessible. Visit this page to learn more: <https://www.varianteffect.org/resources/>

PROTOCOLS

The [AVE protocols.io workspace](https://www.protocols.io/workspaces/atlas-of-variant-effects-alliance/publications) currently includes the following SOPs:

- Saturation Genome Editing (SGE)
- LABEL-seq
- Preparing 8OG:A repair reporter for MUTYH variant functional assays
- High Efficiency Yeast Library Transformation
- DIMPLE library generation and assembly protocol
- Converting ssDNA oligos to dsDNA with T4 DNA polymerase
- sgRNA library re-amplification in liquid culture
- Rapid and robust cloning of sgRNA expression plasmids
- And more!

<https://www.protocols.io/workspaces/atlas-of-variant-effects-alliance/publications>

We encourage all AVE members to share their SOPs. If you would like to contribute a protocol to this collection please view this [video tutorial](#).

EDUCATIONAL VIDEOS

The Alliance also provides educational videos which are free to view on our [youtube channel](#). We now have 200+ videos, with over 80K views, and 1.2K subscribers. The channel includes talks from previous Symposiums, Seminar Series and more! One of our most viewed video to date is an [Introduction to Deep Mutational Scanning](#) by Kenny Matreyek (3.4K views). Another popular video is the [Animated Introduction to Deep Mutational Scanning](#) (2.9K views).

HIGHLIGHTS RECAP

Variant Effect Maps

Over 7 Million variant effect measurements now in [MaveDB](#)

New Resources

- MaveMD <https://www.mavedb.org/mavemd>
<https://www.medrxiv.org/content/10.1101/2025.11.15.25336228v1>
- MAVE Data Analysis Tools <https://www.varianteffect.org/analysis-tools>
<https://doi.org/10.1038/s44320-025-00137-x>
- Variant Effect Predictors <https://www.varianteffect.org/veps>
<https://doi.org/10.1186/s13059-025-03572-z>

Leadership changes

- AVE Executive Committee
 - Jonathan Mill joined (May 2025), J.T. Neal stepped down (Dec 2025)
- AVE Workstreams
 - AMP: Vikas Pejaver appointed as co-chair alongside Joseph Marsh (May 2025)
 - ETS: Leadership transition with Andrew Glazer's departure and Sven Diederichs assuming co-chair role with Alex Nguyen Ba (Nov 2025)

ClinGen AVE Functional Data Working Group (New!)

- In partnership with [ClinGen's Variant Classification Working Group](#) (April 2025)

MAVE Educational Course (New!)

- Held 23–28 November 2025 in conjunction with Wellcome Connecting Science

Virology Interest Group (New!)

- Led by Taha Y Taha (Gladstone Institute) (Inaugural meeting Dec 2025)

Variant Effects Seminar Series (VESS)

- [90 speakers](#) from 11 countries (2021-2025)
- Mariano Martín rotated off the organizing committee

Website Refresh

- Key contributions from Sanger web developers: Paul Bevan, Stephen Robinson

Variants and Us Podcast

- Latest episode: "It's in your blood" with Drs Vijay Sankaran and John Doench
- Committee update: Evelina Tronina departing; Jerome Freudenberg and Katie Partington joining

KUDOS AND SHOUTOUTS

“The Alliance has played an invaluable role in supporting our work at Sanger as we develop a pipeline to deliver variant data at scale. Engagement through the Alliance has created a highly supportive and collaborative forum for open, thoughtful discussions, particularly around data release practices. We have greatly valued the opportunity to exchange perspectives, learn from the experiences of others, and align on shared approaches, and we look forward to continuing this productive and rewarding collaboration. In addition, the VESS seminar series and the annual Alliance meeting have been highly valuable in keeping our teams informed of emerging best practices”

Sónia Gonçalves – Head MAVE Operations | Wellcome Sanger Institute

“A little more than a year ago I found myself taking over a MAVE project in a genetics lab consisting of mainly computational biologists. I joined as a former industry Research Associate accustomed to product and process development at a tissue engineering-focused startup. As the only wet-lab aficionado amongst the dry-labbers, with an eagerness to learn but zero experience in molecular biology, computational biology, or genetics, the AVE Alliance has been an invaluable resource. Through the online resources, Slack channel, and annual Mutational Scanning Symposium, I have accessed protocols, found relevant literature, learned about data analysis pipelines, and made friends and colleagues who have been instrumental in progressing my project.”

Matt Blake – Research Associate (Claw lab) | University of Colorado Anschutz

“The Atlas of Variant Effects Alliance is a welcoming and collaborative community. I’ve had the opportunity to attend three MSS so far, and being able to share my work and receive feedback from experts in the field has been invaluable to my research. I recently discovered The Variants and Us podcast, which covers a range of topics and offers a different way to learn and connect. In addition, the many available MAVE resources helps me keep learning new approaches and perspectives that strengthen my research.”

Rebeca Olvera León – PhD student (Adams Group) | Wellcome Sanger Institute

“The AVE Seminar Series has been one of the most valuable parts of my engagement with the Alliance. The presentations consistently highlight cutting-edge work across the MAVE community and have broadened my perspective on both methodological and biological questions. I’ve also found MaveDB to be an indispensable resource. Its accessibility and organization make it easy to explore datasets and integrate them into ongoing projects. Together, these resources have meaningfully supported my research and strengthened my connection to the broader AVE community.”

Taha Y. Taha – Research Investigator | Gladstone Institute

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WHAT TO LOOK FOR IN 2026

- More 'live events'! e.g. [Fireside Chat with Matthew Hurles and Maitreya Dunham](#)
- 9th Annual [Mutational Scanning Symposium](#) Melbourne, Australia (March 25-27, 2026)
- Upgrades and enhancements to our central data repository
- New interest groups and AVE led initiatives

Save the date

[9th Annual Mutational Scanning Symposium](#)

March 25-27 , 2026

Melbourne, Australia

[10th Annual Mutational Scanning Symposium](#)

Tentative: **June 23-25th 2027**

Seattle, WA USA

OPPORTUNITIES TO ENGAGE AND CONNECT

The Alliance fosters a collaborative and inclusive community committed to the open exchange of MAVE data, protocols, and methodologies. Outputs from our collaborative activities include shared resources that accelerate progress in variant interpretation.

Participation in the Alliance is open to all, and members are actively encouraged to engage through a variety of pathways:

- Join the Alliance (<https://www.varianteffect.org/membership>)
- Attend or present at the monthly Variant Effects Seminar Series ([VESS](#))
- Attend and/or present at the [Annual Mutational Scanning Symposium](#)
- Propose a topical interest group to explore emerging scientific themes
- Join workstreams and committees during open calls
- Share experimental protocols via the [AVE protocols.io](#) repository
- Consider supporting us! <https://www.varianteffect.org/donate>
- Tune into the Variants and Us podcast ([VUSPod](#))
- Explore educational content on AVE's [YouTube channel](#)
- Engage with the community on [LinkedIn](#) and [BlueSky](#)
- Contribute to or utilize the MAVE [resource collection](#)
- Register your gene or project on [MaveRegistry](#)
- Deposit a variant effect map into [MaveDB](#)
- Utilize [MaveMD](#) for clinical insights

[Members](#) of the Alliance receive an invitation to join our Slack channel and are welcome to post in one of the open channels:

- ▶ [#educational_resources](#)
- ▶ [#general](#)
- ▶ [#introductions](#)
- ▶ [#job_announcements](#)
- ▶ [#literature](#)
- ▶ [#mss26](#)
- ▶ [#random](#)
- ▶ [#tips-tricks-troubleshooting](#)

The Atlas of Variant Effects (AVE) Alliance

Research Organization Registry (ROR) # <https://ror.org/00p2ftz29>

Report prepared by: Lara Muffley - Director of Program Operations for the Atlas of Variant Effects Alliance

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Thank You!