

### INTRODUCTION

The Center for the Multiplexed Assessment Phenotype (CMAP), is a Center of Excellence in Genome Science based at the University of Washington and the University of Toronto. We build Multiplexed Assays of Variant Effect (MAVE) and mutational scanning technologies to enable proactive measurement of the functional effects of all possible single-nucleotide variants in a target gene. These methods offer a general, scalable way to understand how variants impact molecular and cellular phenotypes, and to resolve variants of uncertain significance (VUS) that arise when human genomes are sequenced.

CMAP's primary objectives are to 1) develop novel, scaleable and phenotypically complex technologies to assess the functional impact of single nucleotide variants in human genes 2) drive wide adoption of these methodologies 3) train early career experimentalists, clinical geneticists and data scientists to obtain and use large-scale functional data.

The technologies developed by our Center can shed light on the functional consequences of genetic variants and ultimately on the landscape of human genetic variation. By providing inclusive access to training and tools for making, depositing, obtaining, and using variant effects maps we are building a community that can take on the challenge of understanding how all of the billions of possible variants of the human genome impact phenotype and disease. Here we describe CMAP's outreach and education program.



CMAP offers short-term internships, postdocs as well as opportunities for undergraduates. Above: CMAP intern Moez Dawood and Dr. Lea Starita discussing a Saturation Genome Editing (SGE) project. Postdoc Curran Oi presenting work on whole organism Deep Mutational Scanning (DMS) at the CMAP all hands meeting.

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Free educational content is available on our CMAP youtube page which includes presentations from the Mutational Scanning Symposium and Workshop as well as talks from the Variant Effect Seminar Series.

- 1. Department of Genome Sciences, University of Washington, Seattle, WA, USA
- 2. Brotman Baty Institute for Precision Medicine, Seattle, WA, USA
- 3. The Donnelly Centre and Departments of Molecular Genetics and Computer Science, University of Toronto, Toronto, Ontario, Canada 4. Lunenfeld-Tanenbaum Research Institute, Sinai Health, Toronto, Ontario, Canada
- 5. Howard Hughes Medical Institute, Seattle, WA, USA
- 6. Bioinformatics Division, WEHI, Parkville, Australia
- 7. Department of Medical Biology, University of Melbourne, Parkville, Australia

# Variant effect mapping outreach and education program at the **Center for the Multiplexed Assessment of Phenotype**

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## EDUCATION AND OUTREACH RESOURCES FOR VARIANT EFFECT MAPPING

	Amazing MAVE projects	
A V	TRACKED HERE	
I M F Y	MaveRegistry is a collaborative resour Multiplexed Assays of Variant Effect (I	rce for sharing pr MAVE).
R H K	Please cite: <u>Kuang et al. 2021</u>	
DE	Browse Projects	Nominate <sup>-</sup>
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We provide a suite of resources and tools for variant effect mapping. Resources include MaveRegistry, a platform to catalyze collaboration, reduce redundant efforts, allow stakeholders to nominate targets and enable tracking and sharing of progress on ongoing MAVE projects and MaveDB, a hub for the analysis and dissemination of MAVE datasets.

ТҮРЕ	Decription (and URL)
TRAINING (tailored to individuals inte	rested in mastering CM
Technology Dissemination Internships 1-3mo duration	2 interns per year
Application-Focused Apprenticeships (Postdoc level) 1-2yr duration	1-2 fellows per year
Training Fund	Support for predoctora risk, high-reward methors recipients in 2022
Summer Research Program REU (URM undergraduates)	The Department of Ge opportunities for unde year <u>https://www.gs.w</u> edu/academics/summ
EDUCATION	
Mutational Scanning Symposium and Workshop	CMAP co-hosts the an (with 25 speakers, 10 l from over 20 countries Brings together top int Annually <u>https://www.v</u>
Variant Effect Seminar Series	Monthly seminar serie and discuss their resea Held virtually, free to th series
Online resource_ Knowledgebase	Variant effect mapping org/resources
Educational Videos	Free educational conte presentations from Mu Variant Effect Seminar https://www.youtube.co
COMMUNITY, RESOURCES and TOOL	KITS
Atlas of Variant Effects (AVE) Alliance	Expands the communi maps <u>https://www.var</u>
MaveDB	A public repository for
MaveImpute	A web application and effect maps <u>http://imp com/joewuca/imputat</u>
MaveQuest	A web resource for pla https://mavequest.var
MaveRegistry	Collaborative resource //registry.varianteffect
MaveVis	A visualization tool for varianteffect.org https
Enrich2	Software tool for proce mutational scanning e
VARITY	Improved pathogenicit http://varity.varianteff
CMAP Website	Information, recent new edu, <u>https://www.cmap washington.edu/public</u>
CMAP Twitter Account	https://twitter.com/CE



#### AP-developed technologies)

ral and postdoctoral research trainees working on highnods development in the field of functional genomics, 2

enome Sciences, provides summer research ergraduate students. CMAP hosts ~ 5 students per <u>ashington.</u>

<u>ner/gssummer/participants.htm</u>

nnual Mutational Scanning Symposium and Workshop lightning talks, 86 posters and over 362 attending, all s for the most recent hybrid event in Toronto, Canada). ternational experts on mutational scanning. Held varianteffect.org/mss2022

es where career scientists from around the globe share earch related to interpreting human genetic variation. he public https://www.varianteffect.org/seminar-

g resource knowledgebase https://www.varianteffect.

ent on the CMAP YouTube channel. Includes lutational Scanning Symposium, Workshops and the r Series. (over 300 subscribers, and 21,373 views) com/channel/UCdS-1bGBkOvAMYXkFZoAklg

nity of scientists generating and using variant effect rianteffect.org/about

variant effect maps <a href="https://www.mavedb.org/">https://www.mavedb.org/</a>

service for imputing and visualizing missense variant pute.varianteffect.org, https://github.

ion

anning experimental tests of human variant effects rianteffect.org

e for sharing progress on Variant Effect Mapping https: <u>.org/</u>

r variant effect maps in MaveDB <u>http://vis.</u> ://github.com/VariantEffect/mavevis#readme cessing, analyzing, and visualizing data from deep experiments https://github.com/FowlerLab/Enrich2 ity prediction for rare human missense variants

fect.org/ ews, manuscripts <u>https://www.cmap.gs.washington.</u> ap.gs.washington.edu/news, https://www.cmap.gs. ications

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## **MUTATIONAL SCANNING SYMPOSIUM**

The Center's technologies have the potential to transform our understanding of the human genome. Our annual Mutational Scanning Symposium brings together top international experts on mutational scanning, who share their latest work and provide insights on the future of this science. The Symposium began four years ago as a local event in Seattle, and has grown each year, with generous support from funders and lots of work from organizers.



This year, the three day program included keynote lectures by Clare Turnbull and Doug Fowler, as well as 25 invited talks and 10 lightning talks selected from poster abstracts. Most speakers agreed to help us create a community resource by making their talks permanently available on our CMAP YouTube channel. In 2023, the Symposium will be in the UK at the Wellcome Sanger Institute. (Conference Illustrations by Alex Cagan).

## We invite everyone to help solve the VUS problem



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NIH National Human Genome Research Institute