



Global Genomic  
Medicine Collaborative

# 3<sup>rd</sup> International Cohorts Virtual Summit

May 4 – 5, 2020



International 100K Cohort Consortium

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## Summit Objectives

The International Cohorts Summit, conceived by the **Heads of International Research Organizations (HIROs)** chaired by Jeremy Farrar of the Wellcome Trust and Francis Collins of the National Institutes of Health, held an inaugural summit held in March 2018 in Durham, North Carolina, USA, organized by the Global Genomic Medicine Collaborative (G2MC, [www.g2mc.org](http://www.g2mc.org)), with the goal of enabling leaders of large-scale longitudinal cohorts worldwide to share best practices, discuss data sharing, explore standards, discuss common challenges, and explore the potential for a larg(er) collaborative sequencing strategy. From that Summit, the International Hundred K+ Cohorts Consortium (IHCC, <https://ihcc.g2mc.org/>) was formed.

The IHCC now convenes the third International Cohorts Summit to continue this discussion with the intent of developing the scientific agenda to drive the research and collaboration between cohorts. Originally planned as an in-person event in Santiago, Chile, the realities and impacts of the global pandemic that emerged mere weeks before the planned dates required pivoting our agenda to a more focused and concentrated session with relevancy to current issues and that provide the opportunity to leverage the potential of the IHCC.

As a virtual event, the primary objectives of this meeting are:

- To galvanize the IHCC around a visionary charter and path forward (defining the IHCC organization, mission, membership, partnership opportunities, industry engagement).
- To examine how IHCC can rapidly mobilize worldwide cohorts to address the COVID-19 pandemic.
- To introduce the IHCC to a cohorts atlas that can be used to stimulate/enable collaborations among cohorts.
- To engage the entirety of the IHCC membership in developing the key topics to chart a scientific agenda that can only be achieved by assembling cohorts and their data.

We thank you for joining us at this pivotal event, and for contributing to our shared goal of enhanced international collaboration to advance and accelerate the full scientific potential of cohorts from around the world and translate these findings to global health impact.

## Programme Committee

**Philip Awadalla, Ph.D.**, Ontario Institute for Cancer Research, *Canada*  
**Arash Etemadi, M.D., Ph.D.**, National Cancer Institute, NIH, *USA*  
**Catterina Ferreccio, M.D., Ph.D.**, Pontificia Universidad Católica de Chile, *Chile*  
**Mike Gaziano, M.D., Ph.D.**, Johns Hopkins University, *USA*  
**Kelly Gebo, M.D., Ph.D.**, Johns Hopkins Medicine and *All of Us* Research Program, NIH, *USA*  
**Geoffrey Ginsburg, M.D., Ph.D.**, Duke University and G2MC, *USA*  
**Peter Goodhand**, Global Alliance for Genomics & Health, *Canada*  
**Josep Maria Haro, M.D., Ph.D., M.P.H.**, University of Barcelona, *Spain*  
**Kobus Herbst, M.D.**, Africa Health Research Institute and SAPRIN, *South Africa*  
**Norihiro Kato, M.D., Ph.D.**, National Center for Global Health and Medicine, *Japan*  
**Rongling Li, M.D., Ph.D., M.P.H.**, National Human Genome Research Institute, *USA*  
**Paulo Lotufo, M.D., Ph.D.**, University of Sao Paulo, *Brazil*  
**Teri Manolio, M.D., Ph.D.**, National Human Genome Research Institute and G2MC, *USA*  
**Nicola Mulder, Ph.D.**, University of Cape Town, *South Africa*  
**Eric Plummer**, International HundredK+ Cohorts Consortium, *USA*  
**Teji Rakhra-Burris**, Duke University and G2MC, *USA*  
**Gad Rennert, M.D., Ph.D.**, Carmel Medical Center, *Israel*  
**Ricardo Verdugo, Ph.D.**, University of Chile, *Chile*

# International Cohorts Virtual Summit

Hosted by the Global Genomic Medicine Collaborative (G2MC)

**May 4-5, 2020**

## Meeting Objectives

- To galvanize the IHCC around a visionary charter and path forward (defining the IHCC organization, mission, membership, partnership opportunities, industry engagement)
- To examine how IHCC can rapidly mobilize worldwide cohorts to address the COVID-19 pandemic
- To introduce the IHCC to a cohorts atlas that can be used to stimulate/enable collaborations among cohorts
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## Monday May 4, 2020

Day 1/Part 1

### Summit Introduction and Background

**Chair: Geoffrey Ginsburg, M.D., Ph.D.**

11:00 - 11:10 UTC

#### ***Day 1 Welcome and Introductions***

**Geoffrey Ginsburg, M.D., Ph.D.**

Duke University, USA

11:10 - 11:30 UTC

#### ***Keynote***

**Francis S. Collins, M.D., Ph.D.**

Director, National Institutes of Health, USA

### Session 1 – IHCC Work Team Progress – Governance

**Chair: Geoffrey Ginsburg, M.D., Ph.D.**

11:30 - 11:35 UTC

#### ***Overview of Current Progress***

**Geoffrey Ginsburg, M.D., Ph.D.**

Duke University, USA

11:35 - 12:05 UTC

**Charter Work Team Progress Update**

**Mary De Silva, MSc, Ph.D.**

Wellcome Trust, UK

- *Charter Introduction*
- *Charter Discussion and Ratification*

12:05 - 12:50 UTC

**Policy and Bio-Data Sharing**

**Laura Lyman Rodriguez, Ph.D.**

USA

- *Policy Document Introduction*
- *Policy Discussion and Q&A*

12:50 - 13:05 UTC

**BREAK**

13:05 - 14:10 UTC

**Mobilizing IHCC Cohorts to Address COVID-19 Pandemic**

**Keri Althoff, Ph.D.**

Johns Hopkins Bloomberg School of Public Health, USA

**Kelly Gebo, M.D., M.P.H.**

All of Us Research Program, USA

- *Summary of Ongoing Efforts and Participation*
- *Potential Additional Collaborations*
  - **Liz Cirulli, Ph.D.** – Helix, USA
  - **Eileen Scully, M.D., Ph.D.** – Johns Hopkins Medicine, USA
  - **Karestan Koenen, Ph.D.** – Harvard T.H. Chan School of Public Health, USA
  - **Kári Stefánsson, M.D.** – deCODE, Iceland
- Q&A

14:10 - 14:15 UTC

**Summary and Next Steps**

**Geoffrey Ginsburg, M.D., Ph.D.**

Duke University, USA

Adjourn - Total running time: 3 hours 15 minutes  
45-minute BREAK between sessions

# Monday May 4, 2020

Day 1/Part 2

## Session 2 – IHCC Work Team Progress – Science and Technology

**Chair: Laura Lyman Rodriguez, Ph.D.**

**15:00 - 15:05 UTC**    **Session 2 - Welcome and Introductions**  
Laura Lyman Rodriguez, Ph.D.  
USA

**15:05 - 15:50 UTC**    **Data and Infrastructure**  
**Philip Awadalla, Ph.D.**  
Canadian Partnership for Tomorrow Project, Ontario Institute for Cancer Research, Canada  
**Thomas Keane, Ph.D.**  
European Bioinformatics Institute and Global Alliance for Genomics & Health, UK

- *Data and Infrastructure – Update*
- *Data Atlas Pilot Demo*
- *Data & Infrastructure – Next Steps*
- *Q&A*

**15:50 -16:05 UTC**    **BREAK**

**16:05 -16:50 UTC**    **Scientific Strategy**  
**Adam Butterworth, Ph.D.**  
University of Cambridge, UK  
**Hákon Hákonarson, M.D., Ph.D.**  
Children's Hospital of Philadelphia, USA  
**Gad Rennert, M.D., Ph.D.**  
Carmel Medical Center and Technion, Israel

- *Scientific Strategy – Update*
- *PRS Pilot Results*
- *Scientific Strategy – Next Steps*
- *Q&A*

**16:50 -17:00 UTC**    **Summary and Next Steps**  
Laura Lyman Rodriguez, Ph.D.  
USA

Adjourn - Total running time: 2 hours

# Tuesday May 5, 2020

Day 2/Part 1

## ICS3 Day 2 – Scientific Presentations and External Engagement

**Chair: Teri Manolio, M.D., Ph.D.**

- 11:00 - 11:05 UTC** *Day 2 Welcome and Introductions*  
**Teri Manolio, M.D., Ph.D.**  
National Human Genome Research Institute, NIH, USA
- 11:05 - 11:20 UTC** *Keynote*  
**Soumya Swaminathan, M.D., Ph.D.**  
World Health Organization, Switzerland
- 11:20 - 11:50 UTC** *Plans for IHCC COVID-19 Research*  
**Keri Althoff, Ph.D.**  
Johns Hopkins Bloomberg School of Public Health, USA  
**Kelly Gebo, M.D., M.P.H.**  
All of Us Research Program, USA

## Session 3 – Scientific Presentations

**Chair: Teri Manolio, M.D., Ph.D.**

- 11:50 - 11:55 UTC** *Introduction of Data Standards and Infrastructure*  
**Moderators: Philip Awadalla, Ph.D. and Thomas Keane, Ph.D.**
- 11:55 - 12:30 UTC** *Data and Infrastructure Presentations*
- **David Glazer** – Verily, USA
  - **Alison Motsinger-Reif, Ph.D.** – National Institute of Environmental Health Sciences, USA
  - **Goncalo Abecasis, D.Phil.** – Regeneron, USA
  - **Q&A**
- 12:30 - 12:45 UTC** **BREAK**
- 12:45 - 12:50 UTC** *Introduction of Scientific Strategy and Cohorts Enhancement*  
**Moderators: Adam Butterworth, Ph.D., Hákon Hákonarson, M.D., Ph.D. and Gad Rennert, M.D., Ph.D.**
- 12:50 - 13:25 UTC** *Scientific Strategy and Cohorts Enhancement Presentations*
- **Alicia Martin, Ph.D.** – Broad Institute, USA
  - **George Vradenburg / Elias Zerhouni** – CEOi/Johns Hopkins University, USA
  - **Erin M. Ramos, Ph.D., M.P.H.** – National Human Genome Research Institute, USA
  - **Q&A**



13:25 - 13:30 UTC **Introduction of Policy and Bio-Data Sharing Presentations**  
**Moderator: Laura Lyman Rodriguez, Ph.D.**

13:30 - 14:05 UTC **Policy and Bio-Data Sharing Presentations**

- **Benjamin Neale, Ph.D.** – Broad Institute, USA
- **Fruzsina Molnár-Gábor, Ph.D.** – Heidelberg Academy of Sciences and Humanities, Germany
- **Lynsey Chediak** – World Economic Forum
- **Q&A**

14:05 - 14:15 UTC **Summary and Next Steps**  
**Teri Manolio, M.D., Ph.D.**  
National Human Genome Research Institute, NIH, USA

Adjourn - Total running time: 3 hours and 15 minutes  
45-minute BREAK between sessions

## Tuesday May 5, 2020

Day 2/Part 2

Session 4 – External Engagement (Other Consortia and Industry Partners)

**Chair: Peter Goodhand**

15:00 - 15:05 UTC **Overview of Partner and Industry Presentations**  
**Peter Goodhand**  
Global Alliance for Genomics & Health, Canada

15:05 - 15:40 UTC **Partner Presentations**

- **GA4GH – Ewan Birney, Ph.D.** – The Global Alliance for Genomics Health and European Bioinformatics, UK
- **GBC – Eric Green, M.D., Ph.D.** – Global BioData Coalition and Genomic National Human Genome Research Institute, USA
- **ICDA – Rachel Liao, Ph.D.** – International Common Disease Alliance and Broad Institute, USA
- **Q&A**

15:40- 15:45 UTC **Keynote Introduction**  
**Peter Goodhand**  
Global Alliance for Genomics & Health, Canada

15:45 - 15:55 UTC **Keynote**  
**Jeremy Farrar**  
Wellcome Trust, UK

15:55 - 16:15 UTC **BREAK**

**16:15 - 17:00 UTC *Industry Partners – Panel Discussion***

- **Alan Shuldiner, M.D.** – Regeneron, USA
- **Meg Ehm, Ph.D.** – GlaxoSmithKline, USA
- **Phil Febbo, M.D.** – Illumina, USA

**17:00 - 17:30 UTC *Summary, Consensus and Next Steps***

**Geoffrey Ginsburg, M.D., Ph.D., Peter Goodhand and Teri Manolio, M.D., Ph.D.**

Adjourn - Total running time: 2 hours 30 minutes

# Summit Presenters & Session Chairs

## Keynote Speakers



**Francis S. Collins**

Director, National Institutes of Health  
USA

**Francis S. Collins, M.D., Ph.D.** was appointed the 16<sup>th</sup> Director of the National Institutes of Health (NIH) by President Barack Obama and confirmed by the Senate. He was sworn in on August 17, 2009. On June 6, 2017, President Donald Trump announced his selection of Dr. Collins to continue to serve as the NIH Director. In this role, Dr. Collins oversees the work of the largest supporter of biomedical research in the world, spanning the spectrum from basic to clinical research.

Dr. Collins is a physician-geneticist noted for his landmark discoveries of disease genes and his leadership of the international Human Genome Project, which culminated in April 2003 with the completion of a finished sequence of the human DNA instruction book. He served as director of the National Human Genome Research Institute at NIH from 1993-2008.

Before coming to NIH, Dr. Collins was a Howard Hughes Medical Institute investigator at the University of Michigan. He is an elected member of the National Academy of Medicine and the National Academy of Sciences, was awarded the Presidential Medal of Freedom in November 2007, and received the National Medal of Science in 2009.



**Jeremy Farrar**

Director, Wellcome Trust  
UK

**Jeremy Farrar**, Director of Wellcome Trust, was Director of the Oxford University Clinical Research Unit in Viet Nam for 18 years before joining Wellcome in October 2013. His research interests were infectious diseases and global health, with a focus on emerging infections. He has published almost 600 articles, mentored many dozens of students and fellows, and served as Chair on several advisory boards for governments and global organisations.

He was named 12<sup>th</sup> in the Fortune list of 50 World's Greatest Leaders in 2015 and was awarded the Memorial Medal and Ho Chi Minh City Medal from the Government of Viet Nam. In 2018 he was awarded the President Jimmy and Rosalynn Carter Humanitarian of the Year Award. He is a Fellow of the Academy of Medical Sciences UK, the National Academies USA, the European Molecular Biology Organisation and a Fellow of The Royal Society. Jeremy was knighted in the Queen's 2019 New Year Honours for services to global health.



**Soumya Swaminathan**

Chief Scientist, World Health  
Organization  
Switzerland

**Dr. Soumya Swaminathan, M.D., Ph.D.** was most recently WHO's Deputy Director-General for Programmes. A paediatrician from India and a globally recognized researcher on tuberculosis and HIV, she brings with her 30 years of experience in clinical care and research and has worked throughout her career to translate research into impactful programmes. Dr Swaminathan was Secretary to the Government of India for Health Research and Director General of the Indian Council of Medical Research from 2015 to 2017. In that position, she focused on bringing science and evidence into health policy making, building research capacity in Indian medical schools and forging south-south partnerships in health sciences. From 2009 to 2011, she also served as Coordinator of the UNICEF/UNDP/World Bank/WHO Special Programme for Research and Training in Tropical Diseases in Geneva. She received her academic training in India, the United Kingdom of Great Britain and Northern Ireland, and the United States of America, and has published more than 350 peer-reviewed publications and book chapters. She is an elected Foreign Fellow of the US National Academy of Medicine and a Fellow of all three science academies in India. She has previously been on several WHO and global advisory bodies and committees, including the WHO Expert Panel to Review Global Strategy and Plan of Action on Public Health, Innovation and Intellectual Property, the Strategic and Technical Advisory Group of the Global TB Department at WHO, and most recently was Co-Chair of the Lancet Commission on TB.

## IHCC Co-Chairs



**Geoffrey Ginsburg**

Director, Center for Applied Genomics  
& Precision Medicine  
Duke University Medical Center  
USA

**Geoffrey S. Ginsburg, M.D., Ph.D.** is the founding director for the Center for Applied Genomics & Precision Medicine at the Duke University Medical Center and for MEDx, a partnership between the Schools of Medicine and Engineering to spark and translate innovation. He is a Professor of Medicine, Pathology, and BioMedical Engineering. His research addresses the challenges for translating genomic and digital health information into medical practice and the integration of precision medicine into healthcare. In 2017 he received Duke's Translational Research Mentorship Award and is a finalist in the NIH/BARDA Antimicrobial Resistance Prize. He is a member of the Advisory Council to the Director of NIH and is co-chair of the National Academies Roundtable on Genomic and Precision Health and is the founder and president of the Global Genomic Medicine Collaborative, a not for profit organization aimed creating international partnerships to advance the implementation of precision medicine. He recently served as a member of the Board of External Experts for the NHLBI, the advisory council for the National Center for Accelerating Translational Science, chair of the review for Genome Canada's Large Scale Applied Research Competition in Genomics and Precision Medicine, and the World Economic Forum's Global Agenda Council on the Future of the Health Sector. He is a founder of Predigen, Inc and MeTree&You, Inc. He was previously Vice President of Molecular Medicine at Millennium Pharmaceuticals, Inc and a faculty member at Harvard Medical School.



**Peter Goodhand**

Chief Executive Officer, Global Alliance  
for Genomics and Health (GA4GH)  
Canada

**Peter Goodhand** played a key role in the creation of the Global Alliance for Genomics and Health (GA4GH) to accelerate progress in genomic research and medicine and in June 2013, he was appointed Executive Director of the GA4GH for its critical development phase. In 2018, Mr. Goodhand's title was updated to Chief Executive Officer to reflect the changing responsibilities associated with leading a growing organization with an ambitious strategic roadmap. Goodhand is currently Co-Chair of the International 100K+ Cohorts Consortium (IHCC), Co-Chair of the Medical and Scientific Advisory Board of Global Genes, a member of the Global Genomic Medicine Collaboration (G2MC) Steering Committee, and a member of the Occupational Cancer Research Centre Steering Committee.



**Teri Manolio**

Director, Division of Genomic Medicine  
National Human Genome Research  
Institute (NHGRI)  
USA

**Dr. Manolio, M.D., Ph.D.**, directs NHGRI's Division of Genomic Medicine, where she leads programs to develop and implement genomic applications in clinical care. She led the epidemiology programs of the National Heart, Lung, and Blood Institute until 2005 where she was heavily involved in large-scale cohort studies such as the Cardiovascular Health Study, the Framingham Heart Study, and the Multi-Ethnic Study of Atherosclerosis. She moved to NHGRI to lead efforts in applying genomic technologies to population research and clinical care, including the Electronic Medical Records and Genetics (eMERGE) Network, the NHGRI Genome-Wide Association Catalog, and the Clinical Genome (ClinGen) Resource. She continues to practice and teach internal medicine at the Walter Reed National Military Medical Center and is a professor of medicine at the Uniformed Services University of the Health Sciences. She is the author of over 290 research publications and has research interests in incorporating genomic findings into clinical care.

## **Presenters & Session Chairs**



**Goncalo Abecasis**

Vice President for Analytical Genetics  
and Data Sciences, Regeneron  
Pharmaceuticals  
USA

**Goncalo Abecasis, D.Phil.**, is Vice President for Analytical Genetics and Data Sciences at Regeneron Pharmaceuticals. He uses computational and statistical methods to study human genetic analysis at scale, looking for insights into human biology and disease.



**Keri Althoff**

Associate Professor  
Johns Hopkins Bloomberg School of  
Public Health  
USA

**Dr. Keri N Althoff, Ph.D., M.P.H.**, is an Associate Professor of Epidemiology in the Department of Epidemiology, Johns Hopkins Bloomberg School of Public Health. She has a joint appointment in the Department of Oncology at the Johns Hopkins University School of Medicine. She is one of two Principal Investigators for the North American AIDS Cohort Collaboration on Research and Design (NA-ACCORD) of the International Epidemiologic Databases to Evaluate AIDS (IeDEA) project. The NA-ACCORD is a collaboration of >20 longitudinal HIV cohorts, which rely on electronic medical record data and data collected via research study visits for analyses from most of the cohorts. She is an expert in epidemiologic methods that address challenges in the use of cohort collaboration study design. Dr. Althoff also teaches the third term of the three-term course series on epidemiologic methods in research and is the Johns Hopkins University Provost's Fellow for Research Communication.



**Philip Awadalla**

Director of Computational Biology,  
Ontario Institute for Cancer Research  
Canada

**Dr. Philip Awadalla, Ph.D.**, is National Scientific Director of CanPath (Canadian Partnership for Tomorrow's Health), Director of Computational Biology and the Executive Scientific Director of Ontario Health Study at the Ontario Institute for Cancer Research, and Professor of Population and Medical Genomics at the University of Toronto. He is Director of the Genome Canada, Canadian Data Integration Centre and a member of the International Hundred Thousand Consortium Steering Committee. He obtained his doctorate in population and statistical genetics from the University of Edinburgh and awarded NSERC, Killam, and Wellcome Trust Fellowships to pursue his postdoctoral work before taking faculty positions at North Carolina and the University of Montreal. He was the Scientific Director of CARTaGENE, and part of the analysis groups of the 1000 Genomes Program and PCAWG. Major projects include genomics of aging, hematological diseases and cancers and projects include estimating mutation and recombination rates; model-based approaches to identify genetic; and environmental control points for infectious diseases in Africa.



**Ewan Birney**

Director, The European Bioinformatics  
Institute (EMBL-EBI)  
UK

**Ewan Birney, Ph.D.**, is Director of EMBL-EBI, and runs a small research group. Ewan completed his PhD at the Wellcome Sanger Institute with Richard Durbin. In 2000, he became Head of Nucleotide data at EMBL-EBI and in 2012 he took on the role of Associate Director at the institute. He became Director of EMBL-EBI in 2015. Ewan led the analysis of the Human Genome gene set, mouse and chicken genomes and the ENCODE project, focusing on non-coding elements of the human genome. Ewan's main areas of research include functional genomics, DNA algorithms, statistical methods to analyse genomic information and use of images for chromatin structure. Ewan is a non-executive Director of Genomics England, and a consultant and advisor to a number of companies. Ewan was elected an EMBO member in 2012, a Fellow of the Royal Society in 2014 and a Fellow of the Academy of Medical Sciences in 2015.



**Adam Butterworth**

Reader in Molecular Epidemiology  
University of Cambridge  
UK

**Adam Butterworth, Ph.D.**, is a Reader in Molecular Epidemiology at the Department of Public Health & Primary Care at the University of Cambridge. He leads the Health Data Research UK Multi-omics Consortium and co-leads the Scientific Strategy and Enhancements team of the IHCC. His research group focuses on genetic discovery for molecular traits and complex diseases, as well as therapeutic target prioritisation using human genetics.





**Lynsey Chediak**

Lead, World Economic Forum's  
Precision Medicine Team  
USA

**Lynsey Chediak** is a Lead on the World Economic Forum's Precision Medicine Team currently piloting cross-border genomic data access for rare disease diagnosis and treatment. She is the former Founder and CEO of a patient advocacy non-profit where she implemented new models for increased patient engagement and emotional support across hospital systems in the US. She wrote public health policy in the California Governor's Office and in the UK's Parliament, House of Commons extending minimum length of care for rare disease patients. Lynsey was one of 100 entrepreneurs in the USA selected for the Venture for America Fellowship (2014-2016). She holds a BA from Claremont McKenna College; MSc from the London School of Economics and Political Science. Lynsey is a spokesperson for Shriners Hospitals for Children.



**Liz Cirulli**

Principal Scientist, Helix  
USA

**Liz Cirulli, Ph.D.** is a human geneticist who focuses on large-scale analysis of rare variants from high-coverage NGS data and is interested in unusual phenotypes such as synesthesia and differences in face recognition abilities.



**Mary De Silva**

Head of Population Health  
Wellcome Trust  
UK

**Mary De Silva, MSc, Ph.D.**, leads the Population Health team which directs Wellcome's funding of population health research in the UK and in low- and middle-income countries.

Current strategic areas of interest include Wellcome's Longitudinal Population Studies Strategy, catalysing advances in nutrition, health of the public and multi-morbidity research, and strengthening the pathways through which research evidence improves health. She also co-developed and sits on the Programme Board for the Mental Health Priority Area at Wellcome, a new research investment of £200 million.

Before joining Wellcome, Mary was Deputy Director of the Centre for Global Mental Health at the London School of Hygiene and Tropical Medicine. Her research included the design and evaluation of complex interventions to improve mental health in low- and middle-income countries, implementation research to ensure that these interventions are scalable and sustainable, and policy influence work to encourage the translation of evidence into policy and practice. I also co-founded and led the Mental Health Innovation Network (<http://www.mhinnovation.net/>).



**Meg Ehm**

Senior Director, Human Genetics  
GlaxoSmithKline  
USA

**Meg Ehm, Ph.D.**, is a member of Human Genetics group at GSK where she develops and manages external alliances that bring together GSK with academic and industry groups to build innovative capabilities capitalizing on genetic data that will drive the identification of high quality drug targets. Her recent work has focused on use of electronic health record data and genetic information. She has been involved in a variety of collaborations with academic groups such as Kaiser Department of Research and numerous US and European universities as well as for profit companies including 23andMe and DaVita. Prior to this role, she led the statistical genetics group at GSK through a series of progressively more challenging roles. She received her BS degree from Vanderbilt University in mathematics and computer science and MA and PhD from Rice University in statistics. She completed a brief post-doctoral post at North Carolina State University in 2001.



**Phil Febbo**

Senior VP and Chief Medical Officer,  
Illumina  
USA

**Dr. Phil Febbo, M.D.**, has been a director of the company since 2019. Since 2018, Dr. Febbo has served as chief medical officer at Illumina and is responsible for developing and executing the company’s medical strategy to drive genomic testing into healthcare practice in order to improve health outcomes. Prior to joining Illumina, he served as chief medical officer of Genomic Health for five years where he drove the company’s medical strategy, was accountable for the development of evidence supporting GHI’s proprietary tests, engaged with the payer community to drive reimbursement, and served as a corporate officer between 2016 and 2018. Prior to Genomic Health, Dr. Febbo was a Professor of Medicine and Urology at the University of California, San Francisco (UCSF), where his laboratory focused on using genomics to understand the biology and clinical behavior of prostate cancer, and his clinical practice focused on genitourinary oncology. He currently serves on the board of the American College of Medical Genetics and Genomics Foundation. Dr. Febbo holds a Bachelor of Arts degree in Biology from Dartmouth College and an M.D. from UCSF. He completed his internal medicine residency at the Brigham and Women’s Hospital and his fellowship in oncology at the Dana-Farber Cancer Institute.



**Kelly Gebo**

Chief Medical and Scientific Officer  
National Institutes of Health *All of Us*  
Research Program  
USA

**Kelly Gebo, M.D., M.P.H.**, is the Chief Medical and Scientific Officer of the *All of Us* Research Program. In this role, she works with diverse stakeholders to lead the Program’s scientific agenda and guide protocol revisions and data collection processes. She also provides clinical oversight of the Program in collaboration with the institutional review board (IRB) and the *All of Us* team.

Dr. Gebo has clinical, research, and educational experience in the health care and higher education sectors. She is a professor of medicine at Johns Hopkins University and an expert in HIV health services research and clinical outcomes of persons with HIV. Previously, she served as the co-principal investigator of the HIV Research Network, an 18-year clinical cohort study of high-volume HIV sites caring for over 20,000 persons with HIV across the country. She has also served as an American Council on Education Fellow at the University of Pennsylvania and as the Vice Provost for Education at Johns Hopkins.

Dr. Gebo holds a doctorate in medicine from the Johns Hopkins University School of Medicine and a master’s in public health from the Johns Hopkins Bloomberg School of Public Health.



**David Glazer**

Engineering Director, Verily Life Sciences  
USA

**David Glazer** is an engineering director at Verily Life Sciences, where he helps life science organizations use cloud computing to accelerate and scale their work with big data. He is a PI for the Data and Research Center, and a member of the Steering Committee, of the NIH All of Us Research Program, and he serves on the NIH Advisory Committee to the Director. He is co-chair of the Cloud Workstream, and a member of the Steering Committee, of the Global Alliance for Genomics and Health (GA4GH). He previously worked at Google, where he founded the Google Genomics team, and led a variety of platform, product, and infrastructure teams. Prior to joining Google in 2006, he successfully started two companies: Eloquent in 1995 (IPO 2000), which used rich media to power business communications, and Verity in 1988 (IPO 1995), which did full-text search. David grew up in Massachusetts, where he earned a BS in physics from MIT.



**Eric Green**

Director, National Human Genome Research Institute  
USA

**Dr. Eric Green, M.D., Ph.D.**, is the Director of the National Human Genome Research Institute (NHGRI) at the U.S. National Institutes of Health (NIH). He is the third NHGRI director, having been appointed by NIH director Dr. Francis Collins in 2009.

Dr. Green has been at the Institute for more than 25 years, during which he has had multiple key leadership roles. He served as the Institute's scientific director for 7 years, chief of the NHGRI Genome Technology Branch for 13 years, and founding director of the NIH Intramural Sequencing Center for 12 years.

For just over two decades, Dr. Green directed an independent research program that included integral start-to-finish roles in the Human Genome Project and groundbreaking work on mapping, sequencing, and characterizing mammalian genomes.

Dr. Green earned his M.D. and Ph.D. degrees in 1987 from Washington University in St. Louis; coincidentally, the word "genomics" was coined in that same year. During his career, Dr. Green has authored and co-authored over 370 scientific publications.



**Hákon Hákonarson**

Director of the Center for Applied Genomics  
Endowed Chair in Genomics Research  
Professor of Pediatrics at the University of Pennsylvania  
Perelman School of Medicine  
USA

**Hákon Hákonarson, M.D., Ph.D.**, is Director of the Center for Applied Genomics at the Children’s Hospital of Philadelphia (CHOP), and Endowed Chair in Genomics Research and Professor of Pediatrics at The University of Pennsylvania, Perelman School of Medicine. He leads a major commitment from CHOP to genomically characterize 100,000 children. He is a Principal Investigator within the Kids First program and the TopMed genomics program funded by the NIH, and has been the principal investigator on multiple other NIH-sponsored grants. Dr. Hakonarson has previously held several senior posts within the biopharmaceutical industry, directing a number of genomics and pharmacogenomics projects as vice president of Clinical Sciences and Development at deCODE genetics, Inc. He has published over 800 scientific papers, including numerous high-impact papers on genomic discoveries and their translations, and has led several clinical trials based on these discoveries.



**Thomas Keane**

Team Leader  
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**Thomas Keane, M.Sc., Ph.D.**, is the team leader for the European Genome Phenome Archive (EGA) and the European Variation Archive (EVA) at EMBL-EBI. He is responsible for strategic planning for EGA and EVA, is a member of the Global Alliance for Genomics and Health (GA4GH) steering committee, and co-leads the Large-scale Genomics workstream. Before joining EMBL-EBI as Team Leader in 2016, Thomas led the Sequence Variation Infrastructure group in the Computational Genomics programme at the Wellcome Trust Sanger Institute. His research interests are in using genomic technologies to understand biological processes, with a particular focus on rodent models for human disease. He is the scientific lead for the Mouse Genomes Project, a collaboration with the Wellcome Sanger Institute. He also holds an honorary professorship at the University of Nottingham.



**Gun Peggy Knudsen**

Executive Director, domain of Health  
data and Digitalisation, Norwegian  
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**Gun Peggy Knudsen, Ph.D.**, is Executive Director of the domain Health data and Digitalisation at the Norwegian Institute of Public Health (NIPH). The domain is responsible for running and modernising infrastructure for research and knowledge, and has expertise in the research and management of health registries, population based health studies, biobanks and IT/e-health/digitalisation. The domain conducts registry-based research and health analysis, as well as genetics, bioinformatics and other research related to biobank material. Knudsen has a PhD in medical genetics, has extensive experience in administrating epidemiological and genetics research projects, and have in depth knowledge of the cohort data, the biological material, previous and ongoing genetics projects based on material at NIPH and use of data from health registries in combination with other health data. She is also involved in national e-health work, representing NIPH in various committees and boards.



### **Karestan Koenen**

Professor, Harvard T.H. Chan School  
of Public Health  
USA

**Karestan Chase Koenen, Ph.D.**, is a professor of psychiatric epidemiology at the Harvard T.H. Chan School of Public Health (HSPH) and leads the Global Neuropsychiatric Genomics Initiative of the Stanley Center for Psychiatric Research at Broad Institute. The broad goal of this initiative is to advance the genetic analysis of serious mental illness while contributing to global mental health equity by expanding the infrastructure and research findings from large-scale psychiatric genetic epidemiology to Africa and Mexico.

At HSPH, Koenen does research and teaches about trauma and posttraumatic stress disorder (PTSD). Her work on PTSD focuses on three areas. First, she studies why, when exposed to a similar traumatic event, some persons develop PTSD while others are resilient. She is particularly interested in how genes shape risk for PTSD. Much of this work is done through the PTSD working group of the Psychiatric Genomics Consortium that she co-leads with Kerry Ressler and Israel Liberzon. Second, she investigates how trauma and PTSD influence weight gain and alter long-term physical health. Third, she documents global burden of trauma and PTSD through her work with the World Mental Health Surveys. Koenen also co-leads the training program in psychiatric epidemiology and biostatistics.

Koenen's work uses research findings to advocate for evidence-based prevention of PTSD and response to trauma survivors, particularly victims of sexual violence. In May 2011, Dr. Koenen testified before the House Foreign Affairs Full Committee hearing "Peace Corps at 50," about the epidemic of sexual violence and victim blaming culture of the Peace Corps. She has written for the Boston Globe, the Washington Post, the Huffington Post, and the Women's Media Center's Women Under Siege Project, a journalism project founded by Gloria Steinem that investigates how rape and other forms of sexualized violence are used as tools in conflict.

Koenen received her B.A. from Wellesley College, her M.A. in developmental psychology from Columbia University, and her Ph.D. in clinical psychology from Boston University.



**Rachel G. Liao, Ph.D.**, is Executive Director of the International Common Disease Alliance (ICDA) and Scientific Advisor to the Director at the Broad Institute in Cambridge, USA. She oversees ICDA's administrative office and advises other priority projects, including the Variant to Function (V2F) Initiative and the COVID-19 Host Genetics Initiative. She holds a PhD in cancer genomics from Harvard University.

**Rachel Liao**

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**Alicia Martin, Ph.D.**, is an Instructor in Investigation at the Analytic & Translational Genetics Unit at Massachusetts General Hospital and an Associate Member of the Broad Institute of MIT and Harvard. As a population and statistical geneticist, her research examines the role of human history in shaping global genetic and phenotypic diversity. Given vast Eurocentric study biases, she examines the transferability of knowledge gained from large-scale genetic studies across globally diverse populations. She is particularly interested in ensuring that the translation of genetic technologies via polygenic scores does not exacerbate health disparities induced by these study biases. She has published on the effective uses, limitations, and promise of polygenic scores in clinical settings and more broadly. Towards this end, she is also developing statistical methods and resources for multi-ethnic studies and underrepresented populations. She received her Ph.D. in Genetics and M.S. in Biomedical Informatics from Stanford University.

**Alicia Martin**

Instructor, Massachusetts General  
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**Fruzsina Molnár-Gábor**

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**Fruzsina Molnár-Gábor, Ph.D. (Dr. iur.),** is research group leader at the Heidelberg Academy of Sciences and Humanities and lecturer at the Faculty of Law, University of Heidelberg, Germany. Her research focuses on the regulation of biomedicine and biotechnology, including the fields of data law, medical law, international law, and the law of the European Union. She is member of the Ethics and Governance Committee of the International Cancer Genome Consortium – Accelerating Research in Genomic Oncology, the Ethics Working Group of the Human Cell Atlas, the GDPR Forum of the Global Alliance for Genomics and Health and the Junge Akademie of the Berlin-Brandenburg Academy of Sciences and Humanities and the Leopoldina. She has received the Manfred-Fuchs Prize, the promotion award of the VG Wort for her PhD, the Young Scholars Award of the Research Network on EU Administrative Law and the Heinz Maier-Leibnitz Prize 2020 for her interdisciplinary research.



**Alison Motsinger-Reif**

Branch Chief & Senior Investigator  
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**Dr. Alison Motsinger-Reif, Ph.D.,** is Branch Chief and Senior Investigator in the Biostatistics and Computational Biology Branch at the National Institute of Environmental Health Sciences. She received her M.S. in Applied Statistics and PhD in Human Genetics from Vanderbilt University. The primary goal of her research is the development and application of computational methods to detect genetic risk factors of complex traits in human populations. As health sciences increasingly accepts a complex model of phenotypic development involving many genetic and environment factors, her methods development work is focused on strategies that embrace this complexity. Her applied work has focused on understanding complex human phenotypes, with a focus on drug response outcomes and common diseases. She has published over 180 peer-reviewed publications as a result of this work, in a broad range of journals that reflect the interdisciplinary nature of her work.



## **Benjamin Neale**

Director of Genetics, Broad Institute  
USA

**Benjamin Neale, Ph.D.**, is Director of genetics in the Stanley Center for Psychiatric Research at the Broad Institute of MIT and Harvard, where he is an institute member. He is an associate professor in the Analytic and Translational Genetics Unit (ATGU) at Massachusetts General Hospital (MGH), where he directs the Genomics of Public Health Initiative. He is also an associate professor in medicine at Harvard Medical School (HMS). Neale is strongly committed to gaining insights into the genetics of common, complex human diseases. Neale and Mark Daly, both of whom are associated with the Broad Institute and MGH, lead the ADHD Initiative, a collaborative effort that focuses on genomic studies of attention deficit hyperactivity disorder (ADHD).

Neale's research and training have focused heavily on statistical methodology. He has analyzed genetic data from large-scale studies of patients with ADHD, autism, age-related macular degeneration, type 2 diabetes, and metabolic disorders. Neale also analyzed data from the first ADHD genome-wide association study (GWAS) meta-analysis, which combined the results of four studies to boost statistical power. Neale contributed to the development of software tools such as PLINK, one of the most frequently used packages for GWAS analysis. In addition to his roles at both the Broad Institute and MGH, Neale is the head of the ADHD psychiatric genetics GWAS analysis committee and an active member of the broader Psychiatric GWAS Consortium analysis committee, which is charged with analyzing all psychiatric data from these large-scale genome-wide association studies. Neale also led the design of the exome chip, a genotyping array that captures rare coding variation in a cost-effective manner. To date, over 1.5 million exome chips have been sold.

Neale studied at the University of Chicago and Virginia Commonwealth University, earning a B.Sc. in genetics. He went on to earn his Ph.D. in human genetics from King's College in London, UK. Neale completed his postdoctoral training in Daly's laboratory at Massachusetts General Hospital. In addition to many local research collaborations, he also serves as advisor and analyst to international genetic research consortia on psychiatric diseases.



**Erin M. Ramos**

Program Director, Division of Genomic  
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**Dr. Erin Ramos, Ph.D., M.P.H.**, is an epidemiologist and program director in the Division of Genomic Medicine. She received her M.P.H. and Ph.D. in the multidisciplinary field of public health genetics from the University of Washington where her research focused on the genetic epidemiology of Alzheimer's disease and the ethical, legal, and social implications that surround genomics research. She joined NHGRI in 2006 and manages a portfolio of research that includes statistical methods for relating sequence variation to disease and curation and prioritization of genetic variants for clinical use.

Dr. Ramos is the Project Scientist for Clinical Genome Resource (ClinGen), a consortium of more than 500 clinicians and scientists who are building a central resource that classifies the clinical relevance of genes and variants for use in precision medicine and research. She also directs the PhenX program that has produced an online catalog of standard data collection protocols for conducting biomedical research.

Dr. Ramos contributes to genomic data sharing activities across the NIH. She is a member of the trans-NIH Genomic Data Sharing Policy Management Task Force and chaired the Data Access Committee (DAC) for the Genetic Association Information Network (GAIN) to provide access to some of the first genome-wide association studies in dbGaP. Her research interests include incorporating genomic findings into clinical care, genome-wide association studies, gene-environment interactions in complex disease, and ethical, legal and social Implications (ELSI) of genomic research.



**Gad Rennert**

Director, Clalit National Israeli Cancer  
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**Gad Rennert, M.D., Ph.D.**, is the Chairman of the Department of Community Medicine and Epidemiology at Carmel Medical Center in Haifa and professor at the Technion-Israel Institute of Technology and is in charge of the teaching program in public health/epidemiology in its Faculty of Medicine. He is the Director of the Clalit National Cancer Control Center and oversees the genomic medicine developments in Clalit. He received his medical degree from Ben-Gurion University in Israel and a doctoral degree in Epidemiology/Public-Health from the University of North Carolina, Chapel Hill. He authored more than 300 peer-reviewed papers. His research interests are in molecular/genetic cancer epidemiology, cancer prevention and cancer screening and in genomic personalized (precision) medicine. A particular focus of his research involving more than 50,000 people is the interaction between behavioral and environmental risk factors, and germline or somatic genetic variants in the causation of breast, colorectal, gynecological, lung, pancreas and other cancers.



**Laura Lyman Rodriguez**

USA

**Laura Lyman Rodriguez, Ph.D.**, most recently served as the Director of Genome Policy and National Engagement at Geisinger Research. Dr. Rodriguez' role focused on providing policy and ethics leadership for developing a clinical genome sequencing pipeline within standard of care workflows and developing a stakeholder engagement portfolio to extend partnerships with patients, providers, and other health system entities. Previously, Dr. Rodriguez directed the Division of Policy, Communications, and Education at the National Human Genome Research Institute (NHGRI). For most of her NHGRI tenure, her responsibilities included policy development related to NHGRI initiatives, communication and outreach activities to engage the public in genomic science, and programs to prepare healthcare providers for the integration of genomic medicine into clinical care. In addition, Dr. Rodriguez was a primary leader for the development and implementation of NIH genomic data sharing policies. Dr. Rodriguez earned a doctorate in cell biology from Baylor College of Medicine.



**Eileen Scully**

Assistant Professor  
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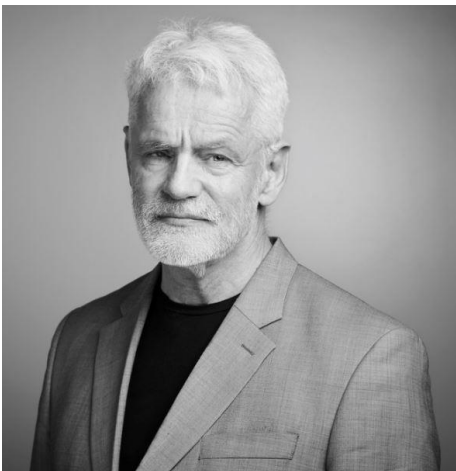
**Dr. Scully, M.D., Ph.D.**, is a clinician trained in infectious diseases and specializing in the care of people living with HIV and an immunologist. Her primary research interests center around the innate immune response to viral infections with a specific focus on sex differences in immune responses. She is an Assistant Professor of Medicine at Johns Hopkins University in the School of Medicine.



**Alan Shuldiner**

Vice President, Regeneron Genetics  
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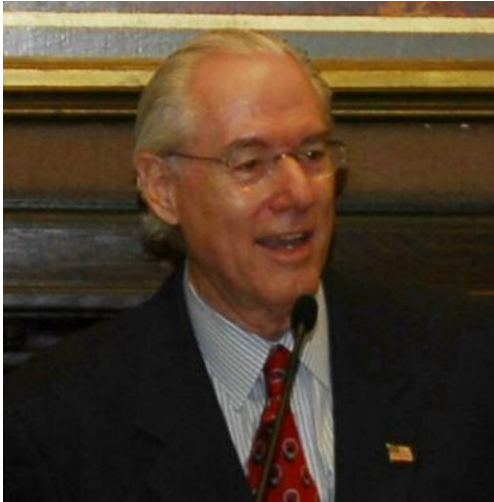
**Dr. Shuldiner, M.D.**, is a physician-scientist with extensive research, clinical, education, and leadership experience both in academia (25 years at the University of Maryland School of Medicine and Johns Hopkins University), and industry (4 years at the Regeneron Genetics Center, Regeneron Pharmaceuticals, Inc.). Leading a large multidisciplinary team of basic scientist and clinical researchers, he has made several seminal discoveries in genetics and genomic medicine that has impacted human health and disease.



**Kári Stefánsson**

CEO, deCODE  
Iceland

**Kári Stefánsson, M.D.**, Dr. Med. founded deCODE in August 1996. Dr. Stefánsson was previously a professor of Neurology, Neuropathology and Neuroscience at Harvard University and Director of Neuropathology at Beth Israel Hospital in Boston, Massachusetts. From 1983 to 1993, he held faculty positions in Neurology, Neuropathology and Neurosciences at the University of Chicago. Dr. Stefánsson received his M.D. and Dr. Med. from the University of Iceland and is board-certified in neurology and neuropathology in the United States. Dr. Stefansson is recognized as a leading figure in human genetics. He has shaped deCODE's scientific approach and been actively engaged in leading its gene discovery work, serving as senior author on most of the company's publications in major scientific journals.



**George Vradenburg**

Chairman & Co-Founder  
UsAgainstAlzheimer's  
USA

**George Vradenburg** is the Chairman and Co-Founder of UsAgainstAlzheimer's (UsA2), a disruptive and catalytic force committed to stopping Alzheimer's by 2020. The UsA2 platform seeks to escalate the fight against Alzheimer's through a broad range of powerful voices from various walks of life. UsA2 serves as the convener of the only industry coalition dedicated to stopping Alzheimer's -- the Global CEO Initiative (CEOi) on Alzheimer's -- as well as the co-convener of a 90+ member coalition of the Alzheimer's-serving community -- Leaders Engaged on Alzheimer's Disease. As a result of his UsA2 work, George was appointed to the World Dementia Council by Prime Minister David Cameron in March, 2014. In 2011, the United States Secretary of the Department of Health and Human Services named George to serve on the National Alzheimer's Advisory Council on Research, Care and Services for the first-of-its-kind National Alzheimer's Strategic Plan. In 2013 George was appointed by Congress to the Long Term Care Commission charged with devising a comprehensive long term support and services plan for the United States. Among other efforts, George has testified twice before the U.S. Congress regarding the Global Alzheimer's pandemic; conceived and supported the Alzheimer's Study Group; and, through the Vradenburg Foundation, has supported the Alzheimer's Disease International World Alzheimer's Reports and the National Institute of Health's Global Alzheimer's Research Summit. George is a member of the Council on Foreign Relations and the Economic Club of Washington. George received his B.A. from Oberlin College, magna cum laude, where he was elected to Phi Beta Kappa, and his J.D. from Harvard Law School, cum laude.



**Elias Zerhouni**

Emeritus Professor  
Johns Hopkins University  
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**Elias Zerhouni, M.D.**, A native of Algeria where he received his basic education and training, his academic career was spent at the renowned Johns Hopkins University and Hospital where he is currently professor of Radiology and Biomedical engineering and senior adviser for Johns Hopkins Medicine. He served as Chair of the Russell H. Morgan Department of Radiology and Radiological Sciences, Vice Dean for Research and Executive Vice Dean of the School of Medicine from 1996 to 2002 before his appointment as Director of the National Institutes of Health of the United States of America from 2002 to 2008. He has authored over 200 publications, holds eight patents and has founded or co-founded 5 start-up companies.

*Note that the following bios and/or photos were retrieved from online sources: Jeremy Farrar, Phil Febbo, David Glazer, Gun Peggy Knudsen, Karestan Koenen, Benjamin Neale, Erin Ramos and Kári Stefánsson.*

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A big thank-you to the sponsors of the 3<sup>rd</sup> International Cohorts Virtual Summit for their generous contribution and continued support!



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*Participants have been pulled directly from the registration website.*

# Participating Cohorts

- 23andMe
- 45 and Up Study
- AstraZeneca Integrated Genomics Initiative
- Barshi Cohort
- BELIEVE cohort study Bangladesh
- Biobank Japan
- BioVu Vanderbilt
- California Teachers Study (CTS)
- Canadian Partnership for Tomorrow
- Cancer Council Victoria Cohort Studies
- Children's Hospital of Philadelphia (CHOP) Biorepository
- Constances Project
- **Davos Alzheimer's Collaborative**
- Dementias Platform UK (data repository)
- ELSA-Brazil Project
- Environmental influences on Child Health Outcomes (ECHO) Cohort
- Environmental Polymorphisms Registry
- EPIC (European Prospective Investigation into Cancer, Chronic Diseases, Nutrition and Lifestyle)
- Estonian Genome Project
- Generations Study (GS)
- Genomics England / 100,000 Genomes Project
- German National Cohort (NAKO)
- Golestan Cohort Study
- H3Africa
- Healthy Nevada
- Israel Genome Project
- Japan Public Health Center-based Prospective Study for the Next Generation (JPHC-NEXT)
- Korea Biobank Project
- Korean Cancer Prevention Study (KCPS-II Biobank)
- Korean Genome and Epidemiological Study (koGES)
- Maule Cohort (MAUCO Study)
- Mexico City Prospective Study
- Million Veteran Program
- Multiethnic Cohort Study (MEC, NCI)
- MyCode Community Health Initiative
- NCI Cohort Consortium
- Newfoundland 100K Genome Project / Sequence Bio
- Newfoundland and Labrador Genome Project

- Newfoundland and Labrador Genome Project
- NHSII (Nurses' Health Study II, NCI)
- Nord-Trøndelag Health Study (HUNT)
- Northern Sweden Health and Disease Study
- Persian Cohort Study
- PLCO (Prostate, Lung, Colorectal and Ovarian Cancer Screening Trial, NCI)
- SAPRIN (South African Population Research Infrastructure Network)
- Saudi Human Genome Program
- Saudi National Biobank
- Shanghai Men and Women's Health Study (2 cohorts)
- SYNCHROS
- Taiwan Biobank
- U.S. Precision Medicine Initiative / *All of Us* Research Program
- UK Biobank
- **UKLWC (UK Collaborative Trial of Ovarian Cancer Screening Longitudinal Women's Cohort)**