

# Update on the *All of Us* Research Program NIH Council of Councils



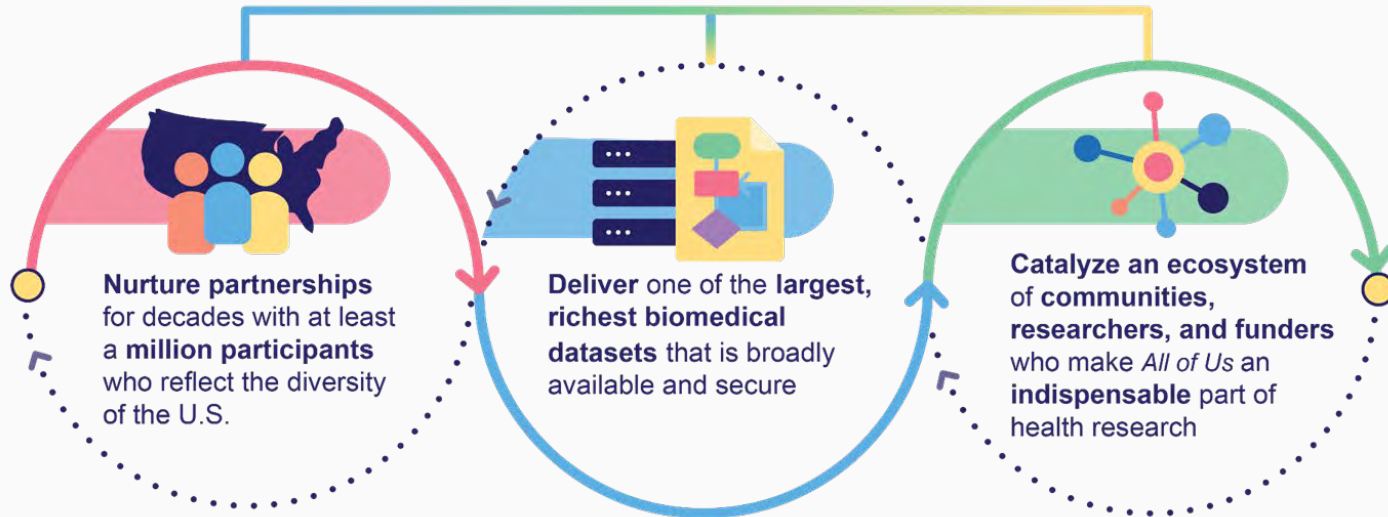
September 7, 2023

Joshua Denny, MD, MS  
Chief Executive Officer

*All of Us* Research Program

# The *All of Us* Research Program Mission

Accelerate health research and medical breakthroughs,  
enabling individualized prevention, treatment, and care for all of us



Made possible by a team that maintains a culture built around the program's core values

# Status of the *All of Us* Research Program (as of September 5, 2023)

## Participant Enrollment

**699,000+**

Participants

**397,000+**

Electronic Health  
Records

**483,000+**

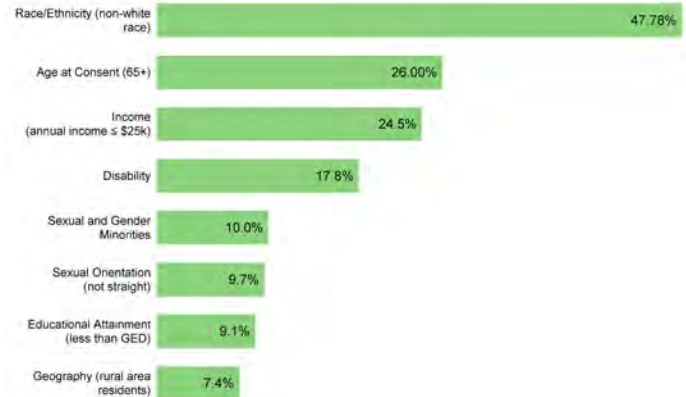
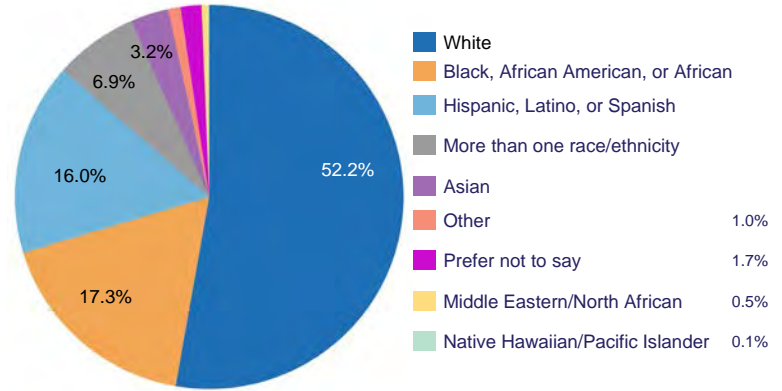
Participants who have  
completed initial steps of the  
program

**500,000+**

Biosamples

## Race and Ethnicity

## UBR Category





# Genomic Return of Results (as of September 5, 2023)

Launched December 2022

## Genetic Ancestry and Traits

Genetic ancestry details for 7 regions, and information on 4 genetic traits

### DNA Results

You'll see all of your DNA results here when they're ready. See [options for your DNA results](#).

#### Genetic ancestry and trait results



##### Genetic ancestry

Genetic ancestry can be very interesting, but you may also learn information you didn't expect. [View Results](#)

[Learn more](#)



##### Bitter taste perception

Learn what you

[View Results](#)



##### Cilantro preference

Your genes play



##### Earwax type

Flaky or sticky



##### Lactose intolerance

Your genes help



The Americas	50%
The Americas	100%
Such as North, Central, and South America	
The Middle East and North Africa	50%
Northern Africa	10%
Such as Morocco, Algeria, and Egypt	
The Middle East	40%
Such as the Arabian Peninsula and Egypt	
Western Asia and the Caucasus	50%
Such as Turkey, Iran, Syria, Iraq, and the	

Country Groups Tracked

Over 134,520 participants viewed their genetic ancestry and/or traits

## Hereditary Disease Risk

Variants in 59 genes associated with serious health conditions that have treatment or prevention options

All Us  
RESEARCH PROGRAM

JANE DOE  
DOB: January 1, 2000  
ID: 123

Specimen: Whole Blood  
Barcode: NDU-GRD-000-000-0000  
Collected: January 1, 2023  
Report date: September 29, 2022

RESEARCH RESULT - Your doctor will need to confirm this result with a clinical test before using it in your care.



### Your result:

Something very important for your health was found in your *BRCA1* gene.

#### What does this mean?

- If confirmed by a clinical DNA test, this result means that you are more likely to get some types of cancers than other people.
- It does **not** mean that you have some types of cancers.
- It does **not** mean that you will definitely get some type of cancers.
- **This result is important** and should not be ignored.

#### IMPORTANT!

Share this report with your doctor.

- This report comes from a research program, so **it is a research result**. Your doctor will need to confirm these results with a clinical DNA test before using them in your care.
- **Do not change your medical care** before this result is confirmed by your doctor.
- **Results provided are from an investigational device.** An "investigational device" is a device that

60,930 participants viewed HDR results

## Medicine and Your DNA

7 genes that can affect how your body metabolizes medicines

All Us  
RESEARCH PROGRAM

DOB: 12/31/2000

Specimen: Saliva  
Barcode: COL-GRD-000-000-0000  
Collected: December 31, 2022  
Report date: September 29, 2022

RESEARCH RESULT - Your doctor will need to confirm this result with a clinical test before using it in your care.



### Medicine and your DNA

#### Our genes affect how we respond to medicine.

They do that in many different ways. Some genes help move medicines to the right part of the body.

#### What is this kind of information used for?

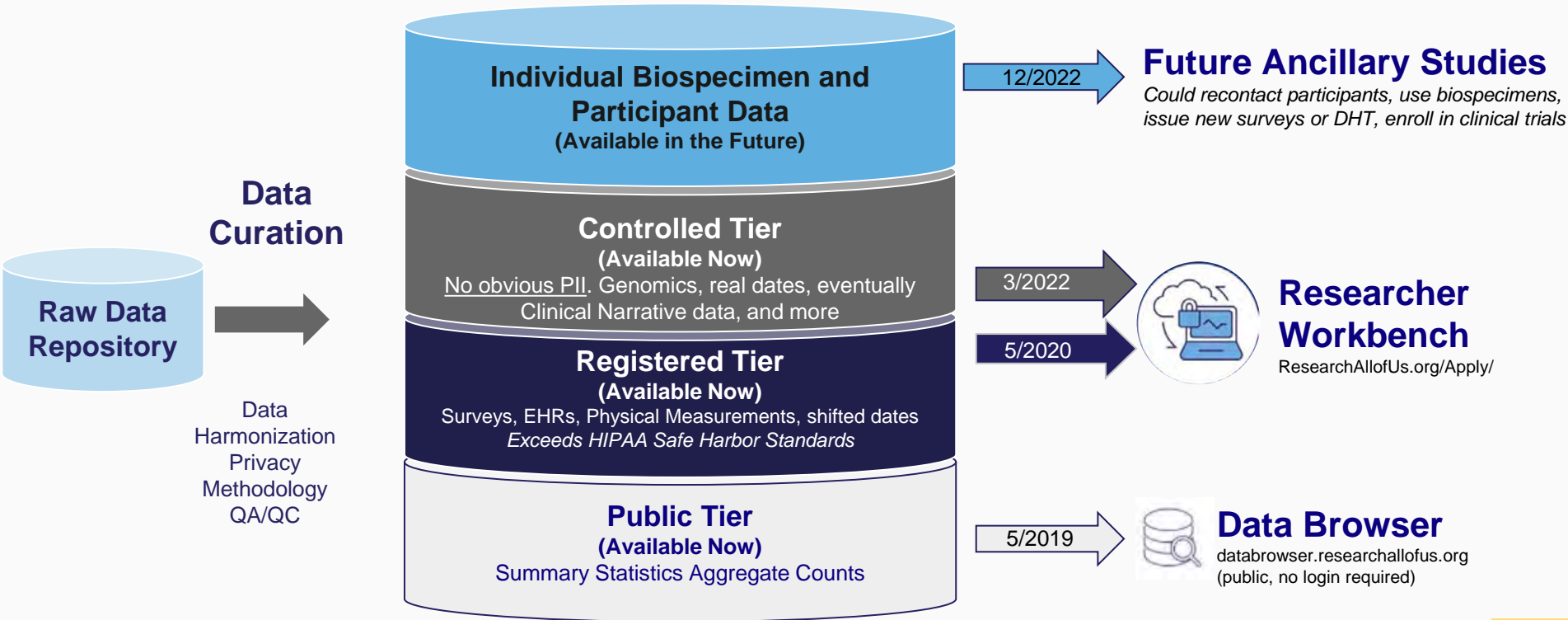
This test looked at a few of the genes in your DNA that can affect how medicines are used. The technical term for this kind of information is "pharmacogenetics."

Doctors and pharmacists use this kind of information when they consider why medicines work differently for different people.

**But doctors and pharmacists don't make decisions based on just DNA.** Some other important considerations can be age, weight, health, diet, and other medicines you are taking at the same time.

57,960 participants viewed PGx results

# Researcher Data Access



# Nearly 250,000 Genome Sequences Available to Advance Precision Medicine

As of April 20, 2023, the *All of Us* Researcher Workbench contains the largest set of whole genome sequences widely available for research.



**413,350+**

Survey Responses



**337,500+**

Physical Measurements



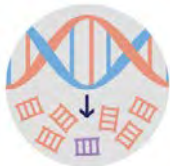
**312,900+**

Genotyping Arrays



**287,000+**

Electronic Health Records



**245,350+**

Whole Genome Sequences



**11,350+**

Structural Variants

NEW! In 2023



**1,000+**

Long-Read Sequences

NEW! In 2023



**15,600+**

Fitbit Records

NEW! Sleep Data

The whole genome sequence dataset includes variation at more than **1 billion** locations, which is nearly **one-third** of the entire human genome



# Researcher Workbench Usage and Diversity (data as of September 5, 2023)



Publications



**6,775+**  
Registered  
Researchers



**6,777+**  
Active  
Projects



**220+**  
Publications  
using *All of Us*  
data



**577+**  
Organizations  
39 Historically Black  
Colleges & Universities  
54 Hispanic Serving  
Institutions



**Top Conditions  
Studied**  
Cardiovascular disease,  
Hypertension, Mental  
Health, Cancer, Diabetes

- **Creating a diverse researcher cohort** that promotes responsible and ethical use of data, returns value to participant communities, and accelerates research impact.
- **Encouraging student assemblies and early-stage investigators** to bring fresh, creative perspectives & innovative research outcomes.
- **Ensuring access for researchers from various institutions / organizations** to establish a truly equitable resource for all.



**Over 72% of our researchers are underrepresented in the biomedical workforce - including over 27% diverse by race and ethnicity**



# Examples of the Scientific Impact of *All of Us*

Article | [Open Access](#) | Published: 10 October 2022

## Association of step counts over time with the risk of chronic disease in the *All of Us* Research Program

Hiral Master, Jeffrey Annis, Shi Huang, Joshua A. Beckman, Francis Ratsimbazafy, Kayla Marginean, Robert Carroll, Karthik Natarajan, Frank E. Harrell, Dan M. Roden, Paul Harris & Evan L. Brittain

*Nature Medicine* 28, 2301–2308 (2022) | [Cite this article](#)

[Open Access](#) Article

## Race, Ethnicity, and Pharmacogenomic Variation in the United States and the United Kingdom

by Shivam Sharma, Leonardo Mariño-Ramírez and I. King Jordan

*Pharmacetics* 2023, 15(7), 1023. <https://doi.org/10.3390/pharmacetics15071023>

Received: 12 June 2023 / Revised: 30 June 2023 / Accepted: 5 July 2023 / Published: 11 July 2023

Research Square

## Brief Communication Quantifying physical activity needed to mitigate genetic risk for obesity

Ida Han, Jeffrey Annis, Hiral Master, Andrew Hughes, Dan Roden, and 3 more

## Otolaryngology—Head and Neck Surgery



Original Research

## Hearing Loss and Sociodemographic Barriers to Health Care Access Using the *All of Us* Research Program

Luis E. Cortina, Andrew Amini, Jalen Benson, Victoria W. Huang, James G. Naples, and 2 more

## International Journal of Dermatology



Correspondence

## The association of cutaneous squamous cell carcinoma and basal cell carcinoma with solid organ transplantation: a cross-sectional study of the *All of Us* Research Program

Annika Belzer, Audrey C. Leasure, MHS, Jeffrey M. Cohen, MD, Sara H. Perkins, MD, and 2 more

First published: 05 May 2023 | <https://doi.org/10.1111/rjd.16700>

## PLOS ONE

## Family and personal history of cancer in the *All of Us* research program for precision medicine

Lauryn Keeler Bruce, Paulina Paul, Katherine K. Kim, Jihoon Kim, Theresa H. M. Keegan, Robert A. Hiatt, Lucia Ohno-Machado, On behalf of the *All of Us* Research Program Investigators

Published: July 17, 2023 | <https://doi.org/10.1371/journal.pone.0288498>

## medRxiv



THE PREPRINT SERVER FOR HEALTH SCIENCES

## Multi-ancestry genome-wide study in >2.5 million individuals reveals heterogeneity in mechanistic pathways of type 2 diabetes and complications

Ken Suzuki, Konstantinos Hatzikotoulas, Lorraine Southam, Henry J. Taylor, Xiayang Yin, Kim M. Lorenz, Ravi Mandla, Alicia Huerta-Chagoya, Nigel W. Rayner, Ozvan Bocher, S.V. Arruda Ana Luiza de, Kyuto Sonehara, Shinichi Namba, Simon S.K. Lee, Michael H. Preuss, Lauren E. Petty, Philip Schroeder, Brett Vanderwerf, Marc Kals, and 2 more

Research article | [Open Access](#) | Published: 11 June 2023

## Using machine learning to develop a clinical prediction model for SSRI-associated bleeding: a feasibility study

Jatin Goyal, Ding Quan Ng, Kevin Zhang, Alexandre Chan, Joyce Lee, Kai Zheng, Kerri Hurler-Kim, Lee Nguyen, Lu He, Megan Nguyen, Sarah McBane, Wei Li & Christine Liu Cadiz

*BMC Medical Informatics and Decision Making* 23, Article number: 105 (2023) | [Cite this article](#)

Original Investigation | Oncology

JAMA Network

August 10, 2023

## Alcohol Consumption Among Adults With a Cancer Diagnosis in the *All of Us* Research Program

Mengyao Shi, MBBS, MPH<sup>1</sup>, Chongliang Luo, PhD<sup>1</sup>, Oluseye K. Oduyale, MD<sup>1</sup>, and 2 more

## Clinical Pharmacology & Therapeutics

Article

## Drug-Induced Liver Injury with Commonly Used Antibiotics in the *All of Us* Research Program

Shaopeng Gu, Govarthanan Rajendiran, Kennedy Forrest, Tam C. Tran, Joshua C. Denry, Eric A. Larson, Russell A. Wilke, and 2 more

First published: 07 May 2023 | <https://doi.org/10.1002/cpt.2930>

AJHG | [ASHG](#) | Supports open access

ARTICLE | VOLUME 110, ISSUE 2, FEBRUARY 02, 2023 | [Download Full Issue](#)

## Functional interpretation, cataloging, and analysis of 1,341 glucose-6-phosphate dehydrogenase variants

Rinsee C. Gock, Nicholas R. Powell, Mattreya J. Durham, and 2 more

[Open Access](#) • Published: January 20, 2023 • DOI: <https://doi.org/10.1002/ajhg.10000>

Original Investigation | Equity, Diversity, and Inclusion

July 31, 2023

## Prevalence of 12 Common Health Conditions in Sexual and Gender Minority Participants in the *All of Us* Research Program

Nguyen K. Tran, PhD, MPH<sup>1,2</sup>, Mitchell R. Lurie, MD, MAS<sup>1,2,3</sup>, Claire E. Schikley, PhD<sup>4</sup>, Samantha Testaye, BA, Siddhartha Nambic, PhD<sup>5</sup>, Sriganesh Chatterjee, PhD<sup>6</sup>, Dawn Kitzowski, MS<sup>7</sup>, Paula Lozano, PhD<sup>8</sup>, Forresta T. Randall, MCRP<sup>9</sup>, Vicki Luu, MSW<sup>10</sup>, Siya Qi, MS<sup>11</sup>, Eli Hindermark, BS<sup>12</sup>, Chloe Eastcum, BA<sup>13</sup>, Anthony T. Pih, PhD<sup>14</sup>, Zubin Dastar, MS, MPH<sup>15</sup>, Mirah E. Ujensky, PhD<sup>16</sup>, Aneesa Filleo, PhD<sup>17,18</sup>, Juno Obedin-Kabiri, MD, MPH, MAS<sup>19,20</sup>, and 2 more

## Nuclear genetic control of mtDNA copy number and heteroplasmy in humans

Rahul Gupta, Masahiro Kanai, Timothy J. Durham, Kristin Tsuo, Jason G. McCoy, Anna V. Kotrys, Wei Zhou, Patrick F. Chinnery, Konrad J. Karczewski, Sarah F. Calyp, Benjamin M. Neale, & Vamsi K. Mootha

*Nature* 620, 839–848 (2023) | [Cite this article](#)

nature

## Higher Hospital Frailty Risk Score Is Associated With Increased Risk of Stroke: Observational and Genetic Analyses

Daniela Renedo, Julián N. Acosta, Andrew B. Koo, Cyprien Rivier, Nanthiya Sujjantarant, Adam de Havenon, Richa Sharma, Thomas M. Gill, Kevin N. Sheth, Guido J. Falcone and Charles C. Matouk

Originally published 22 May 2023 | <https://doi.org/10.1161/STROKEAHA.122.041891> | *Stroke*. 2023;54:1538–1547



[researchallofus.org/publications/](https://researchallofus.org/publications/)

# Funding Opportunities with the *All of Us* Research Program and NIH

## Small Grants to Enhance the Use of the *All of Us* Research Program's Data

### Applications being considered for funding (FY23)

**R03s:** to analyze data in the *All of Us* Researcher Workbench, using standard tools and methods.

**R21s:** to develop new methods, models, and tools and use them to analyze data in the Researcher Workbench.

*Working with the Institutes, Centers, and Offices, the plan is to fund 21 applications for this Small Grants Program – 14 R03s and 10 R21s*

## Active Funding Opportunities

***All of Us* Engagement, Communications, and Enrollment Partnerships** (OTA-22-006) - Rolling White Paper Deadlines

**NHGRI Investigator-Initiated Research in Genomics and Health Equity** (R01 RFA-HG-23-017) - November 8, 2023 Deadline

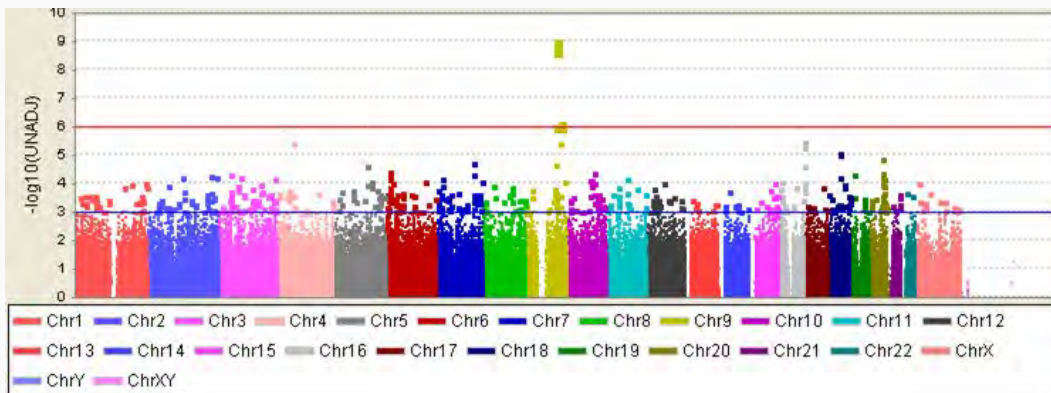
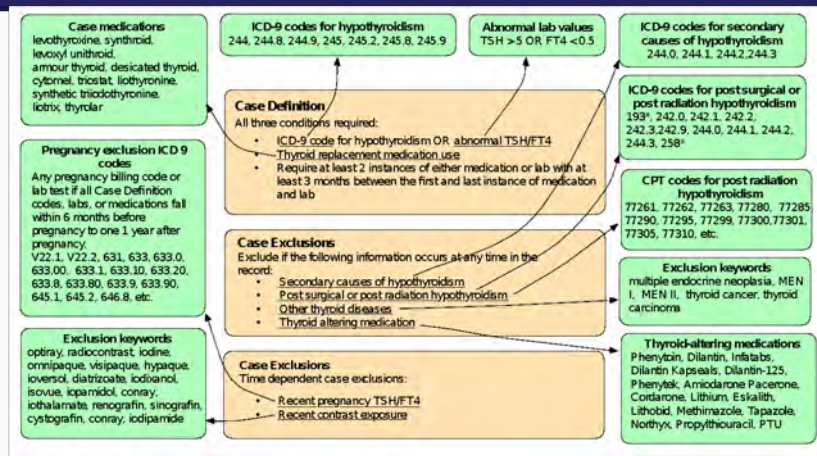
**New Investigators to Promote Workforce Diversity in Genomics, Bioinformatics, or Bioengineering and Biomedical Imaging Research** (R01 RFA-HG-21-041) - February 22, 2024 Deadline



Learn More

# My First Genome-Wide Association Study that Discovered Something New: The Genetic Basis of Primary Hypothyroidism (2011)

- 5 sites worked together to find autoimmune hypothyroidism in EHRs
- Discovered novel genetic factors
- Took ~2.5 years and ~40 people



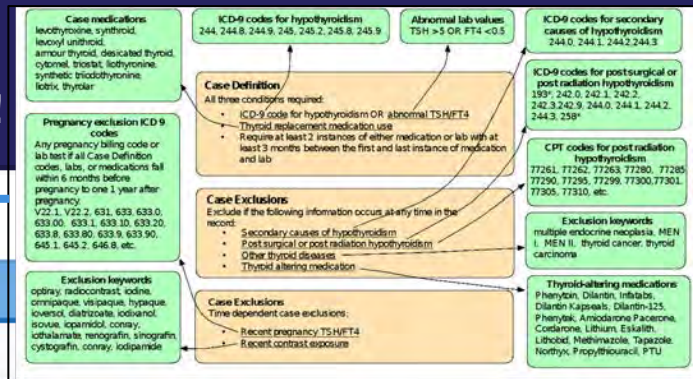
Analyses can be performed using extant data

# You Can Build this Algorithm in the All of Us Cohort Builder!

Workspaces > Cohort Builder Demonstration > Build Cohort Criteria

**All of Us** RESEARCHER WORKBENCH

DATA ANALYSIS ABOUT



Explore Source information on the Data Browser

← Conditions

Q hypothyroidism

There are 26 results in All of Us Registered Tier Dataset v5

Show results as source concepts (ICD9, ICD10)

Name	Concept Id	Source/Standard	Vocab	Code	Roll-up Count	Item Count	View Hierarchy
Hypothyroidism	140673	Standard	SNOMED	40930008			
Acquired hypothyroidism	138384	Standard	SNOMED	111566002			
Postoperative hypothyroidism	137820	Standard	SNOMED	27059002			
Iatrogenic hypothyroidism	134312	Standard	SNOMED	88273006			
Postablative hypothyroidism	132583	Standard	SNOMED	237527007			
Hypothyroidism caused by drug	37016342	Standard	SNOMED	367631000119105			
Subclinical hypothyroidism	4183422	Standard	SNOMED	54823002			
Iodine hypothyroidism	140062	Standard	SNOMED	190279008			
Subclinical iodine deficiency hypothyroidism	4130027	Standard	SNOMED	237567008			

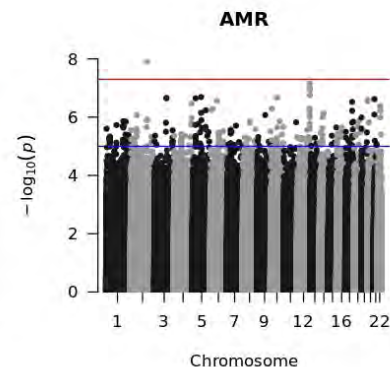
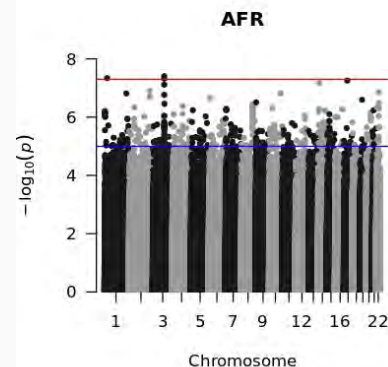
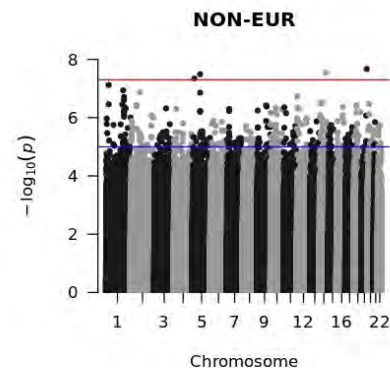
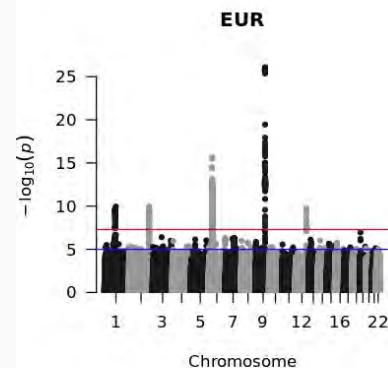
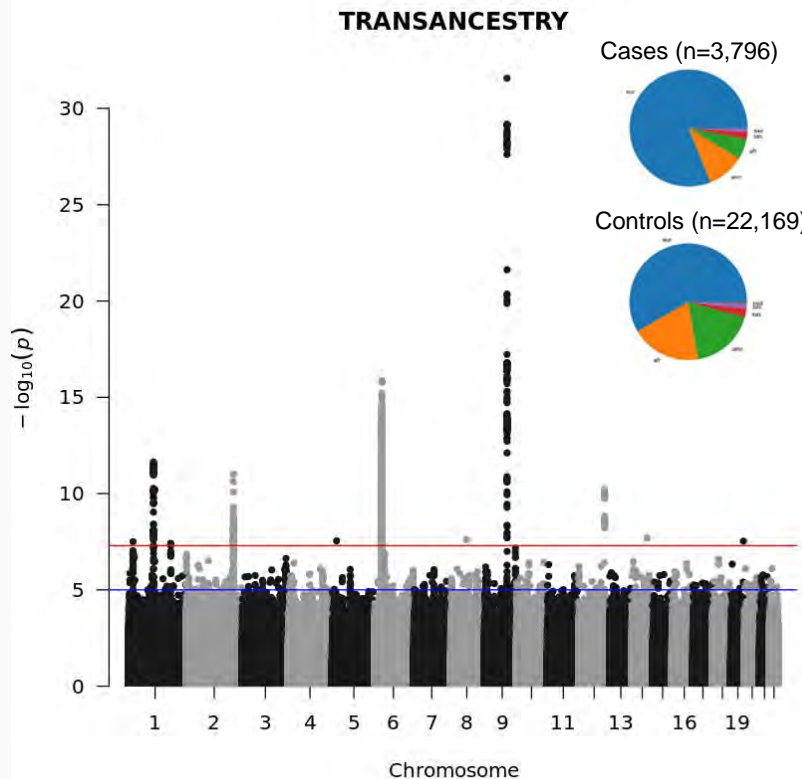
masked

FINISH & REVIEW

Anav Babbar, in preparation



# Primary Hypothyroidism in *All of Us*



Anav Babbar, in preparation



# All of Us Advisory Panel Members



[Learn More](#)



Aaron Abend, M.B.A.



Naomi Allen, D.Phil.,  
MSc, BSc



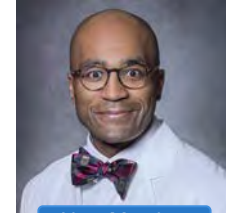
Russ B. Altman, MD,  
PhD



Wendy K. Chung,  
MD, PhD



Marylyn DeRiggi  
Ritchie, PhD



**New Member**  
Dontal Johnson, MD



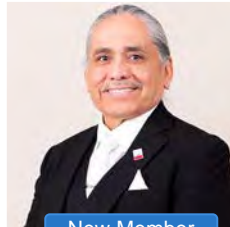
Lovell Jones, PhD



James Lu, MD, PhD

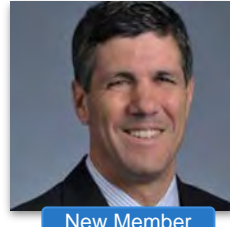


Gary W. Miller, PhD



**New Member**

John Molina, MD,  
JD, LHD

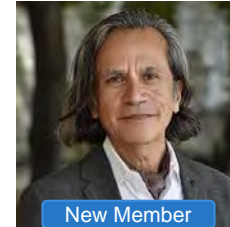


**New Member**

Chris O'Donnell,  
MD, MPH



Elizabeth O. Ofili, MD,  
MPH, FACC

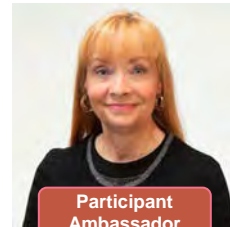


**New Member**

Jesus Ramirez-Valles,  
PhD, MPH

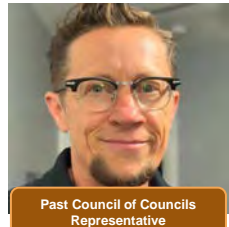


Erica Ramos, MS, CGC



**Participant  
Ambassador**

Elizabeth "Beth"  
Rubinstein

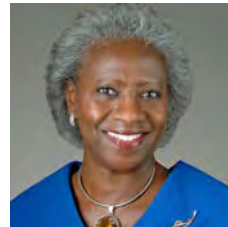


**Past Council of Councils  
Representative**

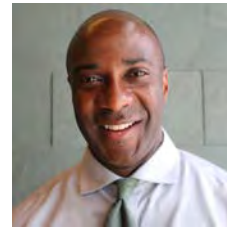
Scout, PhD, MA



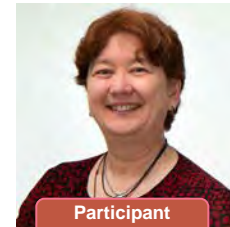
Prashant Shah, MS



Hannah Valentine,  
MD, MRCP



Roberto Vargas, MD,  
MPH



**Participant  
Ambassador**

Karen Wall, EdD, MA



Xiaobin Wang, MD,  
MPH, ScD



## Pediatrics



### More Genomics in late 2024

- About 400,000 whole genome sequences
- About 1,500 long read whole genome sequences



### More Digital Health Technology

- Wearable data from diverse participants

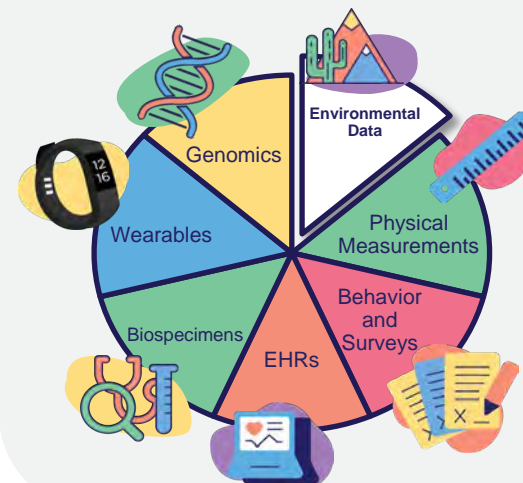


### Collecting New Data

- Life Functioning Survey
- Mental Health and Wellbeing Surveys
- Exploring the Mind cognitive and behavioral tasks

## Ancillary Studies

Incorporating complex questions about how genomic, environmental, and social interactions influence human health





[AllofUs.NIH.gov](https://www.AllofUs.NIH.gov)

[JoinAllofUs.org](https://www.JoinAllofUs.org)

[ResearchAllofUs.org](https://www.ResearchAllofUs.org)



**Thank you!**

@AllofUsResearch  
@AllofUsCEO  
#JoinAllofUs

Four circular icons representing social media platforms: Facebook, Instagram, YouTube, and Twitter.

**Thank you to our 699,000+ participants!**