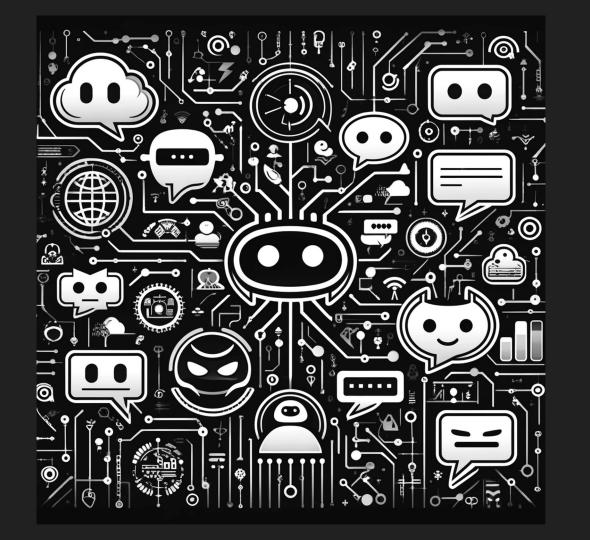
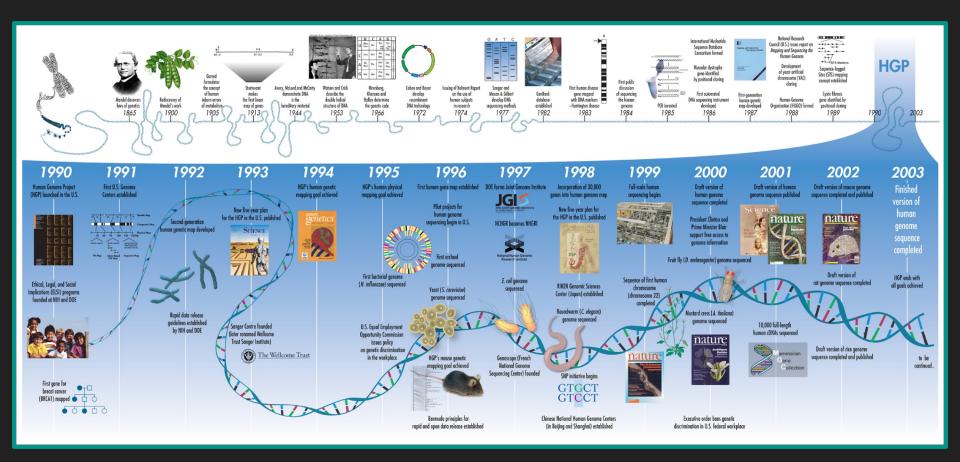




# 21st Century







#### **The Human Genome Project**



Sequenced 92% of the human genome (<400 gaps)\*



Global effort: 20 institutions in 6 countries



\$3 Billion over 13 years (1990-2003)



12 anonymous volunteers

+ innovation to recruit, acquire informed consent, and collect samples



A 1997 Buffalo, New York newspaper advertisement recruiting volunteers to provide blood samples and DNA for the Human Genome Project. (NHGRI History of Genomics Program Archive)

Persons who have undergone chemotherapy are not eligible.

For more information please contact the Clinical Genetics Service 845-5720 (9:00 am - 3:00 pm) March 24 - 26, 1997

```
TTTTTTTTTTTTAGAEGGAGTETEGGTETGTEATEEAGGETGGAGTAEAATGGTGTGATETTGAETEAETGEAAETTETGEETEEEGGGTTEAAGTAATTETEETGEETEAGEETTEEGAGTAG
GGTGeGGeTGeeTeAGGAGTeAeeAeGGeAAeAAGAAeeTGGAeeTGAGeGeAGGTGGTeAGATTeTGGGGeeAGeTTTTTTGGTTTTTAGAGAEGAGGTeTeAeTeTGTTGeeeAGGETGGA
AGGERGGETETGETGEETGGGGGEEEEGTAAEAEETEEETGAGGGETGEEAGAGETEETEATGAEEEEETTGEEATEETETETGEEAGATGETEEEAGGGGGEAAGAAEAAGTETGAAETGAAE
ETGATEGAGGAAAEETGGAEEEAAGAGAGAEETAEAETGEAGAAAGETAGATGEEAGAAAGTEATGEAGAAGGAATEEAAATGTTTGGAGAGAEAAEEEGTTGGAAAGTTTGTTGTTGTTGTTGTTGTTGTTGT
TTEEEAGGETGATETEAAAETEEGGGGETEAAGEGATETGEEEAEETTGGEETEEEAAAGTGTTTGGGATTAEAGGEGTGAGEEAETGEATEEAGETGTTGGTGAGTGTGAGTGTGAGTGTGAGTGTGAEA
EEGGGETGGTTEETEGGTGGGEAGEAGGGGEAGEGTGTETEAGEGGGGATGGGTGGAAGGGGGTTTGGGGETGGTTAATTTGATTTTGGGGTEEETAGGGETGAAGTAGATGGGGAEEEEAETEAA
nnnnnnnnnnnnnnnnnnnnnnnnnnnnnnnnnnnnceGettGeAetGeGGTGGGATAeGGGTGEAGGGAAGeeteAGGGGGGGGGGGGAEAEAGeAeeAGeGeeGTGEG
GGAAGGEGTEATGTGAEGAGGETGEAEAGAAEATGEGTGGTAETGAGETGGGEGTGGGAAGGTGTEAEGTGAEAAGGETGEAEAGAAEATGTGTGGTAETGAGETGGGEGTGGGAAGGEA
AEATGEGTGTGGTAETGAGETGGGEGTGGGAAGGEGTEAEGTGAEAAGGETGTGEAGAAEAEGEGTGTGGTAETGAGETGGGEGTGGGAAGGEGTEAEGTGAAGETGEAEAGAAEAEGEGTG
AAGGTGTEGEGTGAEGAGGETGEGEAGAAEAEGEATGTEATGETGAGETGGGTGTGGGGAAGGEGTEAEGTGAEGAGGETGTGEAGAAEAEGEGTGTGGTAETGAEETGGGTGTGGGAAGGEGTE
GETETGETGETTEATTEATGGGAGAGGEEEAAAGGETTEGGAEGGEGTETGGEAEGEAGTGGGEAEEETETTAEEGEGTGGGGAGGAEGGGTGAAEGAGTGTATETAAGEEAEEGGEAEAGAT
TGTATETAAGEEAEEGGEAEAGATEGEAGTGGGEGEEETETTAEEGEGTGGGGAGGAEGGGTGAAEGAGAGAETGTATETAAGEEAEEGGEAEAGATEGEAGTGGGEGEEETETTAEEGTGTGG
EAEAGAEGETTETGEEAGEEEATGGEEEETTGEGTGEAGAGATGETGGTEETGGGAGAGETGEGGGTGAGAEATGEAGEATEEEGTETEEAAGATGATTEEAAGGAEEETEGGETGTTEATG
TGeTGGGGAAGGTTeAGGTGeTGTTTeeAGAAeGeGGTTTTGGGTeeeGAAGeAGeGGGGGEGGGGGGGGGEGEEAEETGTeeeAGAGeeAeAAeGGAGGeeTeeeGGGAeeGAEGETGGAAGEG
TETGGGGTGTEAEETGTEETEEAGGGEGEETGAGTETEGETEAGGEEEAGETTAGETGAAEEEAEGTEEEEAGETGTGTEEETGEEETGEEETEAEEEETTAGAGEAEEETTTETEEATG
CACCCEAEAEACTGGTGEGAAGGEATGAGGGEAEAEGETGGTGEGAAGGEATGAGGGEAEGEAGGGEAGGGEAAGGEATGAGGGEAEGEAGGGEAGGGEAAGGEATGAGGEAEATGGGGEA
TGEETGGTGGAAAEEEETGGGAGGETGGAGEEAGGAEEAGGGEEEAEGTGTGETTEAEETTGAAGEETGAGGAEAEAGAETETEEGGEAATEAEATAGEEEATGTTGAGGAEGETGEETTEA
TETGGGGGGGETETGTEAGETEEGETEAETTGGEGGETGGTGTGAGGGEETGGGGGEEAGGETGETEGEAGAEEEAGGETTAGTEEEAGGEETGAGEAEAGGTGATGEEEGGEEEGGEEAGTGE
TETTGEETTTATGGGAEGTEAEEAEAAGATGGTGEEAEEGTEGGGEAEEAEAGETEEAGGTAEAGGGTGTTGTGGGGGETGGGATAGGAAGGGEATGETGAGGGEAGGGTGGGEGGEEEEETG
ettteeeAGGGeteAttetGeAeeeGGGGGGetGGGTGGGTGGGTGGetttGeteeeAeeeGtGGATTGAGGetGTGTGAGGGGTGTeAGGeetGGGGAAATGGGGTGGGTAGGTeeeAGG
GAGGGTETTGGTGAGEAAGETTTTTAAEETEAGATAEAGETEGAEATGTGEATTGETTGGGAAAGGTETAEEETGTGETEEEGGEAGGTGGGEGGGGEEEEEAGGAETEGAAGGEGGGGGGAGT
eeteeAceteeeeGGAGeeeetGeGGGeAetetAceGteetetGeeeGTGGActeeeeteAAGGTeAceteeeetTGAGATTTeAGGetnGeGTGeeeetTeAGGAAGeeeeteeetGAcete1
GGTeeeTGEAEAEEAGEETTTeeAAGGAGeeTeeATeTTGGAAATGGETTeeeAEAGAAGGGTeeeTeeATGAEAGAGeeeAEAGGEAGTGTGEAGTATGGGGGTTGGGGGTGAEATGGTGEGG
GEEREEGTGEEERGEETETTTTTETGTTTTETTATGGAGAATAGEEETTEGGTGATEAGAAGAGAEAEAAGAAGGAAAATAAAGATTGGATGAGTETEAGGTEETAGAGATAEGGGGTATGTEEC
GGTERERGETTTGRGTTTERETEGTRAGEEERTERGGATGTETGGRAEETTERGGGETGTGGEEERGGRATGTERGREGETGGRAGEETGGGRAEGGRGGEAGRGEETGETGTEERETGGET
eeeAGGGGAAAGeTGAGGEEAGGGAGEAGEEAGGATGAAGGTEAGGGEETeeeTTGGeeeeAEAGAeeeAGGGEEGETTeeeTeeAGeeeeGGETeeeGTTTGGETGGAGGTGeeeTGGGEEAT
ETTGGTEEAGTGAAGAGGGGAAEAGEAGGGGAAEAGEGTGTGTGAGGGTEEEGEEATGTETEAGGEEETTTEETGGEEEEEGEAETTAGEETTEGGTGEEEATETTEAGGGTEAGGGGAAGEEEAG
GEAGAATEETGTEAGAEAAEEAEEGAEATGGTAAGGETGETETGTGEEEGATGGAGTETETGGTGGGETGGGTAGGTTGAGETGGAAEETGGAGGTGEAATGAEETGEAEGGAGEETG
AGGTGTEAGTGEETAEGGGGGGAAGAGGEEAAAGEEEATGGGGAGTGETGTGEEEEATEEETTGAGEEEEEAGETGTGEEEETTGEAGAEAAAGGGTTETTEAAETGEGATGGTTTEETGGEAETAA
CAAEGEAGGGGAGEEEETEAAEGAGGTGGAGGEGGAGEAGATGATGAAGGAGGEEGAEAAGGATGGGGAEAGGAEEATEGAETATGAGGGTGAGTGGEETGGAGEEETGGGAGEEGTTGGETGG
GEETERETGTEEERGEETGGGTTGGGGGGERAERGGEARAGGEGTGGGGETGGAGAAGGAAAGAGTTTTEETGGEEEAGTEACAGAAAEAGEEEEAGTEATTTETGEEAEGAEAEETGAEACE
AEAGTGETGTGTAEAGGGETGTEAETGTEEATAGGGATGAETGEEAEGTEGEAGGGETGEEGTGGTGGTGGEAGATGTGGETGGGEETGTGEETGETTEEGGAGGEAGAAEEGGAEAEAAE
AGEETGGEETEGGGETGETGGGEETGGETATETETEETGGGETGGEEAGGGGTTGGEETTGGGETEAETEEEAGGAETEGETGTEETEAGEGAGTGEEEEAETGETGAGEGGATEGTAGG
GTG==AEAGG=TG=Te=AG=GGG==TGGGG=AE=TGTGGGGAE=GG=GG=AE=TGGAG=AGG=GGGGEATGGGAE=GG=EGAE=T=GE=AE=EGTG=EE=ATAGGGEGGGGE=AGGE=EE
```

#### Factors of Risk in the Development of Coronary Heart Disease— Six-Year Follow-up Experience

#### The Framingham Study

WILLIAM B. KANNEL, M.D., THOMAS R. DAWBER, M.D., F.A.C.P.,
ABRAHAM KAGAN, M.D., F.A.C.P., NICHOLAS REVOTSKIE, M.D.,
AND JOSEPH STOKES, III, M.D.
Framingham, Massachusetts

Increasingly reliable estimates of the prevalence and incidence of coronary heart disease (CHD) emphasize the importance of this disease as a contemporary health hazard. Cardiovascular disease is

Since it has been established that coronary atherosclerosis is present for many years prior to the development of symptomatic CHD, it seems evident that efforts at prevention must begin many years before the

now the lea nary heart d of all heart in the diag ment of CH decade, no bidity and curred. Thi slight increa which has b decades, wh been substar

Because of manifested a "silent" infa mortality in pital is still of best therape preventive

Received for From the I Framingham, I tute, National Service, U. S. and Welfare, V Presented a The American 1961, Bal Har Requests fo Thomas R. Da tor, Heart Di green St., Fran

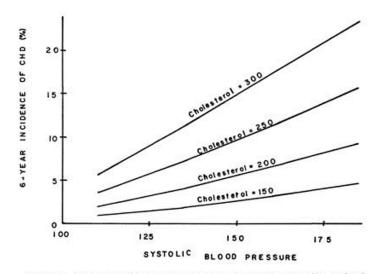


FIGURE 2. Six-year incidence of coronary heart disease according to level of systolic blood pressure at specified serum cholesterol levels (men 45 to 62 years). For explanation, see legends for Figure 1.

#### **Framingham Heart Study**

Enrolled 5,209 men and women in 1948

Some Framingham early discoveries:

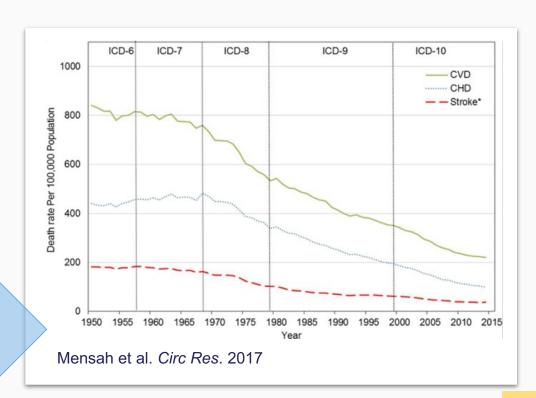
- 1960 Cigarettes increase heart disease
- 1961 cholesterol, blood pressure increase heart disease
- 1967 exercise decreases risk of heart disease; obesity increases it
- 1970 high blood pressure and atrial fibrillation cause stroke

#### The Strength of Large Cohort Studies

# The impact of Framingham (and similar cohorts) has been dramatic

From 1950 - 1996: Heart disease mortality fell 56%, stroke rates fell by 70%

Since 1990, heart disease mortality has continued to fall by 22%



# Could we have a similar experience with precision medicine in the next 40 years?

### All of Us Research Program

National Institutes of Health



May 17, 2024

Chris Lunt
Chief Technology Officer





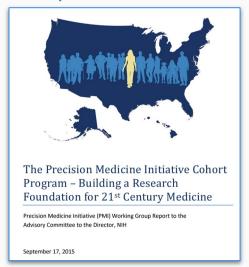






#### What is the NIH All of Us Research Program?

The *All of Us* Research Program is a historic, longitudinal effort to gather data from one million or more people living in the United States to accelerate research and improve health. By taking into account individual differences in lifestyle, socioeconomics, environment, and biology, we hope that researchers will uncover paths toward delivering precision medicine – or individualized prevention, treatment, and care – for all of us.



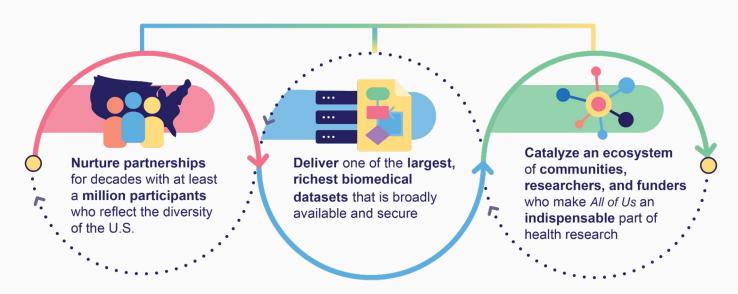


"All of Us is among the most ambitious research efforts that our nation has undertaken!"

NIH Director Francis Collins, M.D., Ph.D.

#### The All of Us Research Program Mission

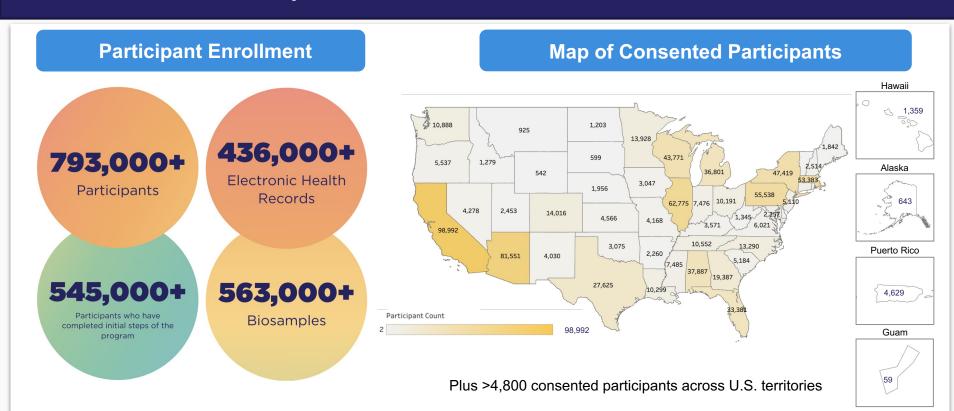
Accelerate health research and medical breakthroughs, enabling individualized prevention, treatment, and care for all of us



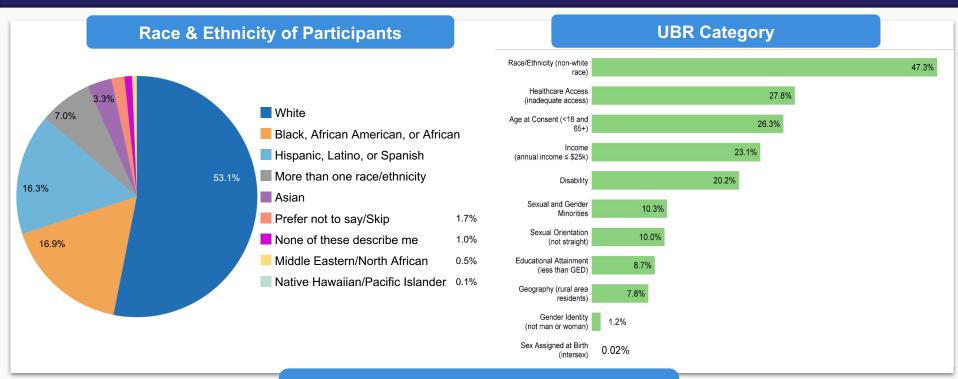
Made possible by a team that maintains a culture built around the program's core values

## **Participants**

#### **Enrolled 793K+ Participants With Continued Growth**



#### **Participant Diversity**



Over 87% of *All of Us* participants are underrepresented in biomedical research

Numbers current as of May 1, 2024

### **Data**

#### Data Types Collected from *All of Us* Participants



#### **Electronic Health** Records

Data types collected from EHR include:

- Demographics
- Vital signs Diagnoses

 Procedures Medications  Doctor and Laboratory Visits



#### **Participant Surveys**

The Basics

Health Care Access & Utilization

Mental Health and Well-

Overall Health Lifestyle

Personal and Family Medical History Social Determinants of Health

Being



#### **Physical Measurements**

- Blood pressure
- Heart rate
- Height
- Weight

- BMI
- Hip circumference
- Waist circumference



#### **Biosamples**

- Blood
- Saliva
- Urine



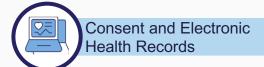
**Wearable Data** 

Fitbit data, including:

- Heart Rate
- Activity (Daily Summary)

- Activity Intraday Steps
- Sleep data

#### Data Collected and Return of Value to All of Us Participants











#### **Return of Value for Participants**

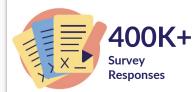
#### Participants may receive:

- Genetic information
- Survey data (comparative)
- EHR and claims data
- Ongoing study updates
- Aggregate results
- Scientific findings
- Opportunities to be contacted for other research opportunities





#### v7 of the Curated Data repository







245K Whole Genome **Sequences** 





300K+ Genotyping



280K+ **Electronic Health** Records



16K **Fitbit** Records

**NEW! Sleep Data** 

#### **Genomic Data Is Paired With Rich Phenotypic Data**



Have Whole Genome Sequences + Electronic Health Records + Physical Measurements + Survey Responses



245.100+

Have Whole Genome Sequences + Physical Measurements + Survey Responses



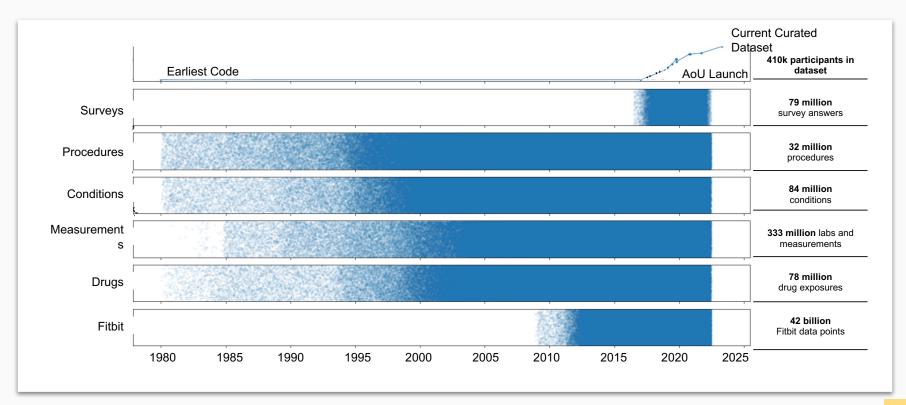
206.150+

Have Whole Genome Sequences + Electronic Health Records



Have Whole Genome Sequences + Fitbit Records Fitbit data may include physical activity, step counts, heart rate, and sleep data

## EHR Data on the Researcher Workbench is Diverse and Longitudinal



#### **Participants Can Receive Four Types of Genetic Research Results**

## Genetic ancestry and traits results





#### 7 regions (20 subregions) and 4 traits

- Sub-Saharan
   Africa
- Europe
- Oceania
- Southern Asia

- Eastern and northern Asia
- The Middle East and North AfricaThe Americas

- Ear wax
- Bitter taste perception
- Cilantro preference
- Lactose intolerance

# Medicine and your DNA Report

#### 7 pharmacogenomics (PGx) genes and 50+ medications

- CYP2C19
- DPYD
- G6PD

- SLCO1B1
- NUDT15

- TPMT
- UGT1A1

## Hereditary Disease Risk (HDR) Report



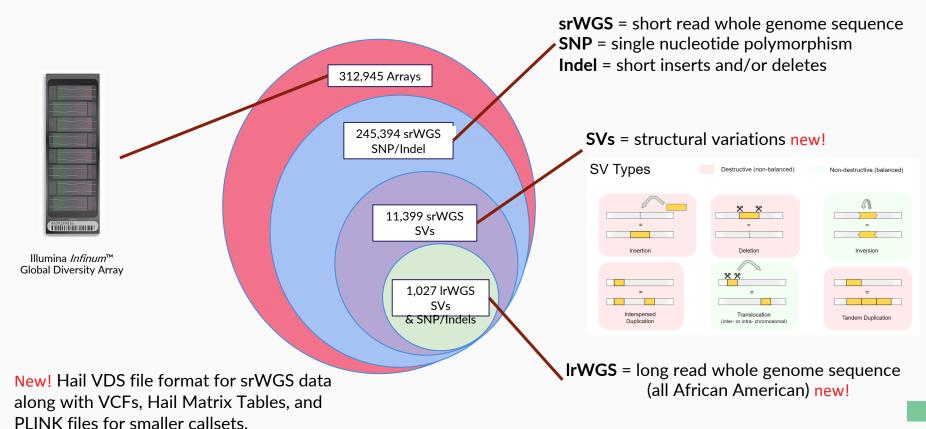
#### 59 genes (SNVs + indels, e.g. no SVs)

- Breast cancer
- Ovarian cancer
- Uterine cancer
- Colorectal cancer
- Prostate cancer

- Melanoma
- Brain cancer
- Pancreatic cancer
- Stomach cancer

- Familial hypercholesterolemia
- Cardiomyopathies
- Arrhythmias
- Arteriopathies

# Genomics

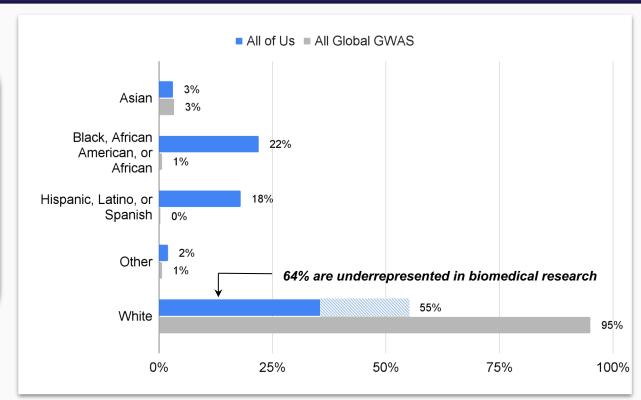


#### All of Us is Enhancing Diversity of Genomic Studies

# Updated genomic data set

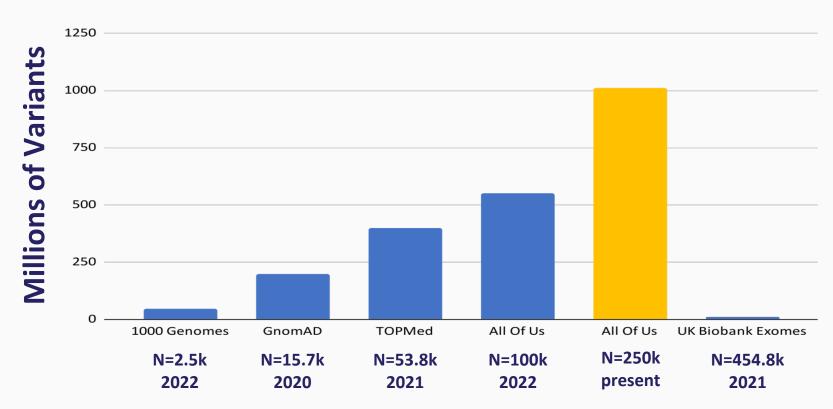
~45% diverse by race/ethnicity

77% underrepresented in biomedical research



All Global GWAS values from www.gwasdiversitymonitor.com. Values current as of November 2023

## **Discovery of Rare Variation**



## All of Us Data Includes Range of Diseases

Conditions	Domain	Participants
Heart Disease	Heart	89,180
Obesity	Endocrine	80,980
Type 1 Diabetes	Endocrine	6,740
Type 2 Diabetes	Endocrine	56,340
Any cancer	Cancer	50,720
Asthma	Pulmonary	50,160
Chronic Obstructive Pulmonary Disorder	Pulmonary	24,940
Epilepsy	Neuro	9,320
Stroke	Neuro	940
Rheumatoid Arthritis	Autoimmune	8,660
Osteoarthritis	Autoimmune	99,880

Conditions	Domain	Participants
Depressive Disorder	Mental Health	82,660
Bipolar Disorder	Mental Health	16,200
Dementia	Mental Health	6,020
Human Immunodeficiency Virus	Infectious Disease	5,500
COVID-19*	Infectious Disease	58,000*
Alcoholism	Abuse	20,380
Opioid Usage	Medication	155,000
Age-Related Macular Degeneration	Eye	4,740
Hearing loss	Hearing	30,400
Falls	Aging/Nursing	4,860

<sup>\*</sup>using combination of diagnosis code, lab test, and COVID survey answers

# Researchers

#### **Researcher Data Access**

# Data Curation

Raw Data Repository

Data
Harmonization
Privacy
Methodology
QA/QC

# Individual Biospecimen and Participant Data

#### **Controlled Tier**

(Available Now)

No obvious PII. Genomics, real dates, eventually Clinical Narrative data, and more

#### Registered Tier

(Available Now)

Surveys, EHRs, Physical Measurements, shifted dates Exceeds HIPAA Safe Harbor Standards

#### **Public Tier**

(Available Now)

**Summary Statistics Aggregate Counts** 

12/2022

#### **Ancillary Studies**

Could recontact participants, use biospecimens, issue new surveys or DHT, enroll in clinical trials



Researcher Workbench

ResearchAllofUs.org/Apply/

5/2019



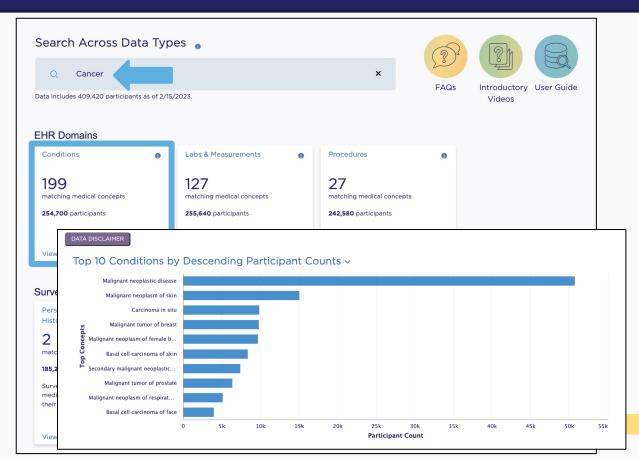
databrowser.researchallotus.org

#### All of Us Research Hub: Public Data Browser

#### **Summary statistics of:**

- EHR Data (Conditions, Drug Exposures, Lab & Measurements, Procedures)
- Genomic Variants
- Survey Questions (including COVID-19 surveys)
- Physical Measurements
- Open Access (no login required)
   Data Browser





## All of Us Researcher Workbench: Access to Row-Level Data for Analysis



#### **CONFIRM YOUR INSTITUTION'S AGREEMENT**

Before you can create an account, your institution must have a Data Use and Registration Agreement (DURA) in place with All of Us. Confirm DURA.



#### **COMPLETE THE MANDATORY TRAINING**

The training focuses on conducting responsible and ethical research using the Researcher Workbench.

Additional training is required to access the Controlled Tier. Learn more.

STEP 2

#### CREATE AN ACCOUNT AND VERIFY IDENTITY

After creating your Researcher Workbench account, you will be asked to verify your identity through login.gov. **Learn more**.



#### SIGN THE DATA USER CODE OF CONDUCT (DUCC)

This agreement outlines the program's expectations for researchers who use the Researcher Workbench and describes how program data may be used.

View the DUCC.

Individual Biospecimen and Participant
Data

(Available in the future)

Controlled Tier (Available Now) 'II. <u>Genomics,</u> real dates, eventually Cli

Registered Tier (Available Now)

Surveys, EHRs, Physical Measurements Exceeds HIPAA Safe Harbor Standards

> Public Tier (Available Now) Summary Statistics Aggregate Counts

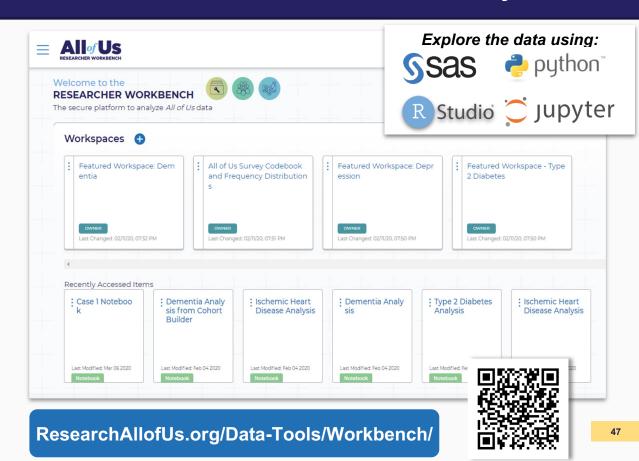


**Researcher Workbench** 

#### All of Us Researcher Workbench: Access to Row-Level Data for Analysis

#### **Researcher Workbench**

- Cloud-based platform
- Graphical tools for creating cohorts and datasets
- Open to researchers at academic, not-for-profit, and health care institutions
- Passport access model with institutional agreements for all researchers
- No separate IRB approval needed



### March 2021, Patrick Wu Defends His PhD Dissertation: Repurposing drugs using gene expression signatures and EHR data

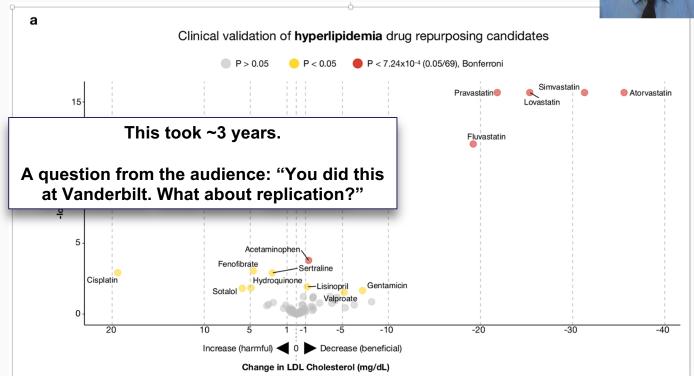
gene expression data identifies drugs that might lower cholesterol



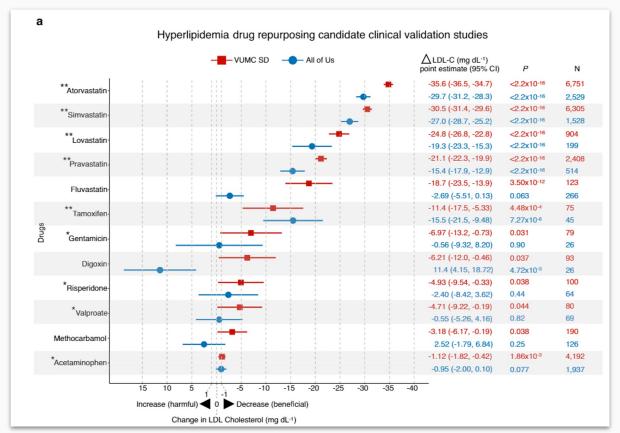
found 69 candidate drugs



study those drugs in the Electronic Health Record data at Vanderbilt



### One Week Later, he emails his advisor this figure including All of Us results...



#### All of Us Research Program's Commitment to Researcher Diversity (as of May 1, 2024)



10,680+ Registered Researchers



10,640+ Active Projects



410+
Publications
using All of Us
data



760+ Organizations

43 Historically Black Colleges & Universities

69 Hispanic Serving Institutions



80 International 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 78% of our researchers are underrepresented in the biomedical workforce - including over 30% diverse by race and ethnicity

#### You can even find *All of Us* at High School Science Fairs now!

High School Students are now using the All of Us Researcher Workbench

5 High Schools

15+ projects currently underway



**Utah's Summer Teacher Institute 2023** 





T.

Research Teal

Can Antidepressants F

on SDOH and cardiovascular risk in diabetics. (Paul FYI – re AMIA and his nursing home proje

project, where he collaborates with local nursing and rehabilitation facilities, by visiting nursing homes a

Jack Cummins

Jack Cummins is an 11th grader with a passion for research, coding, and bioinformatics. He has been selected to present his resea School Scholar and also at the Sigma XI FoRE conference. His research projects applying natural alianguage processing to medically or no predictive modeling and Jack is thrilled to be part of the All of Us community! At Manchester Essex Regional High School, he was and is president of the math team. In addition, he worked with his technology teacher to create a new Python course for the high school a leader in his high school community, as president of the class of 2025 and captain of the tennist team, funder of the French to

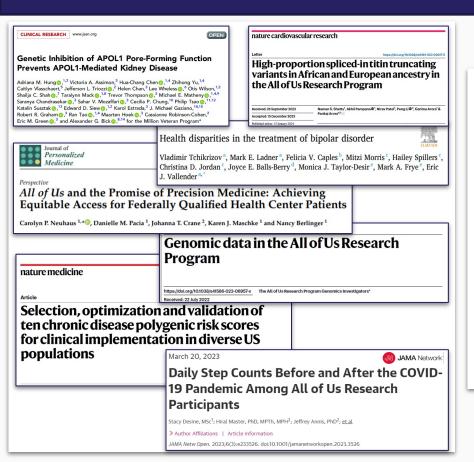
Predictive Analysis of Sjogren's Syndrome: A Machine Learning Analysis

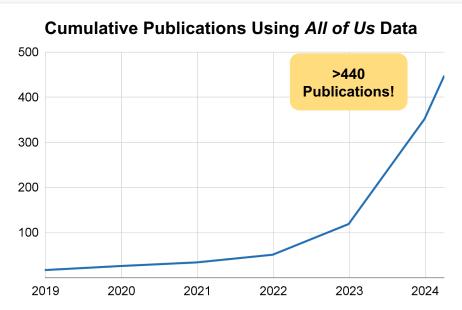
Clonal Hematopoiesis of Indeterminate Potential and Solid Tumors in Breast Cancer

# **Output**

## **Growing Scientific Impact**



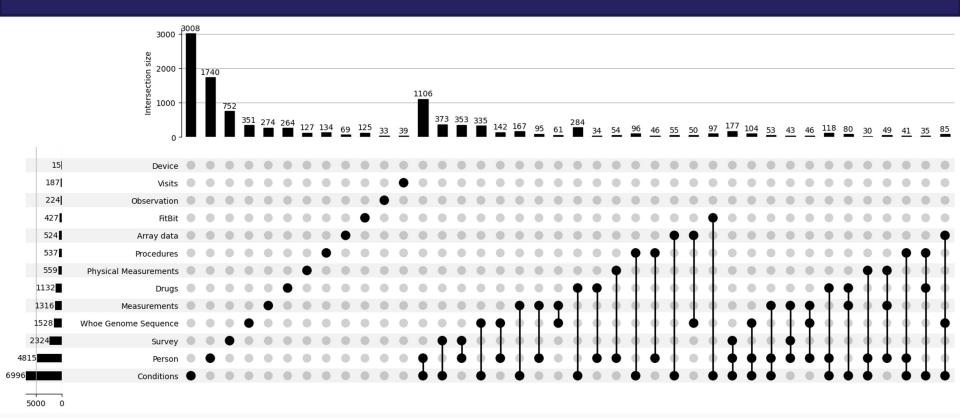




## **Disciplinary spread**



## Data types included in cohorts



Plot restricted to show only intersections with counts of at least 30. Person = Demographics. Device is from EHR.

Created by Lina Sulieman

#### What stories are we telling already about the power of participant data?



This study highlights the importance of **diversity in health research** within underrepresented groups.

Studies like this one helps health care providers move away from "one-size-fits-all" approaches to care.

#### What stories are we telling already about the power of participant data?



Linked data Fitbit devices with data from electronic health records (EHRs).

The researchers analyzed more than 1,700 medical conditions

Taking at least 8,200 daily steps was associated with a lower risk for certain health conditions. This included chronic diseases like high blood pressure, obesity, and diabetes.

The study also uncovered **new links** between daily steps and lower risk for **sleep apnea**, **depression**, and **acid reflux**.

Many people shared **four years or more** of their activity data.

Read more about this <u>here</u>.

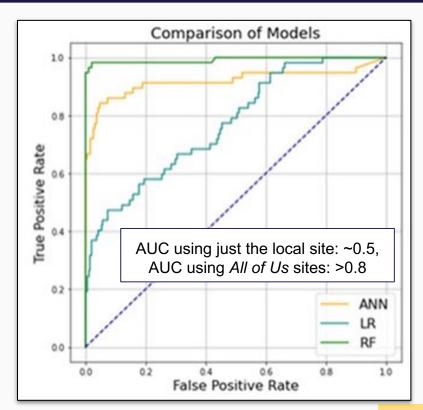
#### Machine Learning in *All of Us* to Predict Glaucoma Outcomes

Predictive Analytics for Glaucoma Using Data From the All of Us Research Program

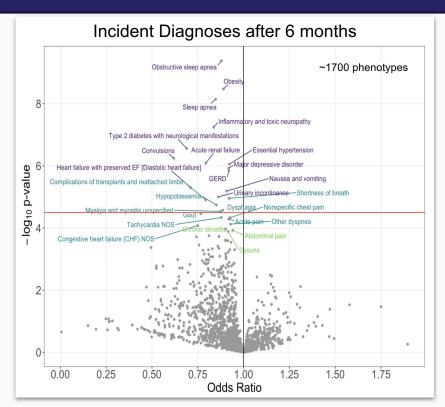


SALLY L. BAXTER, BHARANIDHARAN RADHA SASEENDRAKUMAR, PAULINA PAUL, JIHOON KIM, LUCA BONOMI, TSUNG-TING KUO, ROXANA LOPERENA, FRANCIS RATSIMBAZAFY, ERIC BOERWINKLE, MINE CICEK, CHERYL R. CLARK, ELIZABETH COHN, KELLY GEBO, KELSEY MAYO, STEPHEN MOCKRIN, SHERI D. SCHULLY, ANDREA RAMIREZ, AND LUCILA OHNO-MACHADO, ON BEHALF OF THE ALL OF US RESEARCH PROGRAM INVESTIGATORS

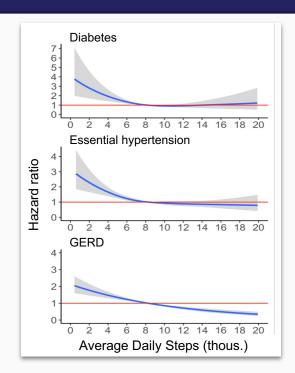
**Conclusion**: Models trained with multisite, multi-vender EHR data *All of Us* data achieved **superior performance** compared to using single-center data.



#### PheWAS of Physical Activity (Step Count) in All of Us



- Analysis uses average of daily steps prior to diagnosis.
- Effect size per 1000 steps, adjusted for age, sex, race



Adjusted for age, sex, race, CAD, cancer, BMI, systolic blood pressure, education level, smoking, alcohol use, and monthly daily steps (time varying)

# **The Future**

## The Scientific Priorities Roadmap for All of Us

INDIVIDUAL/ ADVANCING PRECISION HEALTH, MEDICINE, AND EQUITY: RISK ASSESSMENT, DIAGNOSIS, AND TREATMENT FOR EVERYONE **POPULATION HEALTH** TRANSLATION: GOVERNMENT, COMMERCIAL, HEALTH SYSTEMS, PAYERS, POLICY **HEALTHY AGING AND RETURN OF RESULTS IMPACT** PREVALENT COMMON AND MATERNAL AND CHILD HEALTH **ON INDIVIDUAL AND** RESILIENCE ACROSS THE CAUSALITY & RARE HEALTH CONDITIONS **LIFESPAN** POPULATION HEALTH OUTCOMES MODIFIERS: MENTAL HEALTH, SOCIAL DETERMINANTS, GENETICS, DISPARITIES STRESSORS, AND OTHERS LIFESTYLE, SUBSTANCES, **GENETICS AND EXPOSURES & ENVIRONMENT HEALTH INEQUITY** AND BEHAVIORAL HEALTH **BIOLOGY DRIVERS NEW METHODS TOOLBOX AND PHENOTYPE EXPANSION** WORKBENCH AND DATA CORE DATA: ELECTRONIC HEALTH RECORDS, SURVEYS, PHYSICAL MEASURES, DIGITAL HEALTH TECHNOLOGIES, **GENOMES, ASSAYS** 



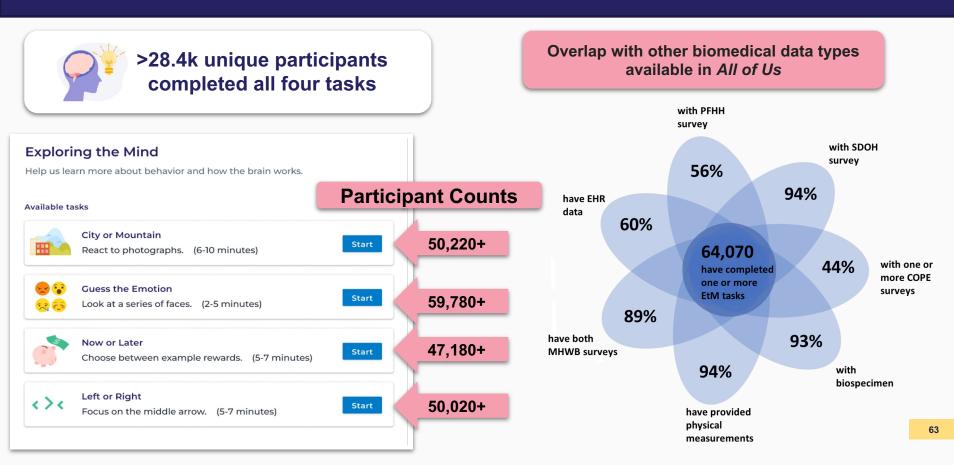
#### New Data (v8) Slated for Release Late 2024 to Researchers

Genomic Increases	Data Quantity
<b>Arrays</b> (from 312,900+)	400K+ Arrays (+27%)
<b>srWGS</b> (from 245,350)	400K+ srWGS (+63%)
Long Read Sequences (from 1K)	2.5K+ Long read sequences (+150%)

\*Data from Mental Health and Well-Being surveys and Exploring the Mind tasks are scheduled for off-cycle release in **January 2025**, and data from participant-mediated EHR is scheduled for release in **March 2025**.

New Data	Data Quantity
Life Functioning Survey	60K+
Surveys from CareEvolution Platform	60K+
Fitbit Data from CareEvolution	10K+
HealthPro Deceased Participant Reporting	4K+
Remote Height & Weight	50K+
Racial and Ethnic Subcategories	620K+
Mental Health and Well-Being Surveys*	110K+
Exploring the Mind Tasks*	35K+
Participant-Mediated EHR*	20K+

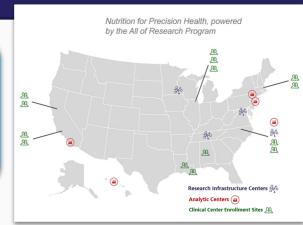
#### All of Us Ancillary Studies: Exploring the Mind with NIMH



#### All of Us Ancillary Studies: Nutrition for Precision Health

#### Over 17,000 participants expressed interest in NPH

Researching how nutrition can be tailored to each person's **genes**, **culture**, **and environment to improve health** 



1

#### **Launched March 2023**

Examine baseline diet and physiological responses to meal challenges

2184 consented, 1679 enrolled, 1546 completed



#### **Launched October 2023**

Examine responses to 3 short-term intervention diets in community dwelling controlled feeding studies

140 consented, 97 enrolled, 22 completed



#### Launched: Fall 2023

Examine responses to 3 short-term intervention diets in domiciled controlled feeding studies

39 consented, 25 enrolled, 6 completed

# Why









SanD







AllofUs.NIH.gov

JoinAllofUs.org

ResearchAllofUs.org





@AllofUsResearch @AllofUsCEO #JoinAllofUs









Thank you to our 790K+ participants!

# **Backup slides**

#### **SAS Studio**

SAS Studio is now available on the *All of Us* Researcher Workbench. SAS Studio has a point-and-click interface and allows users to perform powerful statistical analyses and data visualizations with *All of Us* data.

Researcher Workbench users do not need to pay for an individual SAS license at this time. The software is provided at no additional cost through the Researcher Workbench. Users will still incur computational costs in the cloud, as with all other analyses.

Visit <u>allof-us.org/SAS</u> or scan the QR code to learn more about using SAS Studio.



#### **RStudio**

RStudio is now available on the *All of Us* Researcher Workbench.

RStudio's point-and-click interface and suite of tools, including a variable explorer, an R Markdown editor, a debugger, and an R terminal, make it easier for researchers to use R in the Researcher Workbench.

Visit <u>allof-us.org/RStudio</u> or scan the QR code to learn more about using RStudio.





### Separate harmonization and collection



When storage was expensive, we built databases like a pigeon-hole mailbox

Collection Normalization | is expensive Storage



### 4. COLOR OR RACE

Fill one circle.

If "Indian (American)," also give tribe.

If "Other," also give race.

### 1970 US Census

## -\\_(ツ)\_/-LOL

"An extraordinary memoir . . . It belongs with Atul Gawande's writings and When Breath Becomes Air." —Adam Grant, New York Times bestselling author of Originals

## CHASING MY CURE

A Doctor's Race to Turn
Hope into Action

A MEMOIR

David Fajgenbaum

### Castleman's disease

a rare disorder that involves an overgrowth of cells in lymph nodes

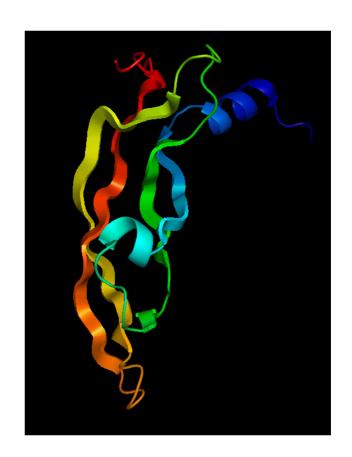
# Abductive reasoning



Cherry angioma

## **VEGF**

Vascular endothelial growth factor



## Collection Normalization is expensive Storage

## Preserve provenance

## Qualitative collection $\longrightarrow$

Analysis-specific feature extraction

## Reduce collection costs with Chatbots



You

You are an expert health researcher performing an interview to collect the personal health history of a study participant. You should be sympathetic and respectful, without offering any health advice or diagnoses. If you see connections between answers, ask follow up questions that can connect the answers. The interview should start by understanding the topics that are of particular interest to the participant. Ask questions one at a time, and acknowledge the answers that are given. Start from the beginning.



Understood. Let's begin the interview. I'll start by understanding the topics that are of particular interest to you:

1. Are there any specific health issues or concerns that you feel are particularly important to share or focus on?

### CL Yo

I have a family history of essential tremors, affecting your father, son, and myself. Notably, my father's tremors worsened with age, impacting his hobbies. My tremors were more prominent when younger but have since reduced. My father had intracranial surgery to implant a deep brain stimulation device, but it was not successful.

### ChatGPT

Thank you for sharing that significant aspect of your family health history. It's important to note the familial pattern of essential tremors and the varied response to treatments like deep brain stimulation.



1. Besides the essential tremors, have you or any family members experienced other

### Reduce harmonization costs with LLMs

(And direct the harmonization toward the specific analysis)



#### ChatGPT

Based on the provided doctor's notes for Mrs. Dev, several conditions from the Observational Medical Outcomes Partnership (OMOP) Common Data Model (CDM) could be considered. OMOP CDM provides standardized codes for various medical conditions. Here are some potential OMOP conditions that may be present based on the symptoms described:

- 1. Major Depressive Disorder: The notes indicate signs of severe depression, such as a depressed mood, slowness in physical movement, and slowed speech and thinking.
- 2. Dysfluent Speech: The description of dysfluent, garbled, and loud speech suggests a possible speech disorder.
- 3. Anomic Aphasia: Difficulty in naming objects may be indicative of anomic aphasia, a type of language disorder.
- 4. Psychotic Disorder: The mention of a psychotic or borderline psychotic process, disorganized behavior, and inappropriate anger might align with a psychotic disorder.
- 5. Cognitive Impairment: The presence of disorganized behavior and difficulty in repeating phrases suggests possible cognitive impairment.

It's important to note that assigning OMOP conditions based on a set of symptoms should be done by a healthcare professional who can conduct a comprehensive evaluation. This list is a suggestive starting point based on the symptoms described and would require further clinical assessment and diagnostic processes to confirm.





