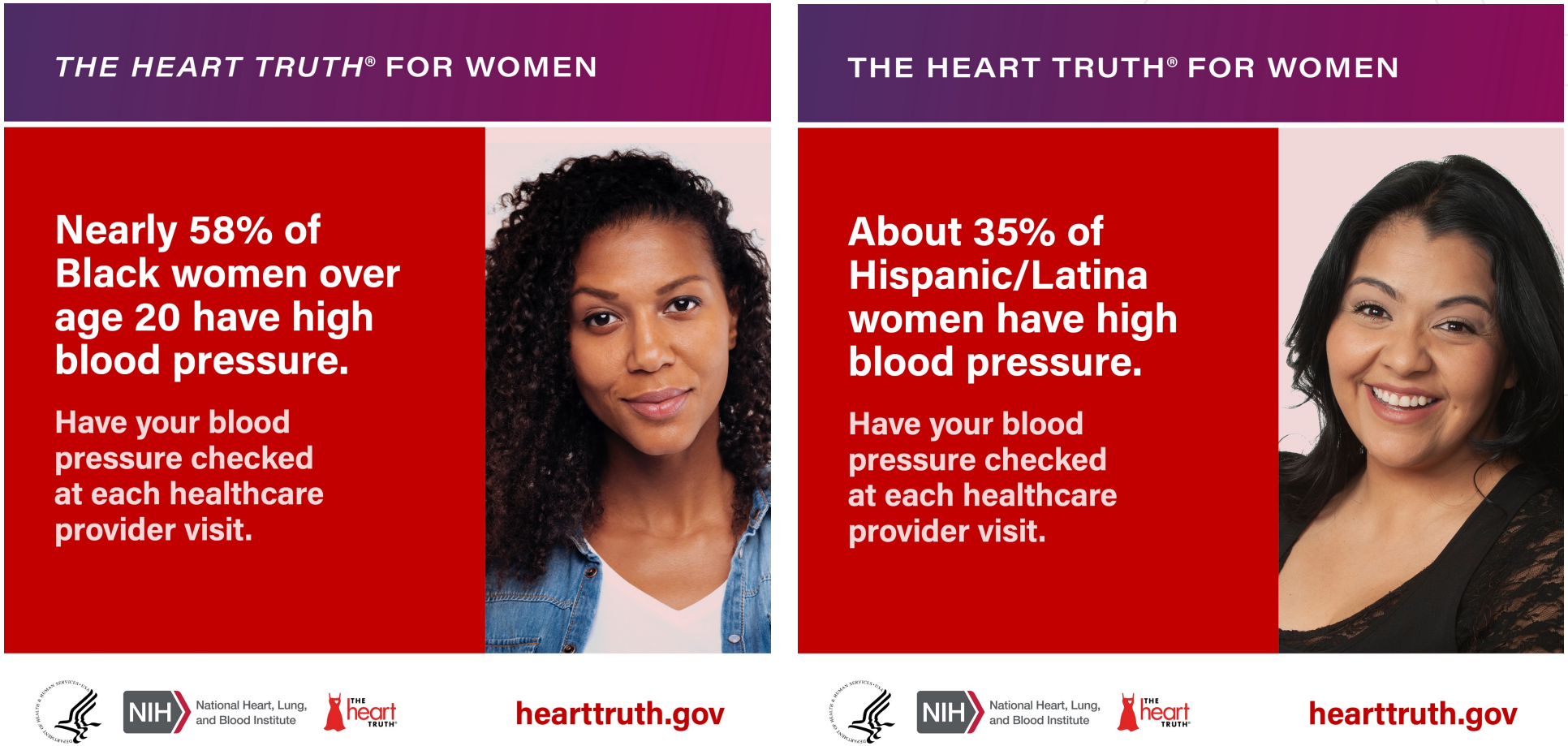
February is American Heart Month



NIH National Heart, Lung, and Blood Institute | BioData CATALYST®

BDC Community Hours will begin shortly

Wednesday February 28th 1PM ET

With Dr. Cera Fisher and Dr. Jose D. Vargas

NIH National Heart, Lung, and Blood Institute | BioData CATALYST®

February is American Heart Month

#OurHearts

eat healthier together

Get heart healthy for life by following the Dietary Approaches to Stop Hypertension (DASH) eating plan. It requires no special foods, provides daily and weekly nutritional goals, and can help lower high blood pressure.

Support your loved ones in their effort to stick to DASH by doing the following together:

- Pick out recipes to try.
- Make a grocery list.
- Cook heart-healthy versions of family favorites.
- Enjoy the meals you've prepared.

nhlbi.nih.gov/DASH

NIH National Heart, Lung, and Blood Institute

THE heart TRUTH®

NIH National Heart, Lung, and Blood Institute

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NIH National Heart, Lung, and Blood Institute | BioData CATALYST®

February is American Heart Month

#OurHearts

are healthier when we move together

Physical activity is a great way to help protect yourself from heart disease and stroke. Keep your heart healthy and aim for at least 2½ hours of moderate physical activity every week.

Doing heart-healthy activities with a friend will keep both of you inspired for the long run.

- Take an online fitness class together like yoga.
- Commit to a walking schedule with a friend or family member, even if you can't walk together.
- If you enjoy the outdoors, try hiking, biking, golfing, or gardening.
- Protect your heart by moving more and get your family and friends to do the same.

NIH National Heart, Lung, and Blood Institute

THE heart TRUTH

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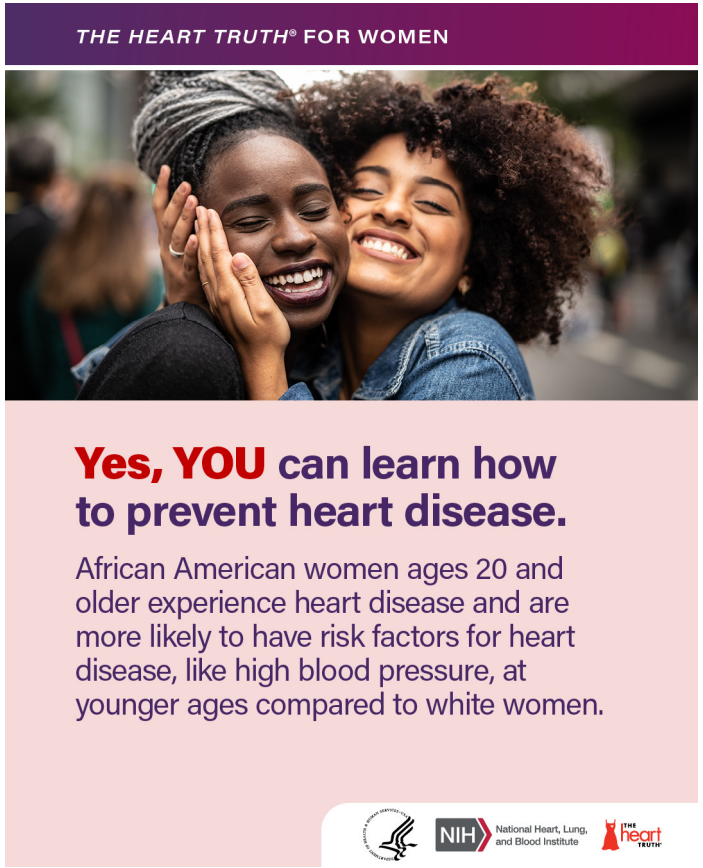

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NIH National Heart, Lung, and Blood Institute | BioData CATALYST®

Let’s get started.

Statement of Conduct

The BioData Catalyst Consortium is dedicated to providing a harassment-free experience for everyone, regardless of gender, gender identity and expression, age, sexual orientation, disability, physical appearance, body size, race, or religion (or lack thereof). We do not tolerate harassment of community members in any form. Sexual language and imagery is generally not appropriate for any venue, including meetings, presentations, or discussions.

Resource: Statement of Conduct

Session Materials and Housekeeping

- Please type your questions into the chat. We will pause every ten minutes or so for questions and discussion.
- Check the chat for relevant links during the session.
- We encourage you to submit unanswered questions, no matter how big or small, to our help desk
- Join the ecosystem: https://biodatacatalyst.nhlbi.nih.gov/contact/ecosystem
- Check your email inbox and the community forum materials by the end of the week. Please pass them on to your colleagues and networks.

Hosts and Support

Jose D. Vargas, M.D., D.Phil
Cardiologist
DC Veterans' Affairs Administration

Cera Fisher, PhD
Community Engagement Manager
Velsera

David Roberson
Community Engagement Manager
Velsera

Nathalie Volkheimer, PhD
User Engagement Specialist
BDC Coordinating Center

Amber Voght
Instructional Design Specialist
BDC Coordinating Center

Expanding Multi-Cloud Platforms on BDC *Powered By Seven Bridges*

Cera Fisher, PhD
Community Engagement Manager
Velsera

Cera Fisher grew up in the exurban wilds of Alabama, where her fascination for the natural world - and bugs - started. She earned Bachelor's and Masters' degrees in the Natural Sciences from Arizona State University, where she was a member of the Embryo project. She earned a PhD from the University of Connecticut in Ecology and Evolutionary Biology. During her postdoctoral research, Cera used bioinformatic methodologies, such as genome sequencing in her projects. Her proficiency in these and related methods, plus a passion for teaching and learning, inform her current work.

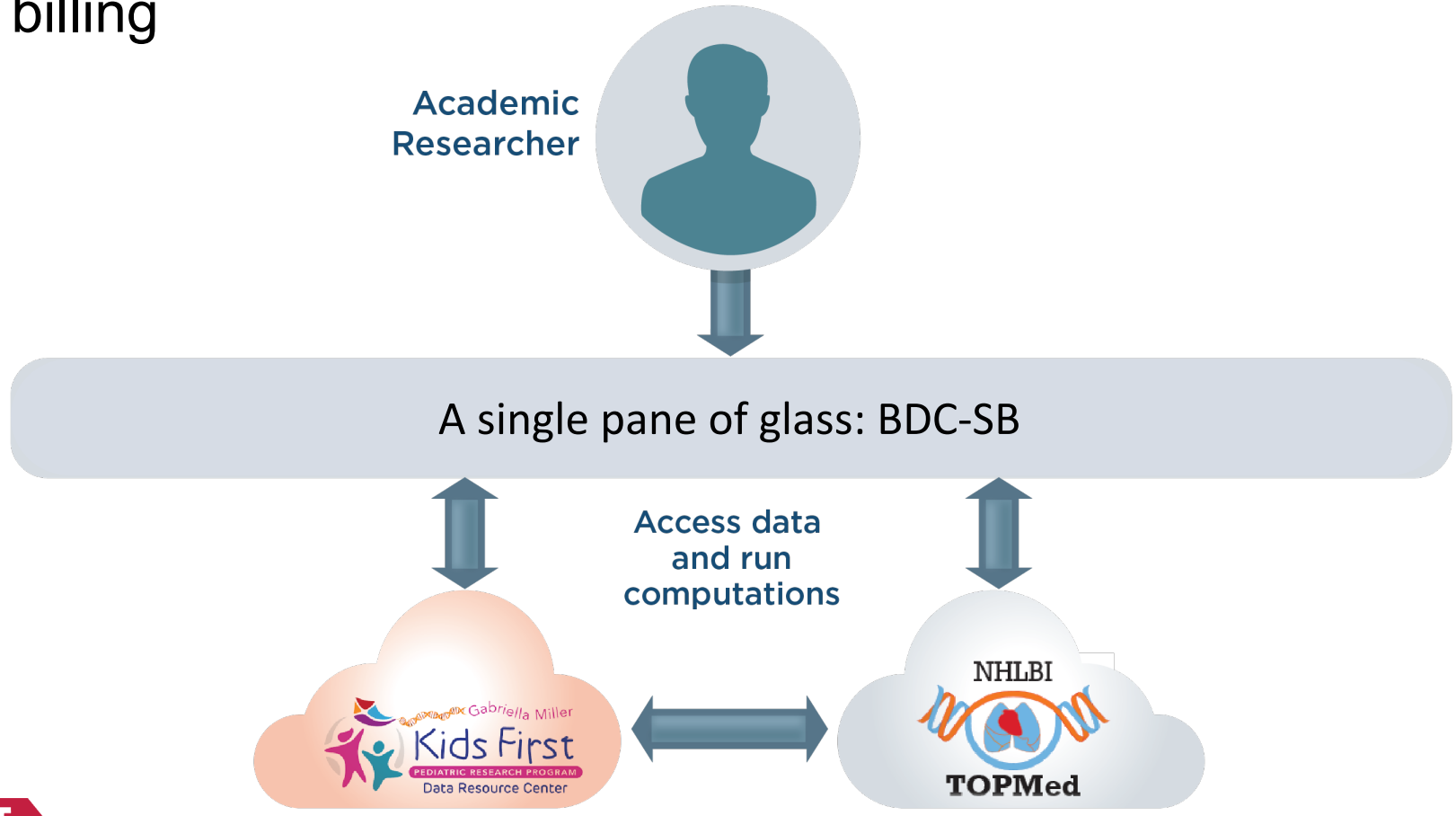
Cera is a Community Engagement Manager at Velsera, where she collaborates with researchers at all stages of their career. She develops tools and approaches to further the impact of scientists on projects like the NHLBI funded BioData Catalyst. Cera enjoys the duality of research: individual focus as well as contribution to a larger team and field. Cera calls upon her vast knowledge of science education, adult training techniques and the research journey on all her projects. And she still loves bugs, computers, science, people and making a difference. Not necessarily in that order.

Cloud platform challenges

- Using data where it lives (if it can't be transferred)
- Transferring data (multiple copies, downloads and egress charges)
- Billing issues (different platform, different charges)

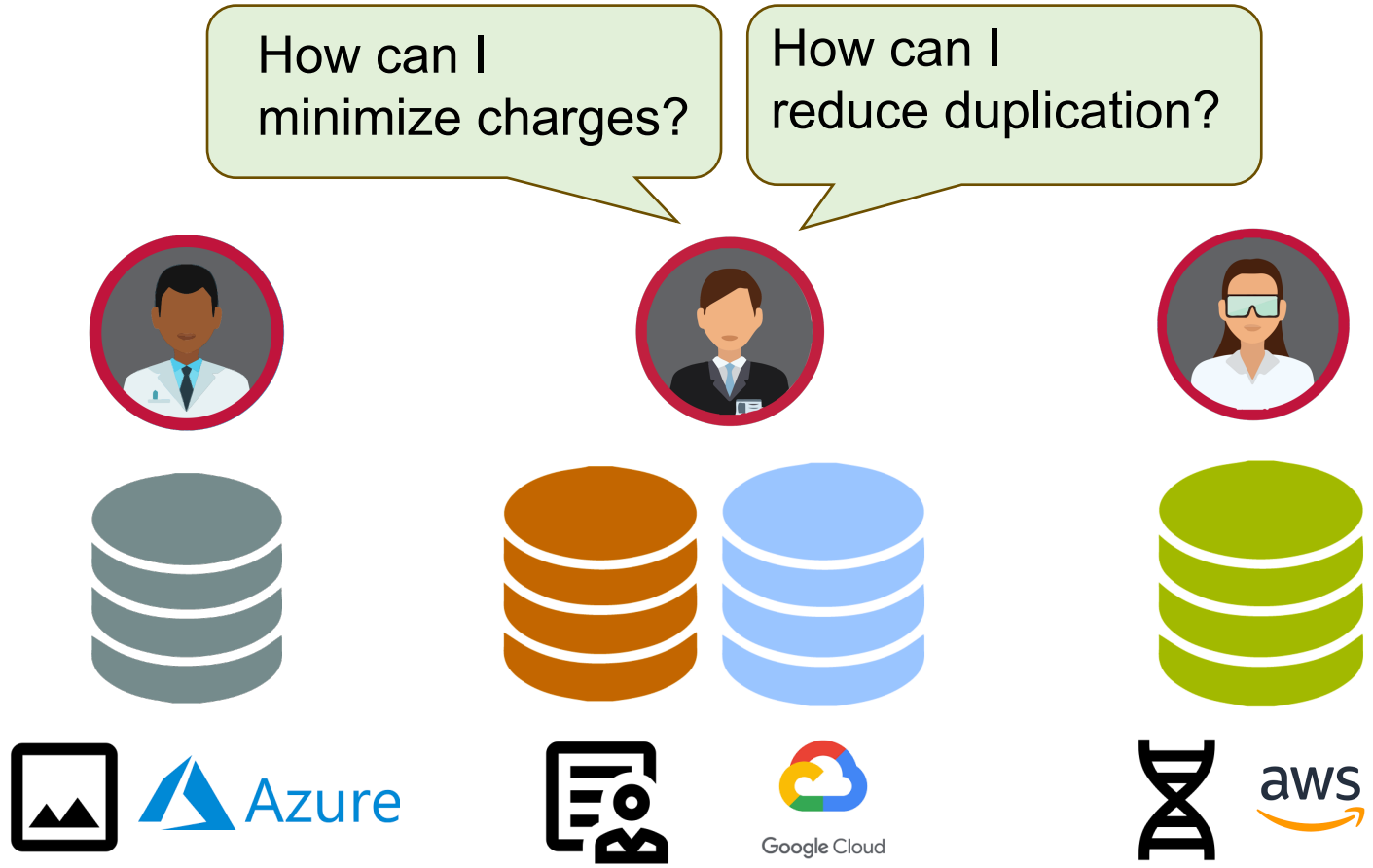
Use data where it lives

- Collaborate in the space where data lives
- Avoid/minimize egress charges
- Unified billing



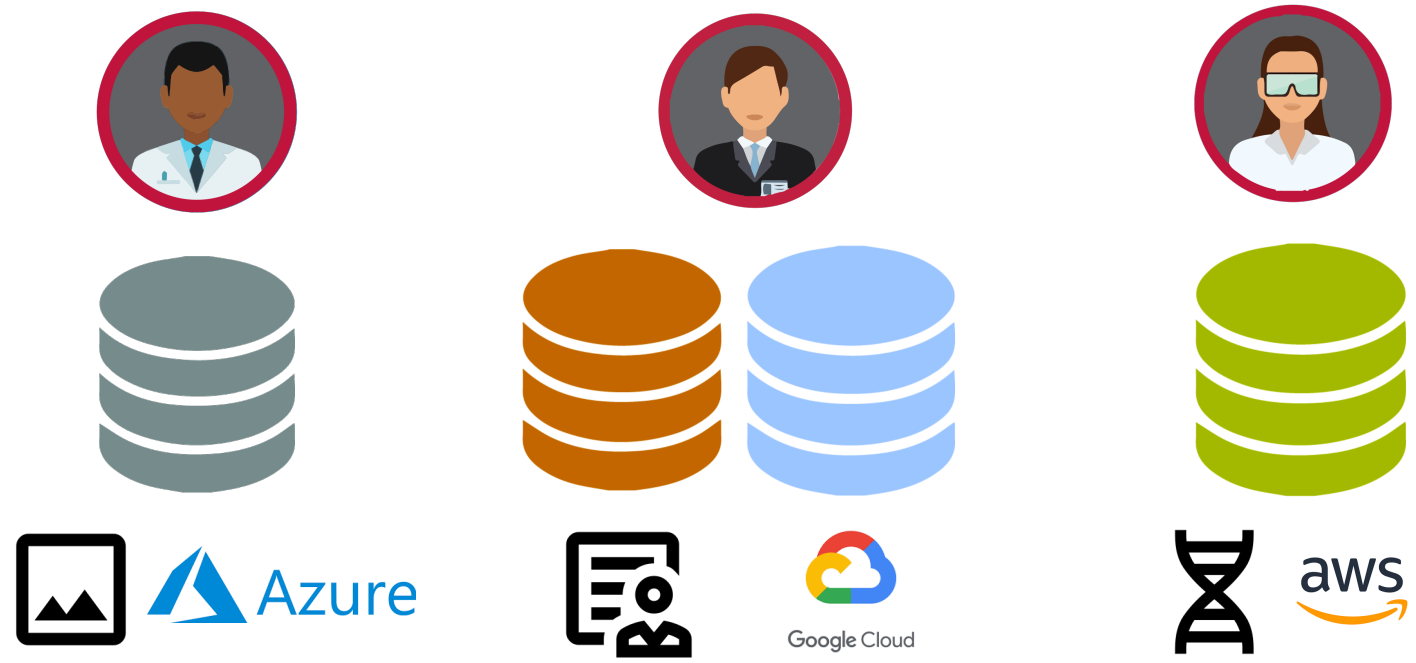
Use Case: Co-Analyzing Data

Dr Collins wants to analyze clinical, genomic and clinical data stored in multiple locations.



Work on data where it lives

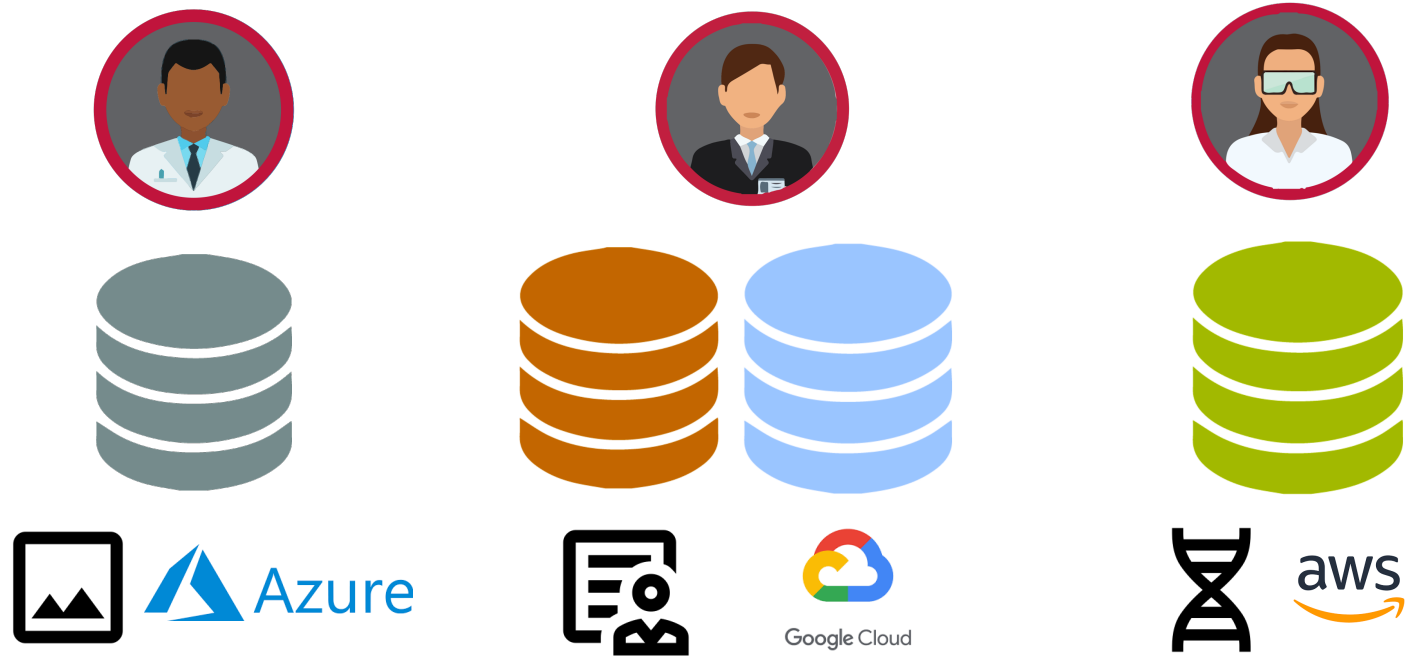
- Big data only *starts* big
- Process down to smaller files
- Store raw data in “cold” data storage



Choose the co-analysis environment

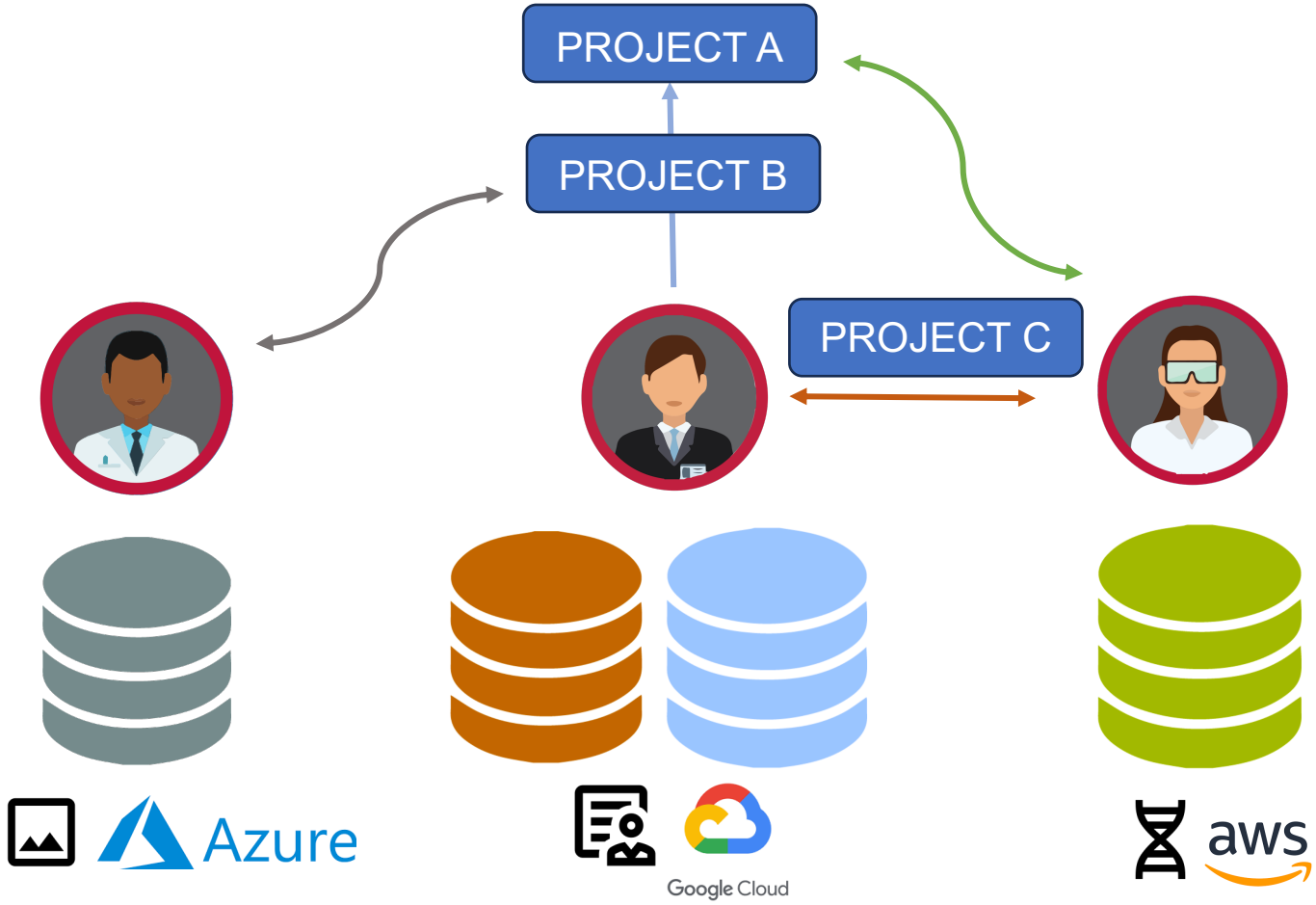
Determine optimal cloud environment priority:

- Security
- Cost
- Comfort



Co-analyze derived files

Move smaller, derived files to chosen location



Utilize three cloud locations with

BDC *Powered By Seven Bridges*

US-west1

US-east-1

South Central US

Demonstration

Insights from GWAS: Linking Genetics and Imaging in Diverse Cohorts

Jose D. Vargas, M.D., D.Phil
Cardiologist
DC Veterans' Affairs Administration

Dr. Jose Vargas currently works at the US Department of Veterans Affairs (VA) and has active collaborations and appointments at Johns Hopkins Hospital, Georgetown University Hospital, The National Institutes of Health (NIH) and Queen Mary University (London).

His research focuses on understanding the genomic basis of cardiovascular disease by leveraging advanced, non-invasive cardiovascular imaging techniques and genomics in large-scale cohorts such as the Multiethnic Study of Atherosclerosis (MESA), the UK Biobank, the Million Veteran Program (MVP), and the Study of Latinos (SOL).

NIH National Heart, Lung, and Blood Institute | BioData CATALYST

A three-part conversation:

- Overview of disparities in current genetic studies
- Importance of diversity in genomic studies
- Future directions in precision medicine

We will pause for questions/discussion after each session. Please put your questions and comments in the chat.

Disparities in current genomic studies

Jose D. Vargas, M.D., D.Phil
Cardiologist
DC Veterans' Affairs Administration

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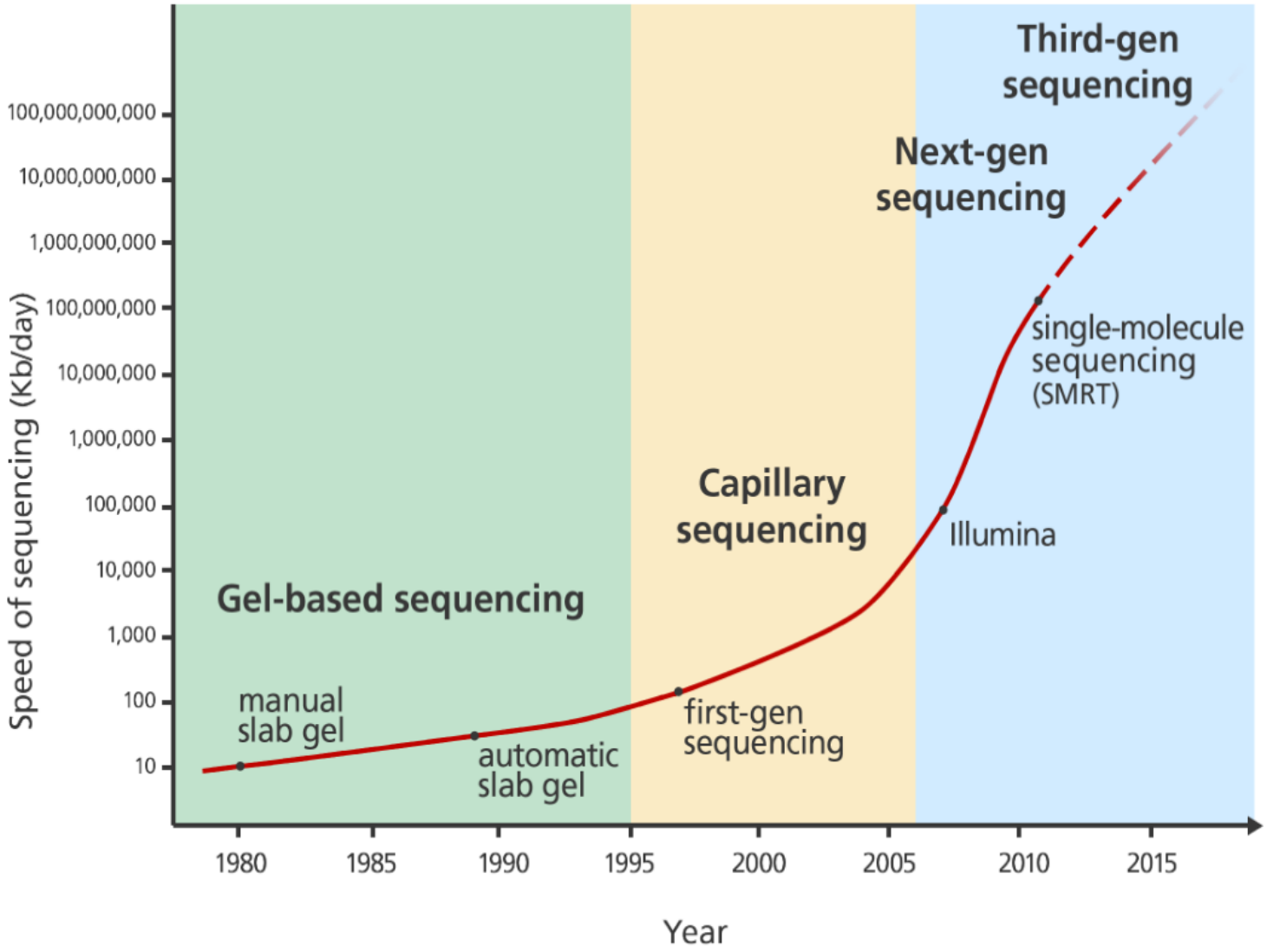
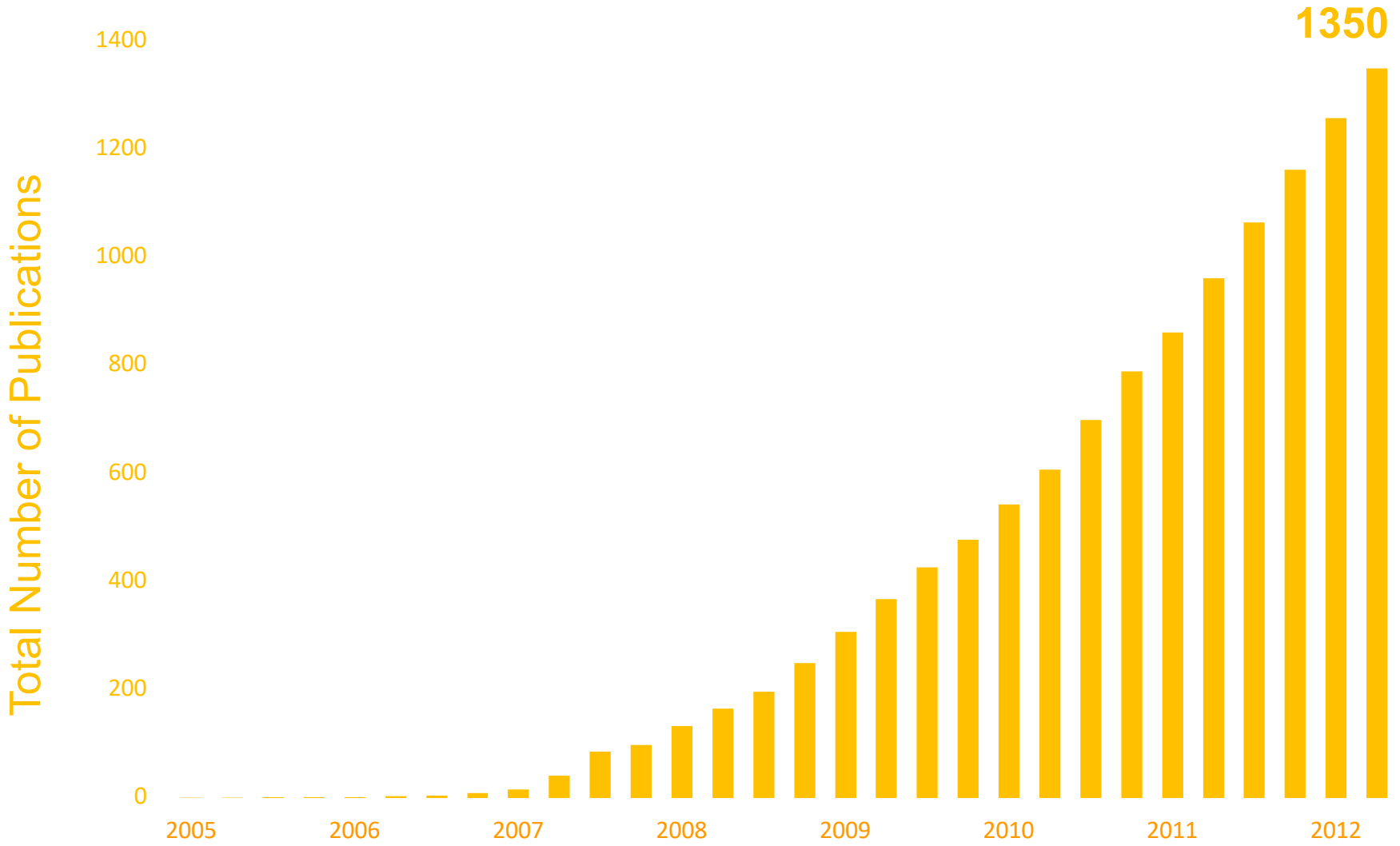
THE PRECISION MEDICINE INITIATIVE

Genome Sequencing Generations



http://ifigure.de/exponential-progress-sequencing-basis-precision-medicine.html

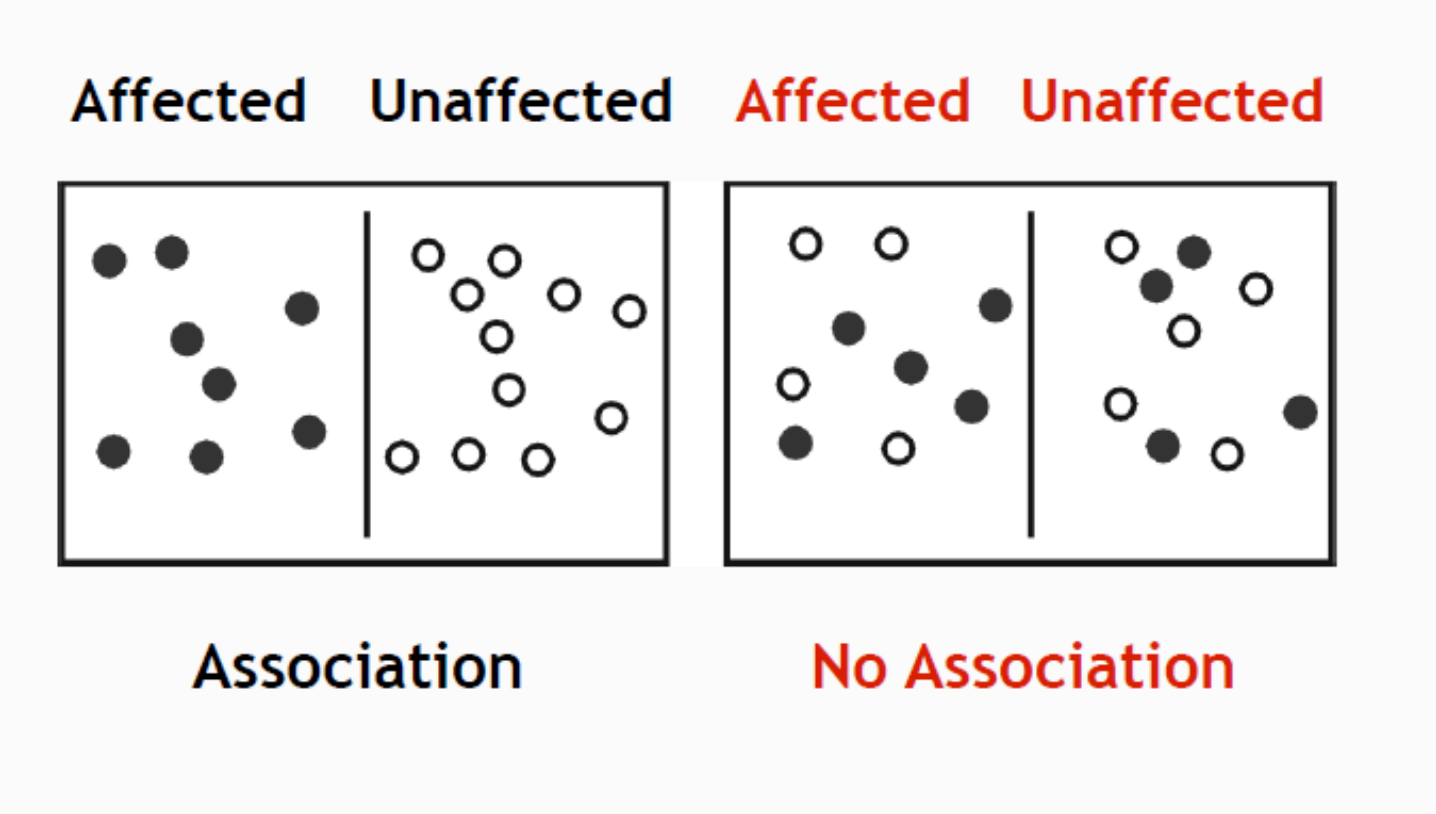
Published Genetic Assocation Studies



www.genome.gov

Genetic Variation

Genome Wide Association Studies



GWAS of CAD

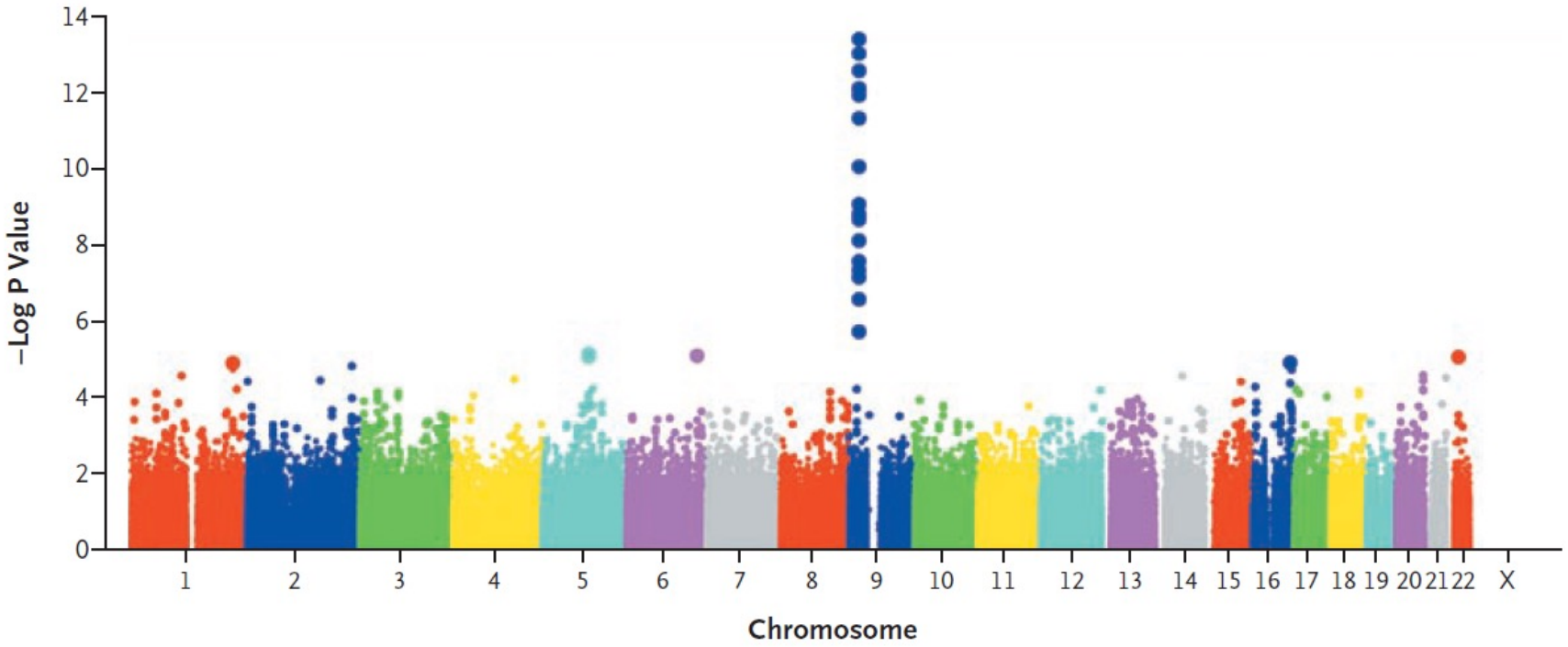
Genomewide Association Analysis of Coronary Artery Disease

Genome-Wide Association Study for Coronary Artery Calcification With Follow-Up in Myocardial Infarction

A Common Variant on Chromosome 9p21 Affects the Risk of Myocardial Infarction
Anna Helgadottir *et al.*
Science **316**, 1491 (2007);
DOI: 10.1126/science.1142842

A Common Allele on Chromosome 9 Associated with Coronary Heart Disease
Ruth McPherson, *et al.*
Science **316**, 1488 (2007);
DOI: 10.1126/science.1142447

GWAS of CAD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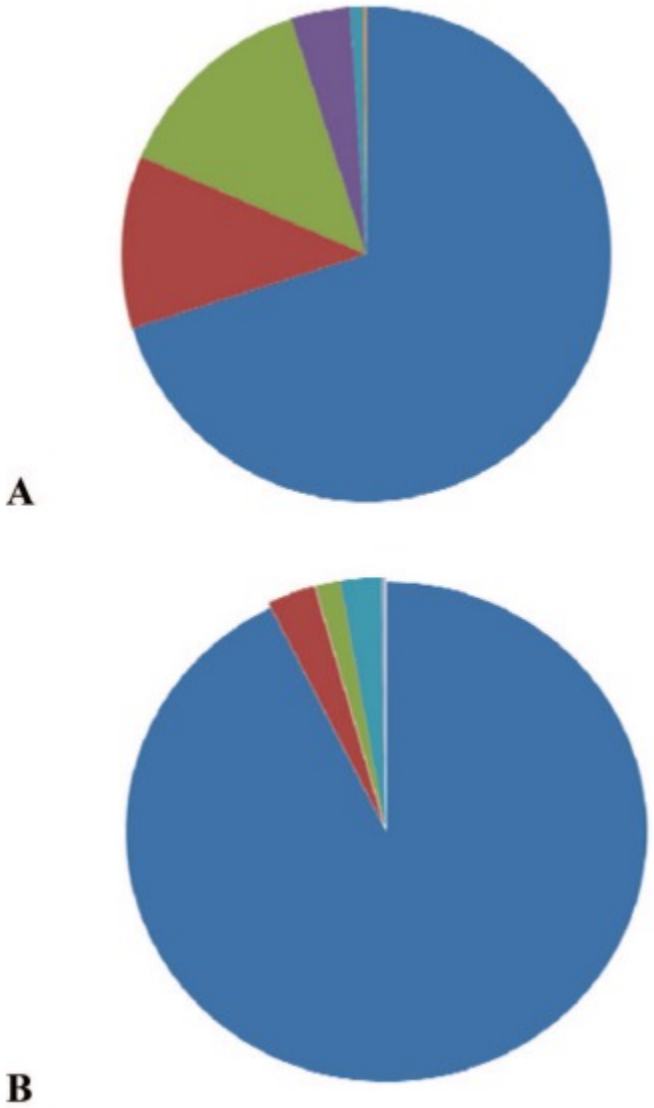
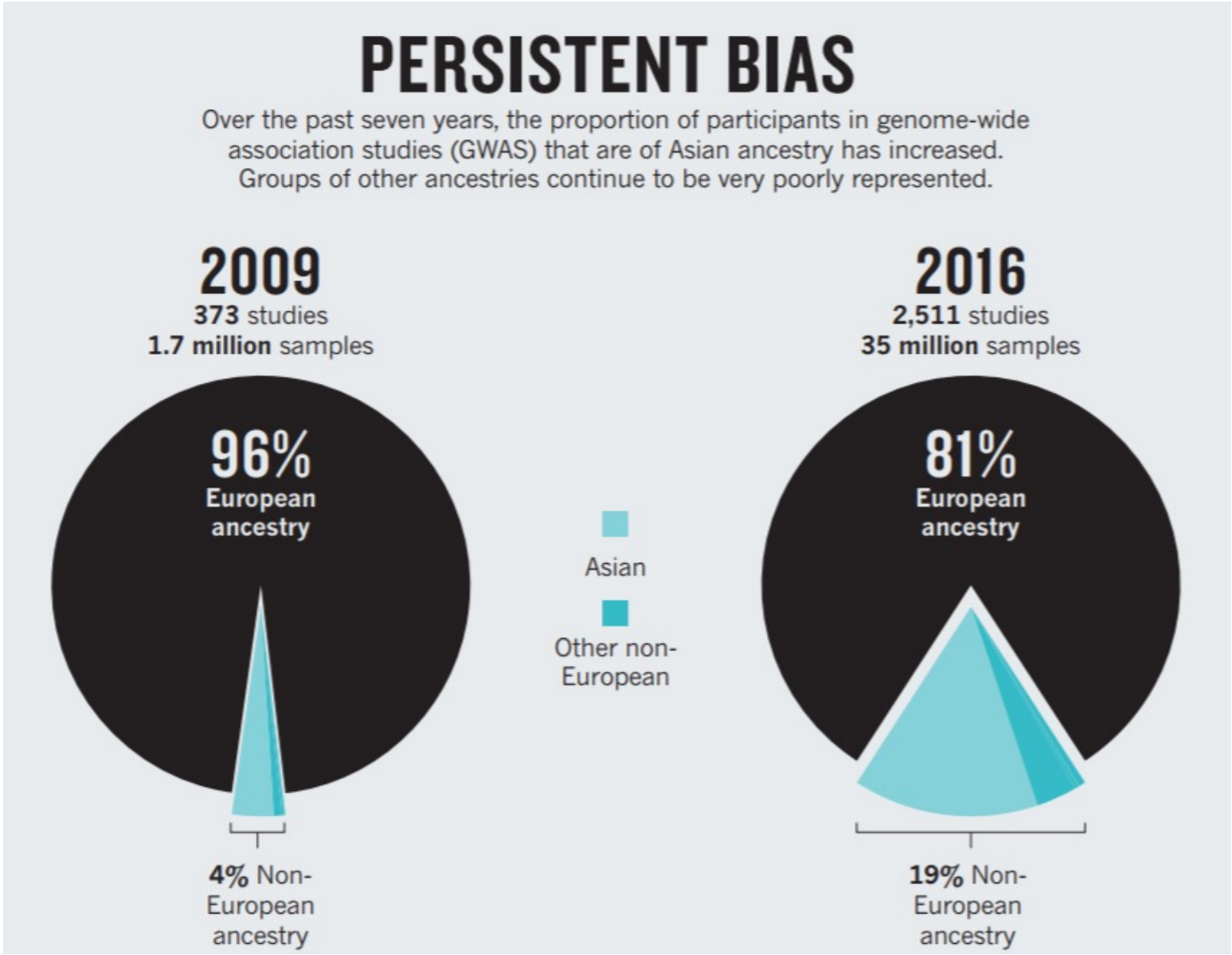
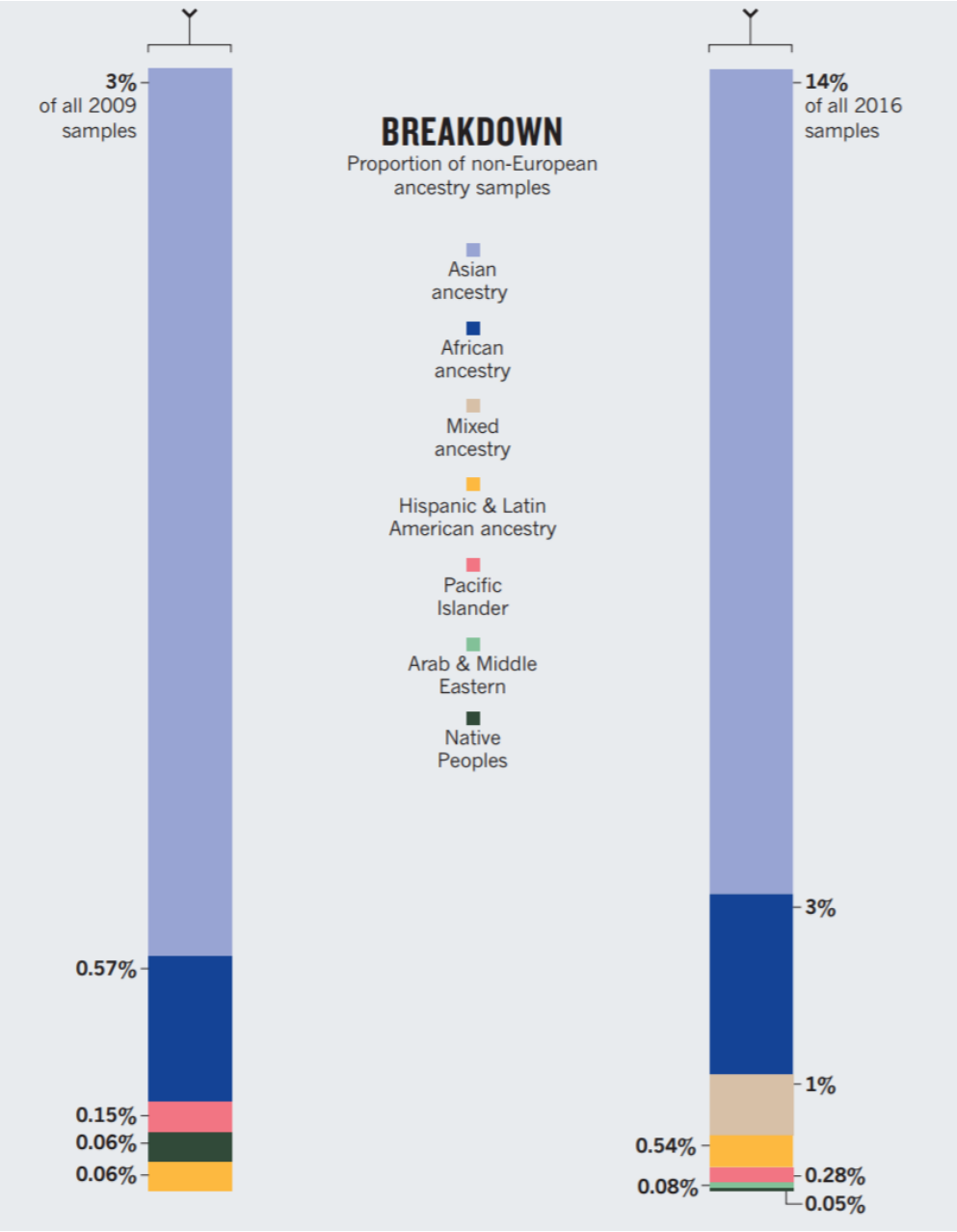
Limited of Diversity in GWAS



Haga SB Genet Med 2010:12(2):81– 84.



Popejoy AB, Fullerton SM Nature 2016 Oct 13; 538(7624) :161–164.



Popejoy AB, Fullerton SM Nature 2016 Oct 13; 538(7624) :161–164.

Pause for questions and comments

The Importance of Diversity in Genetic Studies

Jose D. Vargas, M.D., D.Phil
Cardiologist
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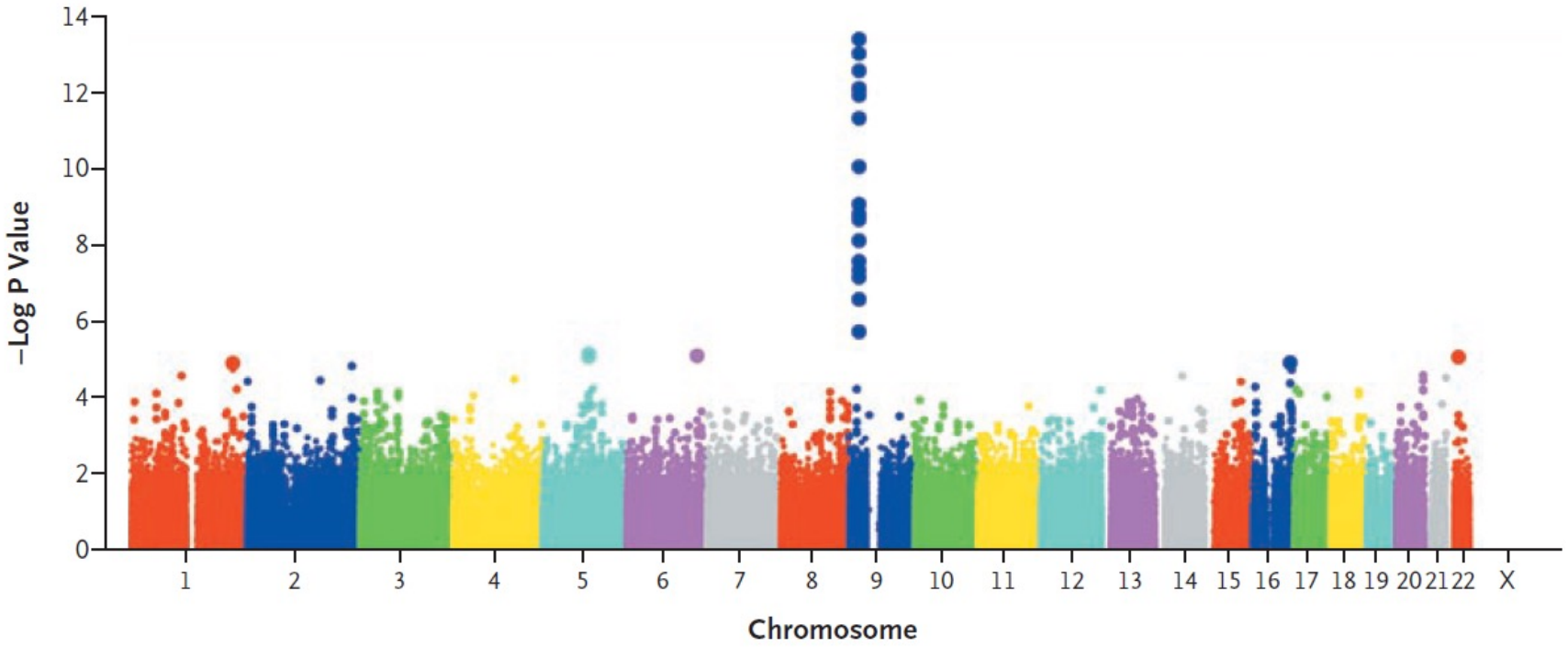
GWAS of CAD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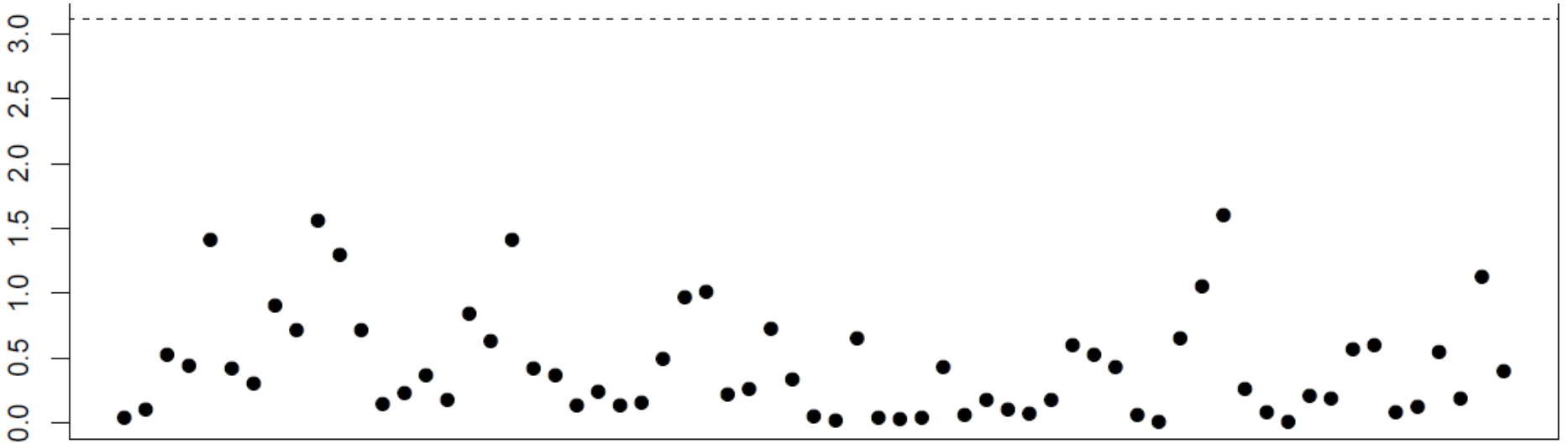
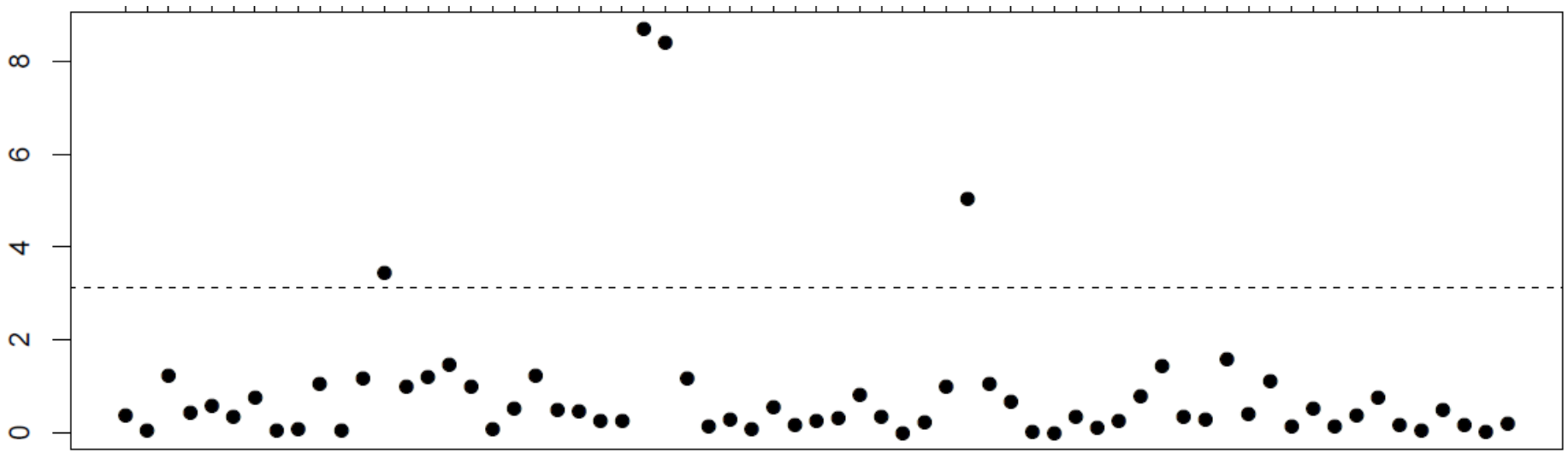
Lack of 9p21 CAD Association in African Americans



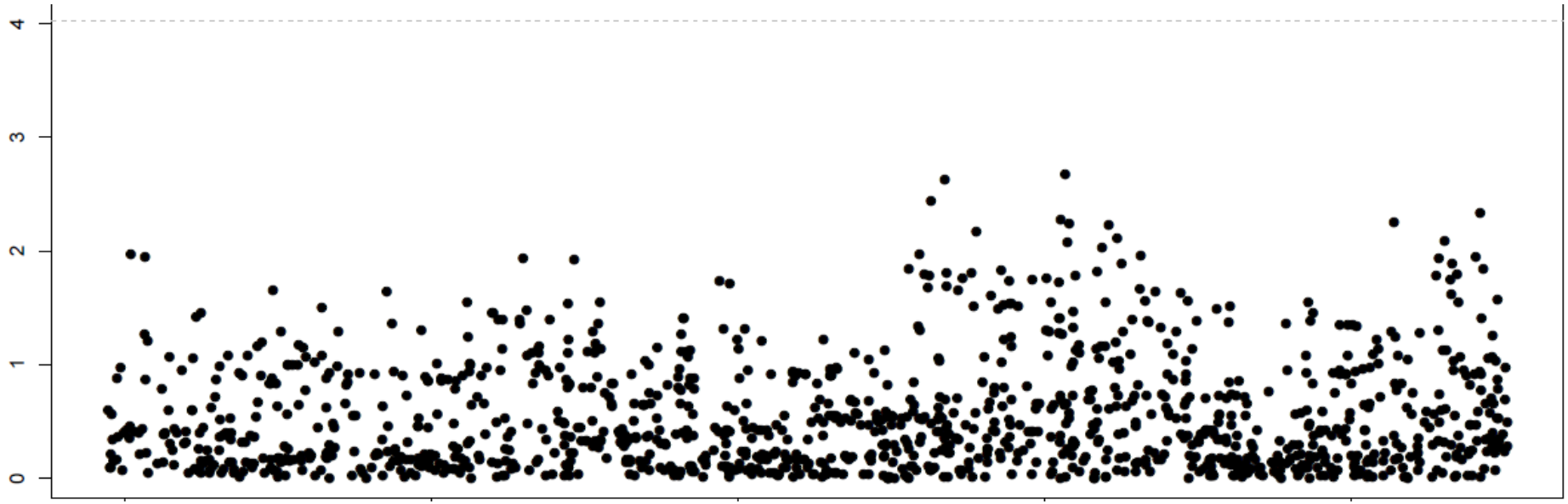
Vargas JD et al. Atherosclerosis. 2016 Feb;245:230-6

Replication of 9p21 CAD Association in Caucasians



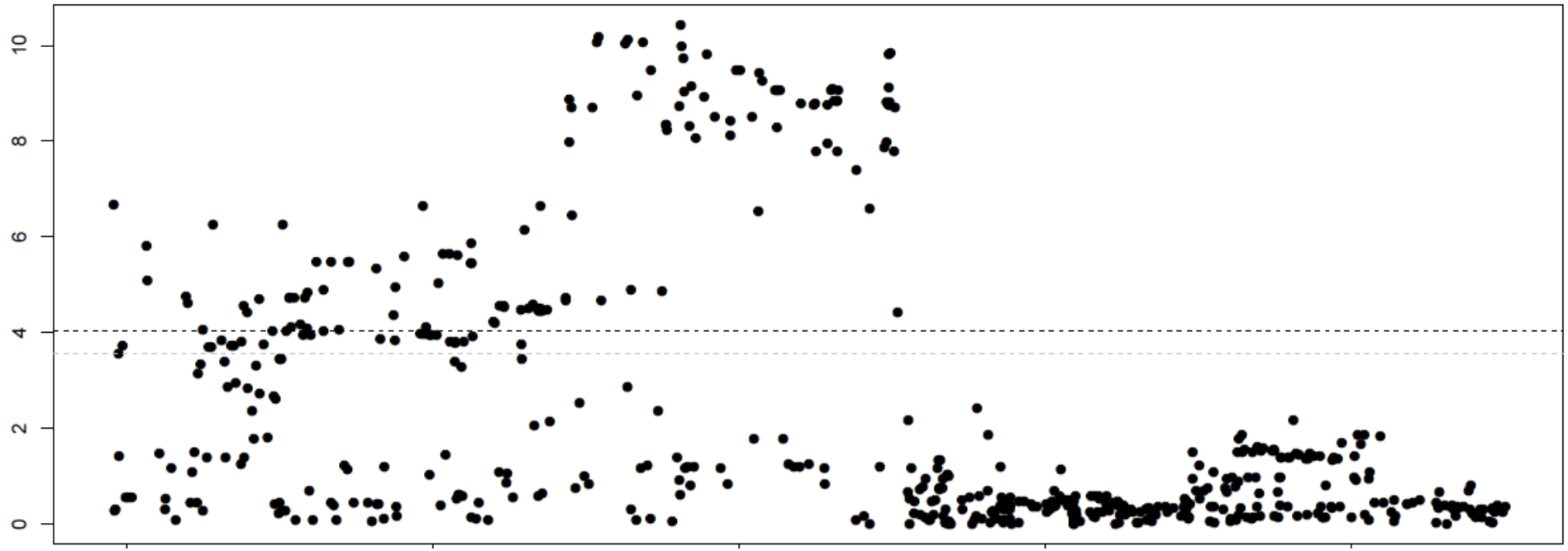
Vargas JD et al. Atherosclerosis. 2016 Feb;245:230-6

Fine Mapping of 9p21 Region in African Americans



Vargas JD et al. Atherosclerosis. 2016 Feb;245:230-6

Fine Mapping of 9p21 Region in Caucasians



Vargas JD et al. Atherosclerosis. 2016 Feb;245:230-6

nature medicine

ARTICLES

https://doi.org/10.1038/s41591-022-01891-3

Large-scale genome-wide association study of coronary artery disease in genetically diverse populations

Catherine Tcheandjieu[1,2,3,4,88] ✉, Xiang Zhu[1,5,6,7,88], Austin T. Hilliard[1,88], Shoa L. Clarke[1,2,88], Valerio Napolioni[8,9], Shining Ma[5], Kyung Min Lee[10], Huaying Fang[11], Fei Chen[12], Yingchang Lu[13], Noah L. Tsao[14], Sridharan Raghavan[15,16], Satoshi Koyama[17], Bryan R. Gorman[18,19], Marijana Vujkovic[20,21], Derek Klarin[1,18,22,23,24,25], Michael G. Levin[20,21], Nasa Sinnott-Armstrong[1,11], Genevieve L. Wojcik[26], Mary E. Plomondon[27,28], Thomas M. Maddox[29,30], Stephen W. Waldo[27,28,31], Alexander G. Bick[32], Saiju Pyarajan[18,33], Jie Huang[18,34,35], Rebecca Song[18], Yuk-Lam Ho[18], Steven Buyske[36], Charles Kooperberg[37], Jeffrey Haessler[37], Ruth J. F. Loos[38], Ron Do[38,39], Marie Verbanck[38,39,40], Kumardeep Chaudhary[38,39], Kari E. North[41], Christy L. Avery[41], Mariaelisa Graff[41], Christopher A. Haiman[12], Loïc Le Marchand[42], Lynne R. Wilkens[42], Joshua C. Bis[43], Hampton Leonard[44,45], Botong Shen[46], Leslie A. Lange[47,48,49], Ayush Giri[50,51], Ozan Dikilitas[52], Iftikhar J. Kullo[52], Ian B. Stanaway[53], Gail P. Jarvik[54,55], Adam S. Gordon[56], Scott Hebbring[57], Bahram Namjou[58,59], Kenneth M. Kaufman[58], Kaoru Ito[17], Kazuyoshi Ishigaki[60], Yoichiro Kamatani[60,61], Shefali S. Verma[62,63], Marylyn D. Ritchie[62,63], Rachel L. Kember[20,64], Aris Baras[65], Luca A. Lotta[65], Regeneron Genetics Center*, CARDIoGRAMplusC4D Consortium*, Biobank Japan*, Million Veteran Program*, Sekar Kathiresan[23,66,67,68], Elizabeth R. Hauser[69,70], Donald R. Miller[71,72], Jennifer S. Lee[1,73], Danish Saleheen[20,74], Peter D. Reaven[75,76], Kelly Cho[18,33], J. Michael Gaziano[18,33], Pradeep Natarajan[23,67,77], Jennifer E. Huffman[18], Benjamin F. Voight[20,62,78,79], Daniel J. Rader[21], Kyong-Mi Chang[20,21], Julie A. Lynch[80,81], Scott M. Damrauer[14,20,62], Peter W. F. Wilson[82,83], Hua Tang[11], Yan V. Sun[84,85,89], Philip S. Tsao[1,73,86,89], Christopher J. O'Donnell[18,33,89] and Themistocles L. Assimes[1,2,86,87,89] ✉

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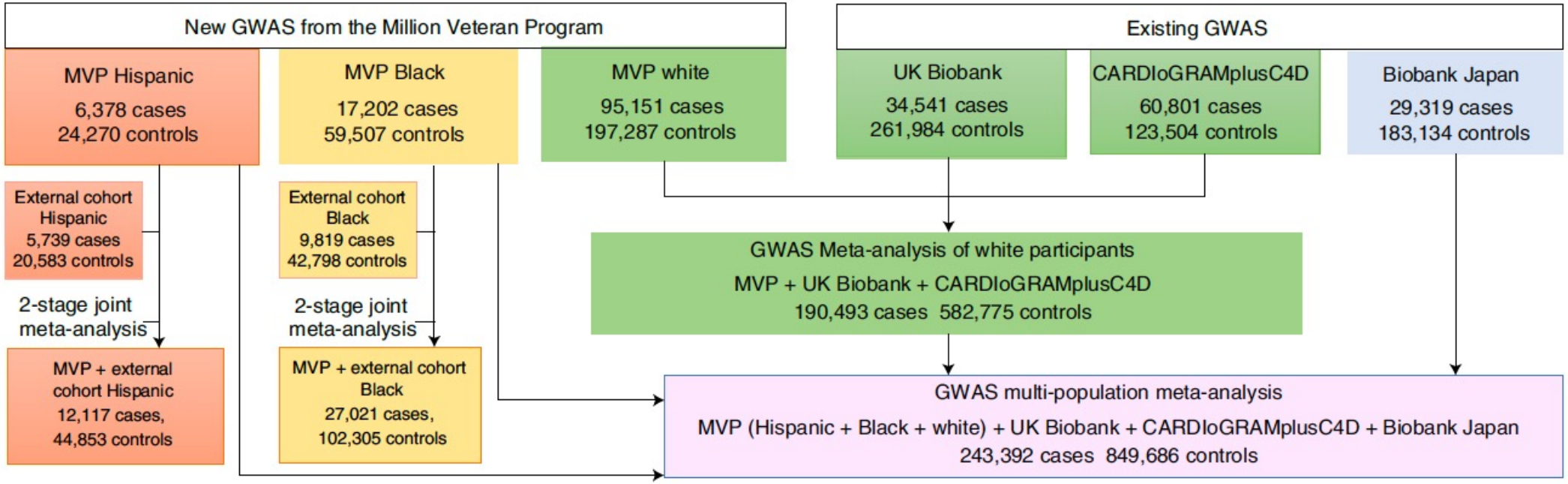
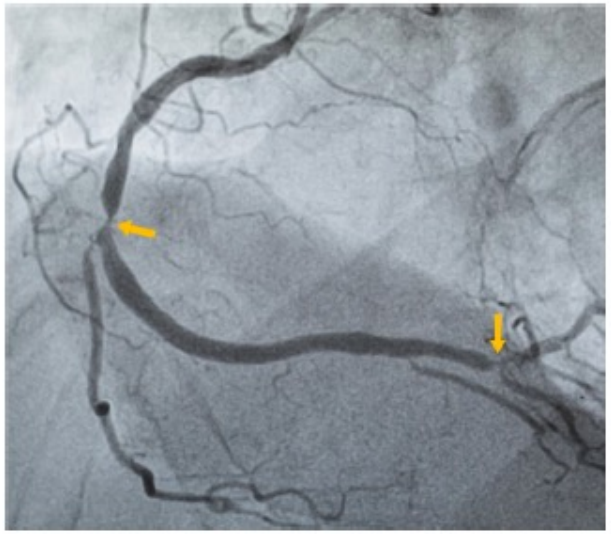
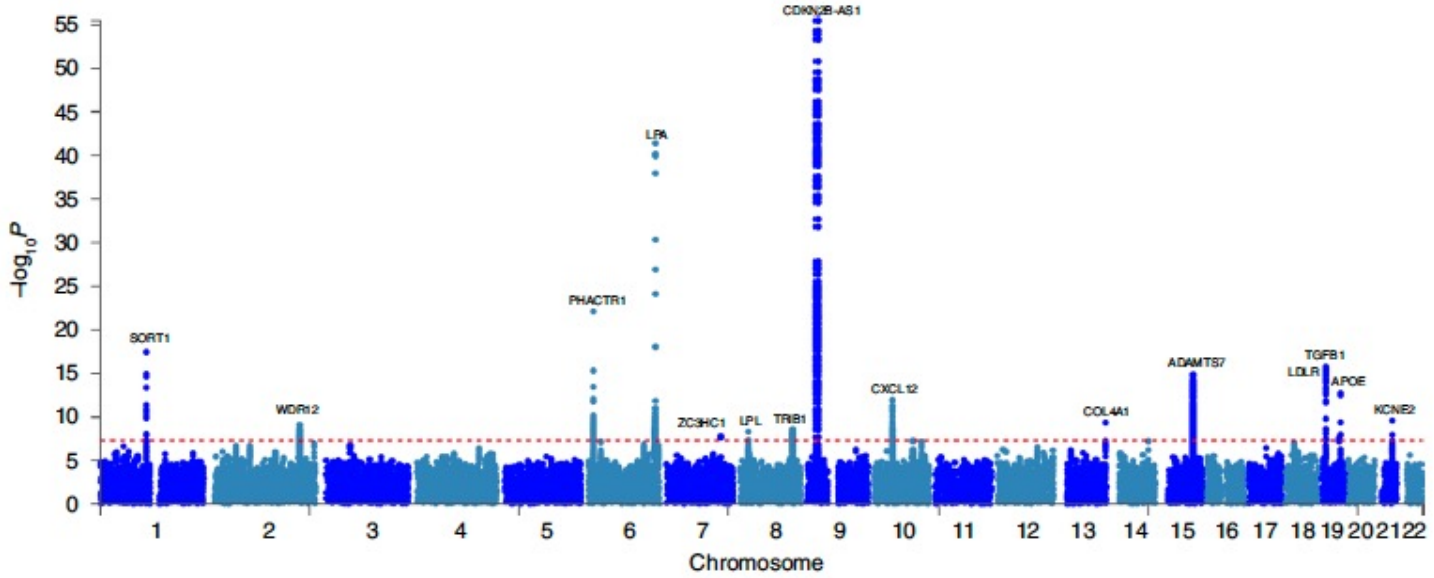

New GWAS from the Million Veteran Program
Existing GWAS
MVP Hispanic
6,378 cases
24,270 controls
MVP Black
17,202 cases
59,507 controls
MVP white
95,151 cases
197,287 controls
UK Biobank
34,541 cases
261,984 controls
CARDIoGRAMplusC4D
60,801 cases
123,504 controls
Biobank Japan
29,319 cases
183,134 controls
External cohort Hispanic
5,739 cases
20,583 controls
External cohort Black
9,819 cases
42,798 controls
2-stage joint meta-analysis
2-stage joint meta-analysis
GWAS Meta-analysis of white participants
MVP + UK Biobank + CARDIoGRAMplusC4D
190,493 cases 582,775 controls
MVP + external cohort Hispanic
12,117 cases,
44,853 controls
MVP + external cohort Black
27,021 cases,
102,305 controls
GWAS multi-population meta-analysis
MVP (Hispanic + Black + white) + UK Biobank + CARDIoGRAMplusC4D + Biobank Japan
243,392 cases 849,686 controls

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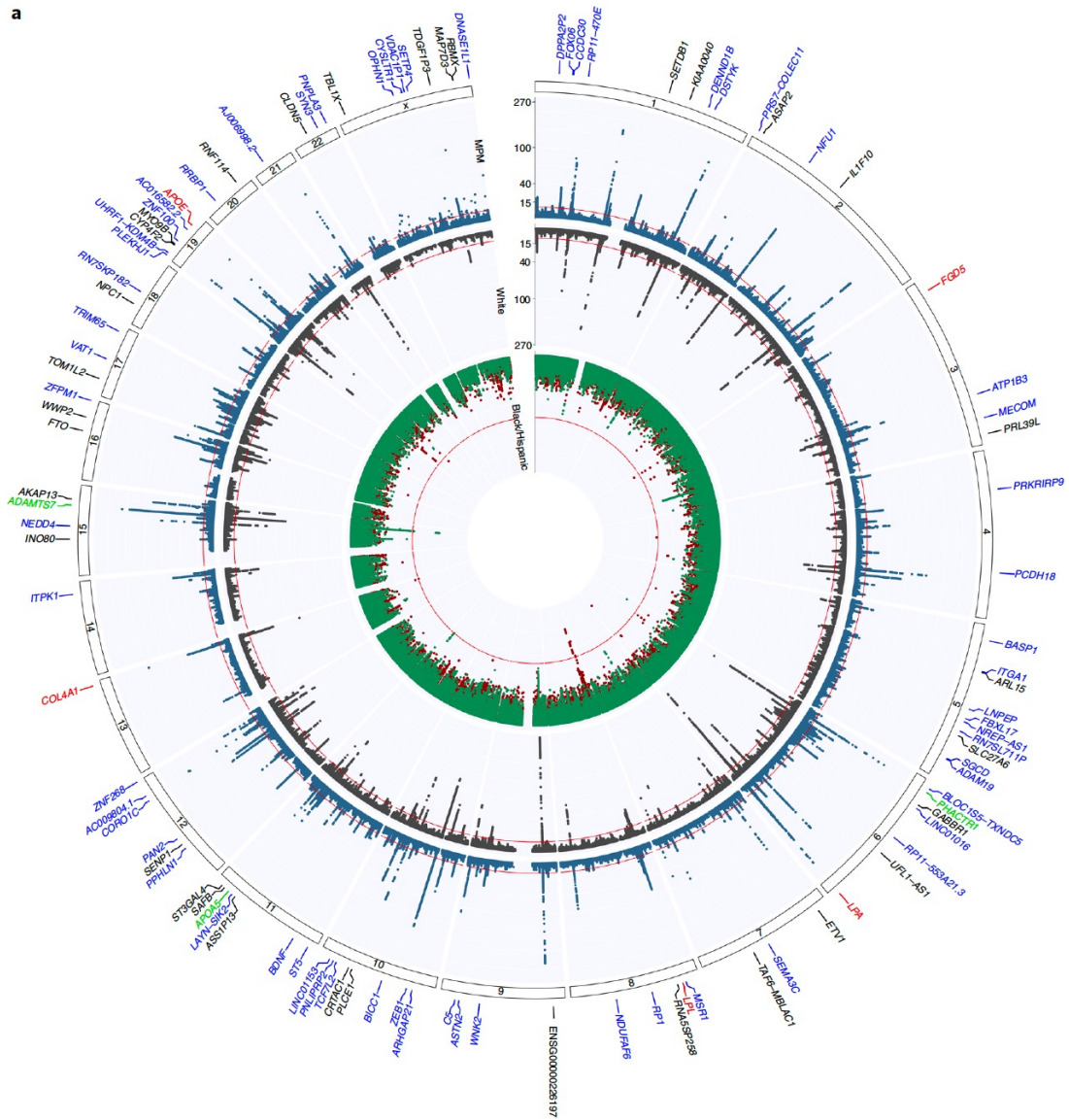
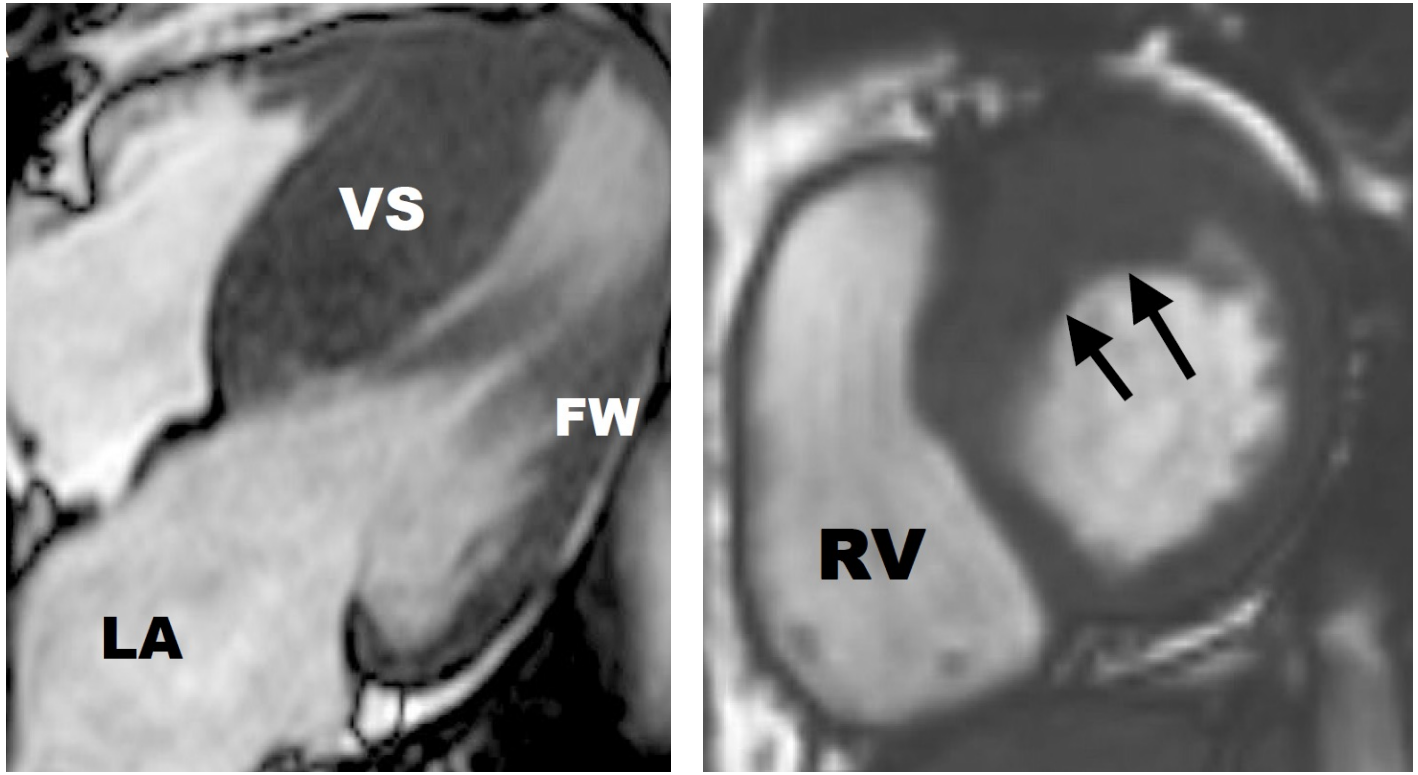
Chromosome



Genetic Misdiagnosis

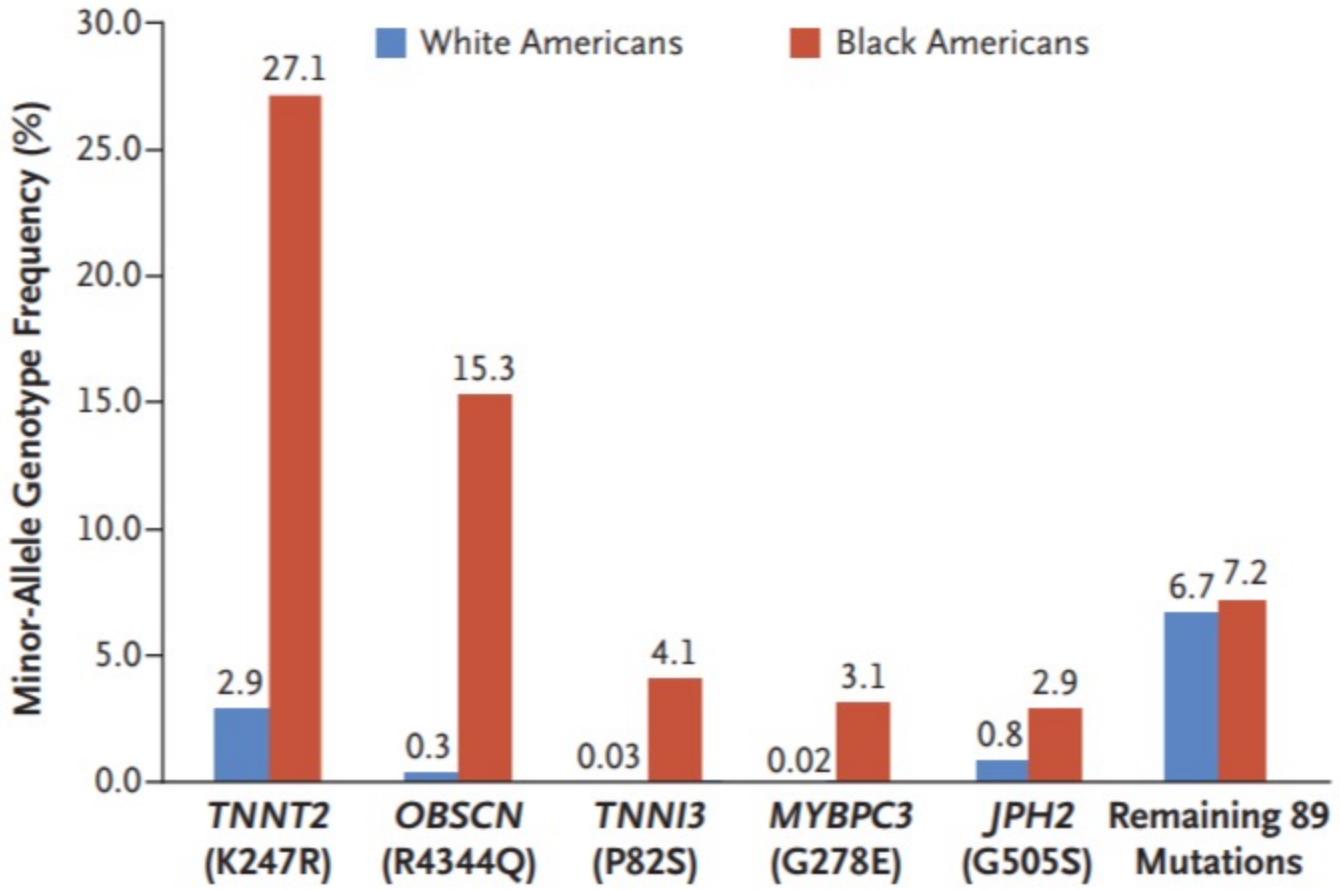


Maron MS JCMR 2012 14:13.

Genetic Misdiagnosis

- Hypertrophic cardiomyopathy consists of unexplained left ventricular hypertrophy
- Can manifest itself as heart failure and/or sudden cardiac death
- A casual genetic variant can be identified in more than one third of patients enabling clinicians to assess risk among relatives.

Genetic Misdiagnosis



Manrai AK NEJM 2016; 375:655-665.

LETTER

https://doi.org/10.1038/s41586-019-1310-4

Genetic analyses of diverse populations improves discovery for complex traits

514 | NATURE | VOL 570 | 27 JUNE 2019

The Population Architecture using Genomics and Epidemiology (PAGE) Study

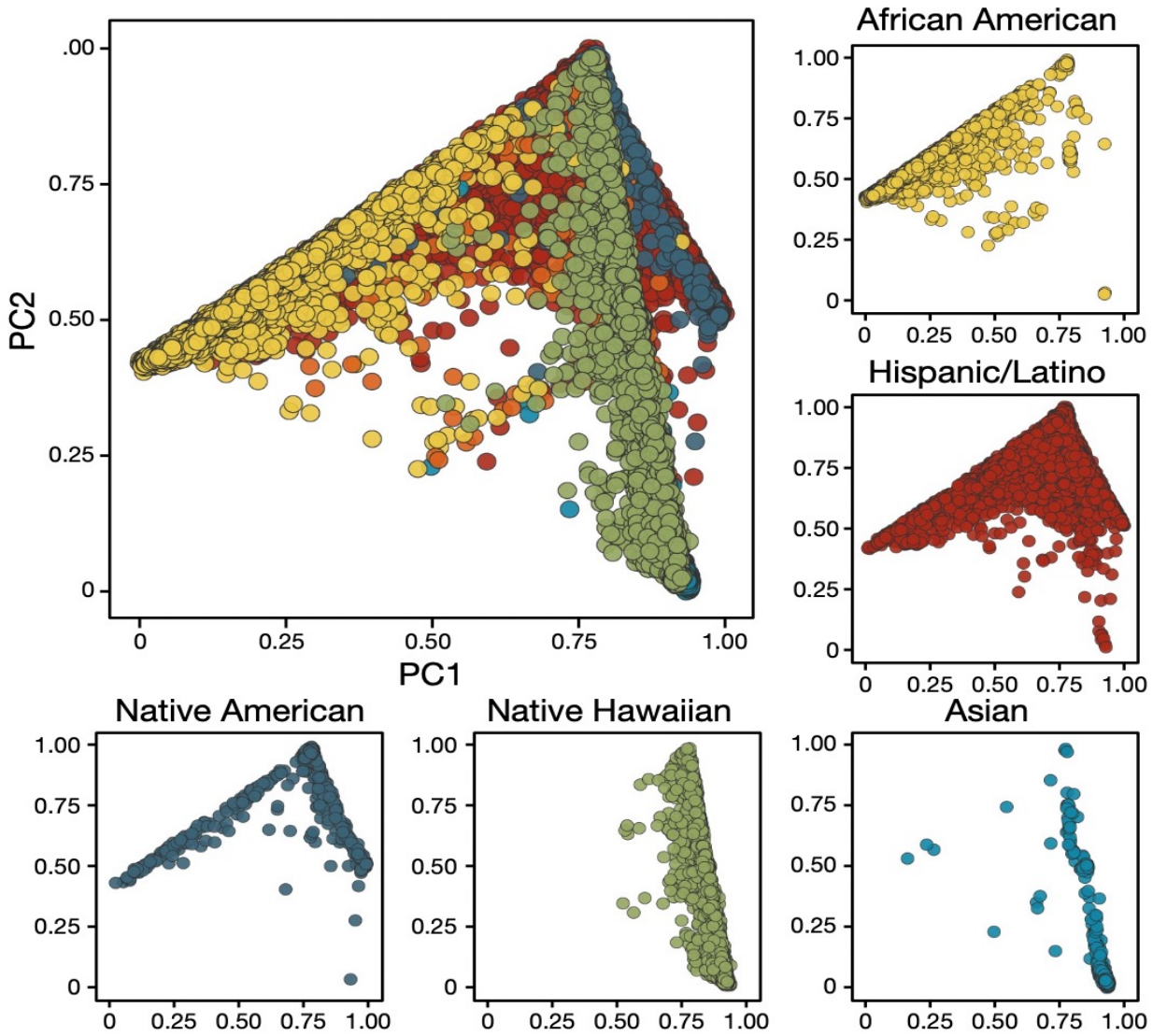
- Established by the National Human Genome Research Institute and the National Institute on Minority Health Disparities

- GWAS of 26 clinical and behavioural phenotypes in 49,839 non-European individuals

The Population Architecture using Genomics and Epidemiology (PAGE) Study

Composed of participants from

- Hispanic Community Health Study/Study of Latinos (HCHS/SOL),
- Women's Health Initiative (WHI)
- Multiethnic Cohort (MEC) and
- the Icahn School of Medicine at Mount Sinai BioMe Biobank in New York City (BioMe)


PC2
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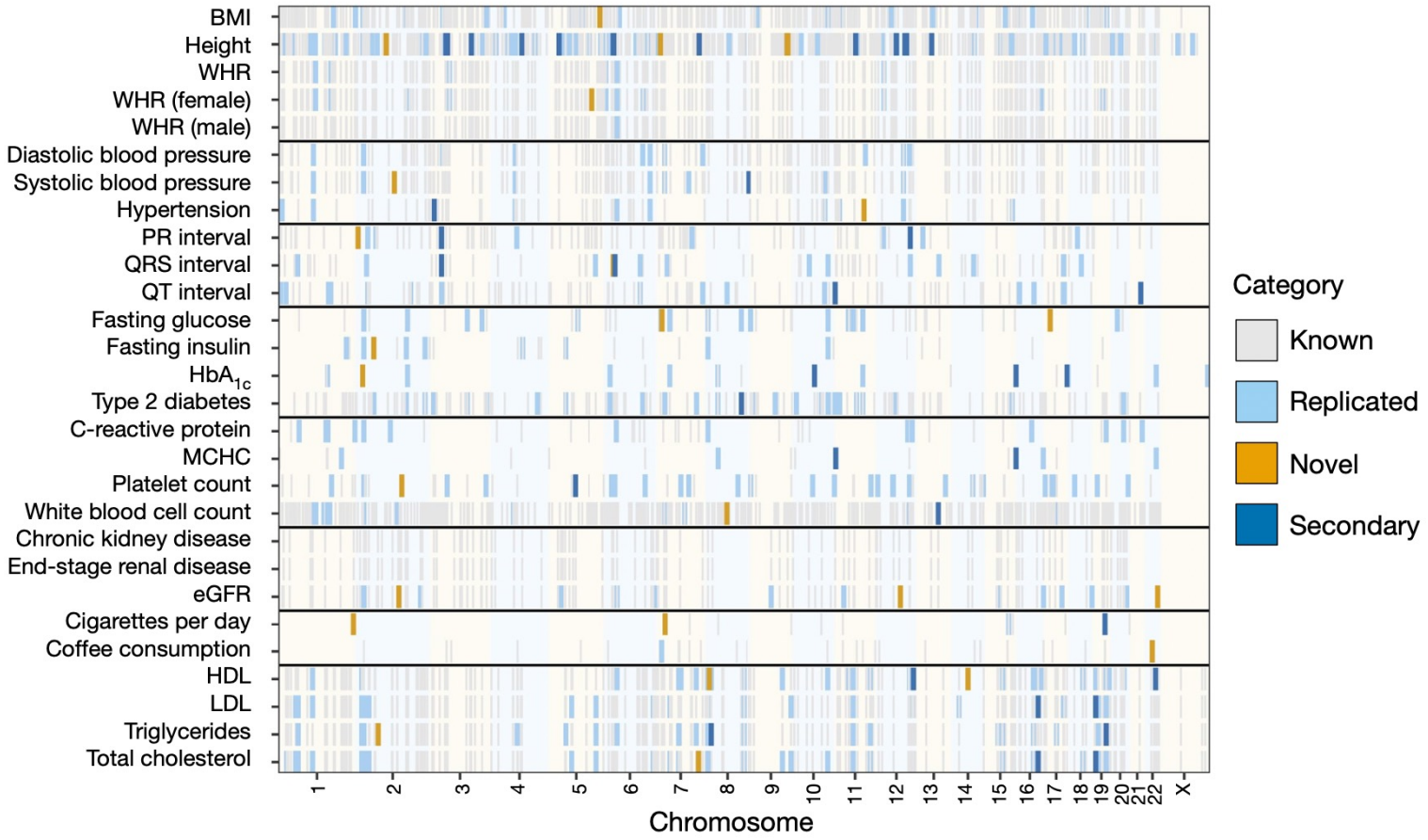
African American
Hispanic/Latino
Native American
Native Hawaiian
Asian

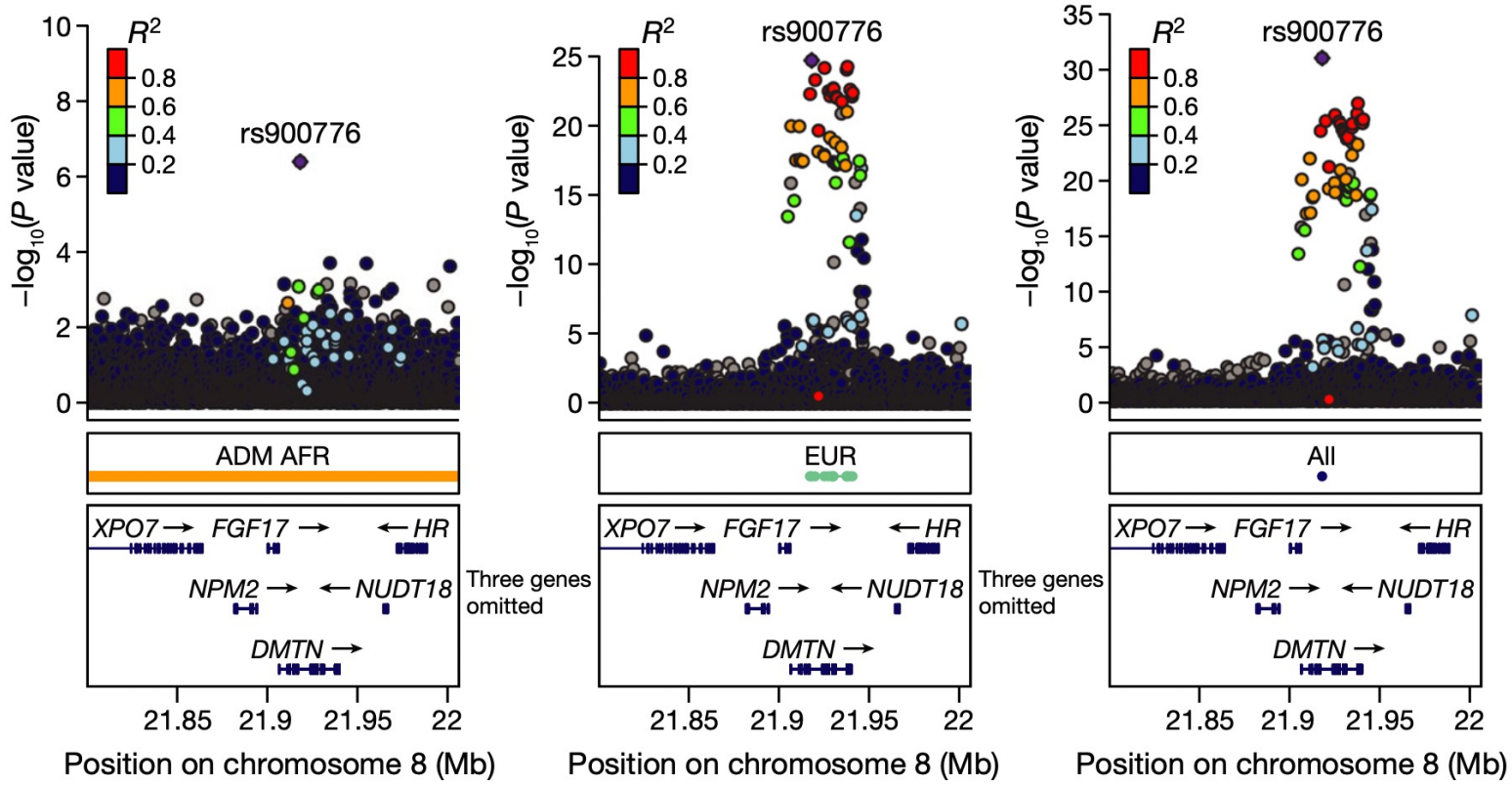


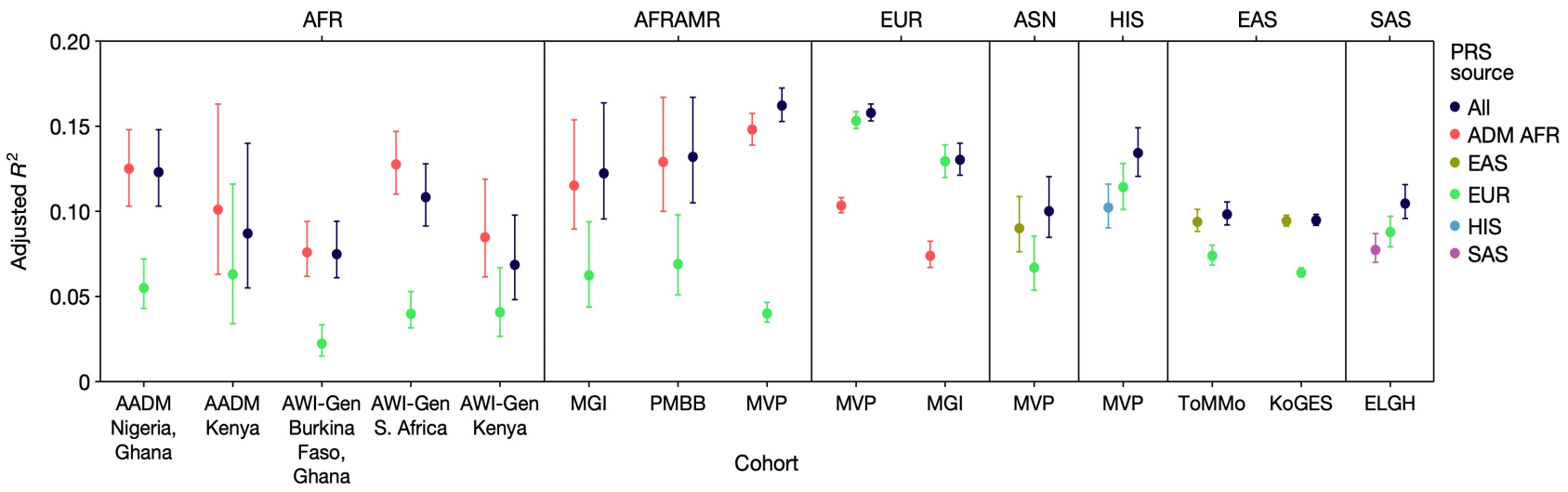
Article

The power of genetic diversity in genome-wide association studies of lipids

Table 1 | Meta-analysis sample size by ancestry group

Ancestry group	Sample size	No. of cohorts	Mean sample size per cohort (range)	No. of variants
European	1,320,016	146	10,928 (173–389,344)	47 million
East Asian	146,492	40	7,448 (150–131,050)	17 million
Admixed African or African	99,432	19	5,330 (473–62,022)	33 million
Hispanic	48,057	10	6,032 (1,496–22,302)	27 million
South Asian	40,963	7	6,413 (1,796–16,110)	17 million
Total	1,654,960	201		52 million





Pause for questions and comments

Future Directions for Diversity in Genetic Studies

Jose D. Vargas, M.D., D.Phil
Cardiologist
DC Veterans' Affairs Administration

Dr. Jose Vargas currently works at the US Department of Veterans Affairs (VA) and has active collaborations and appointments at Johns Hopkins Hospital, Georgetown University Hospital, The National Institutes of Health (NIH) and Queen Mary University (London).

His research focuses on understanding the genomic basis of cardiovascular disease by leveraging advanced, non-invasive cardiovascular imaging techniques and genomics in large-scale cohorts such as the Multiethnic Study of Atherosclerosis (MESA), the UK Biobank, the Million Veteran Program (MVP), and the Study of Latinos (SOL).

Precision Medicine Initiative

- $130 million to NIH for the development of a voluntary national research cohort (one million or more participants)
- $70 million to the National Cancer Institute (NCI), part of the NIH, to scale up efforts to identify genomic drivers in cancer.
- $10 million to FDA to develop the regulatory infrastructure needed in the age of precision medicine.

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H3Africa

Human Heredity & Health in Africa

AN EVOLVING CONSORTIUM

The Human Heredity and Health in Africa (H3Africa) Initiative aims to develop a network of African labs that can do cutting-edge genomics and precision-medicine research with funding from the US National Institutes of Health (NIH) and the Wellcome Trust.

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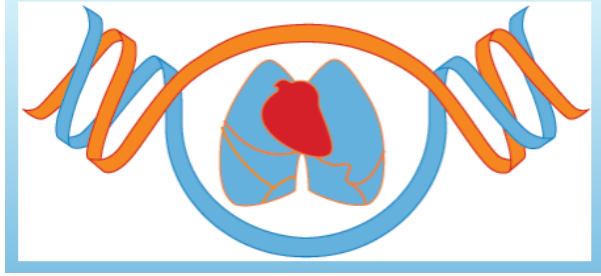
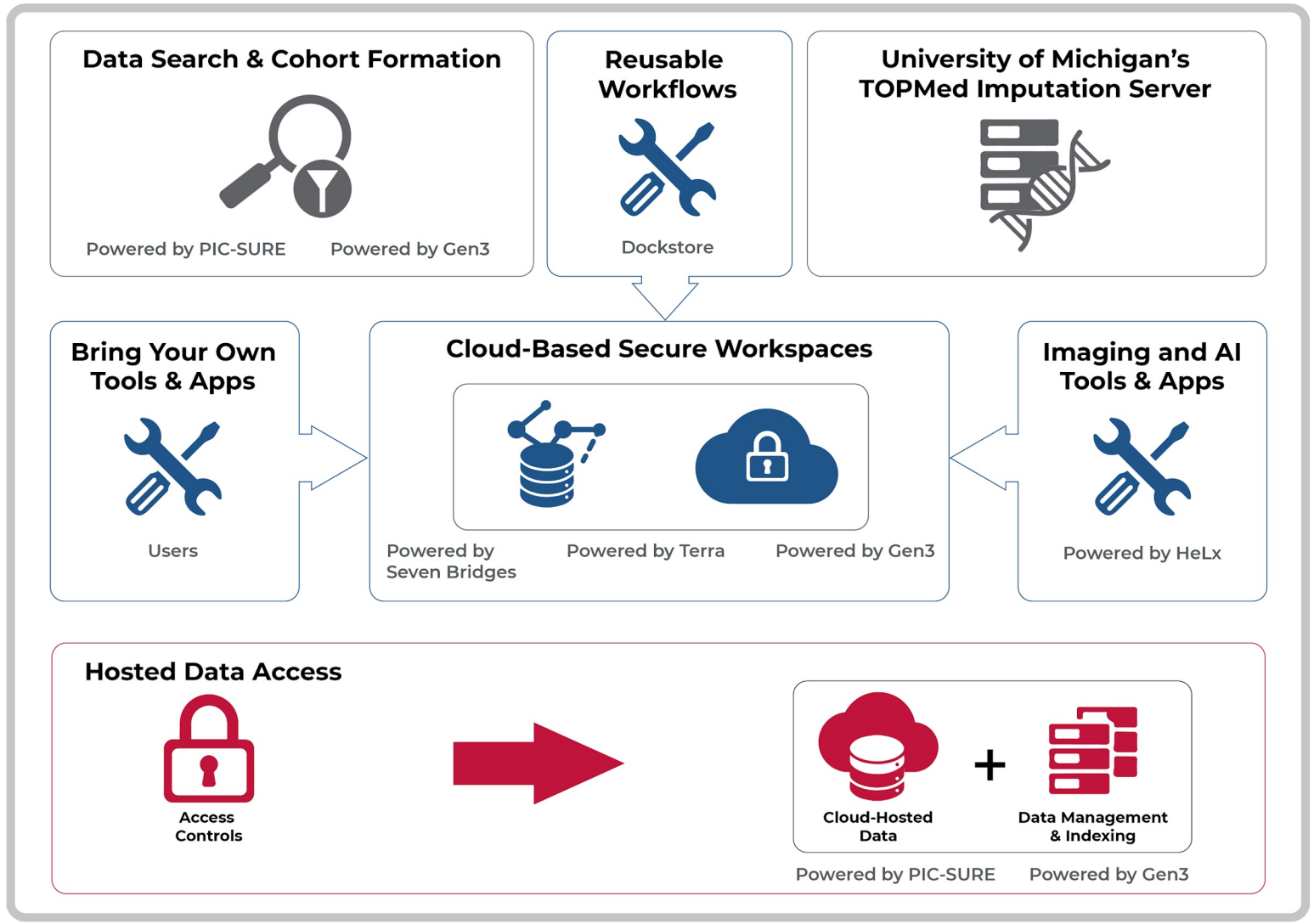


NHLBI Trans-Omics for Precision Medicine Whole Genome Sequencing Program

- The Trans-Omics for Precision Medicine (TOPMed) program, sponsored by the National Institutes of Health (NIH) National Heart, Lung and Blood Institute (NHLBI), is part of a broader Precision Medicine Initiative, which aims to provide disease treatments tailored to an individual's unique genes and environment.
- As of September 2021, TOPMed consists of ~180k participants from >85 different studies with varying designs
- Currently, 60% of the 180k sequenced participants are of predominantly non-European ancestry.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Powered by HeLx
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Cloud-Hosted Data
+
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Thank you.

Coming up
Wednesday March 13th at 1PM ET

Community Hours: BDC Data Update

Sweta Ladwa, MPH
NHLBI BDC Data Management Core
Scientific Program Director

An email invitation is coming to your inbox this week!

Until next time:

- We'll email you the materials from this session. Please share them!
- March is Women's History Month. Join us in celebrating women in science!
- Register for the next BDC Community Hours.
- Use our #BioDataCatalyst on LinkedIn and Twitter/X when/if you post about BDC.