Precision Medicine for All: Ensuring Diversity in Participants and in Practice

Case Western Reserve University
Thursday September 29, 2016

Biorepositories, data collection, and data analysis in US territories

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U.S. Precision Medicine Initiative

"I want the country that eliminated polio and mapped the human genome to lead a new era of medicine ... Tonight, I'm launching a ***new Precision Medicine Initiative*** *to bring us closer to curing diseases like cancer and diabetes -- and to give all of us access to the personalized information we need to keep ourselves and our families healthier."*

President Obama on personalized medicine
State of the Union Address
Jan. 20, 2015

President's 2016 Budget will provide a **$215 million investment**

•**$130 million** to NIH for development of a voluntary national research cohort of a million or more volunteers (through engaged participation and open, responsible data sharing).

•**$70 million** to the National Cancer Institute (NCI), part of NIH, to scale up efforts to identify genomic drivers in cancer and apply that knowledge in the development of more effective approaches to cancer treatment.

"The proposed initiative has two main components: a near-term focus on cancers and a longer-term aim to generate knowledge applicable to the whole range of health and disease."

Francis S. Collins, *N Engl J Med* 2015; 372:793-795

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NIH Precision Medicine Initiative

THE PRECISION MEDICINE INITIATIVE

WHAT IS IT?

Precision medicine is an emerging approach for disease prevention and treatment that takes into account people's individual variations in genes, environment, and lifestyle.

The Precision Medicine Initiative will generate the scientific evidence needed to **move the concept of precision medicine into clinical practice.**

WHY NOW?

The **time is right** because of:

Sequencing of the human genome

Improved technologies for biomedical analysis

New tools for using large datasets

LONGER TERM GOALS

Create a research cohort of **> 1 million American volunteers** who will share genetic data, biological samples, and diet/lifestyle information, all linked to their electronic health records if they choose.

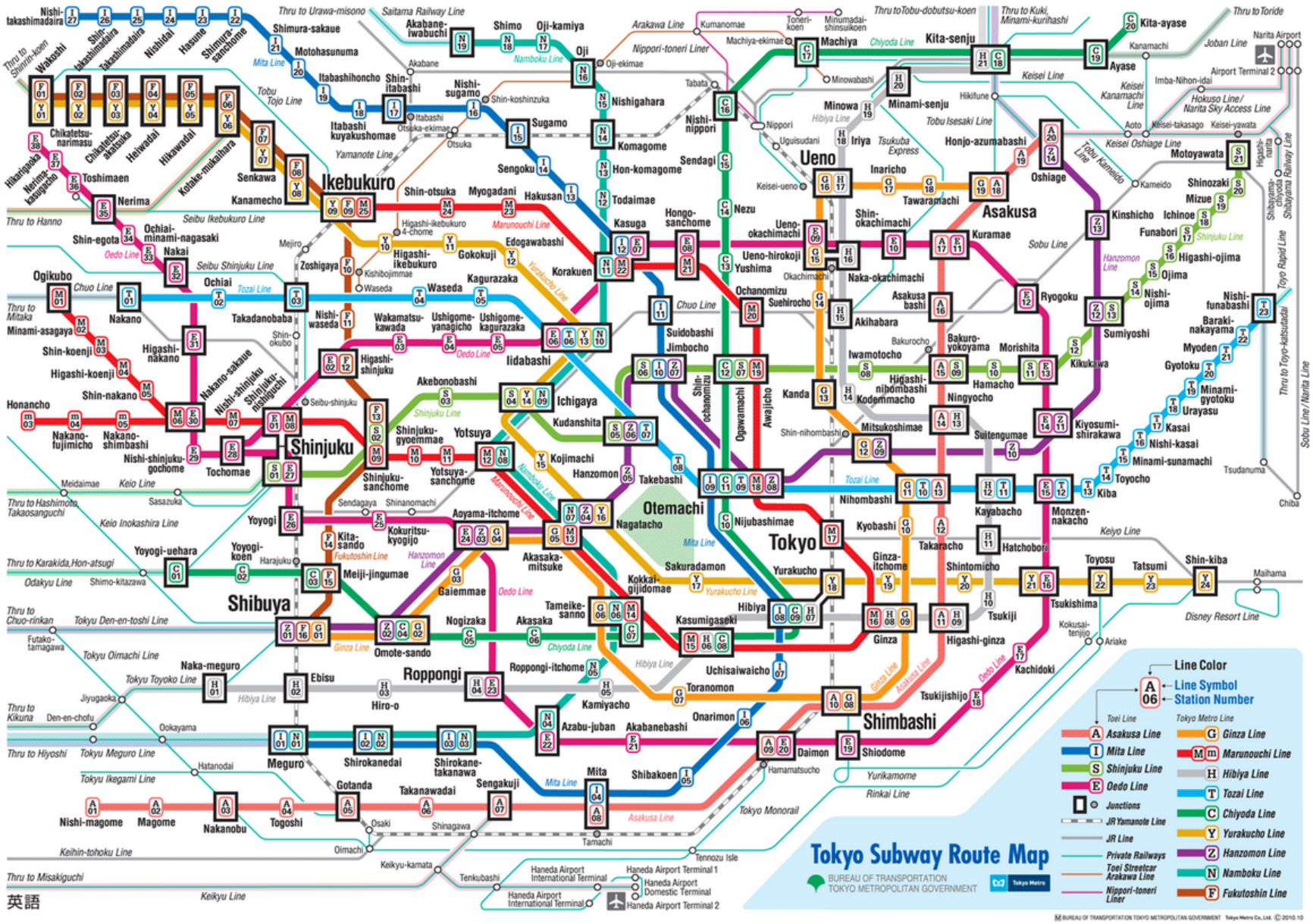
Pioneer **a new model for doing science** that emphasizes **engaged participants, responsible data sharing, and privacy protection.**

Research based upon the cohort data will:

- Advance **pharmacogenomics,** the right drug for the right patient at the right dose
- Identify new targets for **treatment and prevention**
- Test whether **mobile devices** can encourage healthy behaviors
- Lay **scientific foundation** for precision medicine for **many diseases**

https://www.nih.gov/precision-medicine-initiative-cohort-program

A Complex but Robust Infrastructure is needed….



Guiding Principles to PM Efforts

- Creating a dynamic and inclusive governance structure
- Building trust and accountability through transparency
- Respecting participant preferences
- Empowering participants through access to information
- Ensuring responsible data sharing, access, and use
- Maintaining data security, quality and integrity

Bottom line: Samples and data will last for a long time, participants want assurances that these things are protected and utilized appropriately!

Title	ID Number	Earliest Submission Date	Application Due Date
Precision Medicine Initiative® Cohort Program Direct Volunteers Pilot Studies (OTA)* pdf	OT-PM-16-001	November 16, 2015	December 22, 2015
Communication Support for the Precision Medicine Initiative® Research Programs at NIH (OTA)* pdf	OT-PM-16-002	November 16, 2015	December 22, 2015
Precision Medicine Initiative® Cohort Program Biobank (U24)	RFA-PM-16-004	January 4, 2016	February 4, 2016
Precision Medicine Initiative® Cohort Program Coordinating Center (U2C)	RFA-PM-16-001	January 17, 2016	February 17, 2016
Precision Medicine Initiative® Cohort Program Healthcare Provider Organization Enrollment Centers (UG3/UH3)	RFA-PM-16-002	January 17, 2016	February 17, 2016
Precision Medicine Initiative® Cohort Program Participant Technologies Center (U24)	RFA-PM-16-003	January 17, 2016	February 17, 2016

Precision Medicine: Clearly a Hot Topic

- Several PM related Conferences/Forums
- Focus on Participant Engagement

  (must implement into actual practice)
- Precision

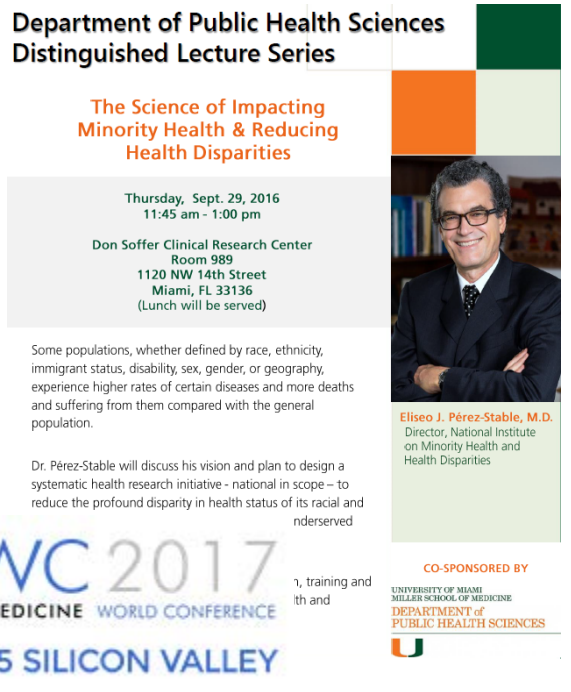
  (genetic information holds promise, but is it ready to deliver in the clinical setting?)
- Phenotype

  (deep and structured, will the existing EHR data work?)

2016 ADVANCES IN GENOME BIOLOGY AND TECHNOLOGY
PRECISION HEALTH MEETING

CHI's 8th International
Leaders in Biobanking CONGRESS 2016
Applying Biospecimen Science to Advance Biomedical Research and Patient Care
September 7-9, 2016
Hilton Baltimore
Baltimore, MD

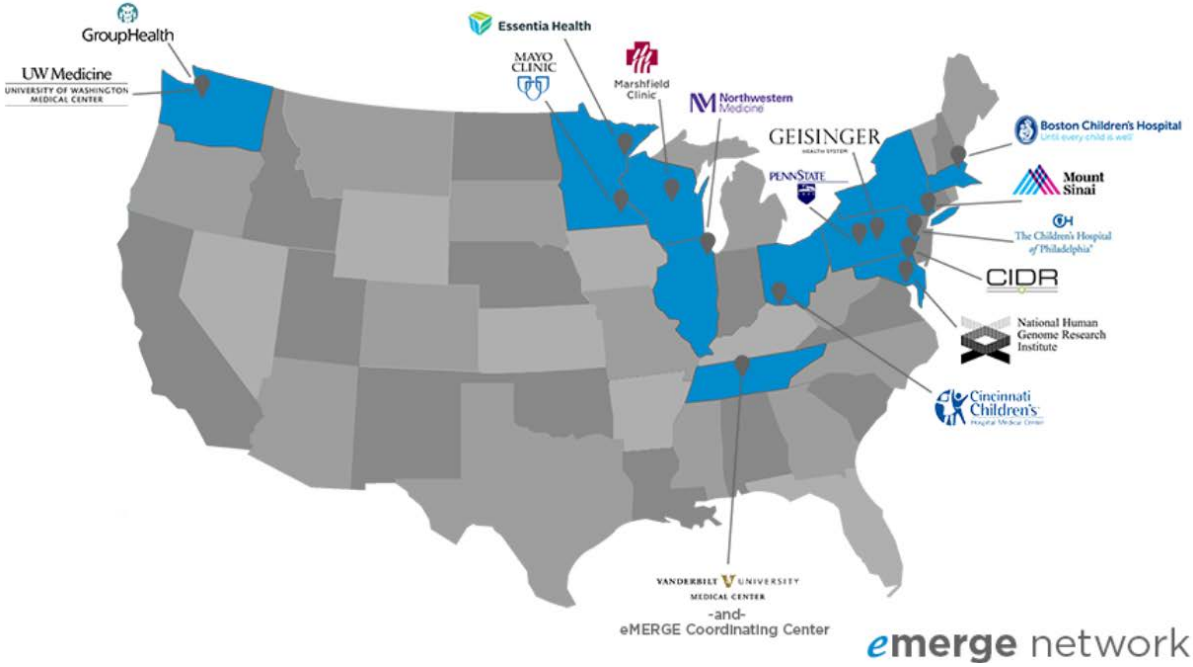
WORLD Precision Medicine Congress USA 2016
THE LARGEST PRECISION MEDICINE EVENT IN THE WORLD FOCUSING ON COMMERCIAL ACTIONABLE GOALS



PMWC 2017
PRECISION MEDICINE WORLD CONFERENCE
Jan 23-25 SILICON VALLEY

Large existing US-based Cohorts

eMERGE



Million Veteran Program (MVP)

MyCode® Community Health Initiative

Geisinger's MyCode project: the link to personalized medicine

CHARGE

Its founding member cohorts include:

- Age, Gene, Environment, Susceptibility Study -- Reykjavik
- Atherosclerosis Risk in Communities Study
- Cardiovascular Health Study
- Framingham Heart Study
- Rotterdam Study

Additional core cohorts include:

- Coronary Artery Risk Development in Young Adults
- Family Heart Study
- Health, Aging, and Body Composition Study
- Jackson Heart Study
- Multi-Ethnic Study of Atherosclerosis

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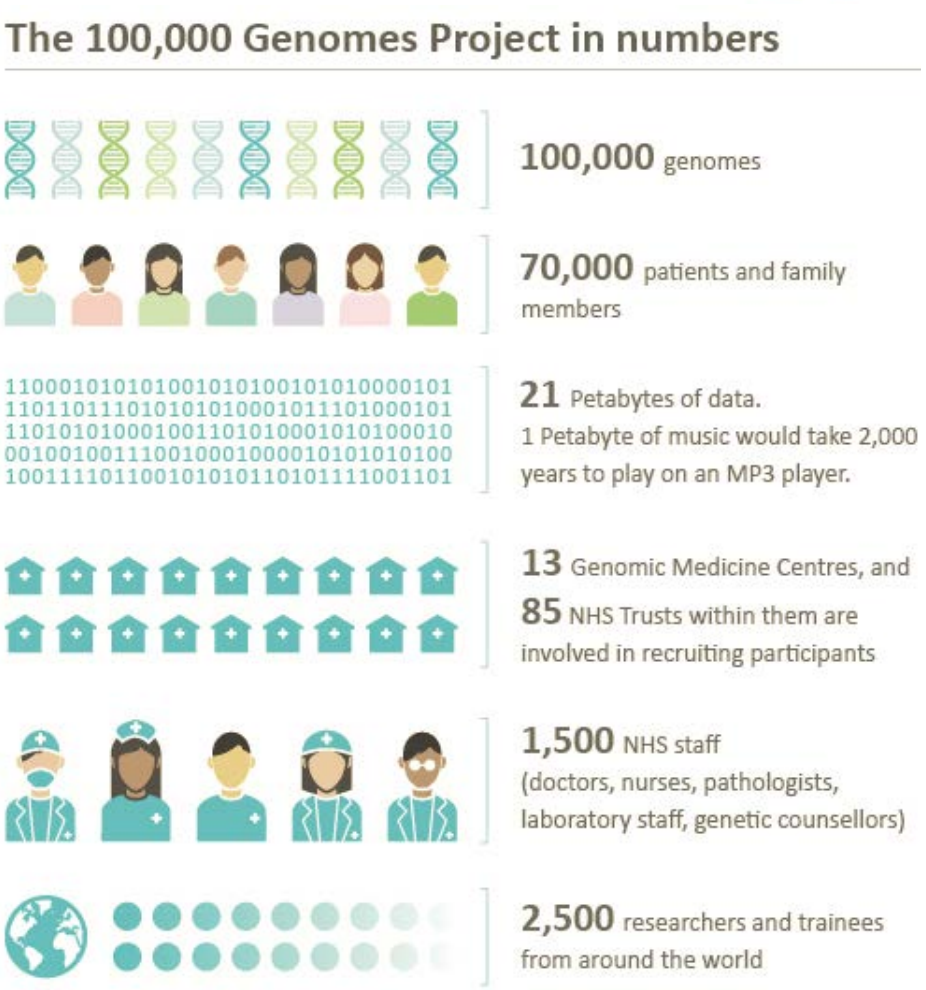
Biobanks: Engines for Genomic Research

About UK Biobank

UK Biobank is a major national health resource, and a registered charity in its own right, with the aim of improving the prevention, diagnosis and treatment of a wide range of serious and life-threatening illnesses – including cancer, heart diseases, stroke, diabetes, arthritis, osteoporosis, eye disorders, depression and forms of dementia. **UK Biobank recruited 500,000** people aged between 40-69 years in 2006-2010 from across the country to take part in this project. They have undergone measures, provided blood, urine and saliva samples for future analysis, detailed information about themselves and agreed to have their health followed.



Genomes Sequenced

1 3 0 4 0

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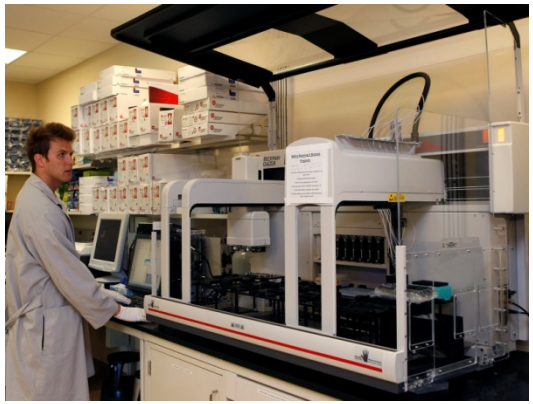
Technical capabilities already exist: HIHG Biorepository Overview

One of the largest academic Biorepositories in the United States.

- ~4,000 square foot facility in BRB
- Contains **>157,000** unique individual participants
- Tracking **1,190,148 total aliquots**
- Supports **75+** individual studies
- Supports numerous national and international disease consortia



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Biorepository

Sample Storage Equipment

- 5 Double-door refrigerators (4°C)
- 2 Freezers (-20°C)
- 23 Ultra-low Freezers (-80°C)
- 2 MVE 1520 Liquid nitrogen dewars (-196°C)

Aliquot Type	Item Count
Blood for DNA	41630
Blood for RNA	19544
Frozen Brain	237
Fixed Brain	130
Tissue Blocks	3555
Tissue Slides	6581
Buffy Coat	3045
Cell Line	25662
DNA	193364
Filter Card	21672
Hair	1
Plasma	77901
RNA	174
Saliva	455
Serum	38884
Tissue	3325
Urine	3401
White Blood Cells	328
Total	**439889**

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Biorepository: Security/Back-ups

A Secure and Comprehensive Facility

- Yokogawa DAQMaster Monitoring System
- Sensaphone (Secondary Monitoring)
- Back-up Air Conditioning
- 10 Ton spot Chiller (tertiary backup)
- Emergency Power

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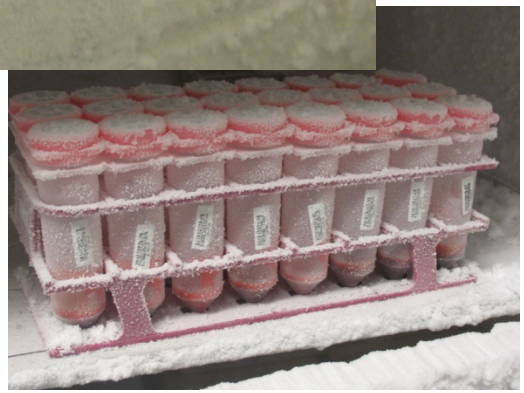
Biorepository: LIMS Tracking/Retrieval

- LIMS tracks all locations for samples, racks, shelves, and freezers
- All samples are ‘coded’ (**no PHI data is used on labels or stored within the LIMS**)
- Samples are retrieved and their locations updated to “checked-out” to a technician and a process
- Samples remain in “check-out” until technician acknowledges their return to storage



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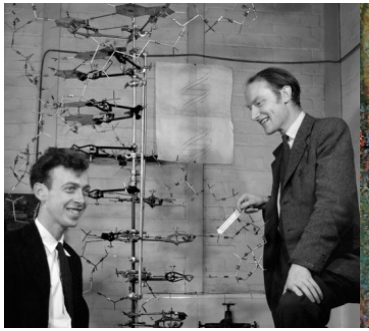
We have the genomic building blocks

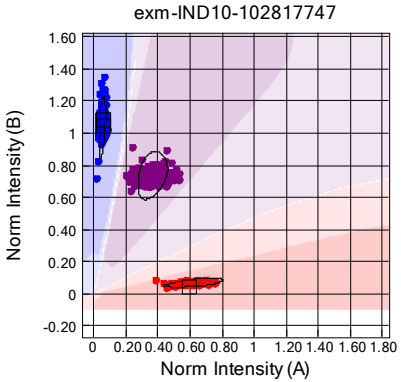


1000 Genomes

A Deep Catalog of Human Genetic Variation

AFFYMETRIX®

illumina®



PACIFIC BIOSCIENCES®

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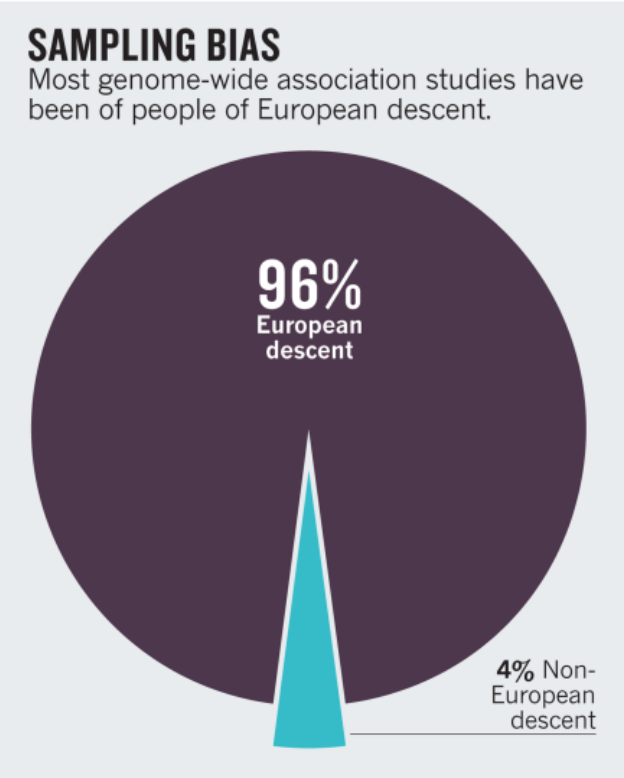
So if we have the tools… why?



(Bustamante 2011)

- Racial and ethnic minorities are underrepresented in genomic research.
- "Those most in need must not be the last to benefit from genetic research."
- What affects participation?

Factors that affect Participation

- TRUST
- Past experiences (either personal or historical examples)

  (e.g. trust between aboriginal communities in Australia and geneticists)
- Personal motivations
  - You or your family/friends are affected by a disease
  - Desire to better understand your own health
- Etc.

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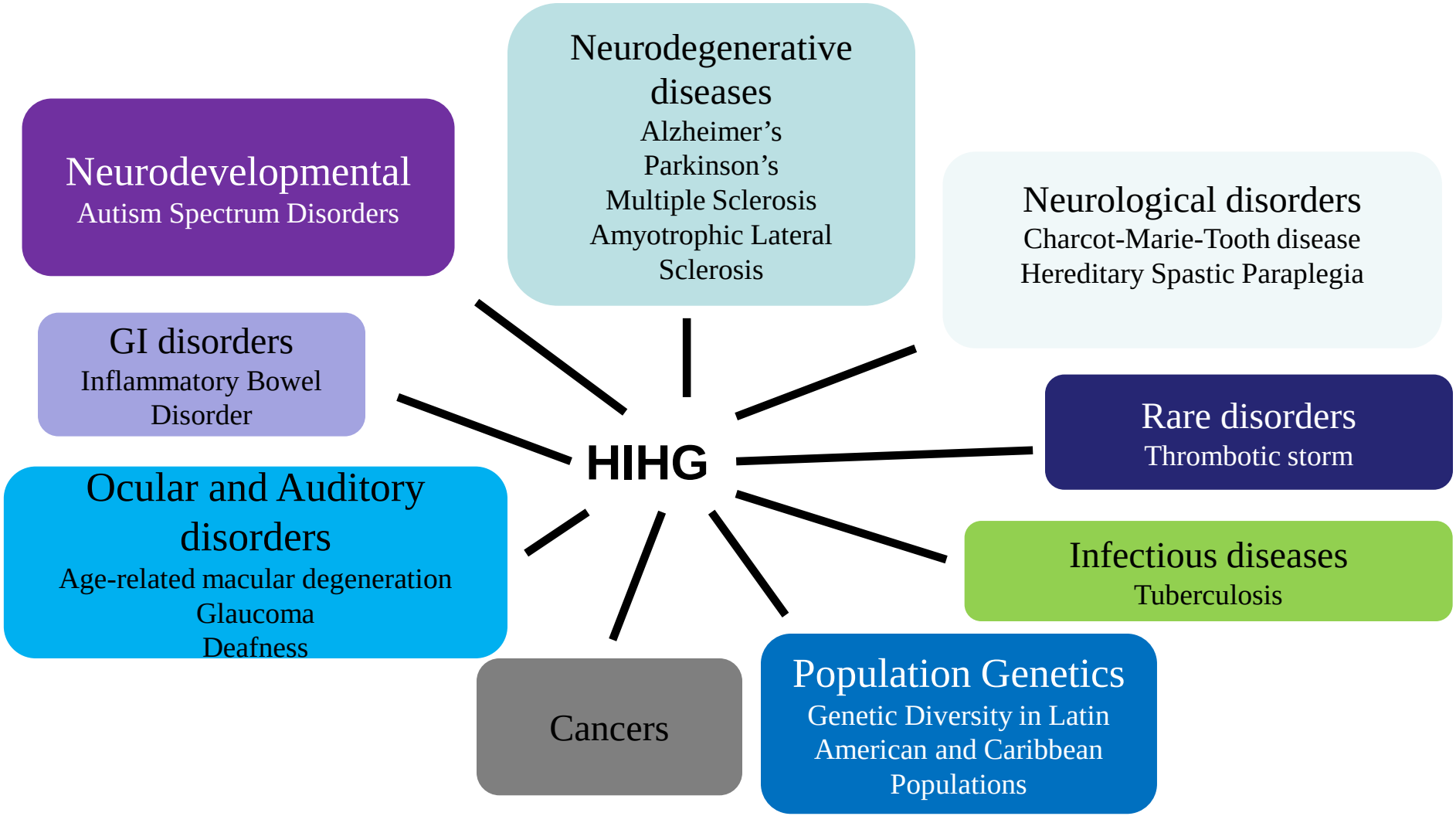
"Geneticists attempt to heal rifts with Aboriginals"
Ewen Callaway *Nature* 2016 Sep 21;537(7621):457-8. doi: 10.1038/537457°.

Are the majority of complex diseases ready for PM?



What samples and data exist for diverse participants groups?

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Examples of ongoing biobanking efforts and complex genetic disease studies of individuals with diverse ancestral and cultural backgrounds

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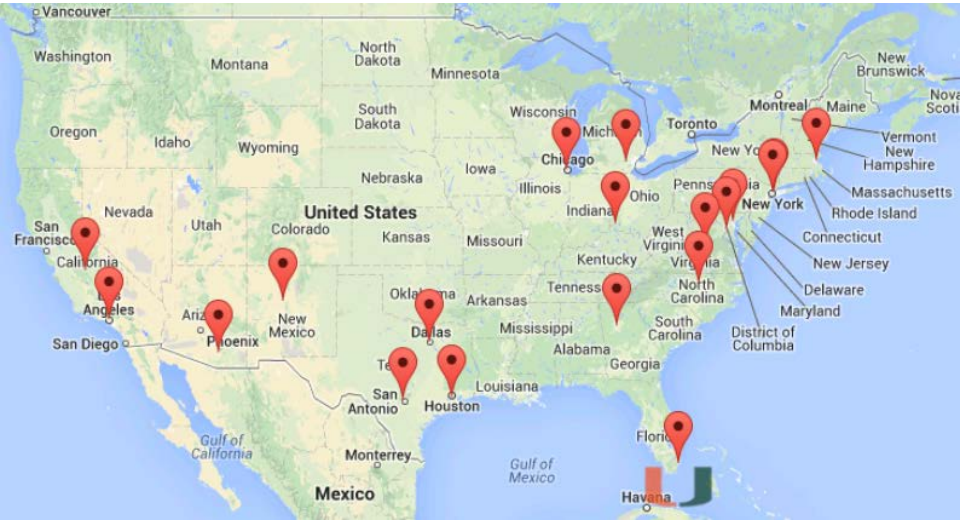
ERICH Study (Stroke)

This study is supported by funds from the NINDS U01-069763 (Dr. Daniel Woo)



- Ethnic/Racial Variations of Intracerebral Hemorrhage (ERICH)
- Large prospective multi-center case-control study focused on identification of genetic and epidemiological risk factors for ICH
- **19 Clinical Recruitment Centers** encompassing **42 sites**
- <u>Goal 6,000 participants</u>
  - 3,000 cases of ICH among white, blacks, and hispanics
  - 3,000 demographically matched controls
- **HIHG serves as central BioBank**
- **>6,000** samples (as of 9/1/16)

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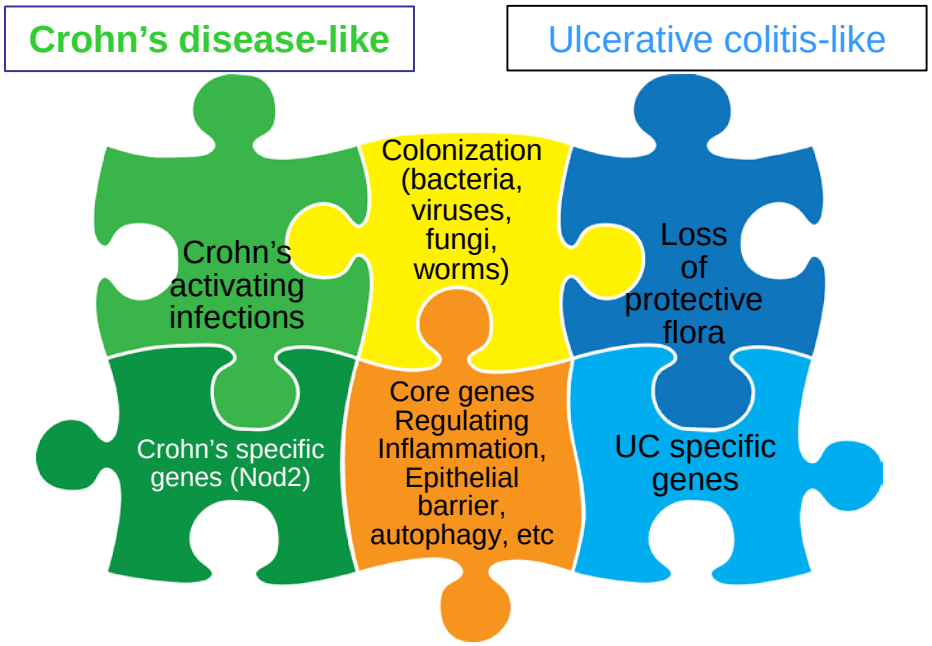
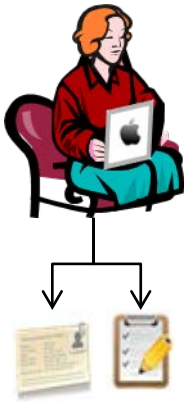
Inflammatory Bowel Disease (IBD)

- Establish sample collection (blood, tissue, DNA)
- Explore genetics, environment, and microbiome
- Collect data electronically within the clinic setting

IBD Pathogenesis





~1890 total study participants
696 Hispanic/ 1,188 non-Hispanic

86% of patients approached agreed to participate and provide a blood sample

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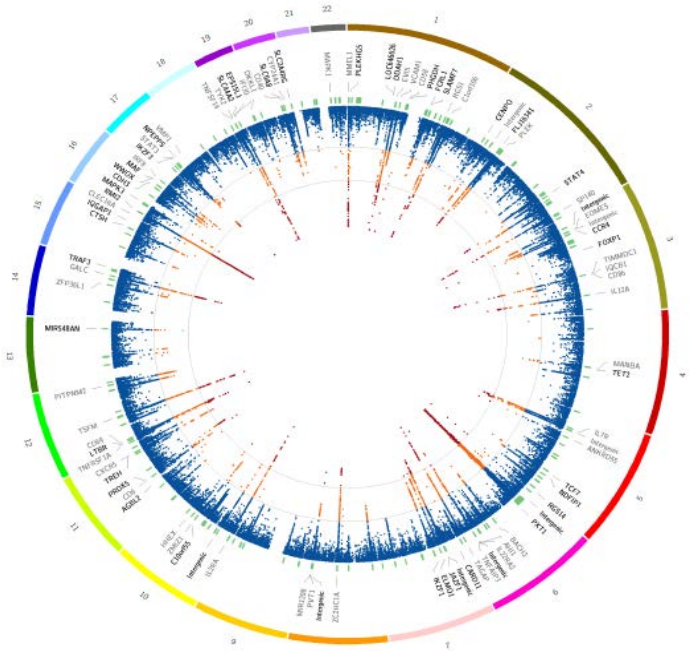
UHealth
UNIVERSITY OF MIAMI HEALTH SYSTEM
Crohn's and Colitis Center

Multiple Sclerosis Genetics

- MS is an unpredictable inflammatory demyelinating disease of the central nervous system
- Affected individuals have a spectrum of chronic symptoms and disabilities
- Genetic association studies in individuals of European descent have identified:
  - 110 autosomal MS risk variants
    - 103 discrete loci
    - Outside Major Histocompatibility Complex (MHC)
    - Explain 18% of the genetic risk
  - 27% of the genetic risk is explained after including the MHC
- International Multiple Sclerosis Genetics Consortium *(IMSGC)* expanded non-MHC variation to include:
  - 200 autosomal non-MHC risk variants
  - 1 X chromosome risk variant



IMSGC. *Nature Genetics*. Nov 2013.


IMSGC
https://www.imsgc.org/
Incidence (per 100,000 inhabitants)
More than 45
Less than 15
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ARTICLES

nature genetics

Meta-analysis of genome scans and replication identify *CD6*, *IRF8* and *TNFRSF1A* as new multiple sclerosis susceptibility loci

Philip L De Jager[1–3], Xiaoming Jia[4], Joanne Wang[5,6], Paul I W de Bakker[3,4], Linda Ottoboni[1–3], Neelum T Aggarwal[7], Laura Piccio[8], Soumya Raychaudhuri[3,9], Dong Tran[3], Cristin Aubin[3], Rebeccah Briskin[2], Susan Romano[1], International MS Genetics Consortium, Sergio E Baranzini[5], Jacob L McCauley[10], Margaret A Pericak-Vance[10], Jonathan L Haines[11], Rachel A Gibson[12], Yvonne Naeglin[13], Bernard Uitdehaag[14], Paul M Matthews[12], Ludwig Kappos[13], Chris Polman[14], Wendy L McArdle[15], David P Strachan[16], Denis Evans[7], Anne H Cross[8], Mark J Daly[3,17], Alastair Compston[18], Stephen J Sawcer[18], Howard L Weiner[1], Stephen L Hauser[5,6,19], David A Hafler[1,3,19] & Jorge R Oksenberg[5,6,19]

Human Molecular Genetics, 2010, Vol. 19, No. 5 953–962
doi:10.1093/hmg/ddp542
Advance Access published on December 9, 2009

European Journal of Human Genetics (2009) 17, 1309 – 1313

www.nature.com/ejhg

ARTICLE

Replication analysis identifies *TYK2* as a multiple sclerosis susceptibility factor

Maria Ban*[,1], An Goris[2], Åslaug R Lorentzen[3,4], Amie Baker[1], Tania Mihalova[5], Gillian Ingram[6], David R Booth[7], Robert N Heard[7], Graeme J Stewart[7], Elke Bogaert[2], Bénédicte Dubois[2], Hanne F Harbo[3], Elisabeth G Celius[3], Anne Spurkland[8], Richard Strange[5], Clive Hawkins[5], Neil P Robertson[6], Frank Dudbridge[9], James Wason[9], Philip L De Jager[10,11], David Hafler[11], John D Rioux[12], Adrian J Ivinson[13], Jacob L McCauley[14], Margaret Pericak-Vance[14], Jorge R Oksenberg[15], Stephen L Hauser[15], David Sexton[16], Jonathan Haines[16] and Stephen Sawcer[1], The Wellcome Trust Case–Control Consortium (WTCCC) and Alastair Compston[1]

The NEW ENGLAND JOURNAL of MEDICINE

ESTABLISHED IN 1812 AUGUST 30, 2007 VOL. 357 NO. 9

Risk Alleles for Multiple Sclerosis Identified by a Genomewide Study

The International Multiple Sclerosis Genetics Consortium*

ARTICLES

Comprehensive follow-up of the first genome-wide association study of mu *KIF21B* and *TMEM39A* a

nature genetics

The International Multiple Sclerosis Genetics

Hum Genet
DOI 10.1007/s00439-010-0789-4

ORIGINAL INVESTIGATION

Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis

International Multiple Sclerosis Genetics Consortium (IMSGC)*

Genetic variation in the IL7RA/IL7 pathway increases multiple sclerosis susceptibility

Rebecca L. Zuvich · Jacob L. McCauley · Jorge R. Oksenberg · Stephen J. Sawcer · Philip L. De Jager · International Multiple Sclerosis Genetics Consortium · Cristin Aubin · Anne H. Cross · Laura Piccio · Neelum T. Aggarwal · Denis Evans · David A. Hafler · Alastair Compston · Stephen L. Hauser · Margaret A. Pericak-Vance · Jonathan L. Haines

Interleukin 7 receptor α chain (*IL7R*) shows allelic and functional association with multiple sclerosis

Simon G Gregory[1,9], Silke Schmidt[1,9], Puneet Seth[2], Jorge R Oksenberg[3], John Hart[1], Angela Prokop[1], Stacy J Caillier[3], Maria Ban[4], An Goris[5], Lisa F Barcellos[6], Robin Lincoln[3], Jacob L McCauley[7], Stephen J Sawcer[4], D A S Compston[4], Benedicte Dubois[5], Stephen L Hauser[3], Mariano A Garcia-Blanco[2], Margaret A Pericak-Vance[8] & Jonathan L Haines[7], for the Multiple Sclerosis Genetics Group

LETTER

doi:10.1038/nature10251

Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis

The International Multiple Sclerosis Genetics Consortium* & the Wellcome Trust Case Control Consortium 2*

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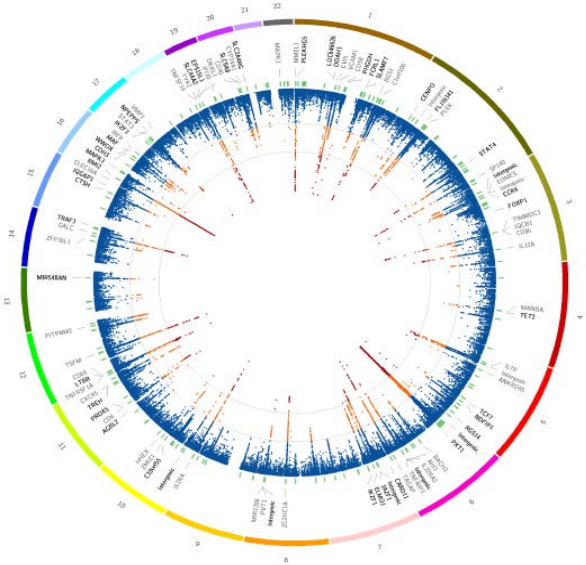
The role of the *CD58* locus in multiple sclerosis

Philip L. De Jager[a,b,c,1], Clare Baecher-Allan[a], Lisa M. Maier[a,c], Ariel T. Arthur[d], Linda Ottoboni[a,c], Lisa Barcellos[e], Jacob L. McCauley[f], Stephen Sawcer[g], An Goris[h], Janna Saarela[i], Roman Yelensky[c,j], Alkes Price[c,k], Virpi Leppa[i], Nick Patterson[c], Paul I. W. de Bakker[b,c], Dong Tran[a,c], Cristin Aubin[a,c], Susan Pobywajlo[a], Elizabeth Rossin[a,c], Xinli Hu[c], Charles W. Ashley[a], Edwin Choy[c,l], John D. Rioux[c,l], Margaret A. Pericak-Vance[f], Adrian Ivinson[m], David R. Booth[n], Graeme J. Stewart[n], Aarno Palotie[c,h], Leena Peltonen[c,h], Bénédicte Dubois[g], Jonathan L. Haines[o], Howard L. Weiner[a], Alastair Compston[g], Stephen L. Hauser[p,q], Mark J. Daly[c,l], David Reich[c,l], Jorge R. Oksenberg[p,q], and David A. Hafler[a,c].

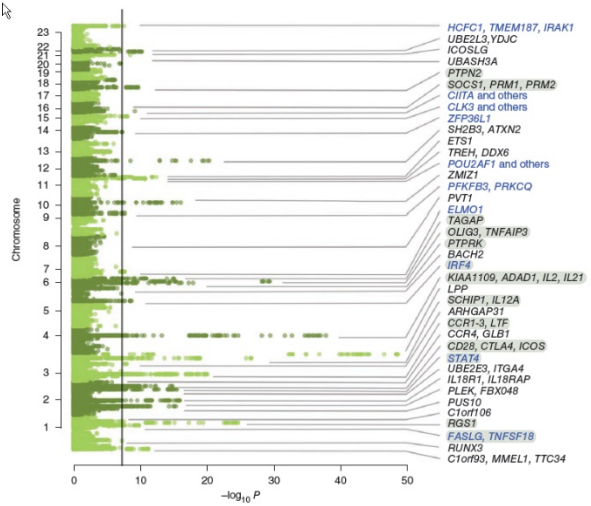
[a]Division of Molecular Immunology, Center for Neurologic Diseases, Brigham and Women's Hospital and Harvard Medical School, Boston, MA 02115; [b]Partners Center for Personalized Genetic Medicine, Boston, MA 02115; [c]Program in Medical and Population Genetics, Broad Institute of Harvard University and Massachusetts Institute of Technology, Cambridge, MA 02139; [d]Department of Medicine and the Nerve Research Foundation, University of Sydney, Sydney NSW 2145, Australia; [e]Division of Epidemiology, School of Public Health, University of California, Berkeley, CA 94720-7360; [f]Miami Institute for Human Genomics, Miller School of Medicine, University of Miami, Miami, FL 33136; [g]Department of Clinical Neurosciences, Addenbrooke's Hospital,

Individual disease results: post-ImmunoChip

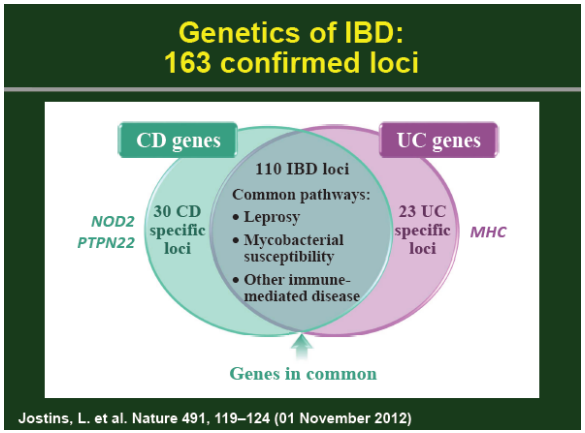
MS (45 new loci/**104** total)



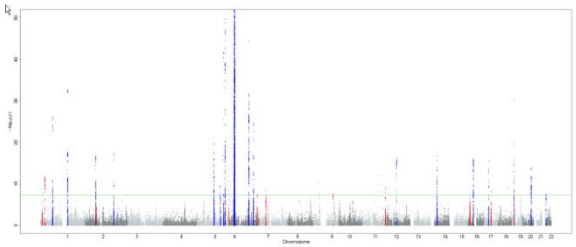
Celiac (13 new loci/**40** total)



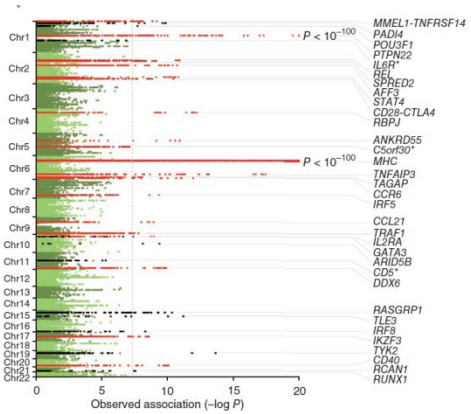
IBD (71 new loci/**163** total)



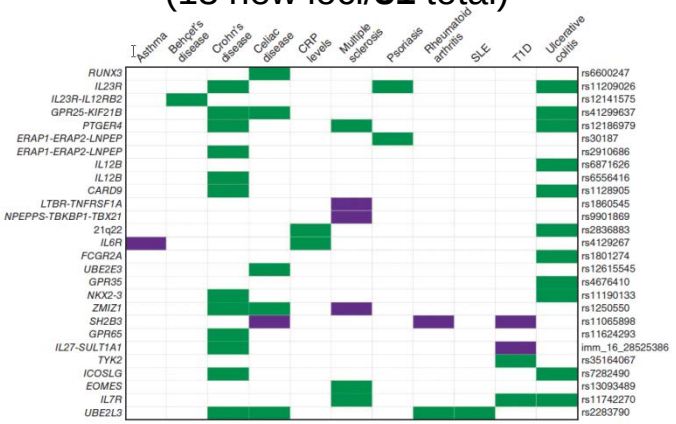
Psoriasis (15 new loci/**36** total)



RA (14 new loci/**48** total)



Ankylosing Spondylitis (13 new loci/**31** total)



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Precision Medicine for All:

This will require us to understand whether our findings in populations of primarily European ancestry extend to other populations affected by disease

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Why study racial and ethnically diverse populations in MS?

Are the risk loci identified in European populations relevant to disease risk in African-Americans or Hispanic Americans?

- Differences in LD structure can help to localize signal (i.e. fine-mapping)
- Understanding etiology of disease
- Ethics – what is the relevance of current scientific knowledge to other populations?
- Potential identification of new loci
- Teasing apart gene-gene and gene-environment interaction in admixed populations
- Better understanding the observed clinical differences across race/ethnic groups

MS: Hispanics and African Americans

Largest minority populations in the US (2010 Census Data)

~16% Hispanic/Latino and ~12% African American

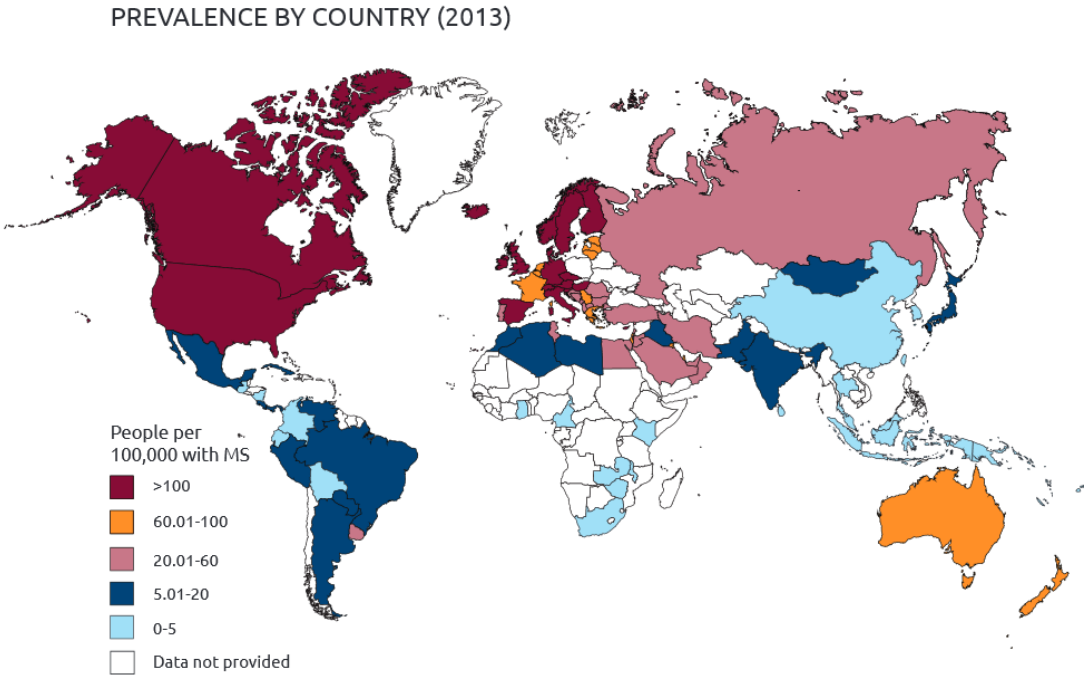
Classically lower MS prevalence than individuals of primarily European descent, but complicated:

- Ethnic heterogeneity
- Lack of consistent diagnostic criteria
- Changing disease awareness

Disease heterogeneity seen:

- African Americans often exhibit greater disease severity (Buchanan R. *Ethn Dis*. 2010)
- Hispanics present more often with Optic Neuritis (Amezcua L. *Mult. Scler*. 2011)

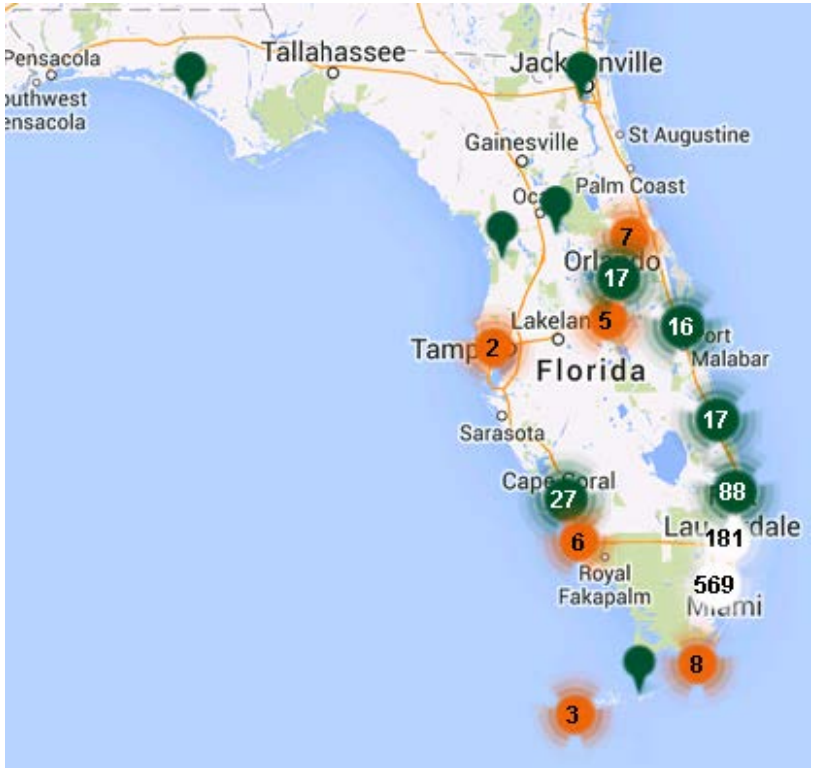
Data from the Multiple Sclerosis International Federation (MSIF)

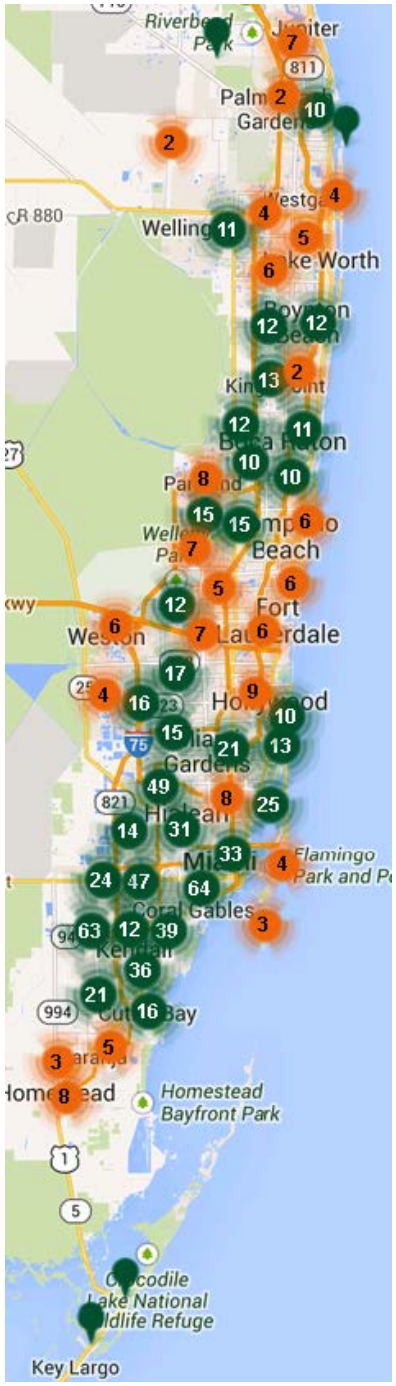


http://www.msif.org/about-us/advocacy/atlas/atlas-of-ms/

Our MS Ascertainment for Genetic Research in Florida

>1,200 participants recruited thus far





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Alliance for Research in Hispanic MS (ARHMS): www.arhms.org

ARHMS Recruitment Centers

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Angel Chinea, MD
San Juan MS Center
Guaynabo, PR 00968

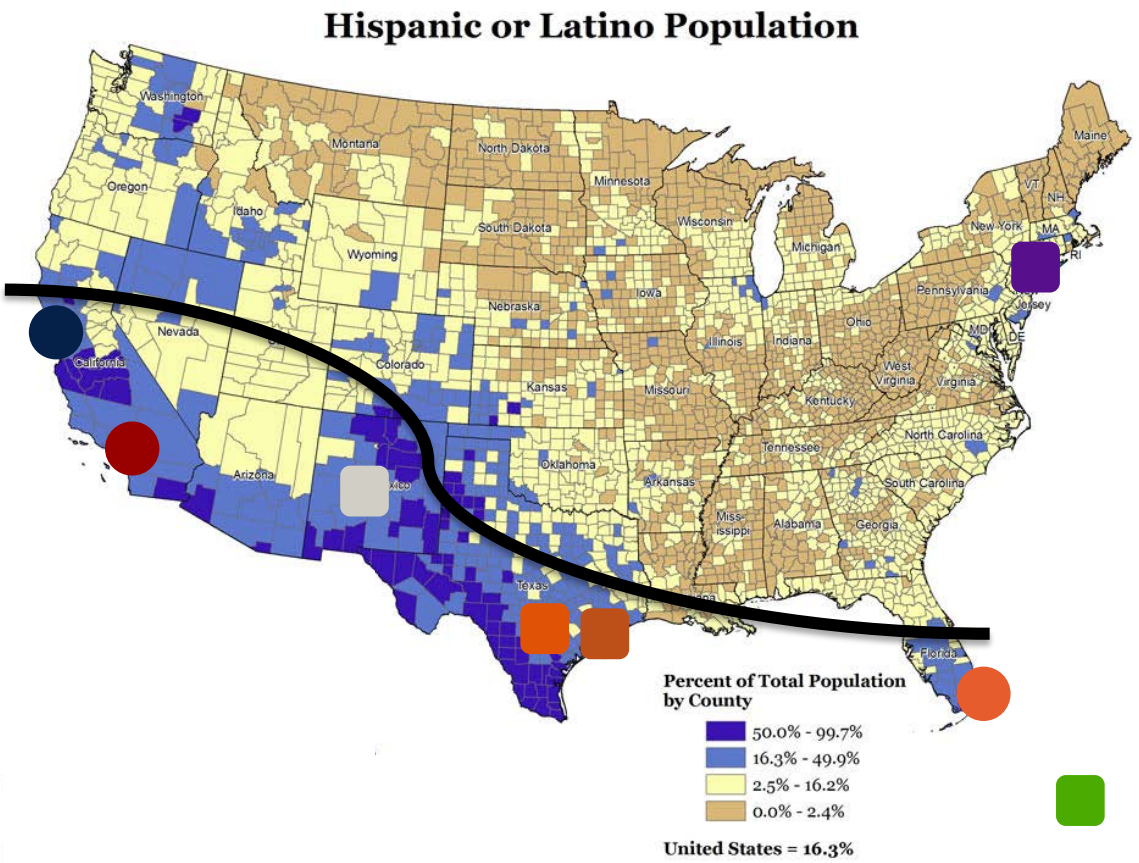

Figure: Adopted from the Rural Health Information Hub (https://www.ruralhealthinfo.org/rural-maps/mapfiles/hispanic.jpg)
Illustrates 2010 Census Summary Data on U.S. Hispanic/Latino population

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Initial Hispanic Dataset

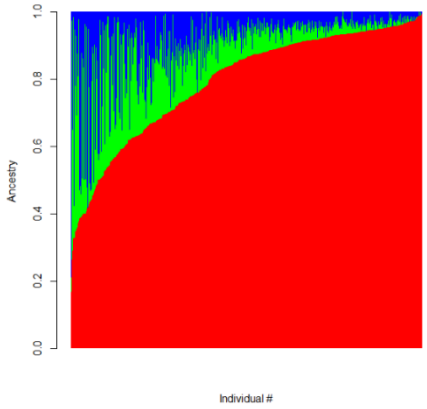
	Hispanic	
	MS Case	Control
	N=1178	N=1330
	N (%)	
University of Miami	561 (48)	1102 (83)
University of California, San Francisco	227 (19)	63 (5)
University of Southern California	196 (17)	
Caribbean Neurological Center	99 (8)	96 (7)
Brigham and Women's Hospital	95 (8)	69 (5)

Genotype data available:

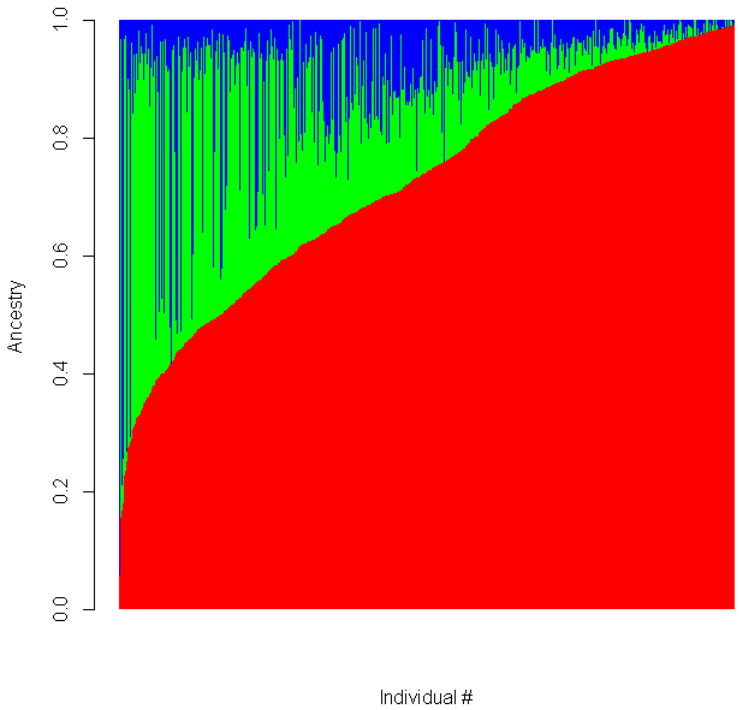
- Illumina ExomeChip+ Beadchip

Global Ancestry: Hispanics

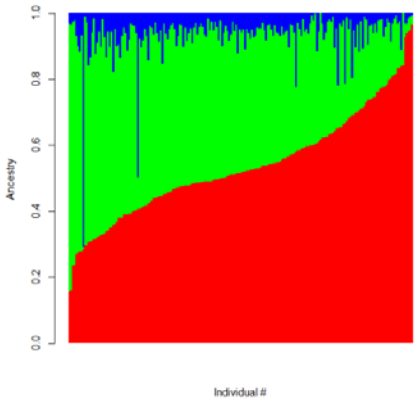
Miami



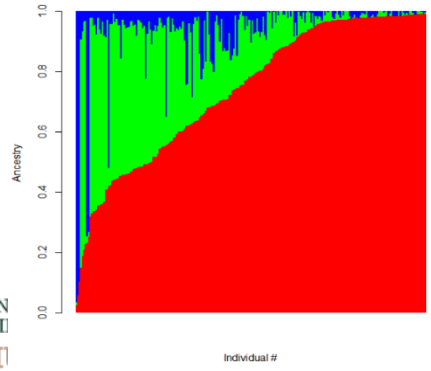
Hispanics



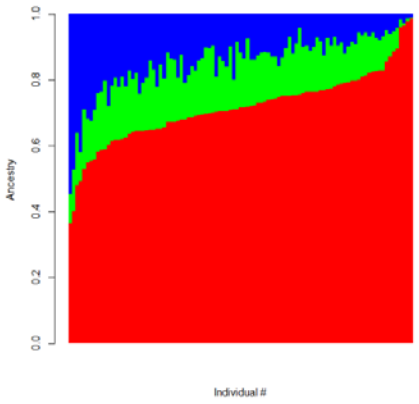
USC



UCSF



Puerto Rico



European ancestry

African ancestry

Native American ancestry

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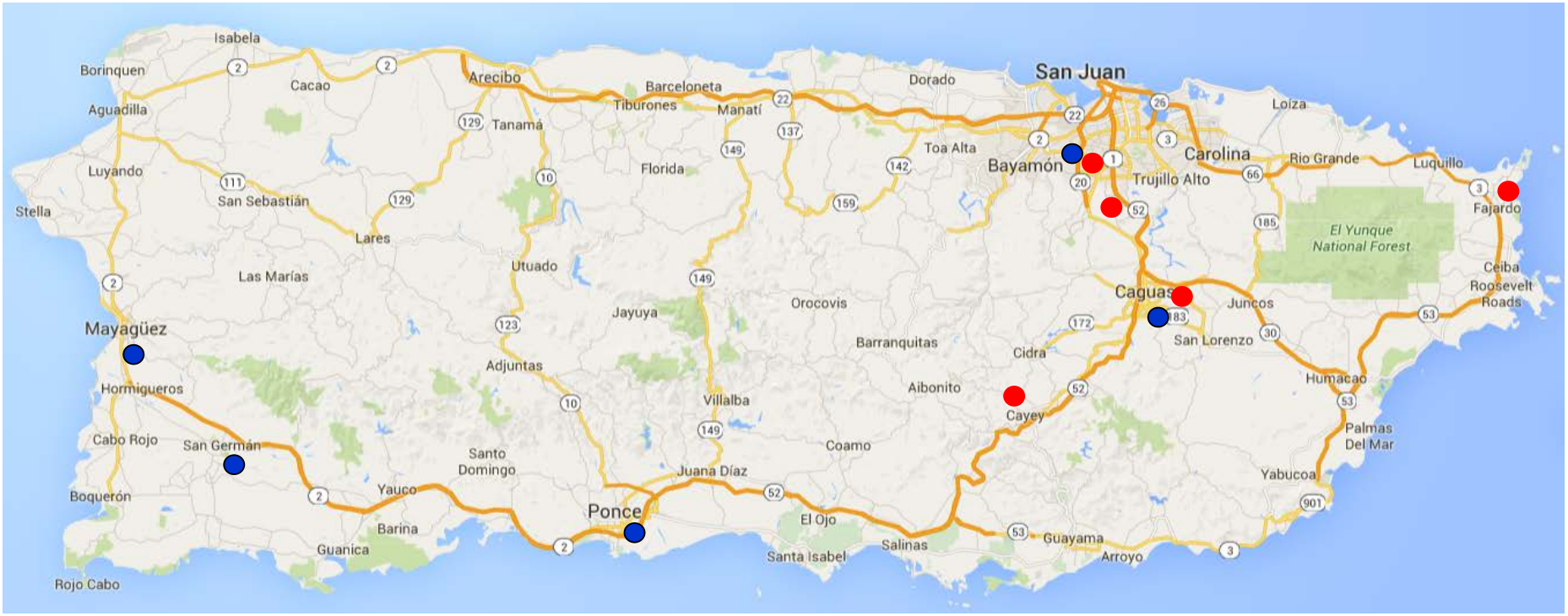
MS Ascertainment in Puerto Rico

- January 2015
- January 2016



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Our Experiences in Puerto Rico

- Building trust: We spent 4 years in discussions and engagement before we collected the first sample
- MS patients and their families are very eager and motivated to participate
- We have a great clinician champion (Dr. Chinea)
- The logistics of sample collection at various sites across the island can be a challenge (especially finding FedEx)
- With each ascertainment trip we have become more efficient in our processes
- Clinical data collection is a little more challenging
- Next Trip November 2016

MS Collaborative Discussions in Cuba

- April 2016

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Some Challenges in Cuba

- Governmental restrictions/limitations on sample and data sharing
- More open to collaboration, but every university is flying in to 'build' collaborations
- Clinicians/Researchers want to grow their expertise and collaborate….. not simply provide samples and data for export to the US
- There are distinct unmet needs of patients, research may be a secondary pursuit in the eyes of many MS patients and their families
- Again, building trust will take time

Pilot Assessment of Motivations for Participation

To investigate the reasons for participation among Hispanic and non-Hispanic enrollees in a genetic risk for disease study.

Study involves a blood draw, family history questionnaire, and medical records review

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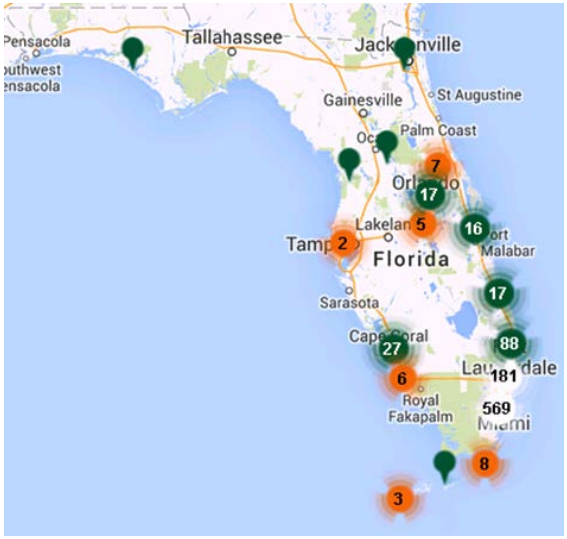
Participants

Participants enrolled via ascertainment for a study of genetic risk for multiple sclerosis.

N=101 individuals (95 agreed to participate)
80% Hispanic
20% M: 80% F



Education ranges	(%)
< high school	5%
High school	24%
Some college/AA	35%
College grad	29%
Post-graduate	8%

Age ranges	(%)
18-25	7%
26-35	17%
36-45	21%
46-55	27%
56-65	20%
>66	8%

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Methods

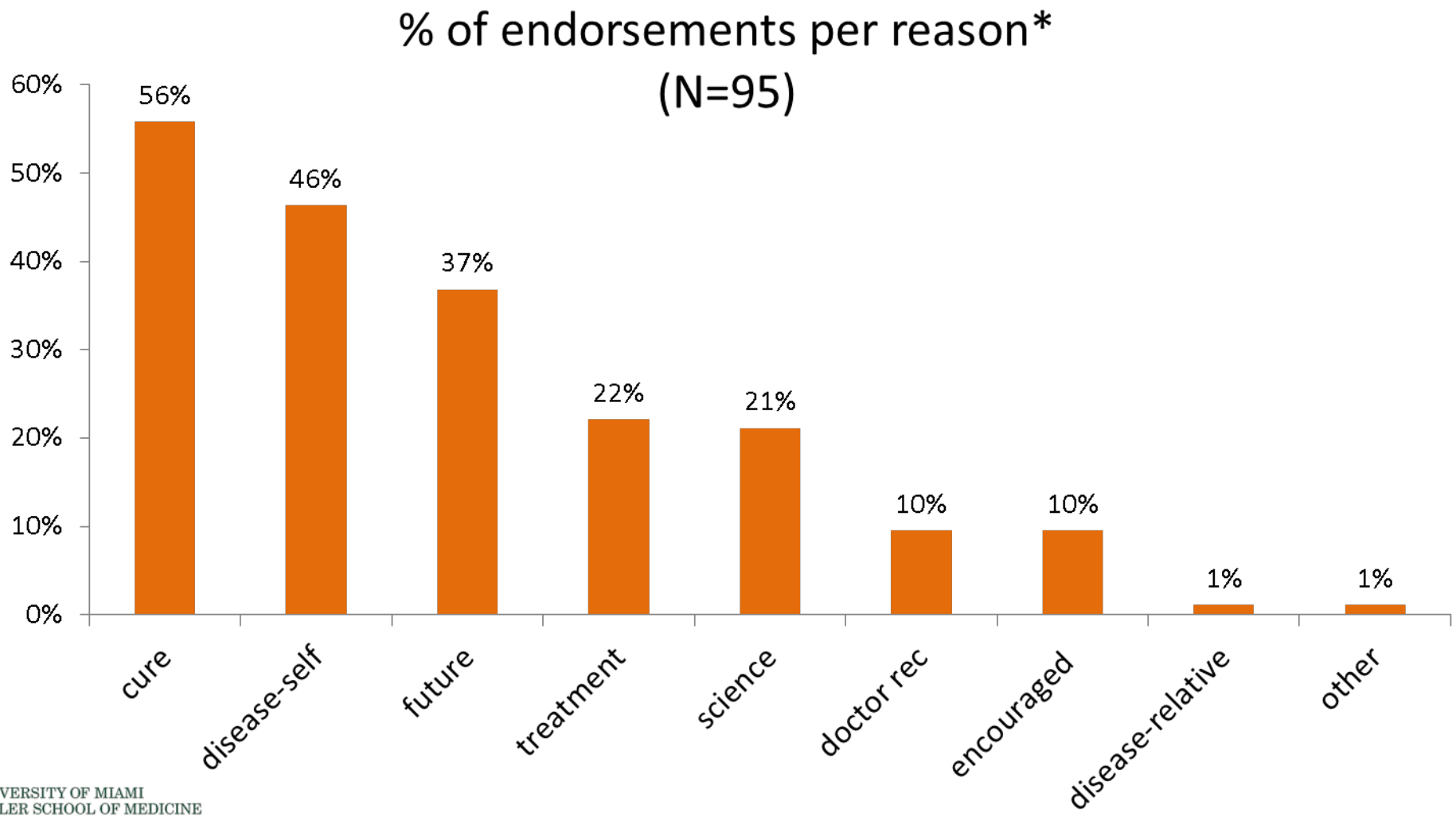
Surveyed reasons for participation

Provided 11 options

- Cure
- Knowledge
- Relative with disease
- Doctor recommendation
- Encouraged by others
- Treatment
- Have MS
- Future generations
- Reimbursement
- Other
- Not sure

"I want to help find a cure for MS."
"I suffer from MS."
"The doctor asked/recommended that I participate."
"To help future generations."

Results



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**not sure* and *compensation* were not endorsed

Results

Comparison of endorsement rates per reason in Hispanics vs. non-Hispanics*

Reason	Hispanic	non-Hispanic	**p-value
Cure for MS	56%	56%	1.00
Have MS	**52%**	**19%**	**0.03**
Help future generations	34%	50%	0.26
Find new treatments	**17%**	**50%**	**0.007**
Improve science	19%	31%	0.32
Doctor recommendation	11%	0%	0.35
Encouraged by others	8%	19%	0.17

**relative with disease* and *other* were not analyzed due to low numbers of endorsements
**Fisher's Exact Test

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Results

Comparison of endorsements in younger vs. older participants (N=52)

Reason	Younger (18-35)	Older (>56)	**p-value
Cure for MS	74%	39%	0.02
Have MS	44%	42%	1.00
Help future generations	30%	39%	0.76
Find new treatments	30%	12%	0.16
Improve science	26%	12%	0.27
Doctor recommendation	4%	15%	0.35
Encouraged by others	13%	12%	1.00

*_relative with disease_ and _other_ were not analyzed due to low numbers of endorsements

**Fisher's Exact Test

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Preliminary Motivation Results

- **Among Hispanics with MS**, participation in genetic research for MS is strongly associated with having MS.

- **Among non-Hispanics with MS,** participation in genetic research for MS is strongly associated with the desire to find new treatments.

- **Among younger individuals with MS,** participation in genetic research for MS is strongly associated with the desire to find a cure for MS.


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EMORY HEALTHCARE
SEEC
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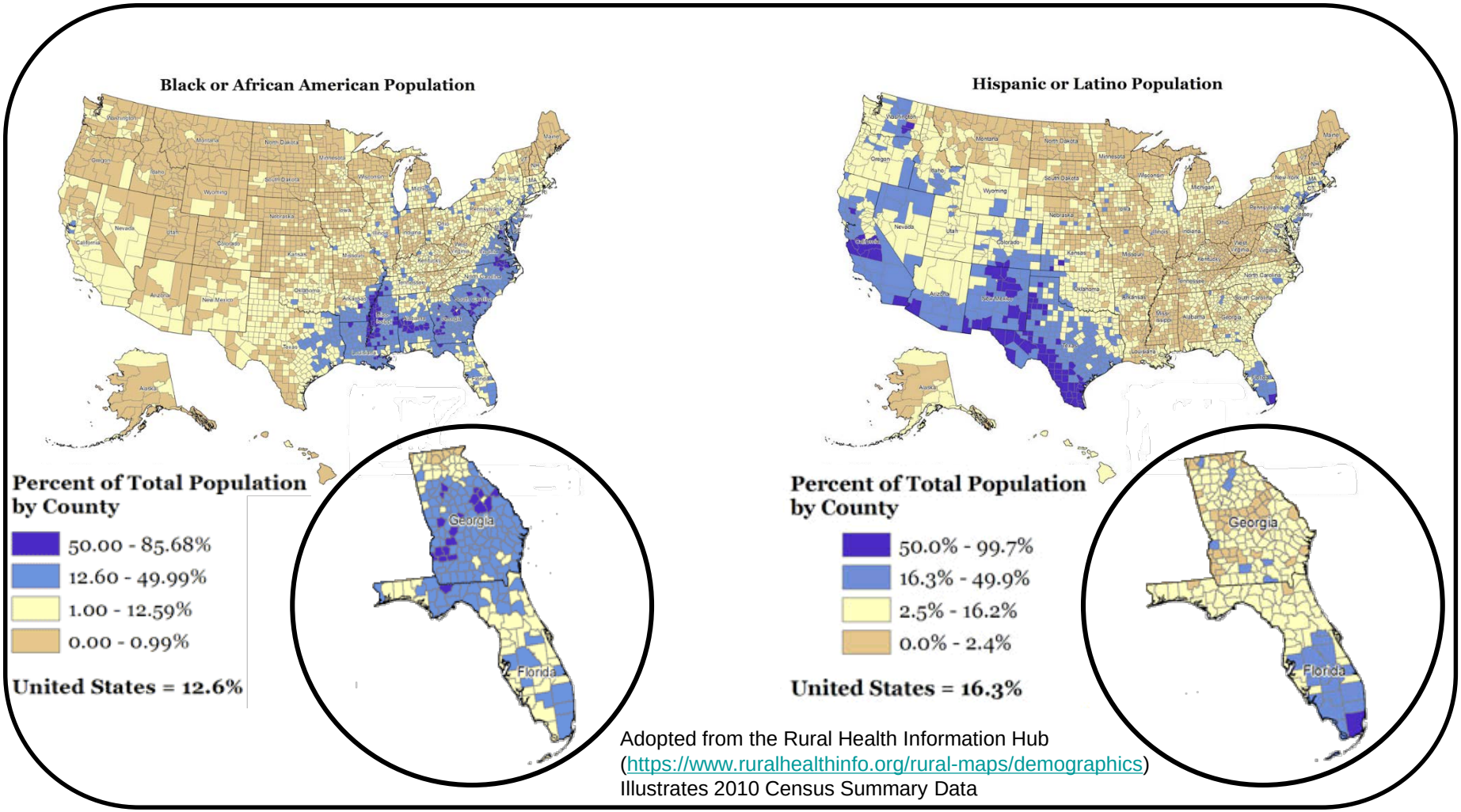
'We SEEC Precision in Medicine'
UF College of Medicine
UNIVERSITY of FLORIDA
MOREHOUSE SCHOOL OF MEDICINE

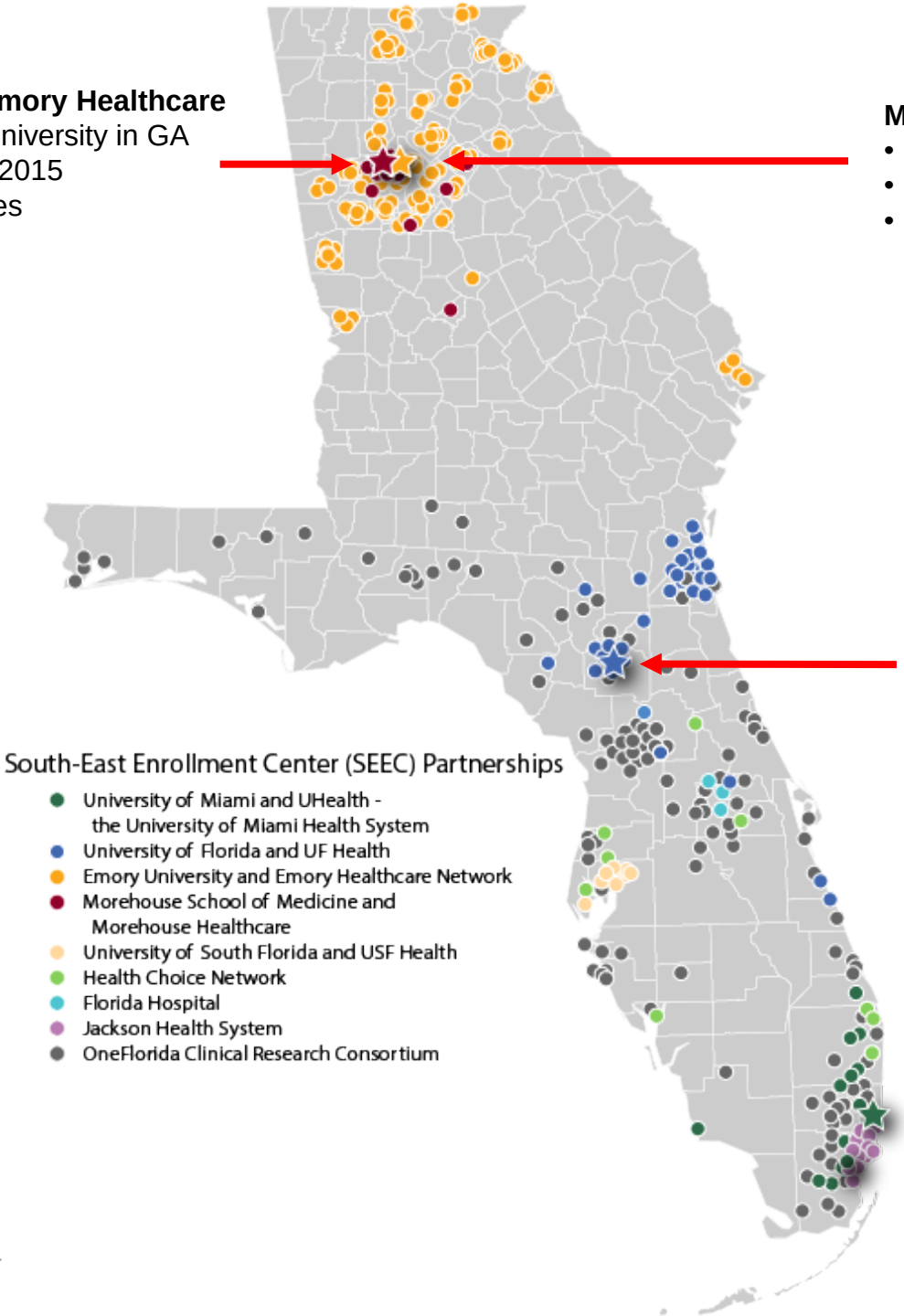
South-East Enrollment Center (SEEC)



Adopted from the Rural Health Information Hub
(https://www.ruralhealthinfo.org/rural-maps/demographics)
Illustrates 2010 Census Summary Data

Emory University/Emory Healthcare
- Largest private university in GA
- 2.8M patients in 2015
- 24,000 employees

Morehouse School of Medicine
- Historically Black College
- 350,000 patients per year
- 1,500 employees



University of Florida Gainesville
- Largest State University in FL
- 1.2M patients with EHR records
- 12,000 employees

University of Miami
- Largest private university in FL
- 1.1M patients per year
- 20,000 employees

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South-East Enrollment Center (SEEC)

Florida and Georgia

- 30 million individuals in GA and FL
- FL is the 3rd most populous state
- GA is the 9th most populous
- Unique climate and environment

Atlanta metro area (6.2M people)

- 30% African-American

North Florida (4.1M people)

- 32% AA and 18% Hispanics

South Florida metro area (5.5M people)

- 70% Hispanic, highly diverse
- Caribbean blacks

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Vanderbilt-Miami-Meharry
Center for Precision Medicine and Population Health

PIs: Consuelo H. Wilkins, Nancy J. Cox, Maria de Fatima Lima, and Roy Weiss

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Vanderbilt-Miami-Meharry Center for Precision Medicine and Population Health

- Foster research using precision medicine approaches to address racial/ethnic health disparities
- Develop novel methods to integrate individual, contextual and environmental data
- Propel novel health disparities research **leveraging genomic and phenotypic data**
- Develop ethical, deliberate, socially and culturally acceptable methods for engaging racial and ethnic minorities and vulnerable populations in precision medicine research

Practical Challenges to Consider

- Patient and Research Community Education
- Resources to more actively engage diverse participants
- General long-term sustainability of these resources (paying for storage of both samples and data over the long haul)
- How will large well-publicized national US efforts affect participation in smaller focused biobank and data collections?

  (i.e. will it help or hurt, how much is dependent on the success of the national effort)
- Diversity isn't only racial/ethnic, how do we engage/include rural participants and those across the SES spectrum?
- Resources to more actively engage diverse participants

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Recent Review Article: Chalmers et al. *BMC Medical Ethics* (2016) 17:39

Some Keys to Success

- Build Trust within your participant community
- Be mindful of how results are reported and respect the participant community
- Well-trained personnel (preferably from the community you are engaging)
- Stay engaged with participants (if they want to be engaged) (i.e. newsletters updating your research progress)
- PATIENCE (building trust and biobanks take time)

Translating Genetic Findings into Precision Medicine for ALL

What can we achieve?

- Better understanding of the pathophysiology of disease
- Identify targets for more ‘precise’ treatments
- Provide better predictions of who might be diagnosed with disease
- Give us improved predictions of the disease course
- Lead to knowledge that can help find a cure or prevent disease

Strong need for large well-characterized (genetics, environmental exposures, treatment history, imaging) cohorts with a diverse set of research biospecimens (DNA, RNA, PBMCs, etc.) and ideally longitudinal engagement of diverse participant populations

Acknowledgements

MS Studies (Hispanics & African Americans)

Alliance for Research in Hispanic MS (ARHMS):
www.arhms.org

University of Miami
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- Patricia Manrique
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- Gary Beecham
- Michael Cuccaro
- Margaret Pericak-Vance

University of Southern California
- Lilyana Amezcua

University of California, San Francisco
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- Noriko Isobe

Universidad Central Del Caribe School of Medicine
- Angel Chinea

IBD Studies (Hispanics)

University of Miami
- Maria Abreu
- Oriana Damas
- Alejandra Quintero

ERICH (stroke) Studies (Hispanics & African Americans)

University of Cincinnati
- Daniel Woo (PI)

Center for Excellence in Precision Medicine and Population Health (Hispanics & African Americans)

Vanderbilt University
- Consuelo Wilkins (PI)
- Nancy Cox (PI)

Meharry Medical College
- Maria de Fatima Lima (PI)

University of Miami
- Roy Weiss (PI)

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Morehouse School of Medicine
- Priscilla Igho-Pemu (PI)

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MS Studies

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NIH/NINDS (R01NS096212)

IBD Studies

NIH/NIDDK (R01DK104844)

Stroke Studies

NIH/NINDS (U01NS069763)

Center for Excellence in Precision Medicine and Population Health

NIH/NIMHD (U54MD010722)

Most importantly our Participants and their Families!

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