Is this the right time to study use of genomic data in health systems?

Carol Horowitz MD, MPH
Icahn School of Medicine at Mount Sinai
-Department of Population Health Science and Policy
-Center for Health Equity and Community Engaged Research

GUARDD
Genetic testing to Understand and Address Renal Disease Disparities

IGNITE
Implementing GeNomics In pracTicE

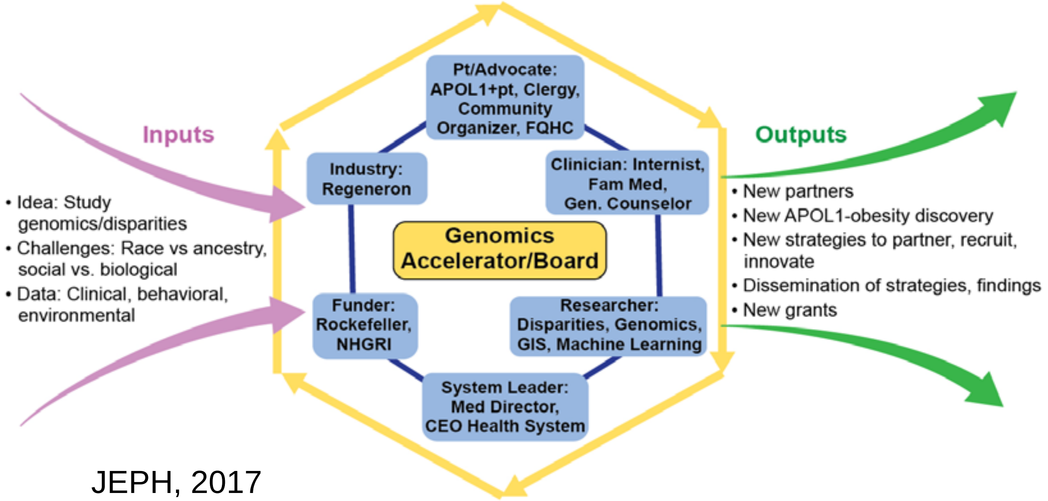
Mount Sinai

THE INSTITUTE FOR FAMILY HEALTH

CHECER

“*If you’re not at the table,*
you’re on the menu.”

- Whose decision is it? Who do we need to convince?
  - – Payer? Provider? Patient? Policymaker? Ourselves?
- Who is at our table? Nationally? In NYC?
- If we don’t study it, will it happen anyway?
  - – If we don’t study it, who will, and in which patients ($)?
  - – Our Board answer: **“***One needs to be vigilant. The research should proceed, but carefully*.”



Inputs

- Idea: Study genomics/disparities
- Challenges: Race vs ancestry, social vs. biological
- Data: Clinical, behavioral, environmental

Pt/Advocate: APOL1+pt, Clergy, Community Organizer, FQHC

Industry: Regeneron

Clinician: Internist, Fam Med, Gen. Counselor

Genomics Accelerator/Board

Funder: Rockefeller, NHGRI

Researcher: Disparities, Genomics, GIS, Machine Learning

System Leader: Med Director, CEO Health System

Outputs

- New partners
- New APOL1-obesity discovery
- New strategies to partner, recruit, innovate
- Dissemination of strategies, findings
- New grants

JEPH, 2017

Culture of understanding: Reflections and suggestions from a genomics research community board. Progress in Community Health Partnerships, 2017

Can genomic-based reach diverse participants and be equitably distributed?

Yes, if intentional, valued, resourced, done carefully.

“*How do you advance science in a good way and not take advantage of the vulnerability of a community?*”

“*The statement that looking at genetic risks by ancestry works against the Black community is traditional and stereotypical. Genomics can be integrated into community health.*”

Who’s rejecting who here?

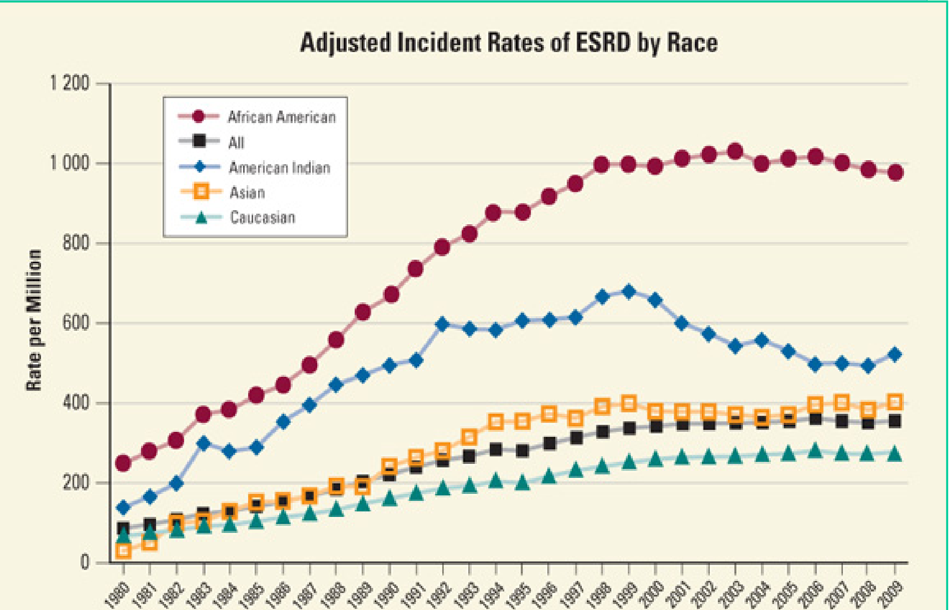
GUARDD

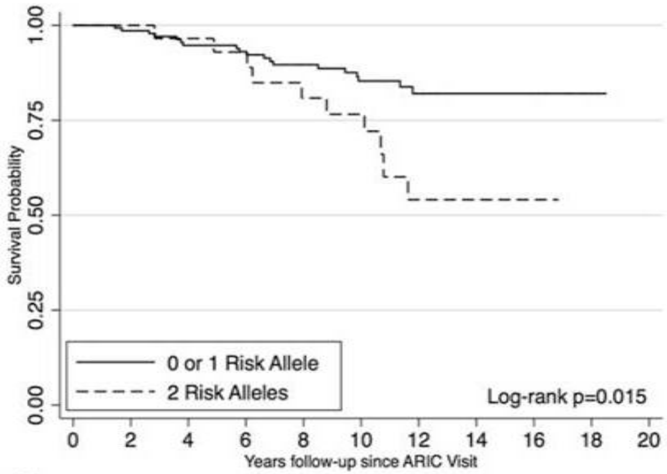
Genetic testing to Understand and
Address Renal Disease Disparities

CHECER

Example: *APOL1* Risk Variants

- African ancestry (AA) 3x odds of kidney failure
- 1/7 AA individuals have genetic variant protective of sleeping sickness
  - 10x odds of kidney failure if hypertension
  - Explains up to 70% of racial disparity in kidney failure





Number at risk											
0 or 1 Risk Allele	147	134	120	112	95	76	41	19	16	4	0
2 Risk Alleles	32	31	27	23	20	17	8	2	1	0	0

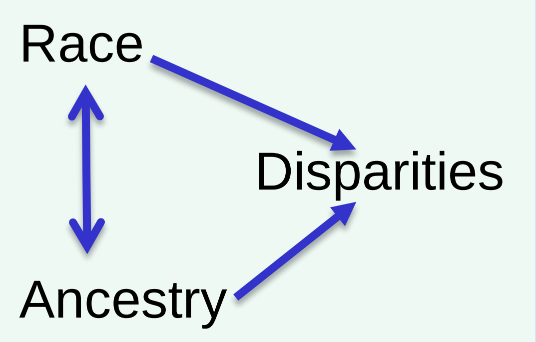
Figure 1. Participants with CKD and two APOL1 risk alleles progressed faster to ESRD than participants with CKD and zero or one APOL1 risk allele. Kaplan–Meier survival curves for ESRD-free survival by number of *APOL1* risk alleles among participants with

Go-No Go Research- Who decides?

- – Genetic ethicist: *“Don’t touch this- you will set the disparities movement back 30 years.”*
- – Elder Mimsie Robinson:

“Now maybe White doctors won’t judge Black people on dialysis as not caring enough or not being compliant. They’ll recognize that there’s more to disease than bad behavior.”

- Where we ended up: NHGRI RCT
  - – To incorporate genomics into primary care
  - – Stakeholder engaged, patient-centered



Race

Disparities

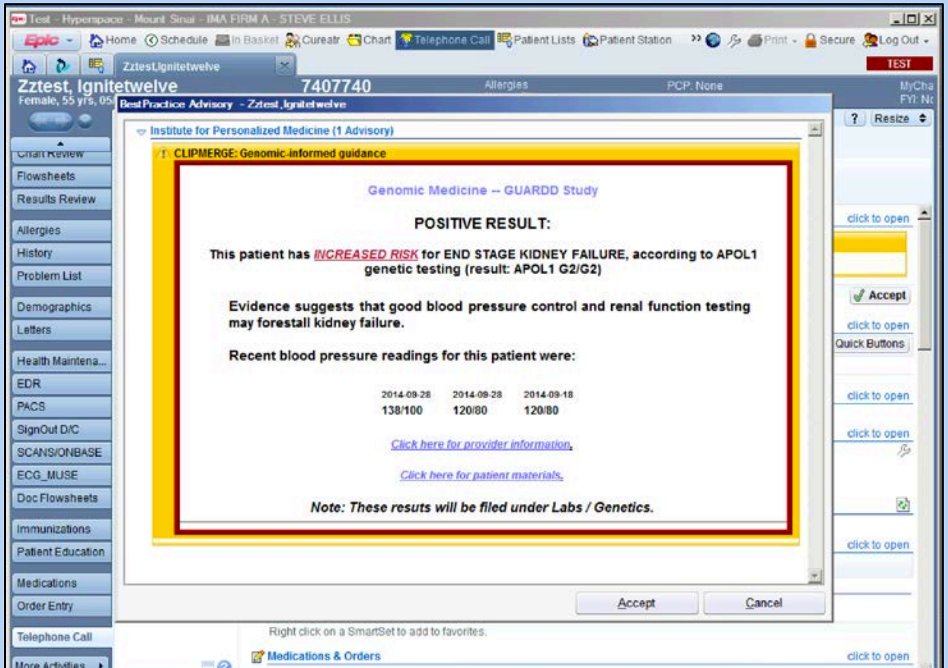
Ancestry

GUARDD

Genetic testing to Understand and Address Renal Disease Disparities

The GUARDD Study Design

- <u>Eligibility:</u> Self-reported AA; 18-70 yrs, HTN, no DM, CKD
- <u>Randomize</u>: *APOL1* testing *vs*. delayed testing in 1y
  - – GC- trained staff to test patients, return results
  - – GC available for patients (never used)
  - – Providers get results as BPAs in EHRs



Best Practice Advisory - Zztest, Ignitetwelve

Institute for Personalized Medicine (1 Advisory)

CLIPMERGE: Genomic-informed guidance

Genomic Medicine -- GUARDD Study

POSITIVE RESULT:

This patient has *<u>INCREASED RISK</u>* for END STAGE KIDNEY FAILURE, according to APOL1 genetic testing (result: APOL1 G2/G2)

Evidence suggests that good blood pressure control and renal function testing may forestall kidney failure.

Recent blood pressure readings for this patient were:

2014-09-28	2014-09-28	2014-09-18
138/100	120/80	120/80

<u>Click here for provider information.</u>

<u>Click here for patient materials.</u>

Note: These resuts will be filed under Labs / Genetics.

Right click on a SmartSet to add to favorites.

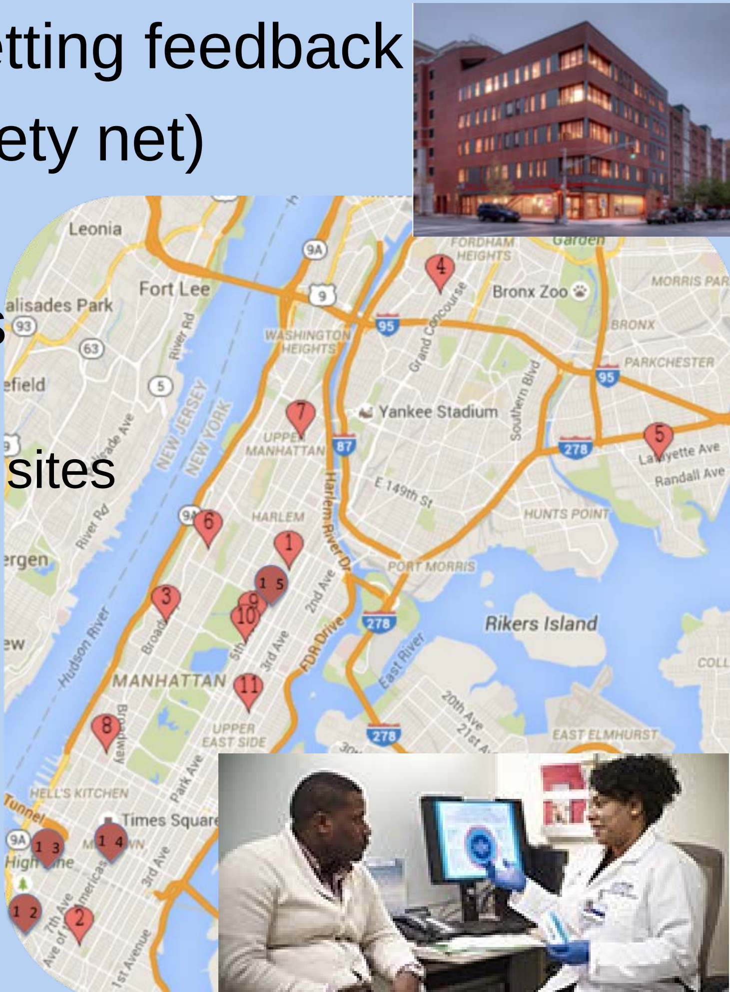
GUARDD

Genetic testing to Understand and Address Renal Disease Disparities

CHECER

Methods

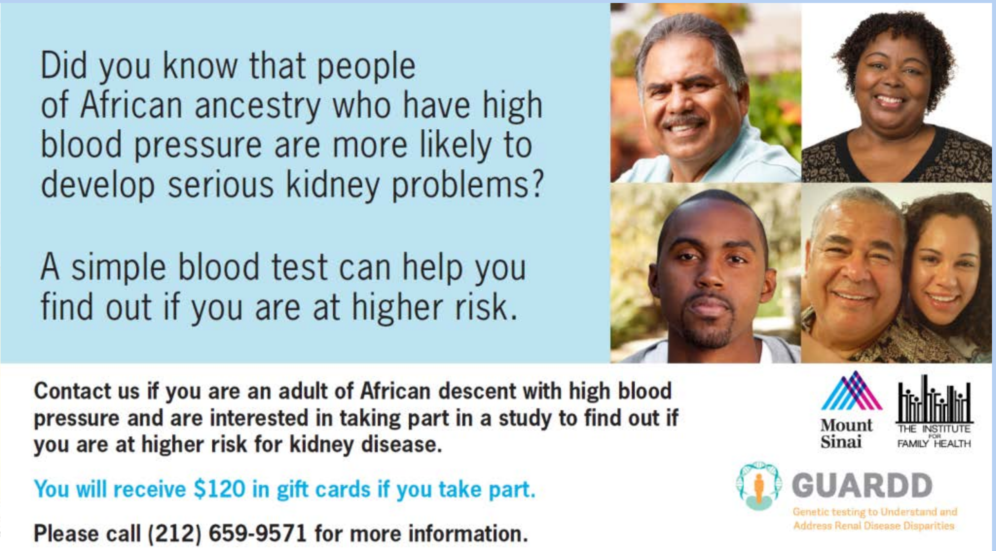
- Community- Clinical- Academic team developed
- Began with formative research (so no surprises)
- Roadshow- introducing GUARDD, getting feedback
  - – 5 fed. qualified health centers (safety net)
  - – 6 neighborhood practices
  - – 4 academic primary care practices
    - Part of system with 4.5m patients,
      13 ac & comm hosp's, 350 outpatient sites
      7000MDs, ~50% non-white
  - – Many genomics/research naïve/concerned- PCP's like me



"*A good invitation to a good party*"

(BTW, who's your invitation meant for?)

- When's good for you? (anytime, anywhere)
- Recruiters- from/of community, build relationships, continuity, help each other
- Recruitment feels like it's from a neighbor (it is)
- Used appropriate, language, literacy, graphics



Did you know that people of African ancestry who have high blood pressure are more likely to develop serious kidney problems?

A simple blood test can help you find out if you are at higher risk.

Contact us if you are an adult of African descent with high blood pressure and are interested in taking part in a study to find out if you are at higher risk for kidney disease.

You will receive $120 in gift cards if you take part.

Please call (212) 659-9571 for more information.

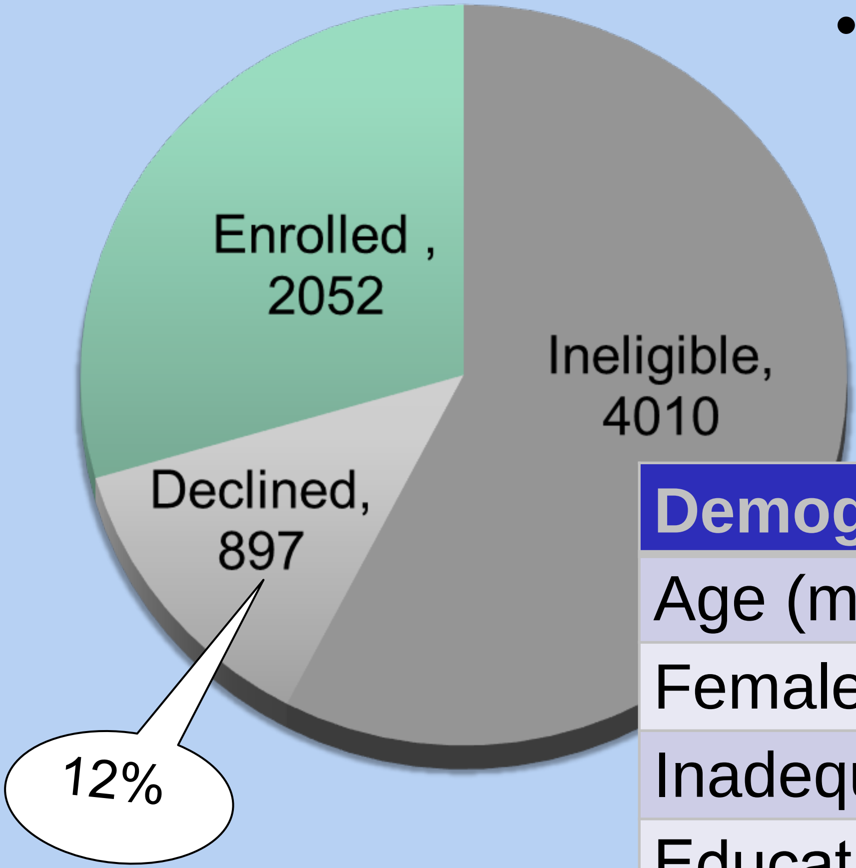
Mount Sinai

THE INSTITUTE FOR FAMILY HEALTH

GUARDD

Genetic testing to Understand and Address Renal Disease Disparities

Results: Completed Enrollment in 2y Retained 93% 3m, 90% 12m



- Difficult to Reach Population
  - Homeless/Mobile
  - Recently incarcerated
  - Competing demands
    - Food/Jobs
    - Professional Conflicts

Demographics (100% AA)	%
Age (mean, range)	53 (18-70)
Female (same as sample)	67
Inadequate Health Literacy	20
Education: ≤ HS degree	44
Income <$30,000	53
Non-adherent to BP meds	48
Uncontrolled BP (≤140/90)	47

GUARDD
Genetic testing to Understand and Address Renal Disease Disparities

Provider Survey n=486 response 89%
48% non-White: Know your target audience

Item	
Trained: Had Formal Genetics Course	79%
Experienced: Ordered genetic test in last year	36%
Prepared: For patients who had genetic test for a chronic disease	25%
Concerned: Re insurance discrimination Don’t trust genetic testing companies	53% 76%
Pt Response: Motivate behavior change (prediction) Will try harder to control BP	34% 69%
Race/Ancestry: Clue for who needs genetic test Genes are partly reason for racial/ethnic disparities	75% 81%

Patient Survey (n=2052) : Genomic Beliefs, Concerns

Item	%
Had previous genetic test	11
Understand genetic testing	12
Good idea to get genetic testing to assess chronic disease risk (genes effect this risk)	96
Want children tested for APOL1 variants	78
Worried about the privacy of results	11
Think doctors would discriminate against people with genetic chronic disease risk	5

GUARDD
Genetic testing to Understand and
Address Renal Disease Disparities

CHECER

3 month survey (no diff. than baseline)

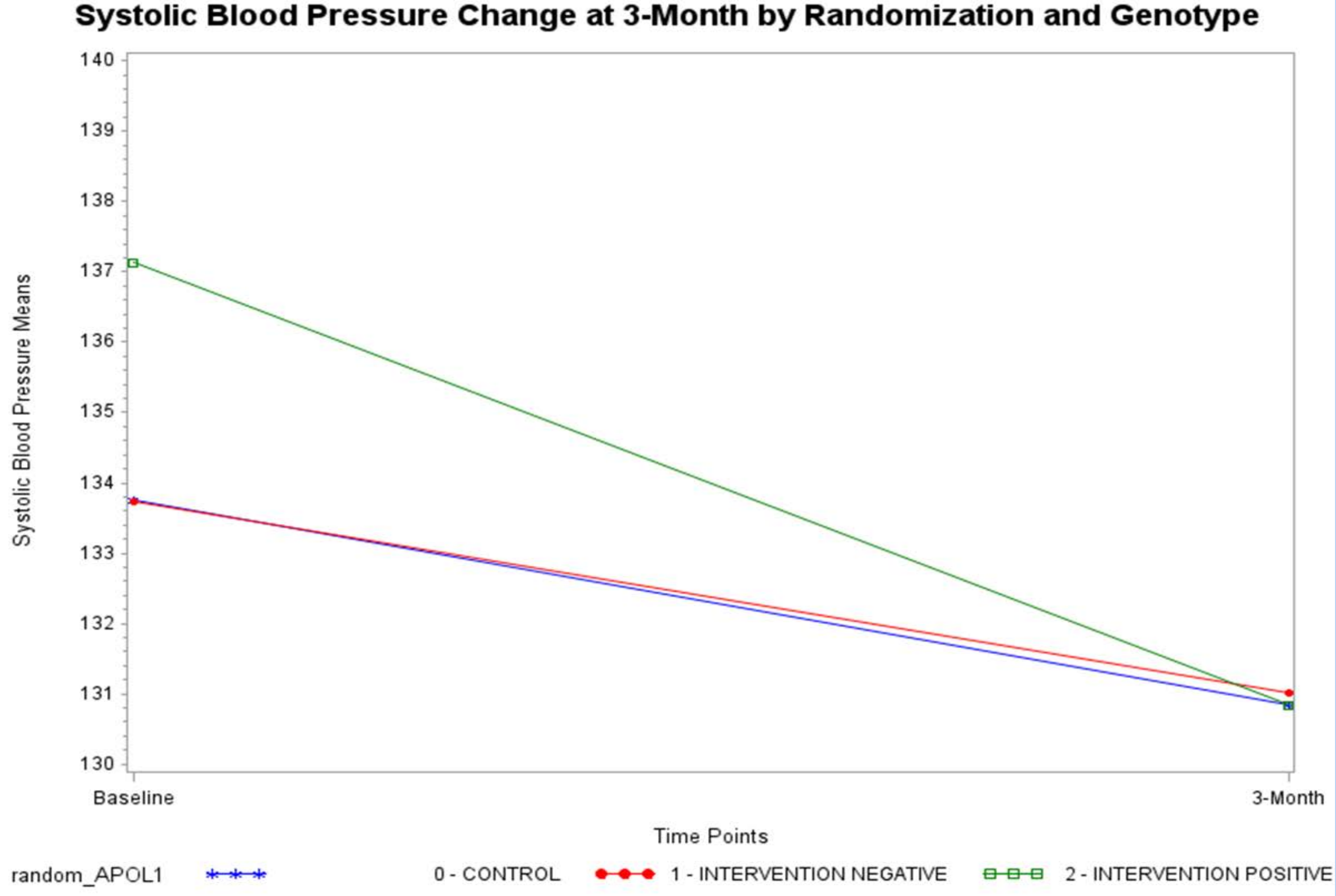
Item	%
Don’t trust their doctor	45
Doctors don’t treat B&W patients equally	67
B more likely to have chronic diseases than W	88
- Genetics contributes to this disparity	82
How docs view you think if disparity is genetic	
- No change in view	63
- More negative	13
- More positive	24
Would get tested again	95
Satisfied w/timing, type, amount of info they got	>95%

GUARDD

Genetic testing to Understand and Address Renal Disease Disparities

CHECER

Study had positive results (some docs were right)



Systolic Blood Pressure Change at 3-Month by Randomization and Genotype

GUARDD

Genetic testing to Understand and Address Renal Disease Disparities

Associated with self-reported improvements in BP med use

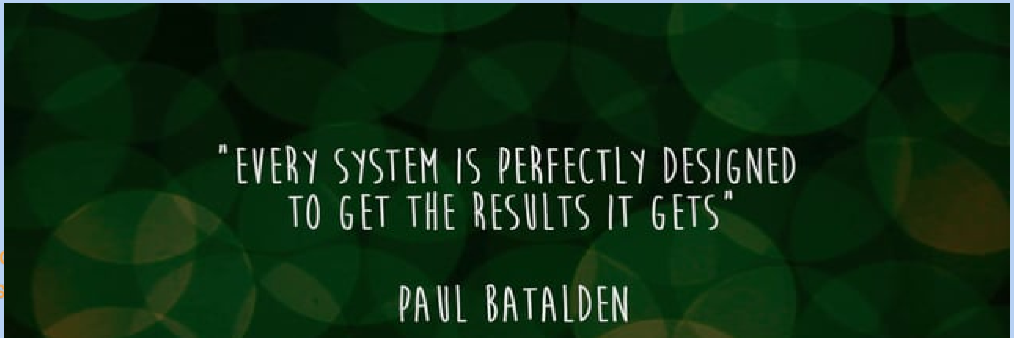
CHECER

“You want to learn about us? Include us.”

Diverse populations/sites should not be an add-on, afterthought, or funding strategy. If we MUST diversify, we will.

“The culture of understanding is far more important than the culture of fear, and the culture of understanding has no color. *Our success came from willingness and ability of all stakeholders to challenge their views about race, racism, ancestry, genomics and research.”*

“This voice needs to be harnessed. *People will become engaged when someone who looks like them is at the helm.”*



"EVERY SYSTEM IS PERFECTLY DESIGNED TO GET THE RESULTS IT GETS"

PAUL BATALDEN

GUARDD

Genetic testing to Understand an
Address Renal Disease Disparities

CHECER

Conclusion: Do it now, do it right.

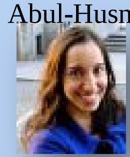
Make diversity/engagement a priority from the get go

Thanks to, and from our Genomics Board


Neil Calman


Carolyn Caddle-Steele
Pt & Funder
(Rockefeller Fund)


Kadija Ferryman
Anthropology


Noura Abul-Husn
Industry
(Regeneron)


Crispin Goytia
Patient Engagement


Mel Gertner
Pediatrician. Children's Advocate


Mimsie Robinson
Community Organizing


Eric Gayle
Clinical Medicine


Lynne Richardson
Clinical Trials, Disparities


Gregory Chisolm
Faith Community


Warria Esmond
Community Health

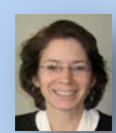

Randi Zinberg
Genetic Counseling


Aniwaa Owusu-Obeng
PGX


Carol Horowitz

Program Management


Michelle Ramos


Diane Hauser

GUARDD
Genetic testing to Understand and Address Renal Disease Disparities

CHECER
Center for Health Equity and

Thanks to our partners and funders

NHGRI IGNITE Implementing GeNomics In pracTicE

NCATS Conduits INSTITUTES FOR TRANSLATIONAL SCIENCES

And thank you!

GUARDD

Genetic testing to Understand and
Address Renal Disease Disparities