

# Precision Health for All

## NIH's Precision Medicine Initiative<sup>®</sup>

### Cohort Program

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National Institutes of Health

**NHGRI Council Sept 12, 2016**

# KNOW ENOUGH TO BE DANGEROUS...AND APPRECIATIVE

## BOX 5

### Genomics and society



Effectively examining the societal implications of genomic advances requires collaborations involving individuals with expertise in genomics and clinical medicine and experts in bioethics, psychology, sociology, anthropology, history, philosophy, law, economics, health services research and related disciplines.

**Psychosocial and ethical issues in genomics research.** These include ensuring appropriate protection of human research participants and addressing the perceptions of risks and benefits of participating in genomic studies; expanding the diversity of research cohorts; incorporating biological ancestry markers and self-identified race and ethnicity as variables in genomic studies; accomplishing effective community engagement; and including vulnerable populations (for example, children and the disabled) and deceased individuals in genomics research.

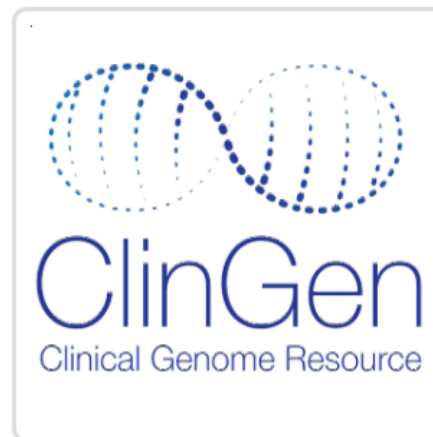
**Psychosocial and ethical issues in genomic medicine.** These include communicating with patients about the uncertainty and evolving nature of predictions based on genomic information; interpreting information from direct-to-consumer genetic tests; ensuring fair access to genomic medicine; assessing the effectiveness of genomically informed diagnostics and therapeutics; using genomic information to improve behaviour change interventions; addressing issues associated with pre-implantation, prenatal and postnatal genetic diagnoses; and determining how constructs of race and ethnicity relate to the biology of disease and the potential to advance genomic medicine.

**Legal and public policy issues.** These include intellectual property in genomics; insurance reimbursement for genomic services; regulation of genetic testing; regulatory and non-regulatory approaches for dealing with direct-to-consumer genetic testing; the regulation of pharmacogenomics and genomics-based therapeutics; protection against genetic discrimination and stigmatization; and uses of genomics in non-medical settings.

**Broader societal issues.** These include the implications of increasing genomic knowledge for conceptualizing health and disease; for understanding identity at the individual and group levels, including race and ethnicity; for gaining insights about human origins; and for considering genetic determinism, free will and individual responsibility.



Learn from your pioneering work on  
EMRs and biorepositories!



Continue to drive data sharing,  
curation, & trust in “meaningful use” of variant data!

Understand how to situate  
this new knowledge  
in our culture!

# APPRECIATIVE OF YOUR WORK MORE THAN YOU KNOW

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First week of very imprecise chemo, summer 1989, Chapel Hill at age 19

## **WORLD FELL APART: Sept 11, 2001, Portland**

*Exhausted. Quick. Much more later. Like I need to write this to remember, but planes destroyed WTC in NYC and Pentagon attacks. Saw horrors wish I could un-see. Wonder if more will hit tonight.*

*Ash and I can't sleep—talking mostly about fear and, in a way, glad this whole ride has meant we never brought kids into this insane world. Am not afraid of dying in a terror attack (though I do worry bombings are now in our daily lives as in many parts of world). **Cancer will certainly save me from an explosion—there's one benefit!***



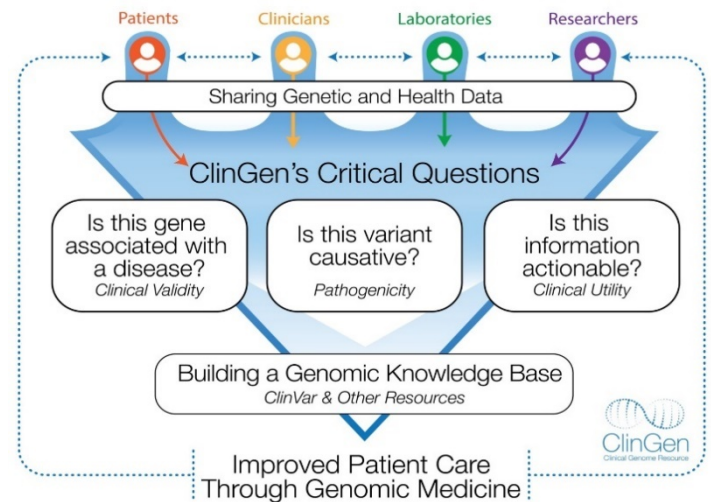
Have kept a journal since 3<sup>rd</sup> grade

# GOOD, CONSISTENT ADVICE FROM MANY LEADERS IN GENOMICS...

*One thing Rehm would definitely not recommend? Spending all of the Precision Medicine Initiative budget—President Obama proposed \$215m to start—on sequencing.*

*“I would argue you need to **spend more time focusing on the effective collection of phenotypes** to then correlate that [genomic] data to,” she says, advocating for smaller pilot projects to successfully link genotypes and phenotypes before sequencing the whole cohort.*

*The best way to design infrastructure, she believes, is to define projects where you are seeking answers to specific questions, and **use the questions and expected answers to drive the building of infrastructure.***



# A review & status update of PMI CP

# THE PRECISION MEDICINE INITIATIVE® COHORT PROGRAM

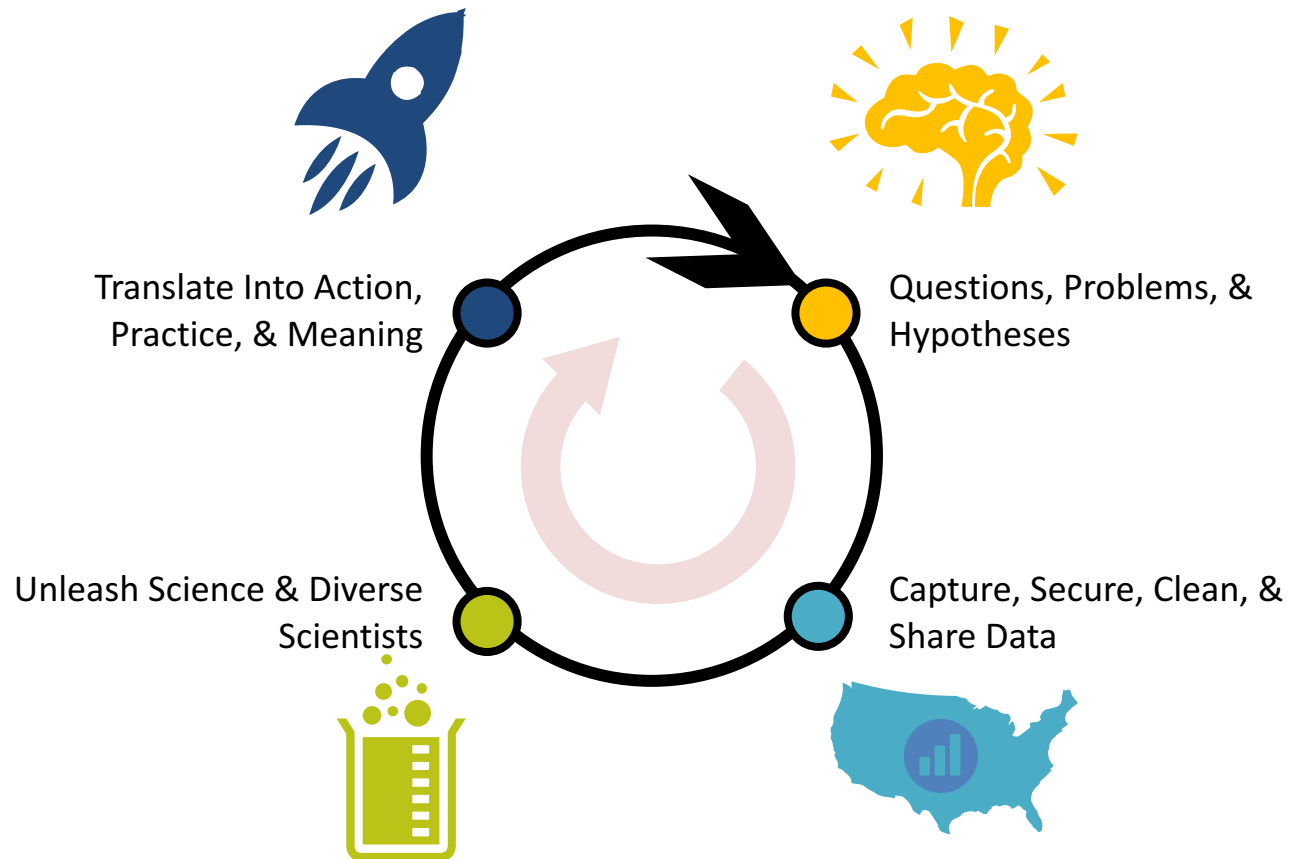
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- One million or more volunteers, reflecting the broad diversity of the U.S.
- Opportunities for volunteers to provide data on an ongoing basis
- Data shared freely and rapidly to inform a variety of research studies



# MISSION: ACCELERATE SCIENCE & BREAKTHROUGHS THAT DRIVE TOWARDS PRECISION HEALTH FOR ALL!

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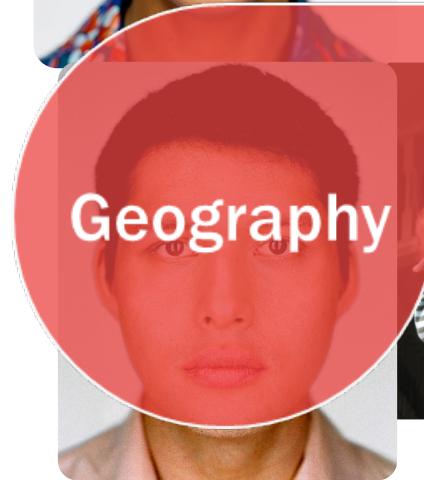
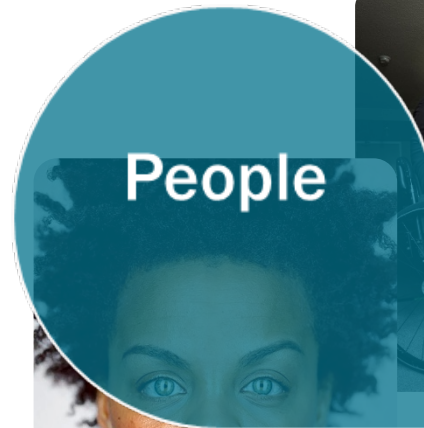


# A TRANSFORMATIONAL APPROACH TO DIVERSITY

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Reflecting the country's rich diversity to produce meaningful health outcomes for historically underrepresented communities

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# A TRANSFORMATIONAL APPROACH TO PARTICIPATION

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Participants in the PMI Cohort Program will be true partners—not patients, not subjects—in the research process

Involved in every step of program development

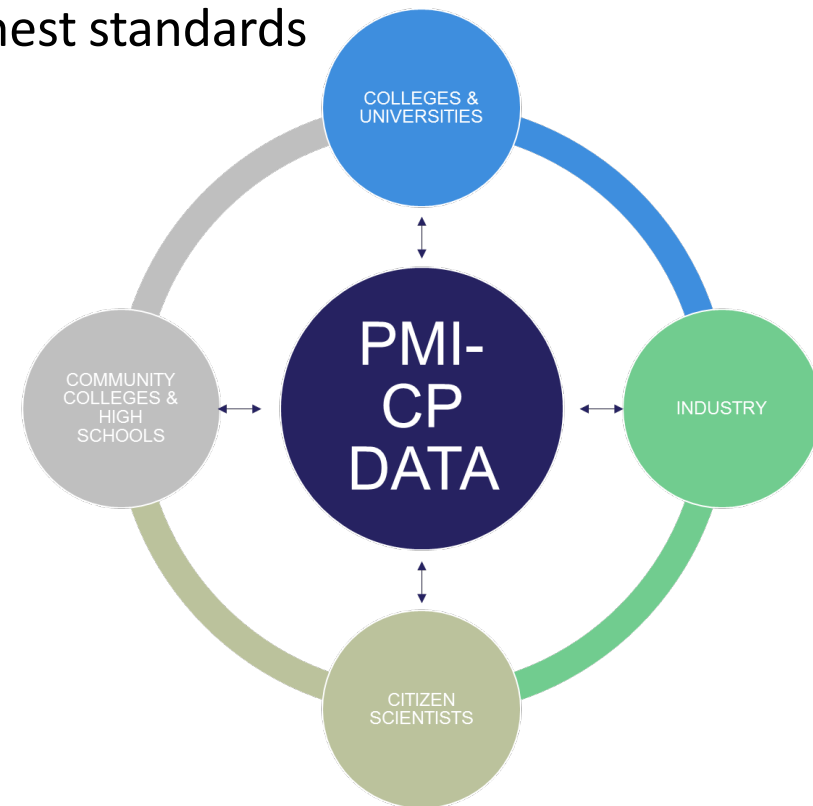
- What data we collect
- What lab analyses we do
- What research is conducted
- How data gets returned



# A TRANSFORMATIONAL APPROACH TO DATA ACCESS

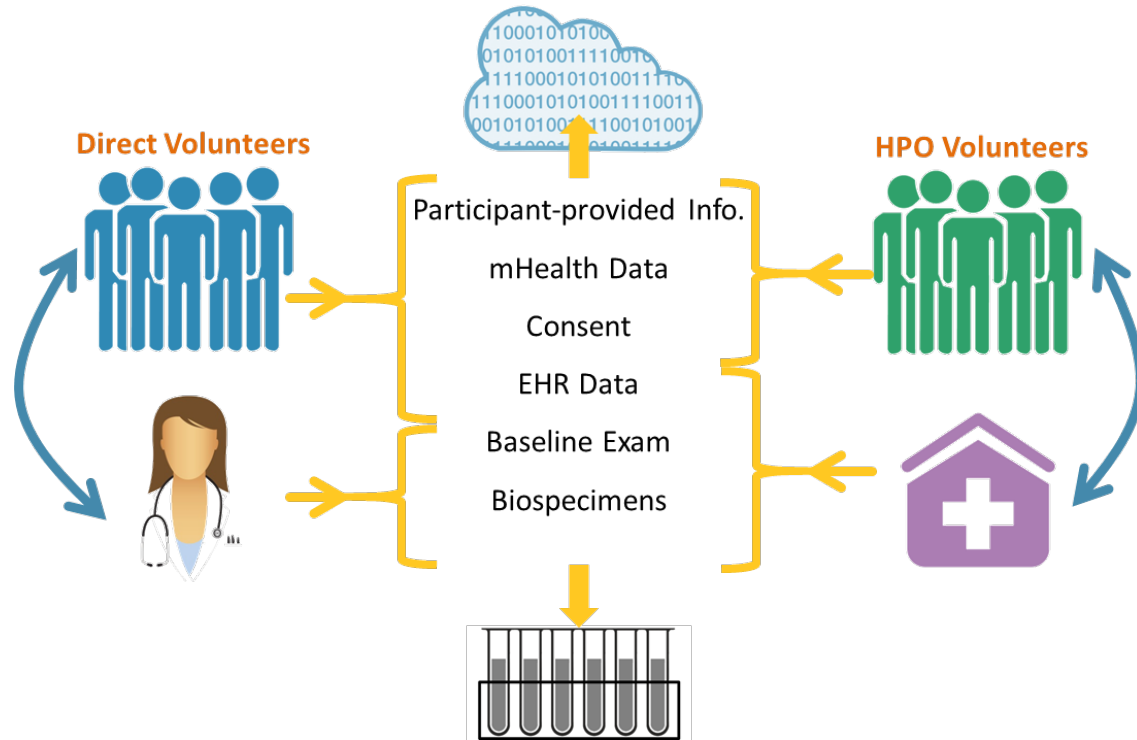
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- Data sharing will be swift to both researchers and participants
- Data collection will start small and will grow over time
- Privacy and security will adhere to the highest standards
- Will invest to level the playing field so diverse researchers can play



# TWO METHODS OF ENGAGEMENT

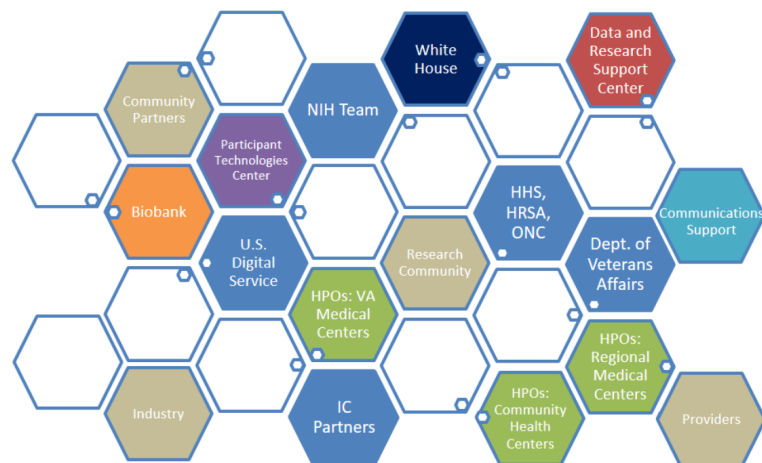
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# RIGHT NOW: BUILDING INTERDISCIPLINARY PLATFORM TEAM

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- **Act as one team with diverse participation**
  - To deliver 1M engaged volunteers & rich, diverse data over long period of time
- **33 awardee partners so far:**  
11 Working Groups / “Departments” . . . Steering & Exec Committees
  - Biobank, Data and Research Support Center, Health Care Provider Organizations, and Participant Technologies Center
- **Hire core NIH leadership team**
  - Deputy, CTO, CMO, Comms Chief, User Experience Chief . . .
- **Develop transition plans with multiple government agencies** to maintain & grow mutual support
- Build robust **community partners network**
- **Use known industry methods:** user-centered design, platform development process



# RIGHT NOW: TESTING & PILOTING PIECES OF THE PROGRAM

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- **Rolling out personas** built upon ethnographic work in homes/focus groups
- Building diverse **5,000-person community** of beta testers
- **Testing consent language & 5 survey modules** and defining next 12 modules
- Iterating **website, smartphone, 1-800#, & data center infrastructure**
- Getting **feedback on content** for educating the public and participants
- Testing **name, brand, look and feel**
- Experimenting with **recruitment methods**
- Drafting **full protocol for V1 launch**
- **Cybersecurity testing**

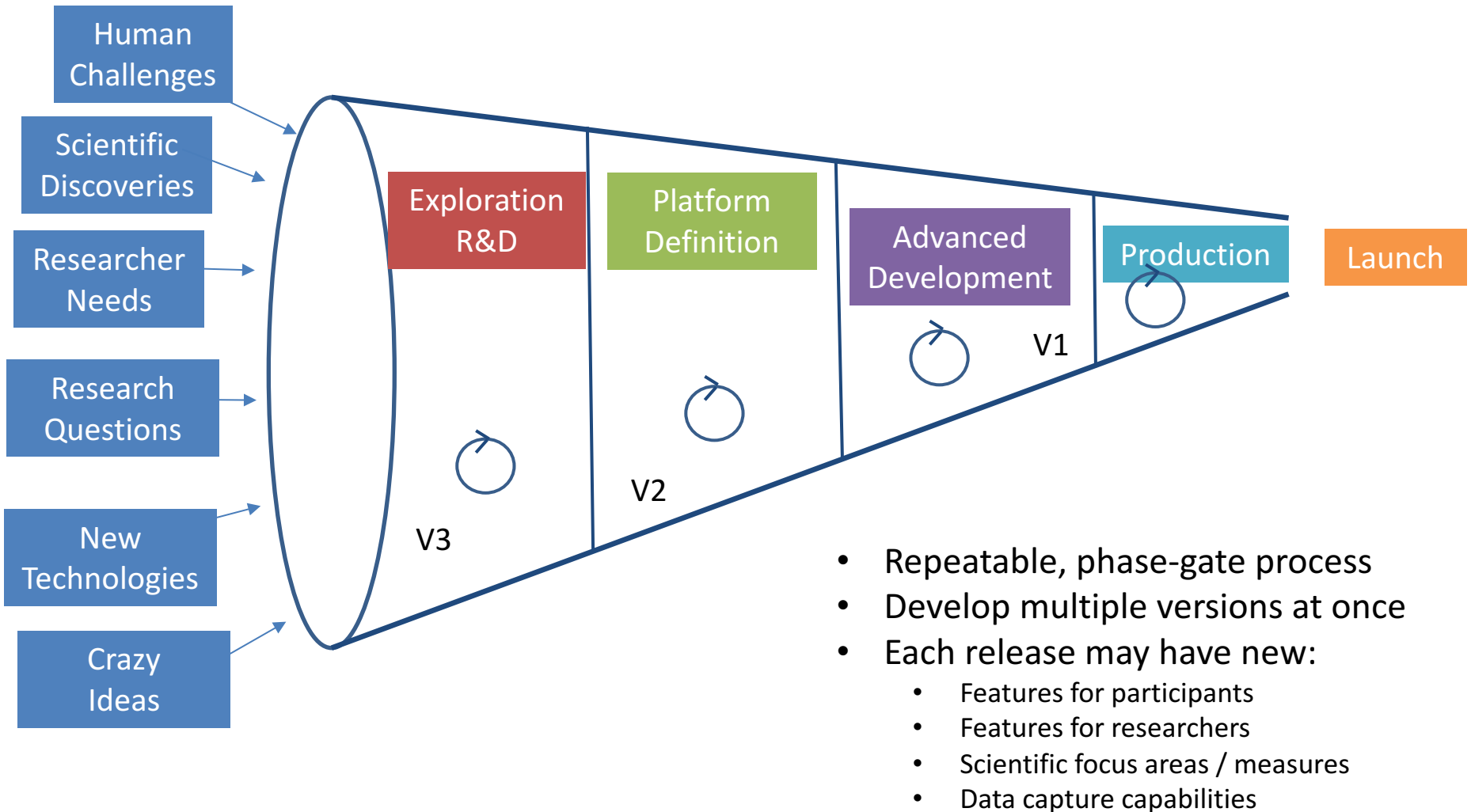
**Not For Me** Jasmine, 34



*“ I worry about the government knowing more about me than I do about myself. Do I want people to have access to my data? Even if means I could be healthier? Even if it means they could catch my cancer sooner? I’m not sure.”*

What I mean by “platform approach”

# PLATFORM DEV. LIFECYCLE: INNOVATION TO IMPLEMENTATION



# DEFINING OUR TARGET “LANDING ZONES”

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- Must have clear definitions of the “product” to build reliable schedules
- Cannot solve for “features” and “schedule” and “resources” at same time
- Landing zones allow for tradeoffs of time and money

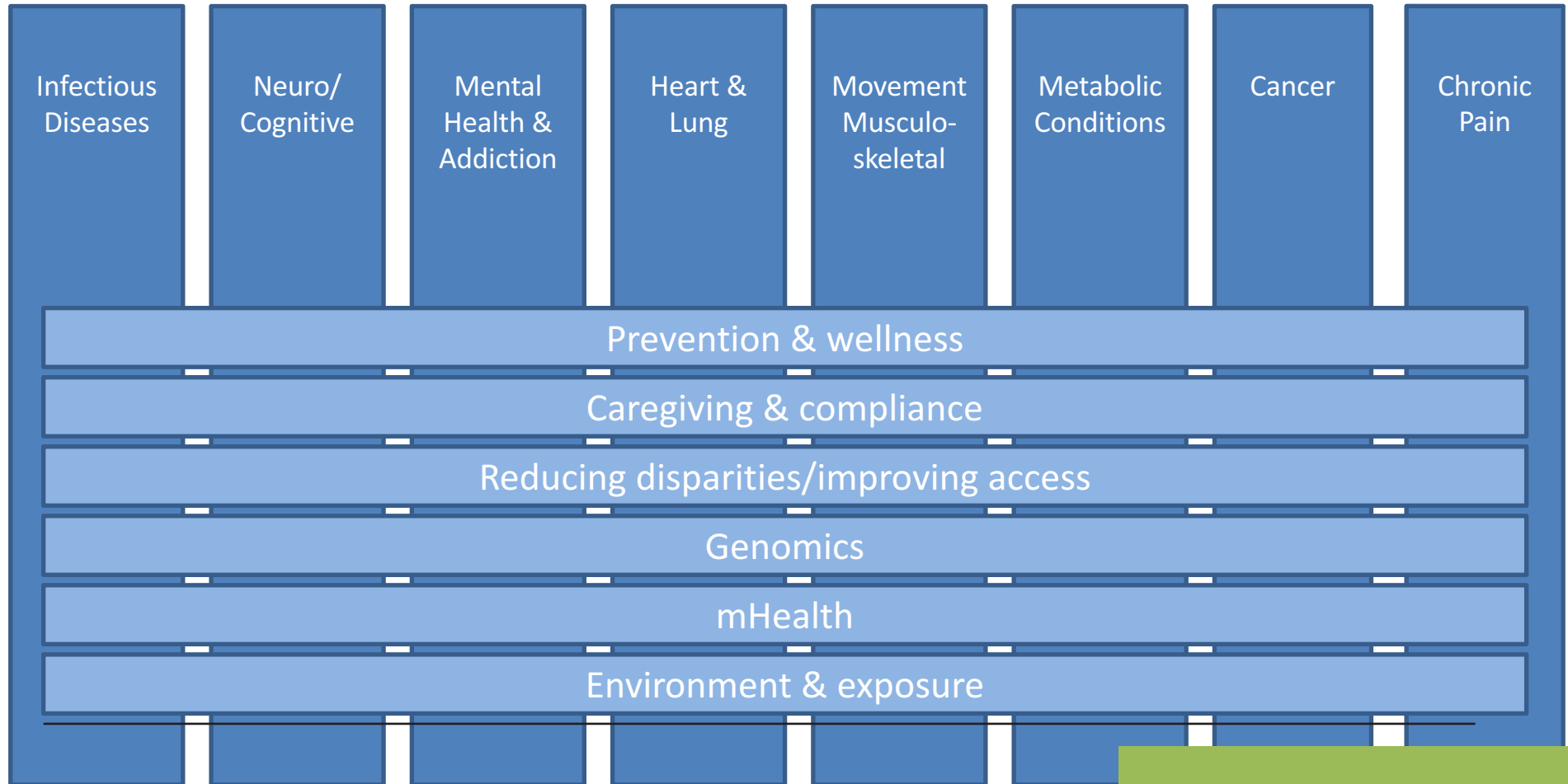
Departments/Work Streams	Minimum	Goal	Stretch Goal
User experience: Participants Researchers Providers Community Partners			
Research questions/protocols			
Technologies/touchpoints			
Security & privacy			
Communications/messaging			
Outreach/partnerships			
Policy & legal			

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# SAMPLE FRAMEWORK FOR RESEARCH QUESTIONS & REQUIREMENTS

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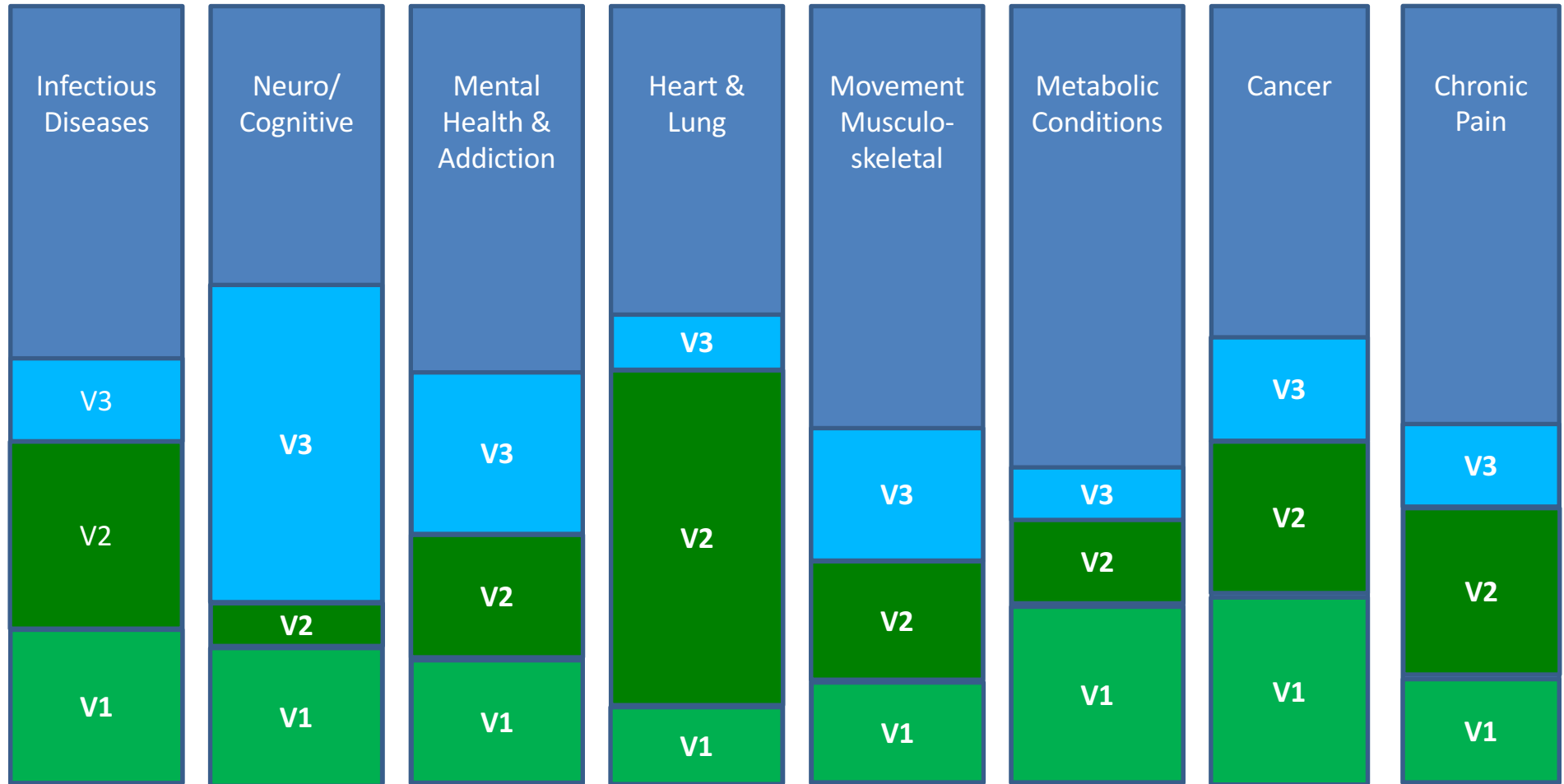


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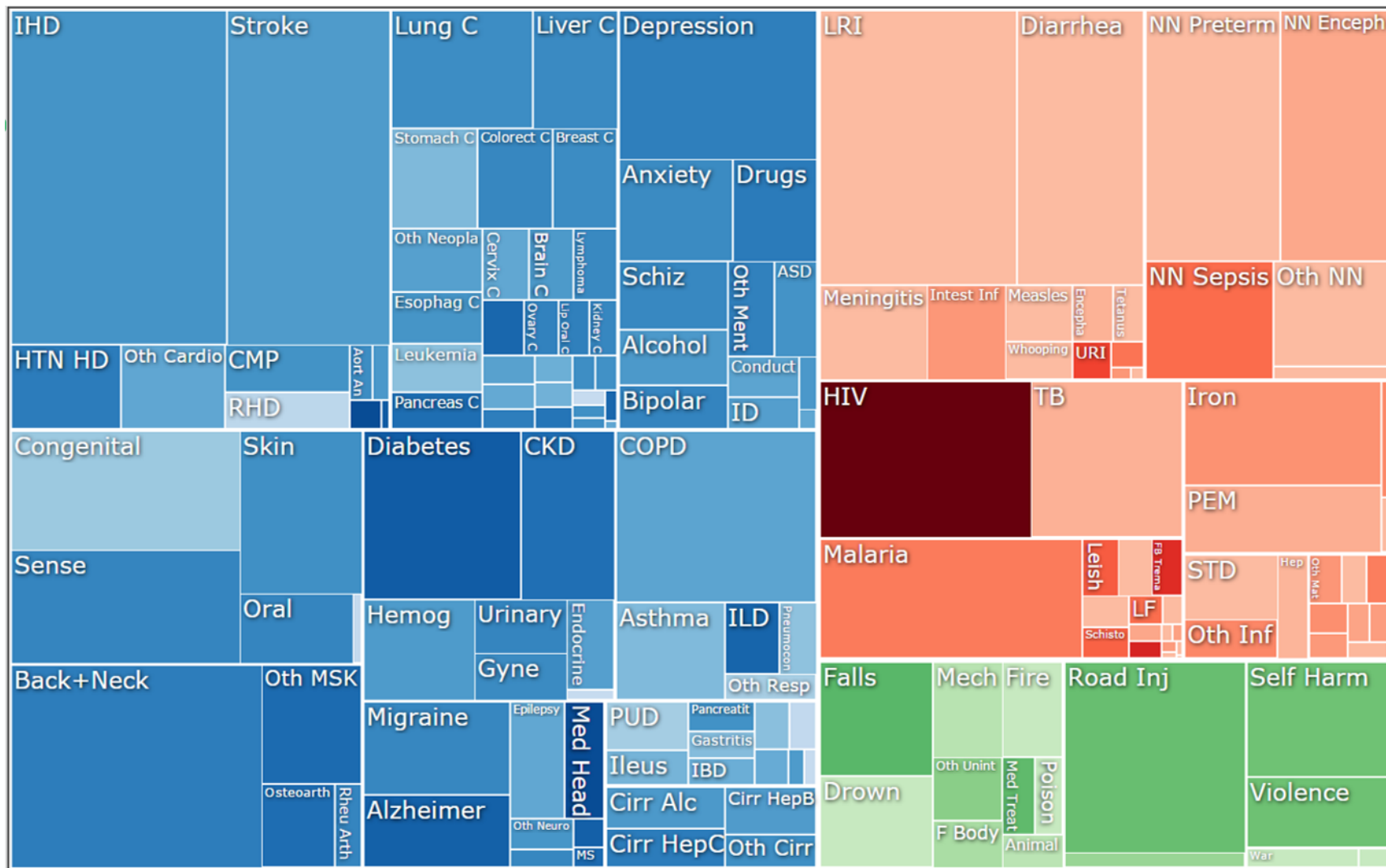
**Examples of Scientific Bodies of Knowledge**

# VERSIONS IN TIME: DIFFERENT REQUIREMENTS/RES. QUESTIONS

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# ULTIMATELY WANT TO FOCUS ON WHERE HUMAN NEED IS GREATEST



Credit: Causes of DALYs globally, 2013. Dr. Christopher Murray, Institute for Health Metrics and Evaluation, University of Washington <http://vizhub.healthdata.org/gbd-compare>

## NEXT UP: RESEARCH QUESTIONS WORKSHOPS

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- **PREMISE TO ADOPT FOR THE WORKSHOPS:** Imagine PMI CP is only to accelerate your body of thought and research in a particular domain!
  - **THREE BIG QUESTIONS:**
    1. NEAR TERM: what are low-hanging fruit questions/measures for which the scale of PMI CP could help accelerate knowledge & breakthroughs?
    2. MID-TERM: what kinds of questions might this CP answer where additional work selecting among measures/instruments is needed?
    3. LONG TERM: what kinds of questions are ripe for CP of this size but for which we need fundamental science & tech to develop the instruments?
  - **KINDS OF ATTENDEES:**
    - Research experts, co-funders, participant/advocacy orgs, providers, etc.
  - **WHAT WE'LL DO:**
    - Factor analysis of common needs across multiple knowledge domains = our “versions”
    - Partner with domain stakeholders to grow funding for unique areas = sub-studies
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Closing thoughts . . .

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# KEY OPPORTUNITIES FOR NHGRI COLLABORATION

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- Defining our genomics roadmap and plan in detail
- Research question workshops—genomic strategy on each domain
- Find ways to drive more ethical, legal, social implications research as we *go/grow*
- Explore what CLINGEN becomes over time as PMI CP capacity kicks in

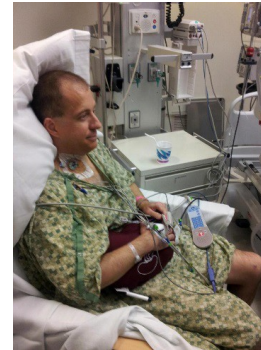


# FROM LUCK FOR THE FEW TO PRECISION HEALTH FOR ALL

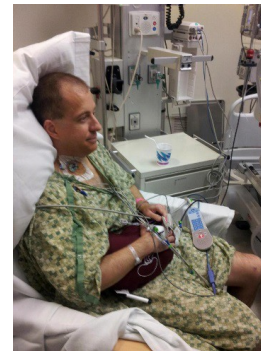
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“Luck & Pluck”



Routine  
Care

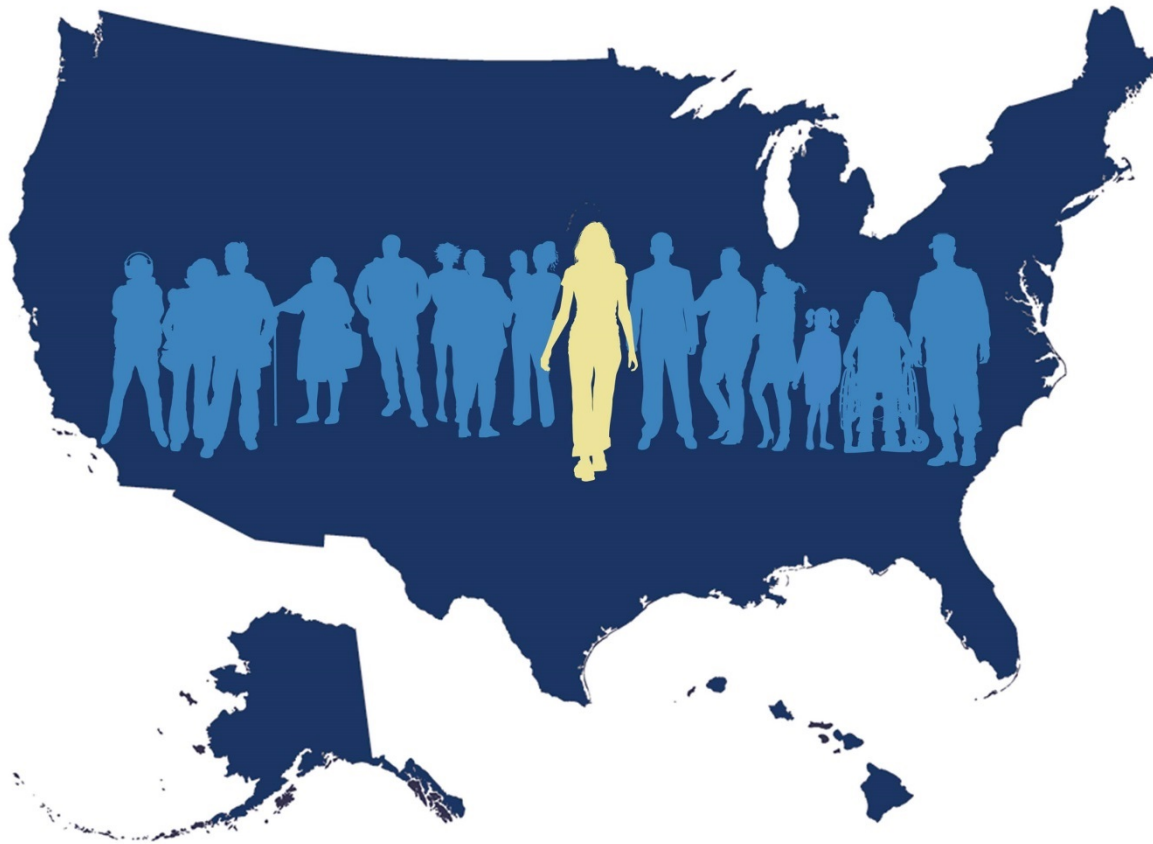


LOOK FORWARD TO WORKING WITH YOU MAKE THIS DREAM A REALITY!

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# QUESTIONS?

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Sign up for updates at: <https://www.nih.gov/precisionmedicine>

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