## Equity in Genomics and Precision Public Health

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## Equity in Genomics and Precision Public Health

"The global applicability of genomic knowledge, fair and even access to genomic services such as testing and counseling, and unbiased implementation of genomic medicine." NHGRI

#### Scientific challenges for precision public health

Frank Kee,<sup>1</sup> David Taylor-Robinson <sup>2</sup>

#### ABSTRACT

The notion of 'precision' public health has been the subject of much debate, with recent articles coming to its defence following the publication of several papers questioning its value.

Critics of precision public health raise the following problems and questionable assumptions: the inherent limits of prediction for individuals; the limits of approaches to prevention that rely on individual agency, in particular the potential for these approaches to widen inequalities; the undue emphasis on the supposed new information contained in individuals' molecules and their 'big data' at the expense of their own preferences for a particular intervention strategy and the diversion of resources and attention from the social determinants of health.

In order to refocus some of these criticisms of precision public health as scientific questions, this article outlines some of the challenges when defining risk for ind the limitations of current theory and study design precision public health; and the potential for unir harms. spotlights targeted or personalised interventions for lifestyle and behavioural change following individual genetic or digital profiling, for example, using smartphone apps. This version of PPH is our main focus. While the Lancet editor warns against 'uncritical techno-optimism' with regard to PPH, he also decries unsustainable 'pilotitis', an excess of pilot studies leading to delayed or foregone PPH benefits,<sup>1</sup> and stresses the need for workforce training if the potential of PPH is to be realised.1 This position leaves us rather confused and raises still further questions. Pilot studies are usually the necessary antecedents of robust scientific studies of mechanisms, efficacy or effectiveness, and it is precisely in this realm that the evidence for PPH remains questionable or at least undercooked. In spite of this, PPH is front and centre in the UK Health Secretary's forward strategy for person-

#### Precision public health: pitfalls and promises

We were delighted that Richard Horton (Oct 27, 2018, p 1504)<sup>3</sup> continued the conversation we<sup>2</sup> aimed to start about precision public health. However, Horton incompletely characterises our assessment<sup>3</sup> of precision public health. He argues that precision public health does not sideline political and social influences on health. In fact, he argues

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argue<br/>labelSome public-health researchers are embracing data and technology to target smalladop<br/>two<br/>narregroups with precise health interventions. Others fear that these tactics could fail<br/>millions.

non-, part of public health's core mission

## "Precision" Public Health — Between Novelty and Hype

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✓ ▲ Authors: Merlin Chowkwanyun, M.P.H., Ph.D., Ronald Bayer, Ph.D., and Sandro Galea, M.D., Dr.P.H. Author Info & Affiliations

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# Why does equity in genomics and PPH matter?

# **KFF** Health News

### Genetics Studies Have a Diversity Problem Researchers Struggle to Fix

Research underway in South Carolina aims to better understand how genes impact health risks such as cancer and heart disease. Project leaders seek a diverse pool of participants but, so far, have struggled to recruit Black residents.

#### EXHIBIT 3

Numbers of genomewide association studies and genotype and phenotype studies, by disease area and study population demographic group, 2017



**SOURCE** Authors' analysis of data from the Genome-Wide Association Study Catalog and the database of Genotypes and Phenotypes (dbGaP). **NOTES** Underrepresented minorities are explained in the text. GWAS is genome-wide association study.

• Limits the benefits of newly developed tools for assessing risk, prognosis, and therapeutic benefit

- Inadequate risk models
- Issues estimating population prevalence (high rates of uncertain significance and novel deleterious mutations)

#### 81% of European ancestry

Enrollment of racially and ethnically diverse participants is essential for promoting health equity in genomics-informed preventive services and clinical care.

# Disparities in Implementation of Genomics

#### Disparities documented in BRCA1/2 testing for:

- Awareness of testing discussion with health care provider
- Referrals to genetics
- Specialists and centers
- Rates of testing
- Time to surgery and chemotherapy
- Rates of risk-reducing mastectomy
- Adherence to endocrine therapy

"What is the value of modernizing technologies to pinpoint the next big outbreak or genomic marker if individuals who are the most vulnerable to its potential impact are wary of getting screened—or unable to do so—because of systemic barriers to care?" Choate, 2023

## What are best approaches for improving equity in genomics and PPH?

## Shifting to a Strengthsbased Approach

- Recruitment strategies do not adequately address engagement preferences of diverse populations
- Require more robust stakeholder engagement and high-touch, relationship centered efforts
- Community capacity building and equipping already established, trusted groups

"...many African Americans don't take part because they just aren't asked." **KEEP IT (Keeping Each Other Engaged via IT):** Training for Community Health Workers about Hereditary Breast and Ovarian Cancer among Black Women

Three days with four-hour sessions each date 26 total participants

#### Modules included:

- Hereditary breast and ovarian cancer
- Family history, family history collection tools
- Communicating about hereditary breast and ovarian can
- Genetics 101
- Genetic Testing and Counseling
- HIT (Telehealth and Telegenetic Platforms/EHR/Patient Port
- Ethical, Legal, and Social Issues
- The CHWs Role in cancer care and prevention
- Supportive Resources
- Locating Resources and Making Referrals

American Cancer Society **HBOC Knowledge Score:** 11 HBOC knowledge questions (correct/incorrect)

GLAC Measure of Word Familiarity: 8 words (7-point Likert scale)

GLAC Measure of Comprehension: 8 words (correct/incorrect)

Health IT Competencies: 4 questions about self-rated abilities (5point Likert scale)

**Attitudes Toward Genetic Testing:** 5 questions about perceived importance (4-point Likert scale)

All measured at pre-course, post-course, and three-month follow-up

	Pre- Training	Post- Training	Three- Months
HBOC Knowledge Score	62% correct	72% correct	67% correct
GLAC Measure of Word Familiarity	5.8	6.5	5.9
GLAC Measure of Comprehension	84% correct	88% correct	85% correct
Health IT Competencies	3.9	4.5	4.5
Attitudes Toward Genetic Testing	3.7	3.9	3.7

Discussed HBOC with 17 individuals at three-months follow-up

## "A Good Invitation to a Great Party"

#### Stakeholder Engagement

- Formative work
- Developing engaging, culturally and literacyappropriate strategies and materials
- Addressing problems that arise

#### **Study Branding**

- Build study buzz and recognition
- Facilitate participant outreach to study team

#### Efficient Participant Identification

- Variety of settings and multi-channel opportunities to meet people where they are
- Identify broad and diverse patient phenotypes

What role does implementation science have in advancing equity in genomics and PPH?

Implementation science comprises the key methodologies for implementing and sustaining genomic and precision public health interventions.

Selecting frameworks, methods, and interventions that are agnostic to the impacts of structural issues can inadvertently exacerbate inequities.

## Extending an Antiracism Lens to the Implementation of Precision Public Health Interventions

- 1. Stakeholder engagement
- 2. Conceptual frameworks and models
- 3. Development, selection, or adaptations of evidence-based interventions
- 4. Evaluation approaches
- 5. Implementation strategies
- 6. Individual research and research contexts

#### **BOX 1**— Recommendations for Implementing an Antiracist Framework in Precision Public Health Interventions

Implementation Science Components <sup>10</sup>	Recommendations		
Stakeholder engagement	Obtain input from communities, particularly those from racial and ethnic minority groups.		
	Ensure interventions and policies are aligned with community perspectives.		
	Implement higher-engagement strategies for greater community involvement in decision-making.		
Conceptual frameworks and models	Seek out and use implementation tools to advance antidiscrimination and antiracism efforts.		
	Incorporate structural racism and other contextual factors into conceptual models.		
	Measure perceived racism and racial discrimination and recognize their impact on implementation.		
	Use transdisciplinary theories to understand the mechanisms that perpetuate health disparities.		
Development, selection, or adaptations of evidence-based interventions	Involve communities in identifying and prioritizing evidence-based interventions.		
	Include evidence-based strategies to address the impact of racism on implementation of precision public health programs.		
Evaluation approaches	Assess the effectiveness of precision public health approaches by race and ethnicity.		
	Use validated measures and self-report to assess racial equity and racism, including qualitative methods to amplify the voices of those with lived experiences of racism.		
Implementation strategies	Focus on multilevel implementation strategies that address structural racism.		
Individual research and research context	Ensure responsible training and engagement of researchers grounded in Public Health Critical Race Praxis.		
	Support and advocate for policies, systems, and structures that promote and sustain diversity in precision public health teams.		

## Selection of Conceptual Frameworks and Models



## **Use of Implementation Strategies**

 Test the influence of implementation strategies on health disparities to understand which strategies will be most effective at improving health equity

 Selection and adaptation of implementation strategies with community members

## 2024 Precision Health Research Symposium Advancing Equity through Precision Health

## SAVE THE DATE NOVEMBER 15TH, 2024

Hosted by the Precision Public Health Network (R13CA261073)



Scan for more information



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"If we want more evidence-based practice, we need more practice-based evidence." LW Green (2006), American Journal of Public Health

