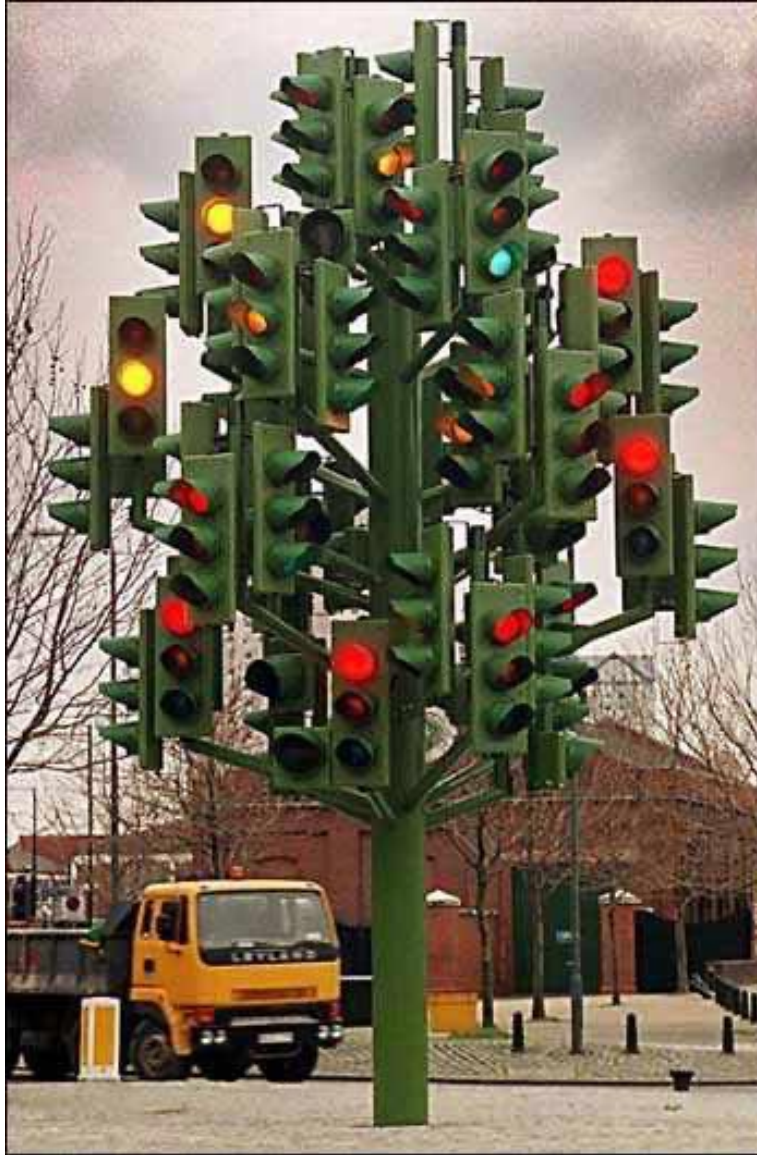


Research Directions on Pre- testing Phase

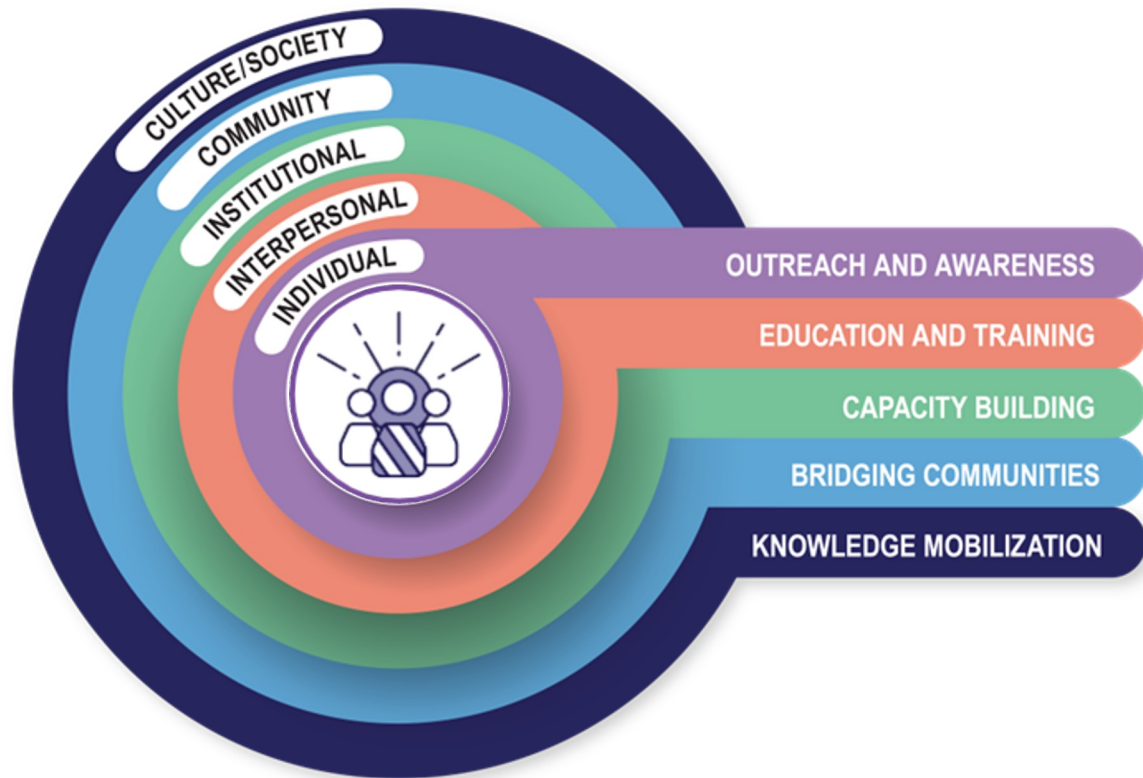


Research Directions on Pre-testing Phase

- Engagement Science
 - Applied to patients
 - Applied to clinicians
 - Applied to other stakeholders



Researcher Engagement Framework



Outreach and Awareness

Interactions at the level of the individual researcher to increase awareness, including participation in outreach activities or receipt of materials and information about the program, its data types, and tools available for research.



Education and Training

Activities that cultivate team science, mentorship, training, and educational support to advance use of the Researcher Workbench.



Capacity Building

Engagement of institutions and partners including minority-serving institutions (i.e., HBCU, HSI, etc.) and community-based organizations to identify needs and assets, build capacity, enhance research activity, and actively utilize program resources.



Bridging Communities

Catalyze a community of multiple researcher audiences (i.e., K-12 students and teachers, undergraduate and graduate students, post-doctoral students and early-stage investigators, established investigators, and community & citizen scientists) and participants to find value in the Researcher Workbench.

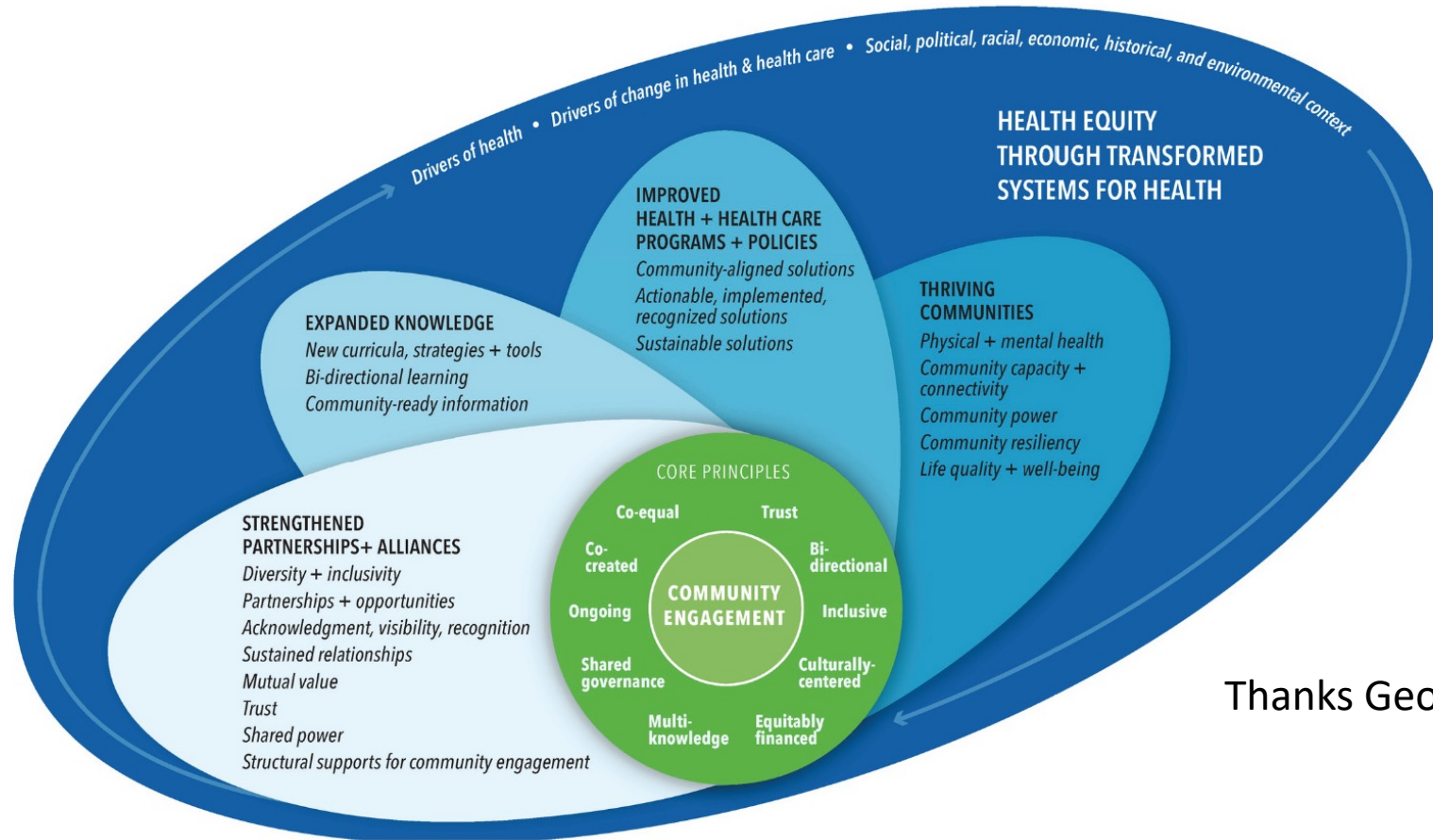


Knowledge Mobilization

Dissemination of findings through publications, presentations, or curricula development to communities, participants, and researchers to mobilize and translate knowledge.



Research Directions on Pre-testing Phase



Thanks George Mensah



FIGURE 1 | A Dynamic Relationship: Achieving Health Equity and Systems Transformation through Meaningful Community Engagement

Research Directions on Pre-testing/screening Phase

- Standardized approach to assessing the chain of probability to inform inclusion or exclusion of genes/variants for population screening
 - Practical Probabilistic Model of Population Screening (Biesecker)
 - Evidence-based Medicine 2.0 (Weaknesses of GRADE Calonge)
 - Context
 - Qualitative data
 - Role of analogy
 - Fit for purpose approach for population screening in genomic medicine
- Standardization of outcomes and cost
 - Cost outcomes from different stakeholders (QALYs, PMPM)



Research Directions on Pre-testing/screening Phase

- Definition of thresholds of evidence:
 - Clinical Utility (benefits and harms) to inform inclusion or exclusion of genes and variants
 - Different perspectives on utilities (payers, USPSTF, healthcare systems)
 - Harms include false reassurance
 - Clinical test performance (Sensitivity, Specificity, PPV, NPV, # needed to screen)
 - Penetrance and natural history of conditions identified by genomic screening
 - Penetrant for what? Which penetrant phenotype drives utility/outcomes/costs?
 - Timeline to realize benefit



Research Directions on Pre-testing/screening Phase

- Research on what is needed for comprehensive and equitable implementation of population screening
 - Pre-implementation research using an evidence-based framework (e.g., CFIR, RE-AIM)
- Focus on equity across multiple dimensions (e.g. innovation equity, deployment equity, contextual equity) and equity metrics in the context of population genomic screening through the many perspectives of different populations and communities
- Research pilot studies for population screening for near-Tier 1 conditions to provide evidence to potentially move to Tier 1
 - Newborn screening pilot model as potential example
- Engage with prevention research community to co-develop genomic prevention research projects



Research Directions on Pre-testing/screening Phase

- Population genomic screening in research settings compared with implementation in public health settings. Different rules, regulations, policies etc.
 - Exploration of these differences within the context of implementation research
- Research into developing a learning (sharing) network to facilitate shared knowledge
 - How do we move from successful projects to widescale implementation
- Research into problems associated with using peripheral blood (mosaicism, clonal hematopoiesis of indeterminate potential) or other samples
- Research programs studying the biology of penetrance
- Research into appropriate population for study design. Move away from disease-based cohort to non-selected (genome first) cohort

