

Why is it so hard to see health benefits from genomic screening?

Alanna Kulchak Rahm, PhD, MS, CGC

Genome XV

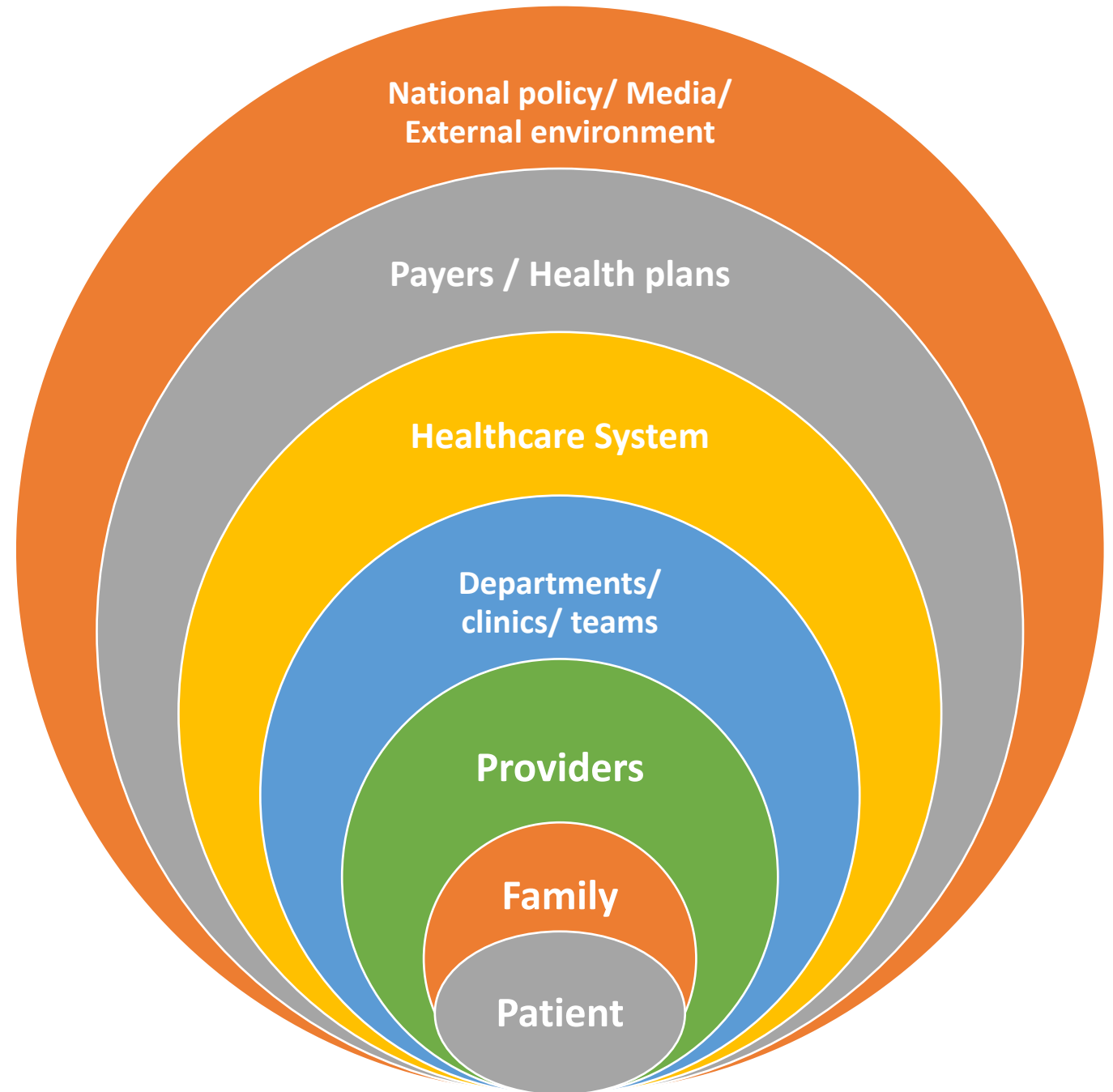
November 9, 2023

Because....

- Access, increased participation in research, and increased testing are not enough
 - Engagement with the information by clinicians and affected individuals through care practice
- Multi-level Barriers and facilitators
 - Communication
 - Billing/Access to services
 - Ongoing engagement and co-development of programs



Implementing genomic information into care to achieve equitable and population-level health benefit is a multi-level issue



Even if 100%
effective, impact
depends on:

- (1) Adoption
- (2) Training
- (3) Fidelity (Implementation)
- (4) Access (Reach)
- (5) Sustainability (Maintenance)

50% threshold for each step=
 $.5 * .5 * .5 * .5 * .5 =$
3% benefit



VOLTAGE DROP

The diminished overall effect of an intervention, process, guideline, test, drug, etc previously shown effective in a trial (a highly controlled environment) when utilized in the real world (an uncontrolled environment)



What is most relevant to the stakeholders?

Even if **genomic screening** was 100% effective at testing and identifying everyone in the population with a result, **health benefit** depends on:

If only **50%** of systems **adopt practices to alert clinicians** of medical management changes



Adoption

& only **50%** clinicians work with individuals to **change medical management**



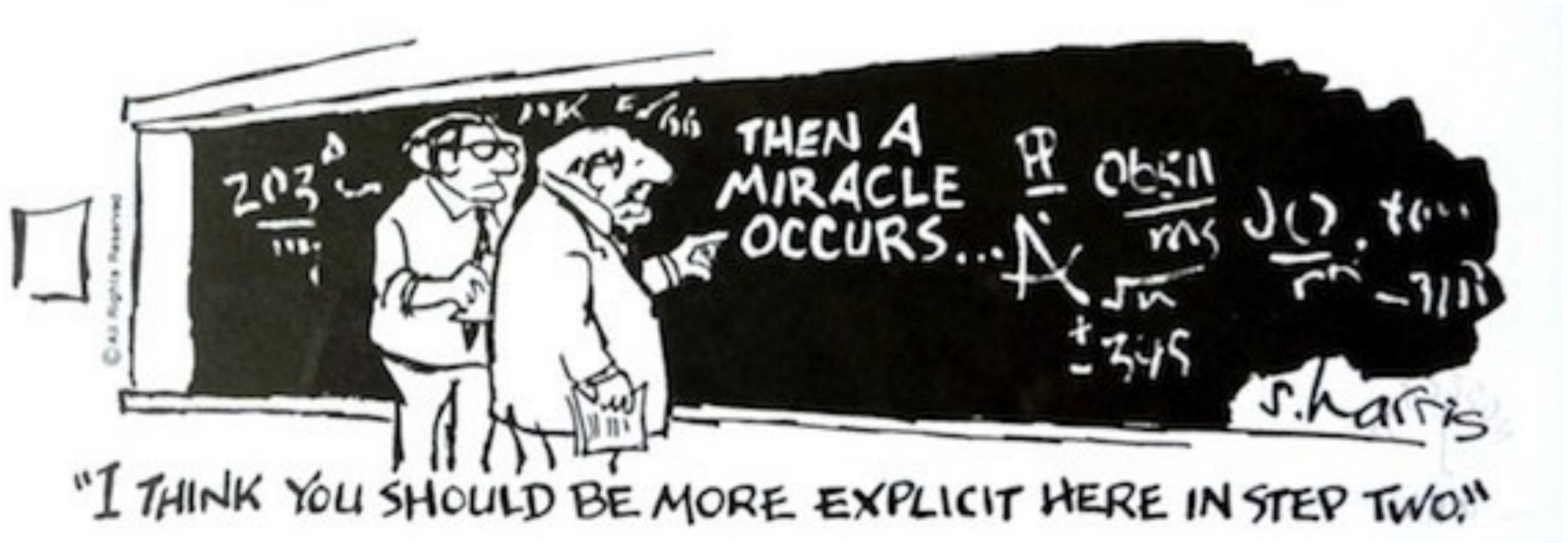
Fidelity

& only **50%** of individuals **follow the guidelines**

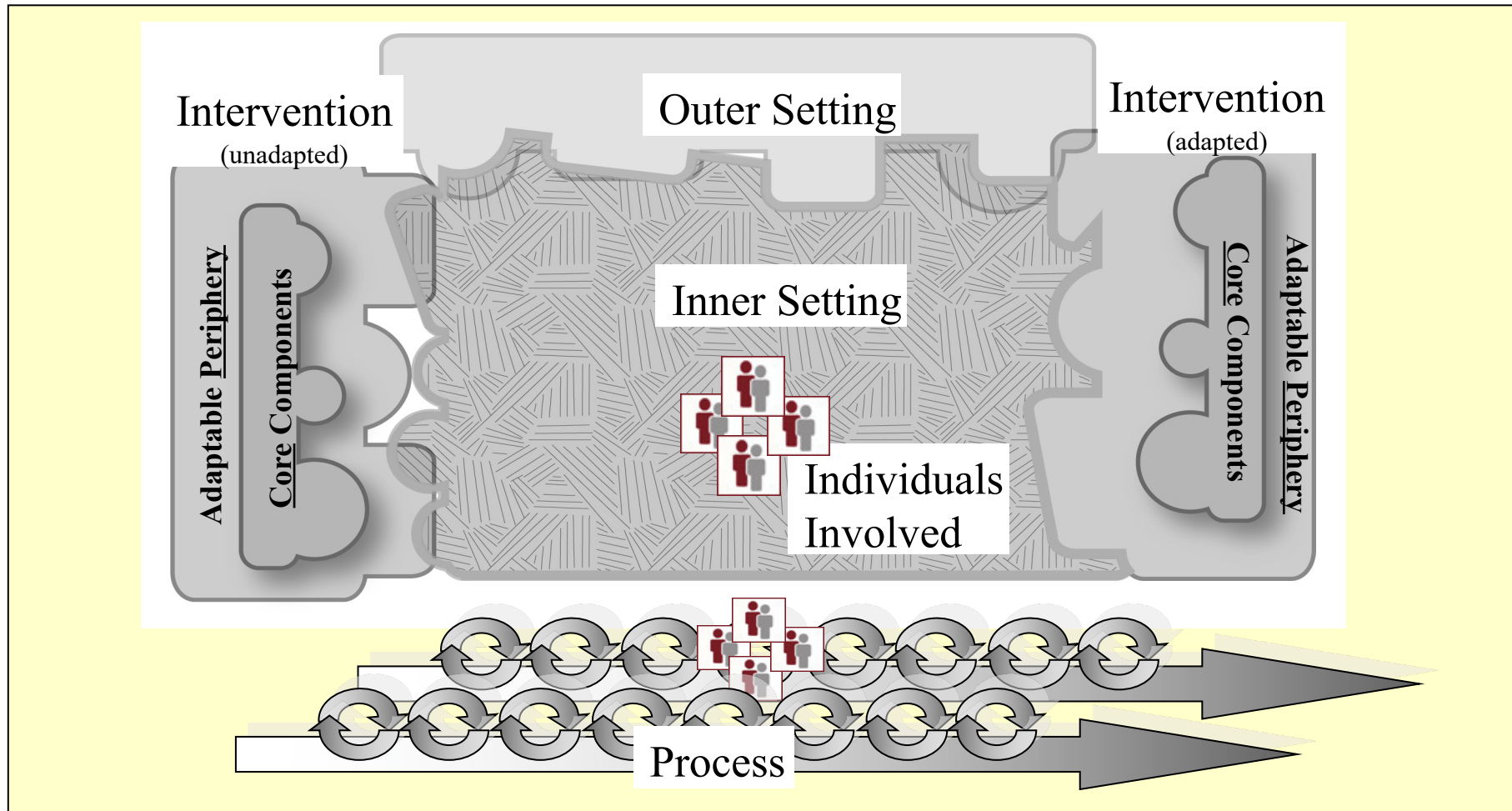
The **overall impact (health benefit)** is reduced to **12.5%** ($.5 * .5 * .5$)

IMPELEMENTATION SCIENCE.....

What works for who, when, and under what conditions/in what contexts



Consolidate Framework for Implementation Research (CFIR)



Engagement – a core component



Co-creation with those who will benefit most



Engagement across spectrum of Implementation



Mitigate biases from big data

Different Engagement Options For Different Preferences

There is a role for everyone. Find the role(s) that are right for you and get engaged.

Donor,
Funder,
Sponsor



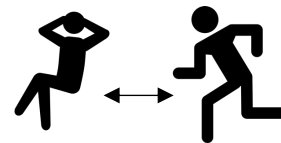
Volunteer



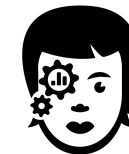
Advisor,
Improver,
Innovator



Passive or Active
Research Participant



Co-Investigator
Co-Creator



Overseer



Importance of addressing equity to realize health benefits of genomic screening



- Lack of heterogeneity in the field and the patients/clients we serve
- Equitable benefit requires equitable implementation
- Equitable implementation does not mean “one size fits all”
- Structural, societal, and other multi-level factors impact implementation and exacerbate inequity when not identified and addressed

Recommended IS Components to Incorporate Equity into Precision Medicine and Public Health



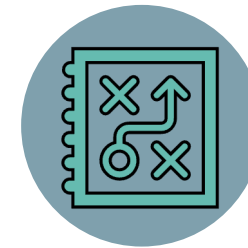
Stakeholder
Engagement



Evaluation Approaches



Models &
Frameworks



Implementation
strategies



Develop, Select, Adapt EBPs

Addressing Context and Promoting Equity with RE-AIM

Reach

- Are all reached equitably?
- Who is not reached?

Effectiveness

- Is health impact equitably experienced?
- Do certain groups experience higher burden or negative effect?

Addressing Context and Promoting Equity with RE-AIM

Adoption

- Were low resource settings able to adopt?
- What adaptation is needed to facilitate adoption?

Implementation

- Who did not deliver the program and why?
- Did all have the capacity/resources to implement?

Maintenance

- Is the program equitably maintained?
- Are long-term outcomes equally experienced?
- Do all settings have capacity /resources to maintain the program over time?



Population Screening Pilot: Adoption & Implementation

What is the ADOPTION of population screening among providers to whom it is available and how consistently do they use it?

- Test available to all patients in 3 self-selected clinics (1 per region)
- Clinics trained and provided EHR tools for documentation/ordering
- 56 clinicians ordered at least one test
- One physician = 462 orders
- One Physician = 142 orders
- Another 8 clinicians ordered between 13 – 32 tests

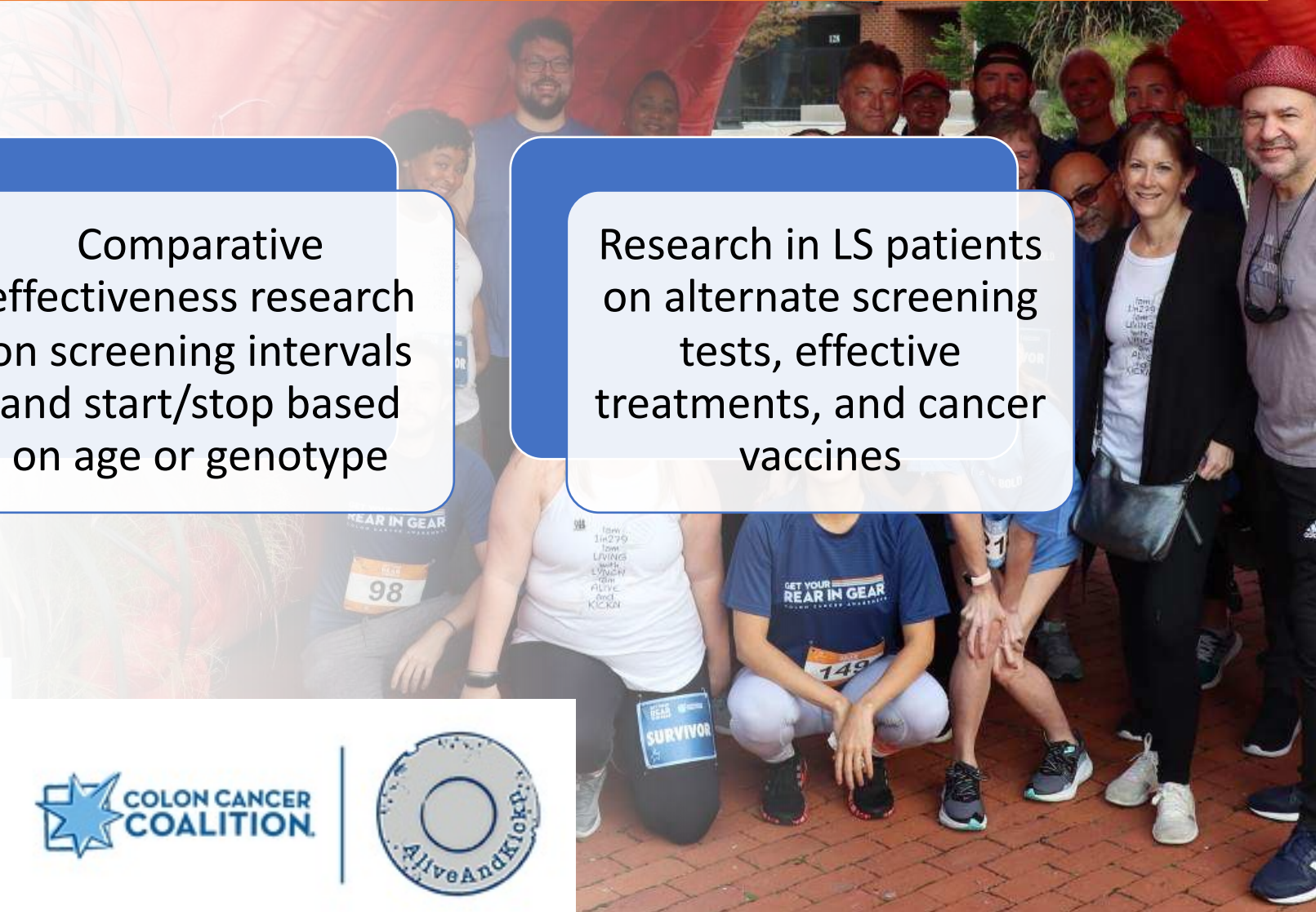
Authorizing provider type	Number of Providers		
	Clinic 1	Clinic 2	Clinic 3
Physician	20	3	4
Resident / Fellow	17	NA	9
Mid-level provider	2	NA	1

EXAMPLE - Cross-Cutting Needs for Patient Stakeholders

Education, understanding, and facilitation of insurance coverage for screening colonoscopy

Comparative effectiveness research on screening intervals and start/stop based on age or genotype

Research in LS patients on alternate screening tests, effective treatments, and cancer vaccines



Traceback interventions (in process)

UXXXX

PI: Rahm, Henrikson, Jonas

Geisinger



Identify eligible patients



Outreach via letter /
patient portal



Reminder calls,
scheduling from genetic
counseling assistant



Usual care for
counseling, testing,
follow-up, cascade testing

12%

KP Mid-Atlantic States



Identify eligible patients



Outreach via letter /
patient portal



Reminder calls, genetic
test order from nurse



Usual care for
counseling, testing, follow-up,
cascade testing (link to video)



Nurse: family sharing
calls, video

37%

KP Washington



Identify eligible patients



Outreach via mailed letter
from KWPA genetics ±
link to video



Outreach, reminder
call(s), scheduling from
KPWA genetics

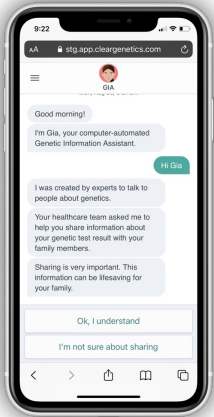


Usual care for
counseling, testing,
follow-up, cascade testing

31%

Implementation Science in a Nutshell **with** Engagement and Equity

Curran GM. Implement SciCommun. 2020



**THE
THING**

**Co-create with
stakeholders**



**Does THE
THING Work**

**Are benefits
experienced
differently?**



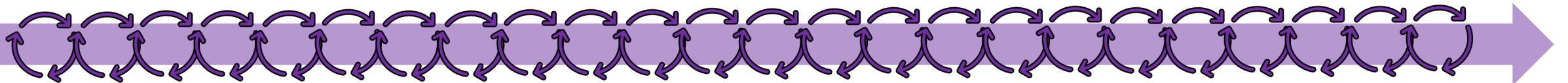
What we do to help
people/ places **DO THE
THING**

**Adapt strategies with
stakeholders for different
settings and people**



How well they
DO THE THING

**Could all settings /
clinicians do THE
THING the same?**



Conclusion and Opportunity - Achieving the Promise of Genomic Screening



Implementation Science provides the tools to achieve the goal to provide equitable benefit for all



Ongoing **Engagement and Co-creation** facilitate equity and improves impact of implementing screening programs



Achieving health benefit from genomic screening is a **Journey**

*“Increasing health equity is within reach, if we commit to building capacity and continue to collaborate with multidisciplinary researchers, practitioners both in health and non-health sectors, policy makers, and **most importantly individuals with lived experiences**, with a reframe of our theories, frameworks, and methods with equity at the forefront. **Beginning with the acknowledgement that health is not distributed across all populations equally, we can work collectively to promote a science for implementation that benefits all, with no one left behind.**”*

Adsul P, Chambers D, Brandt HM, Fernandez ME, Ramanadhan S, Torres E, Leeman J, Baquero B, Fleischer L, Escoffery C, Emmons K, Soler M, Oh A, Korn AR, Wheeler S, Shelton RC. Grounding implementation science in health equity for cancer prevention and control. *Implement Sci Commun.* 2022 Jun 3;3(1):56. doi: 10.1186/s43058-022-00311-4. PMID: 35659151; PMCID: PMC9164317.