



UNC

**ESHELMAN SCHOOL
OF PHARMACY**

Barriers and Facilitators to
Effective & Equitable Cascade
Screening

Megan C Roberts, PhD

Hereditary Conditions

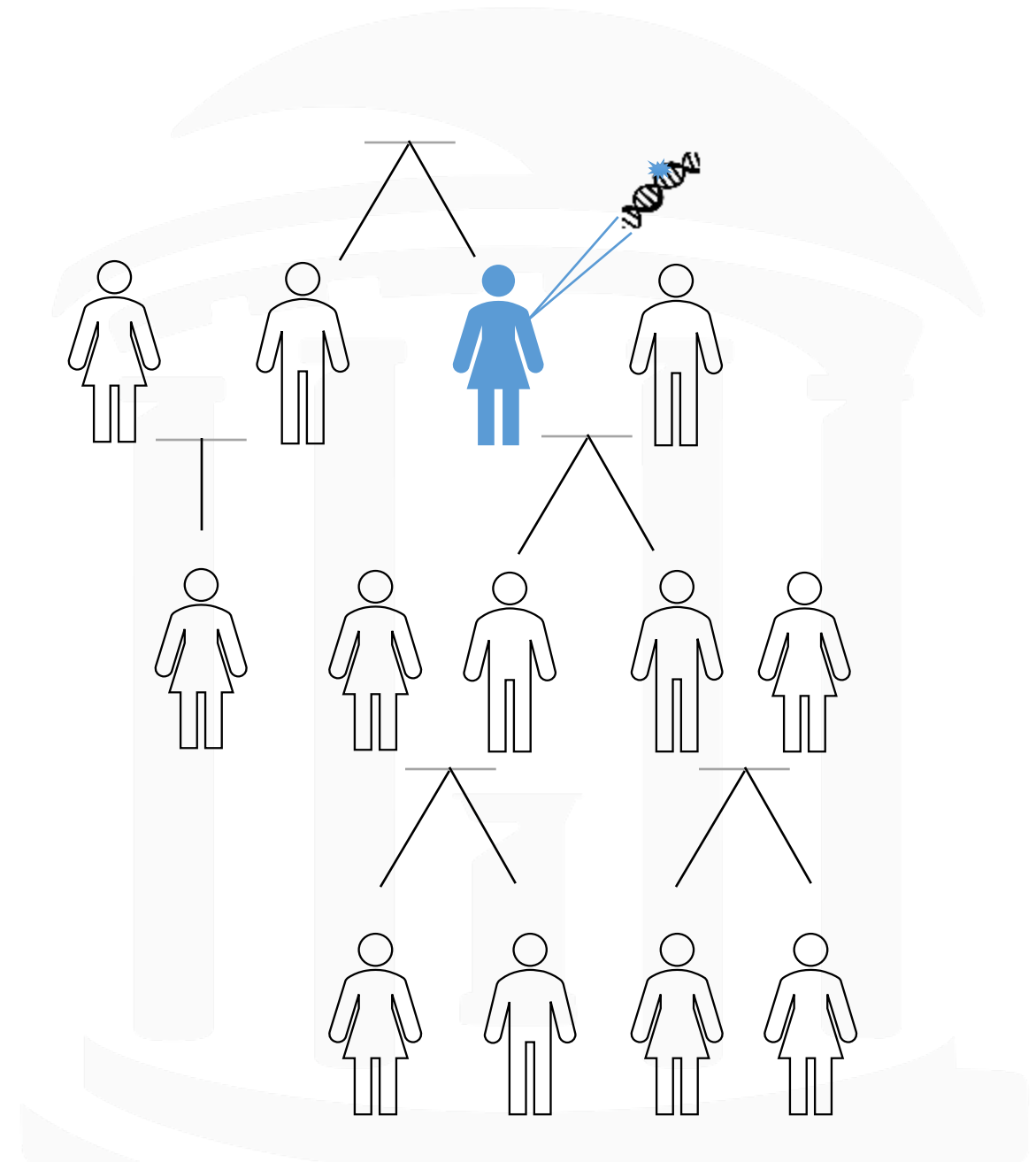
Approximately 3% of us have rare genetic variants causing hereditary conditions that are *preventable* or *treatable* if diagnosed before symptoms develop.

- Lynch syndrome
- Hereditary Breast and Ovarian Cancer Syndrome
- Familial hypercholesterolemia
- Others

Murray, Evans and Khoury 2019; <https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm>

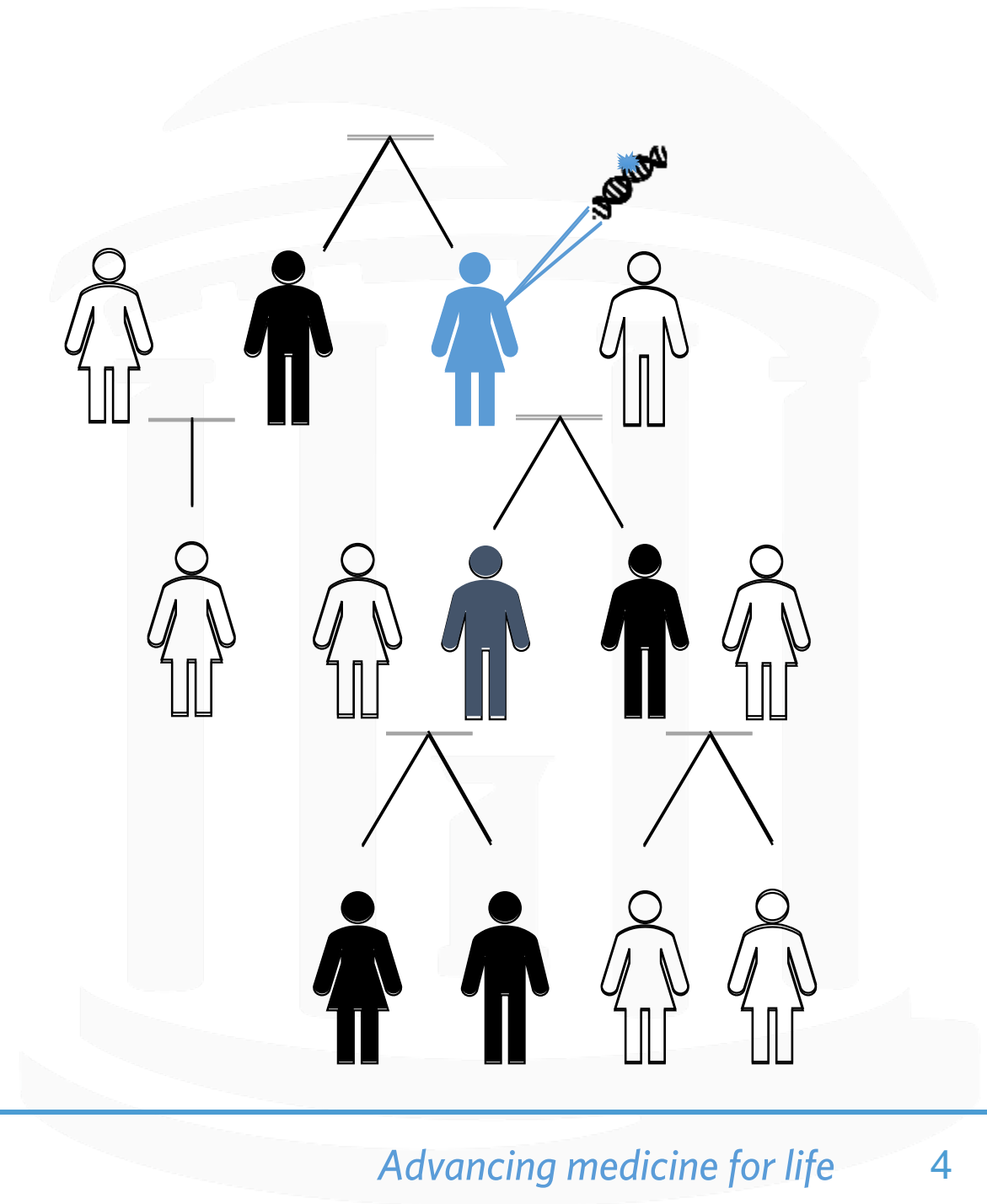
Cascade screening

Active process of identifying and testing relatives of individuals with a known hereditary condition

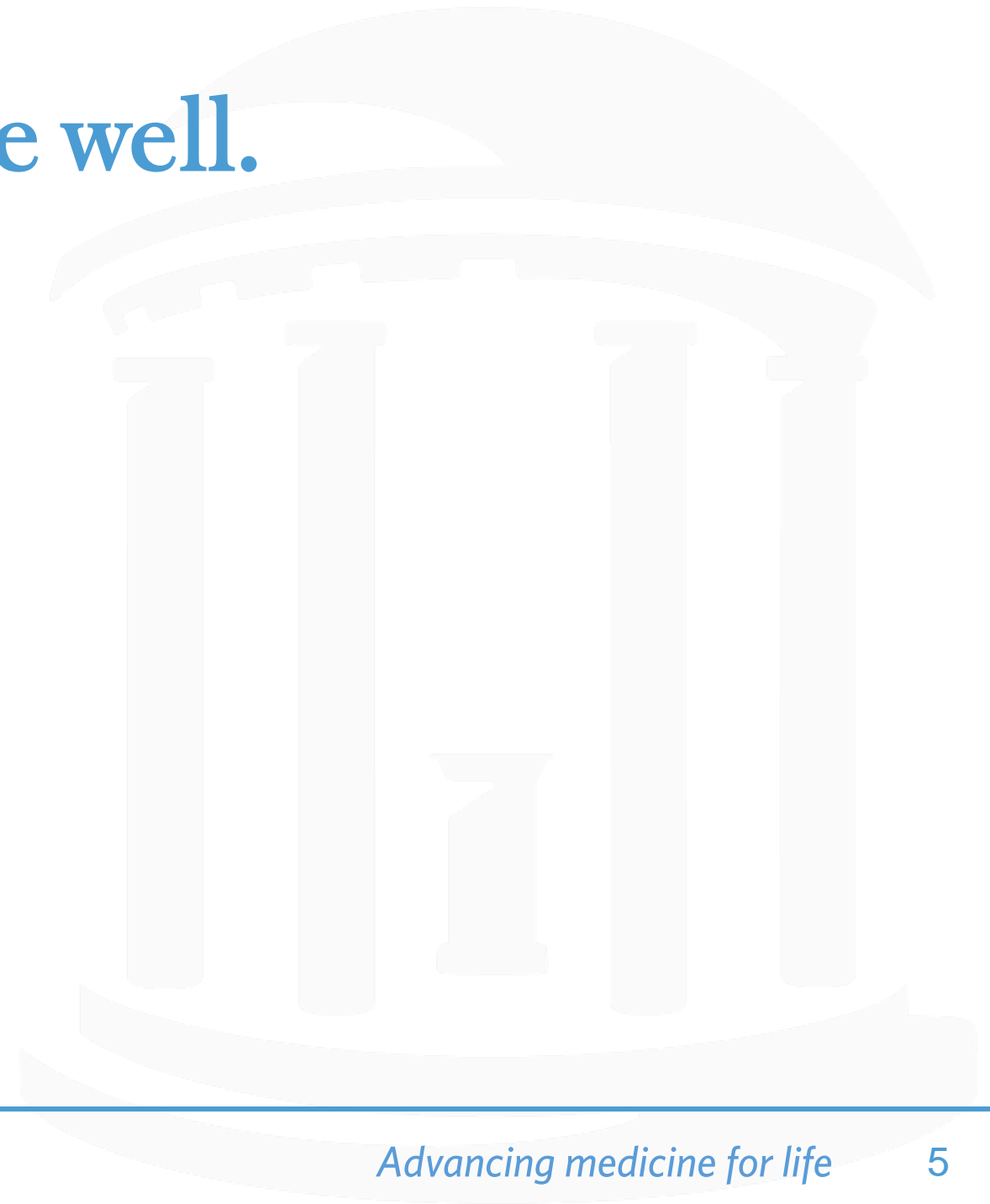


Cascade screening

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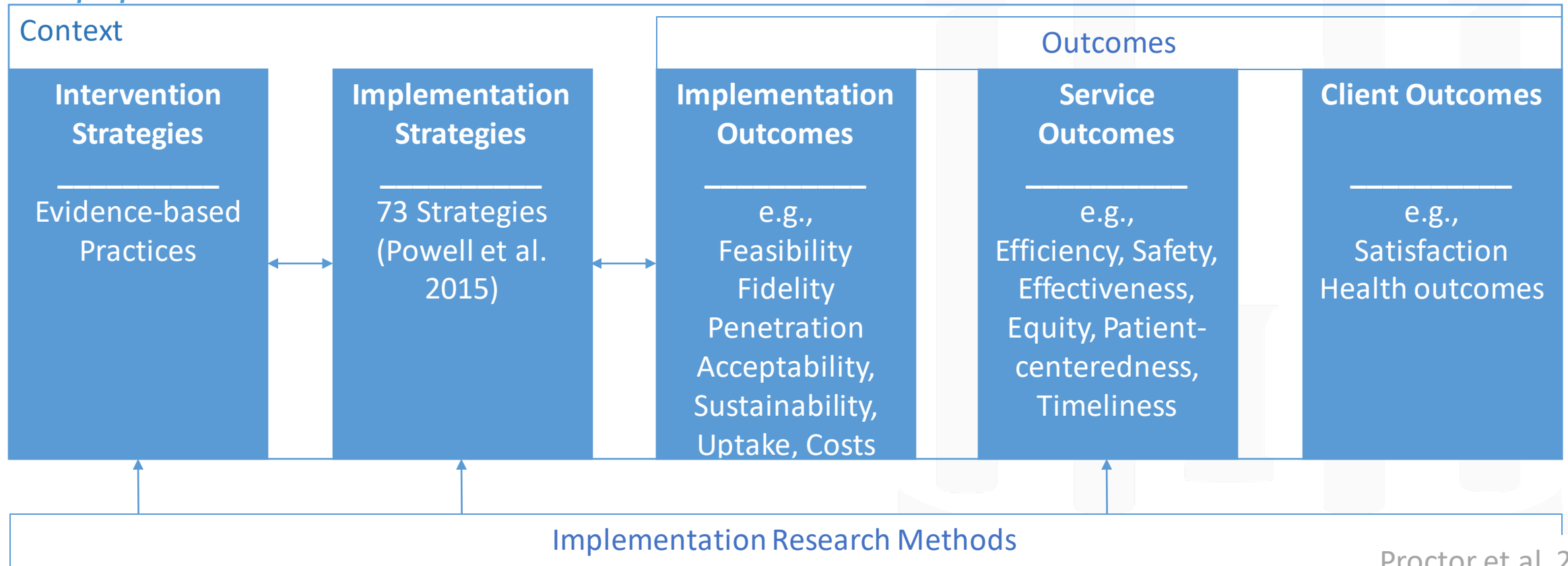


Currently, this isn't done well.



Implementation Science

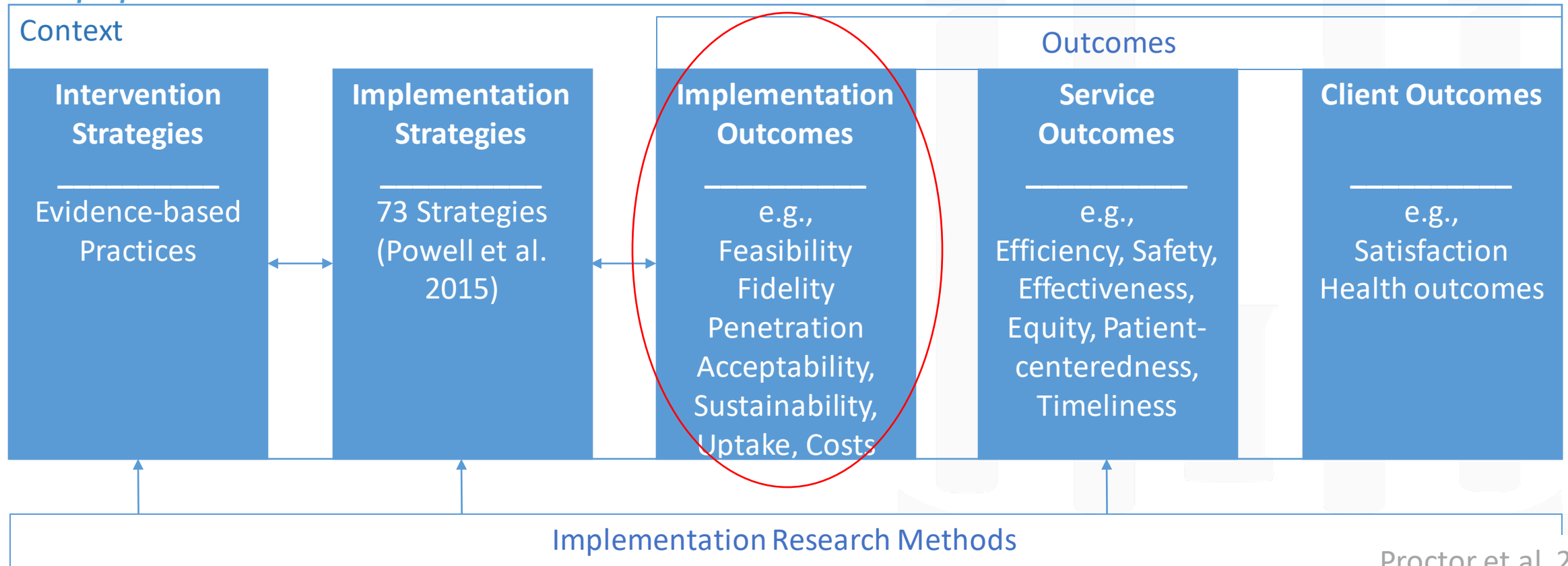
The study of methods to promote the adoption and integration of evidence-based practices into routine health care and public health settings to improve the impact on population health.



Proctor et al. 2009

Implementation Science

The study of methods to promote the adoption and integration of evidence-based practices into routine health care and public health settings to improve the impact on population health



Reviews

PRECISION MEDICINE

By Megan C. Roberts, W. David Dotson, Christopher S. DeVore, Erica M. Bednar, Deborah J. Bowen, Theodore G. Ganiats, Ridgely Fisk Green, Georgia M. Hurst, Alisdair R. Philp, Charité N. Ricker, Amy C. Sturm, Angela M. Trepanier, Janet L. Williams, Heather A. Zierhut, Katherine A. Wilemon, and Heather Hampel

Delivery Of Cascade Screening For Hereditary Conditions: Review Of The Literature

ABSTRACT Cascade screening is the process of contacting people who have been diagnosed with certain hereditary conditions. The purpose is to identify, inform, and manage those at risk. We conducted a scoping review to obtain a broad overview of the literature on cascade screening for hereditary conditions.

- Reviewed literature 2018

- Reviewed literature from 1990-2017
- Described 45 states and regulations

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<https://doi.org/10.1038/s41431-020-00725-5>

ESHG

REVIEW ARTICLE

Barriers and facilitators for cascade testing in genetic conditions: a systematic review

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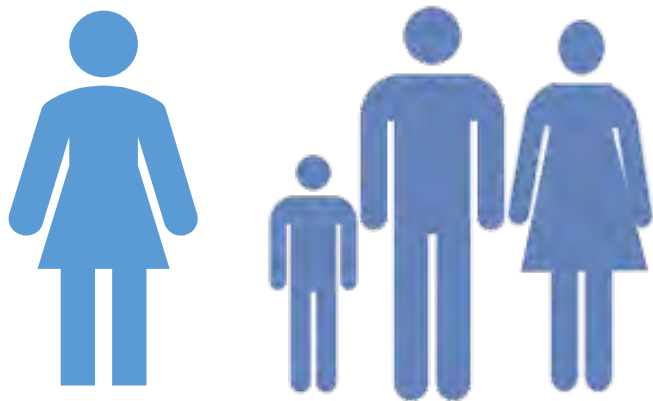
Abstract

Cascade testing is the process of offering genetic counseling and testing to at-risk relatives of an individual who has been diagnosed with a genetic condition. It is critical for increasing the identification rates of individuals with these conditions and the uptake of appropriate preventive health services. The process of cascade testing is highly varied in clinical practice, and a

Qualitative Study

PATIENTS (n=20)

- Adults with LS
- Family members of adults with LS



PROVIDERS (n=20)

- Genetic counselors
- Gastroenterologists
- Colorectal surgeons
- Oncologists
- Primary care physicians
- Gynecologic oncologists



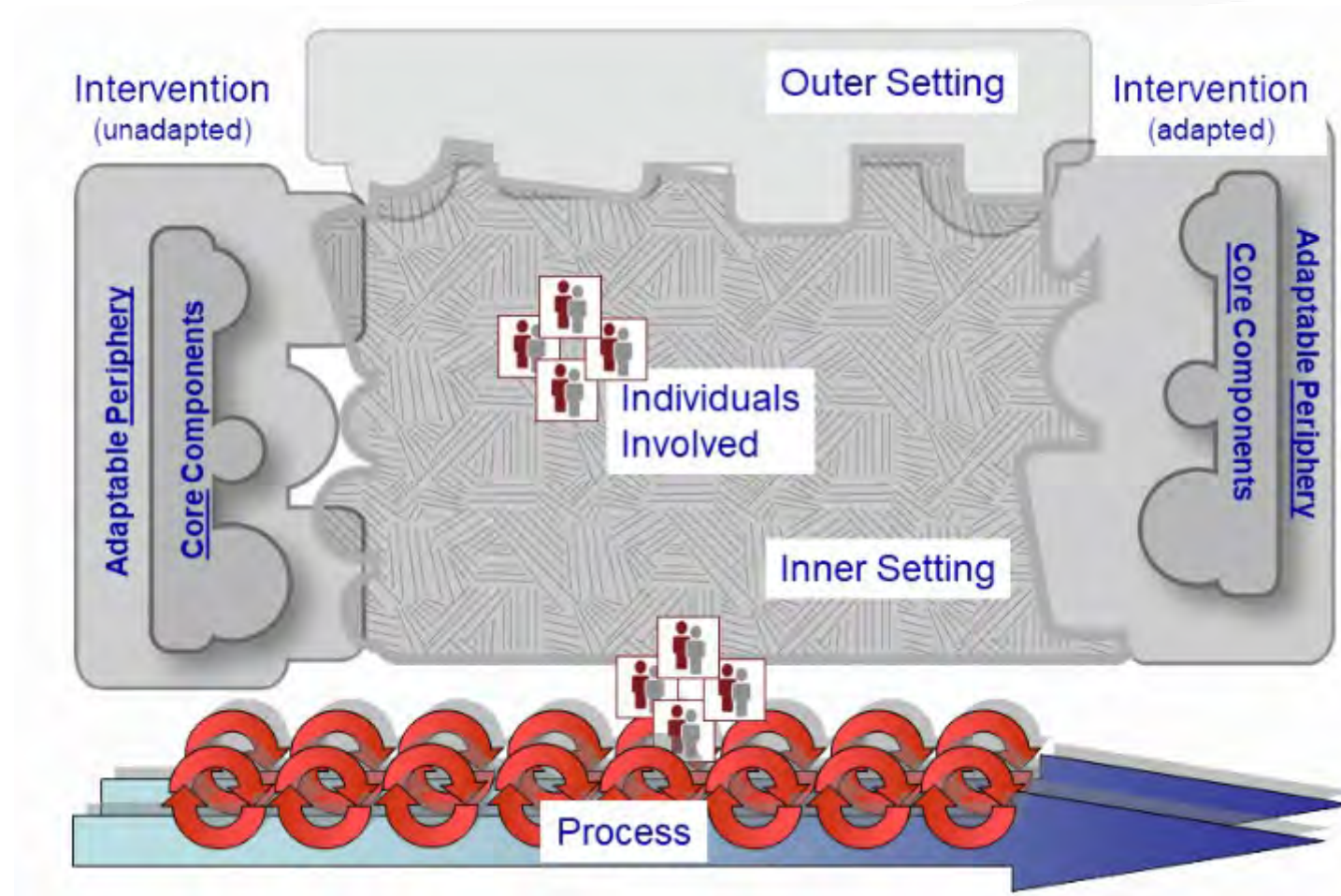
ADMINISTRATORS (n=20)

- Genetics clinic managers
- Genetics labs
- Patient advocates
- Payers



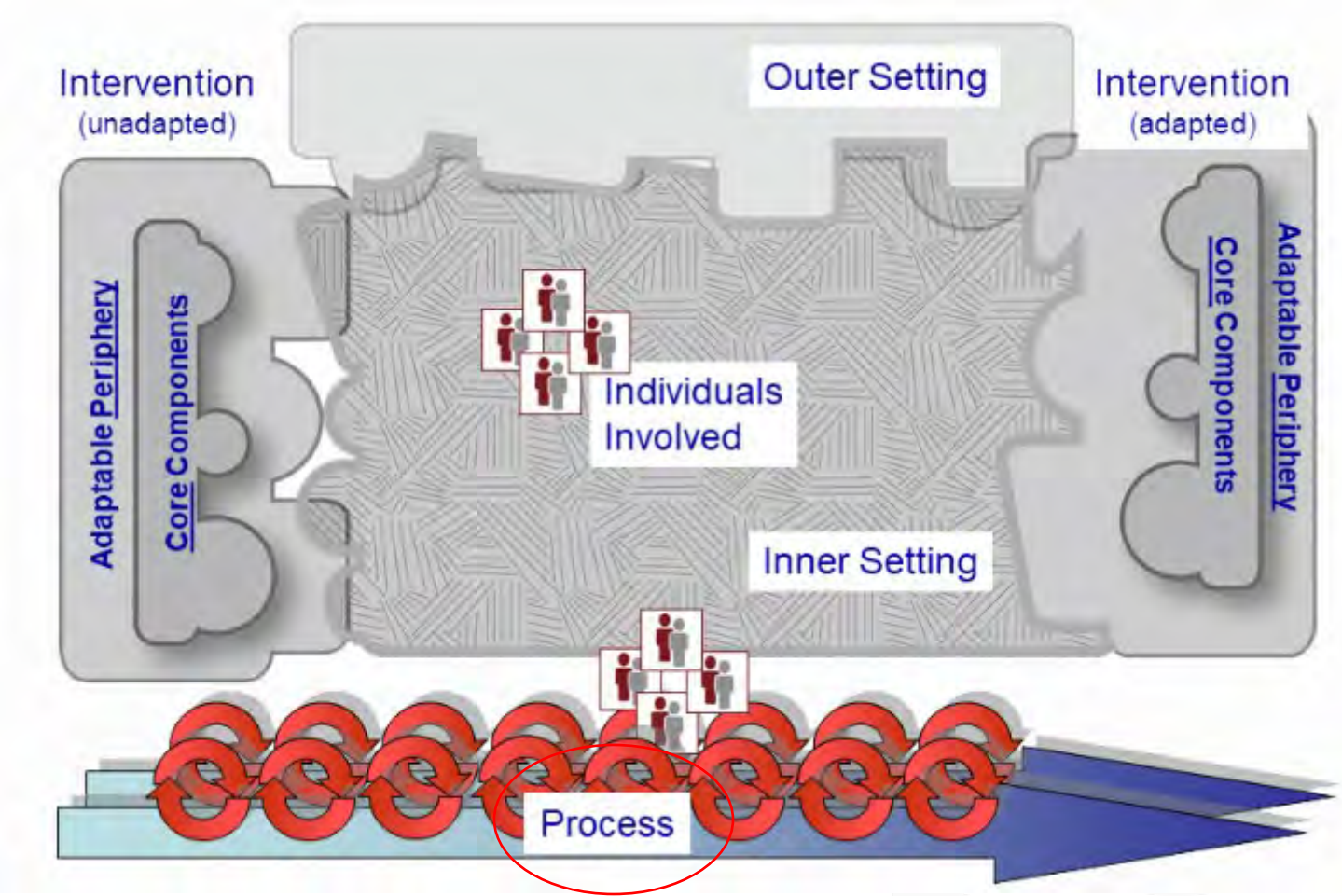
Srinivasan et al. 2020

Findings-Consolidated Framework for Implementation Research (CFIR)



Damschroder et al. 2009; cfirguide.org

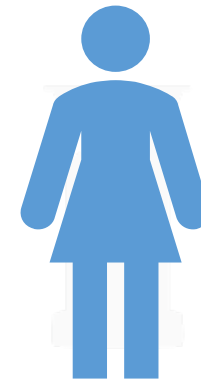
CFIR



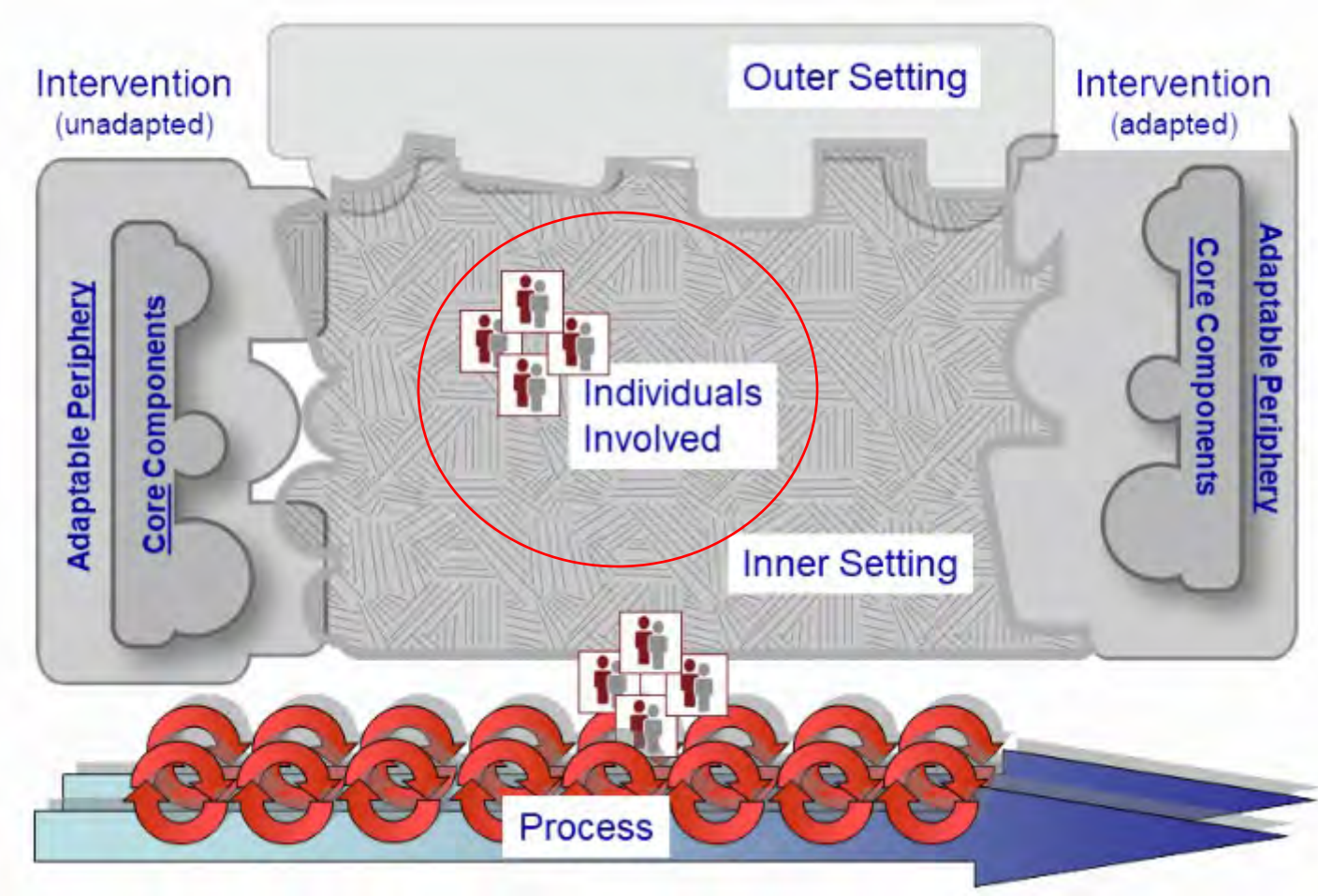
Damschroder et al. 2009; cfirguide.org

Barriers to Cascade Screening Processes

Lack of processes



CFIR

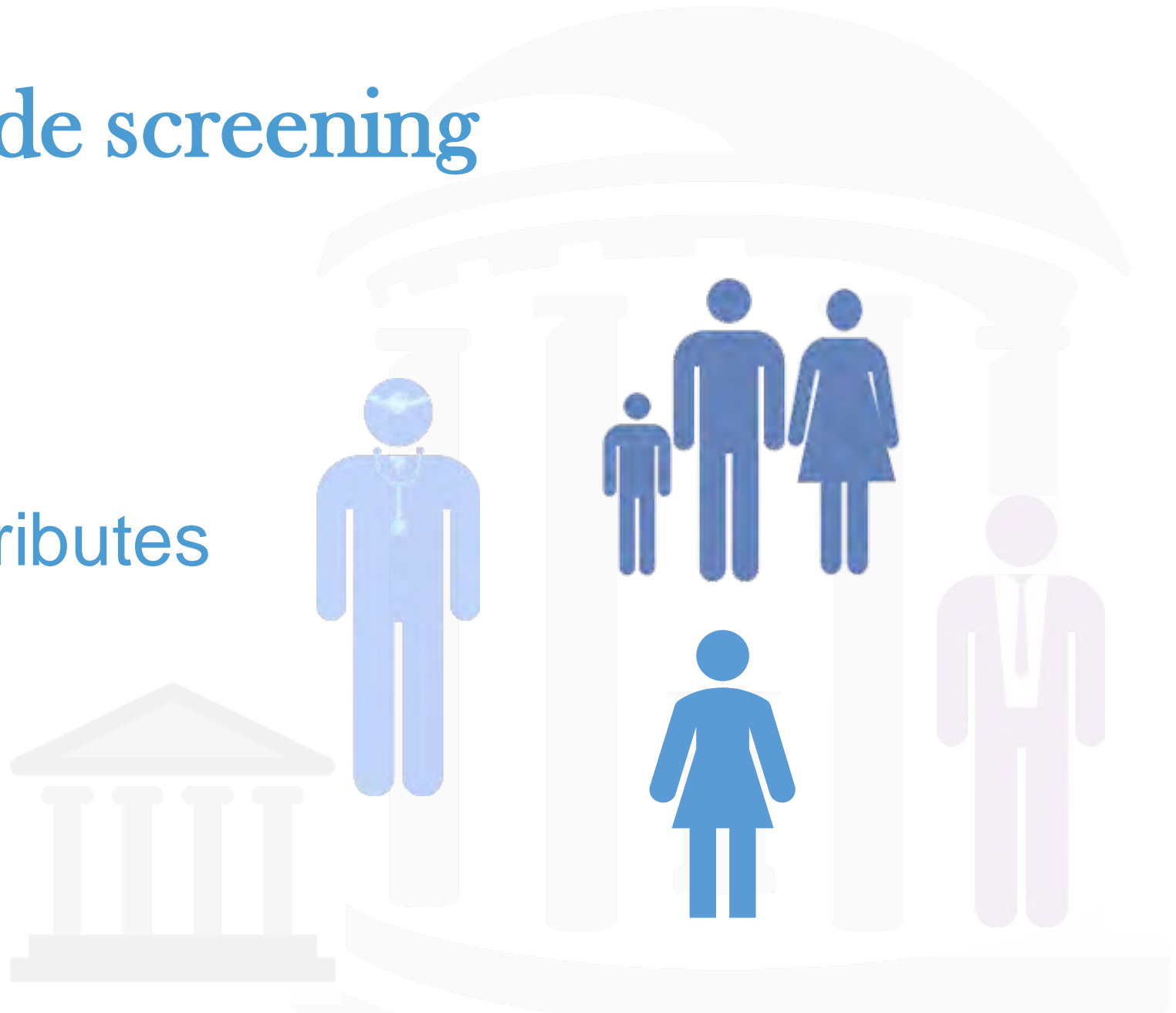


Damschroder et al. 2009; cfirguide.org

Barriers to cascade screening

Individuals Involved

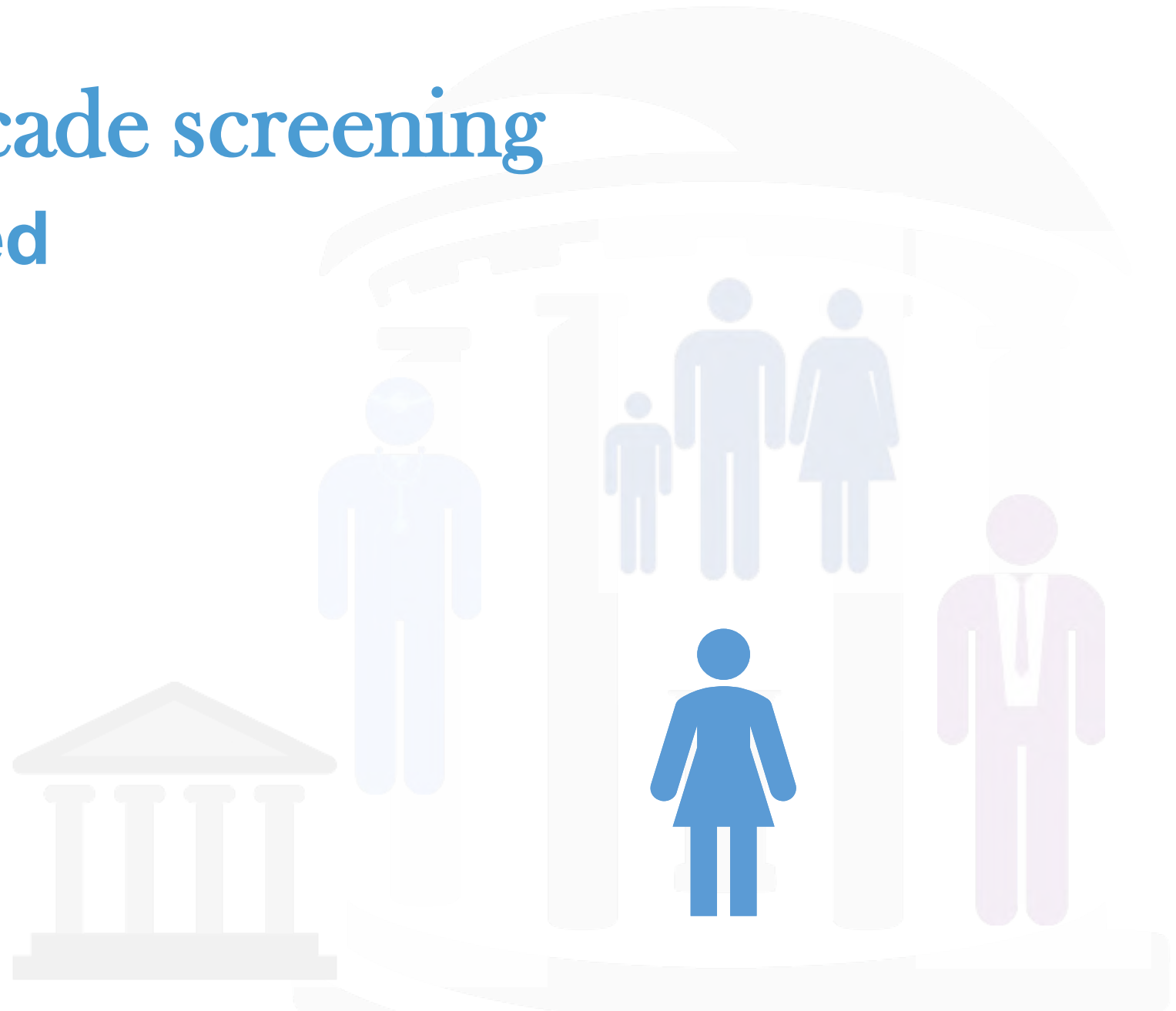
- Knowledge
- Self-efficacy
- Other personal attributes



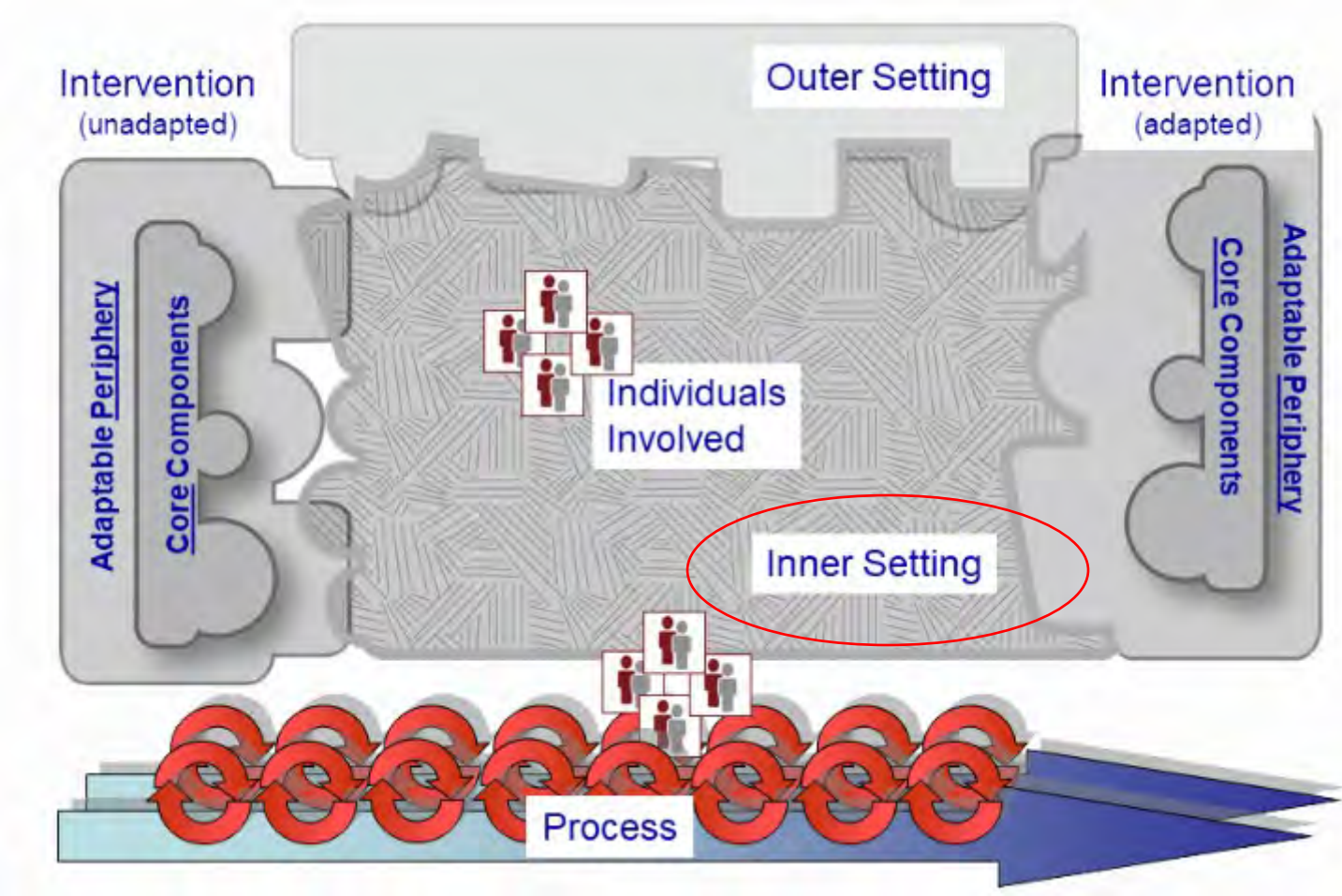
Enablers for cascade screening

Individuals Involved

Beliefs



CFIR



Damschroder et al. 2009; cfirguide.org

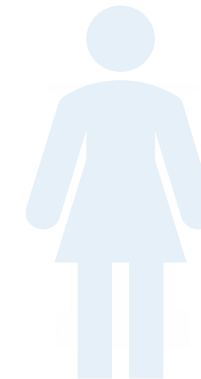
Barriers to Cascade Screening

Inner Setting

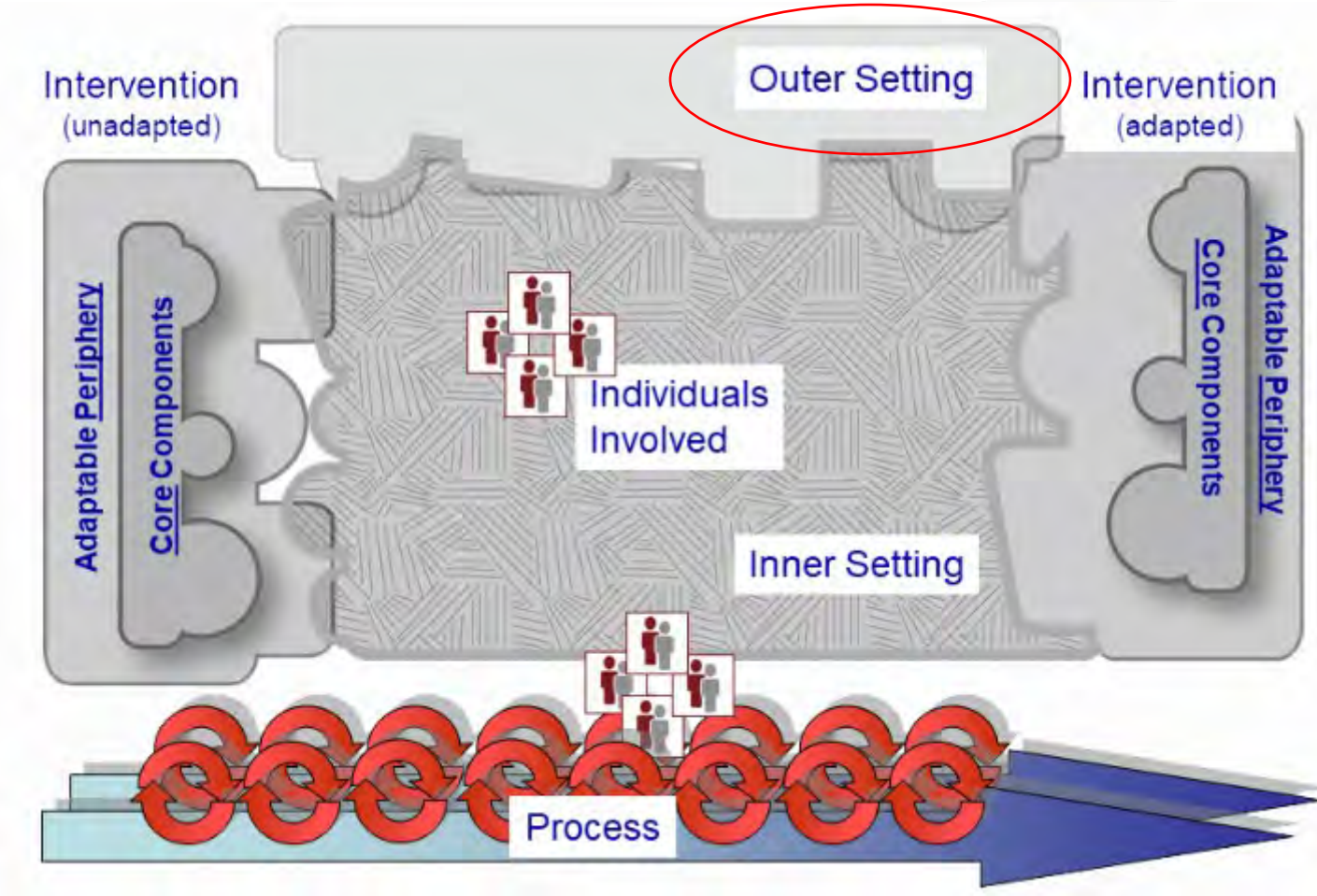
Compatibility

Relative priority

Available resources



CFIR

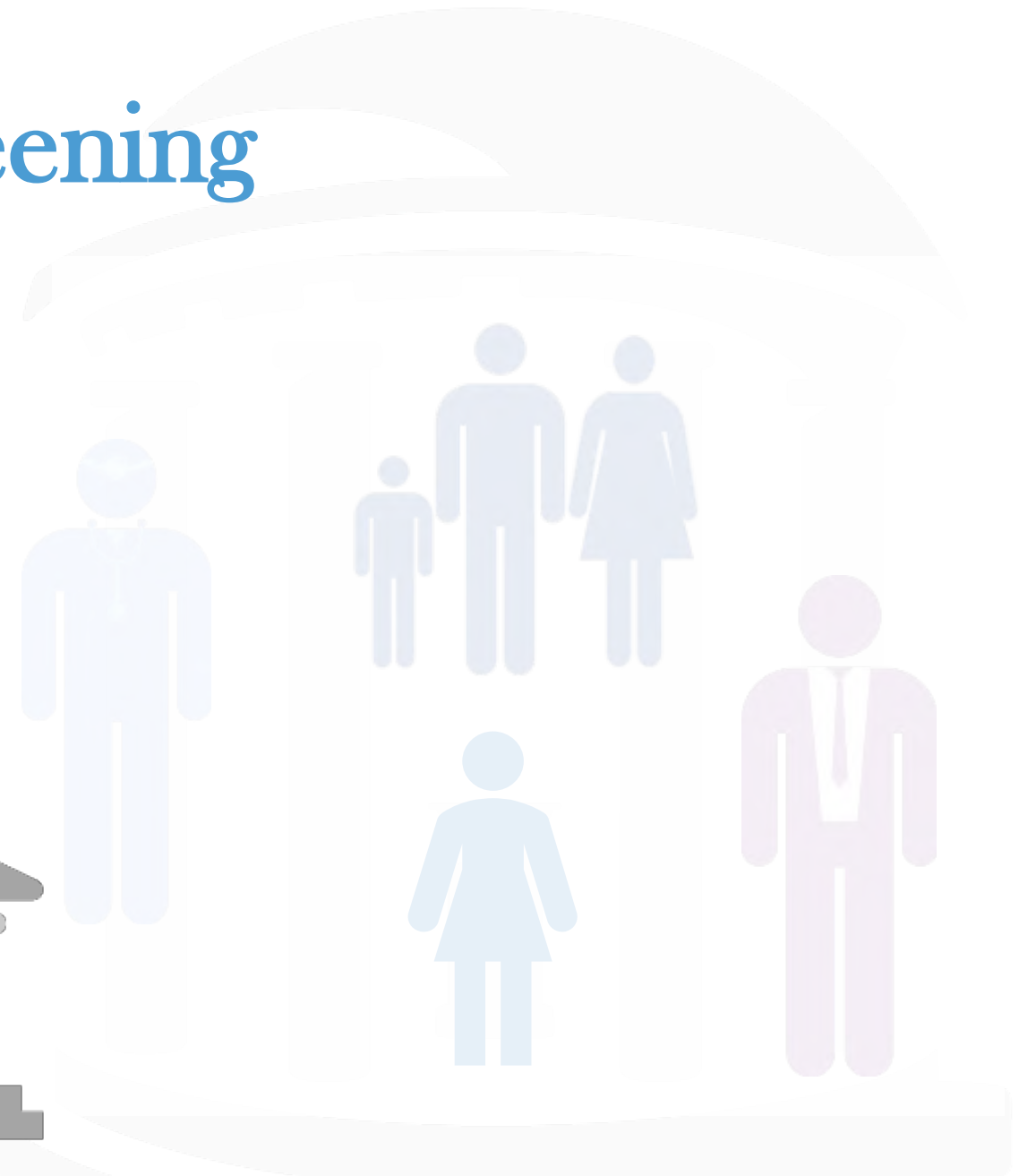


Damschroder et al. 2009; cfirguide.org

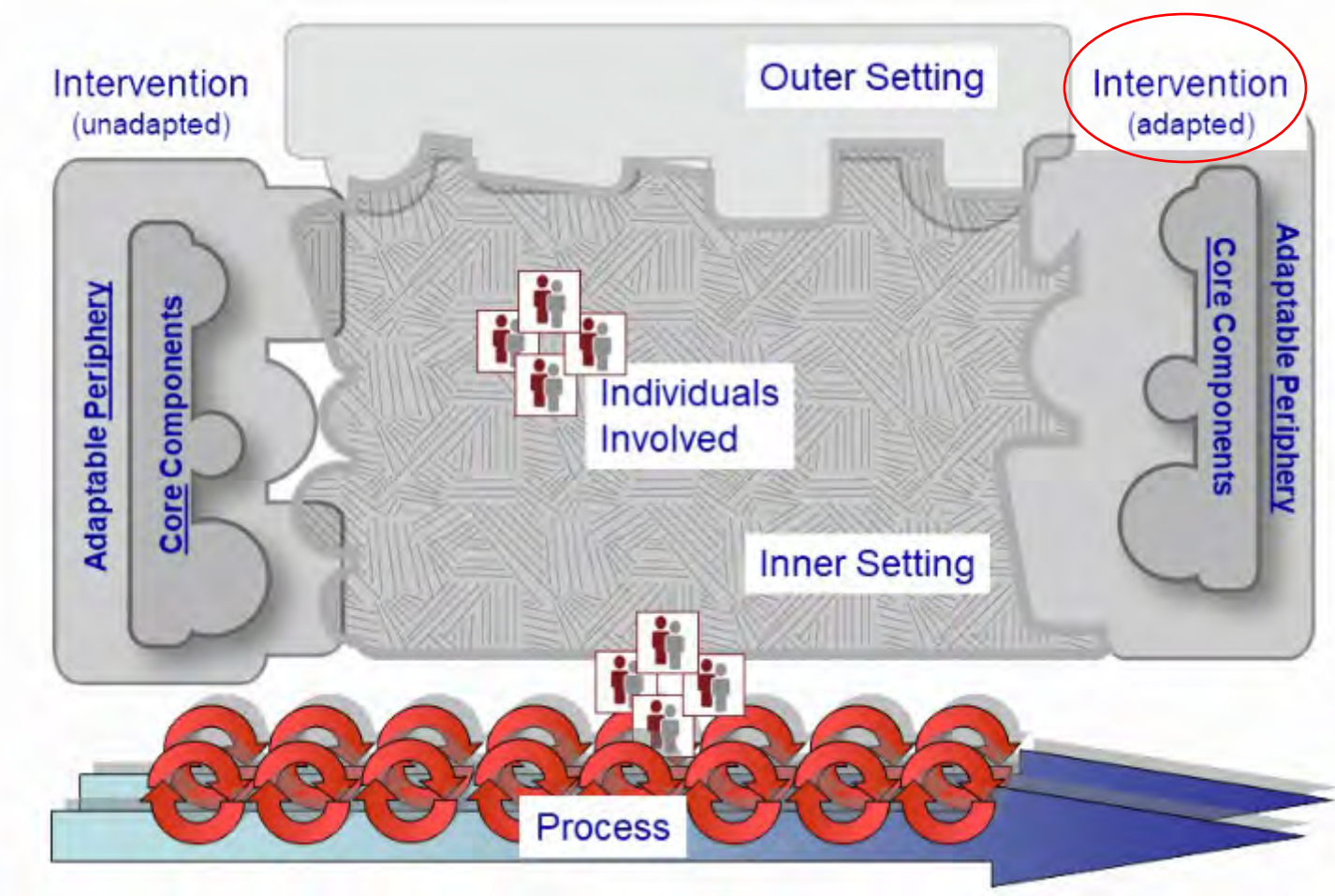
Barriers to Cascade Screening

Outer Setting

External Policies



CFIR



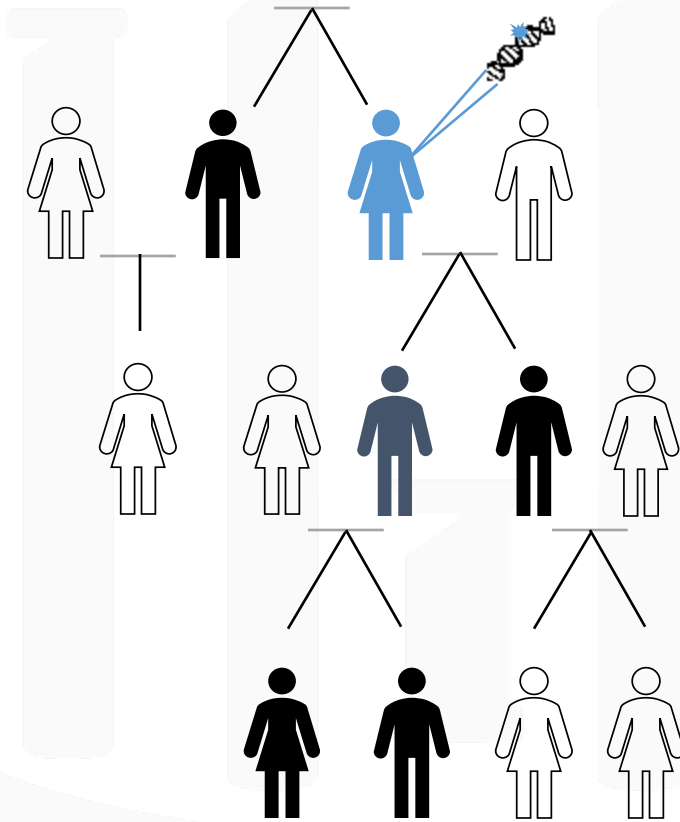
Damschroder et al. 2009; cfirguide.org

Barriers to Cascade Screening

Intervention Characteristics

Costs

Complexity



Gaps in the Literature

Address geographic and communication barriers

Inclusion of implementation measures, methods and frameworks

Improved diversity of study populations

Utilization of rigorous study designs

Increase the number of US studies

Roberts et al. 2018

Acknowledgements

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Alisdair Philp

Daniel Reuland

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Katherine Wilemon

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Sarah Wright

Heather Zierhut



Thank You.

