

Health Disparities in Implementation of Genomic Medicine: Challenges and Opportunities

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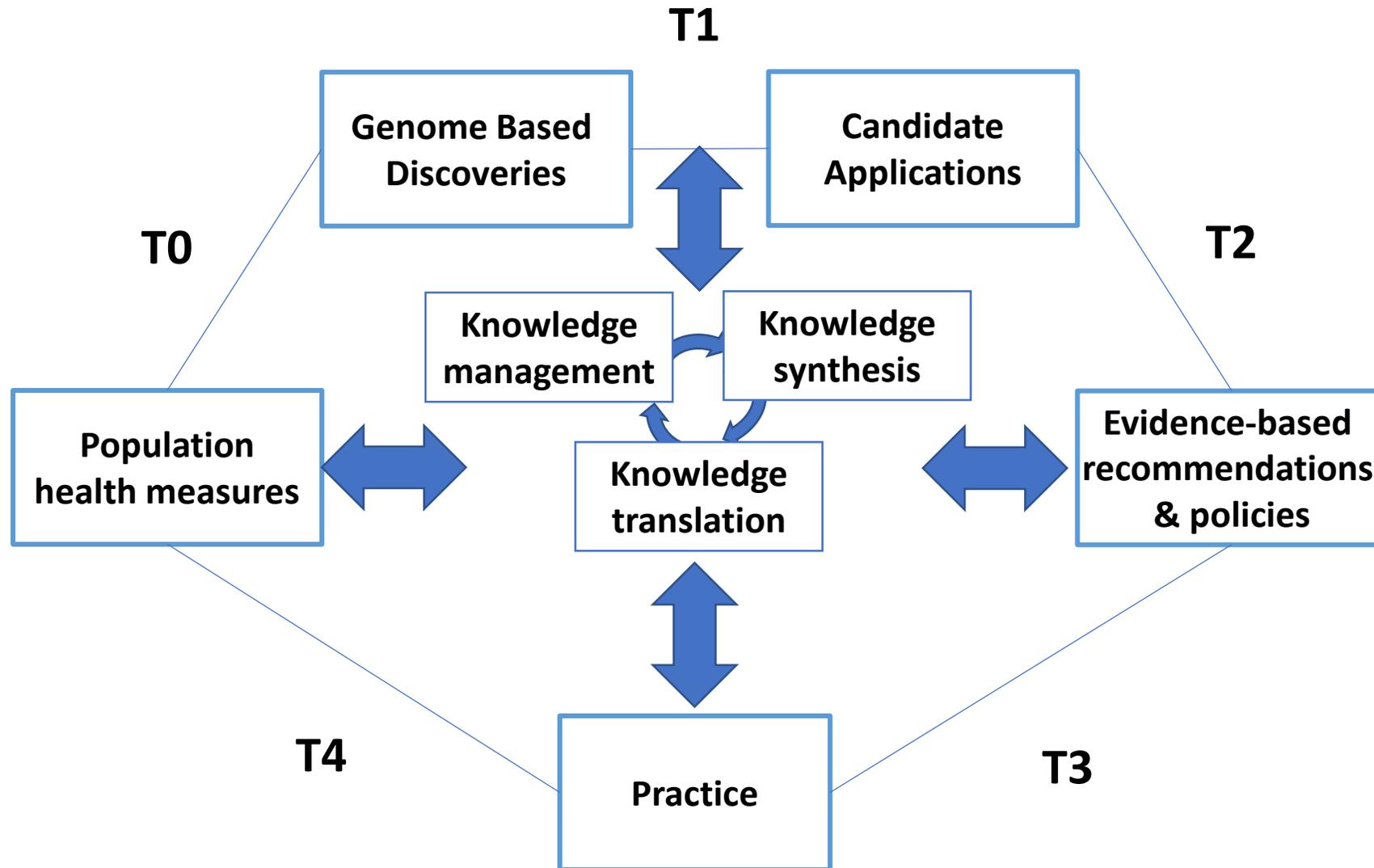
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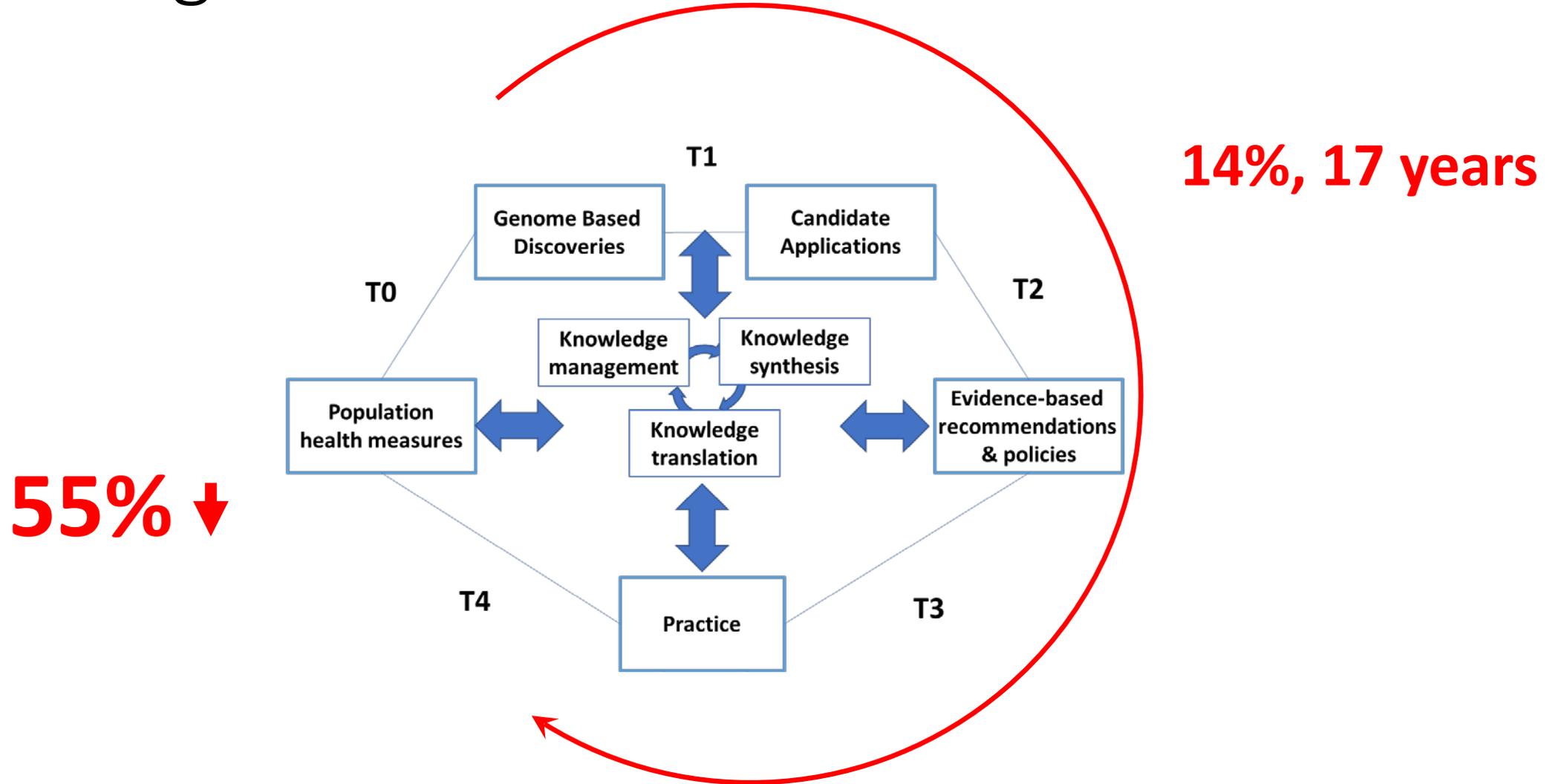
Objectives:

- Review health disparities in the implementation of genetic testing and cascade screening for hereditary cancers and heart disease
- Describe how implementation science approaches can be used to address health disparities

Translational Genomic Research



Time Lags in Translational Research



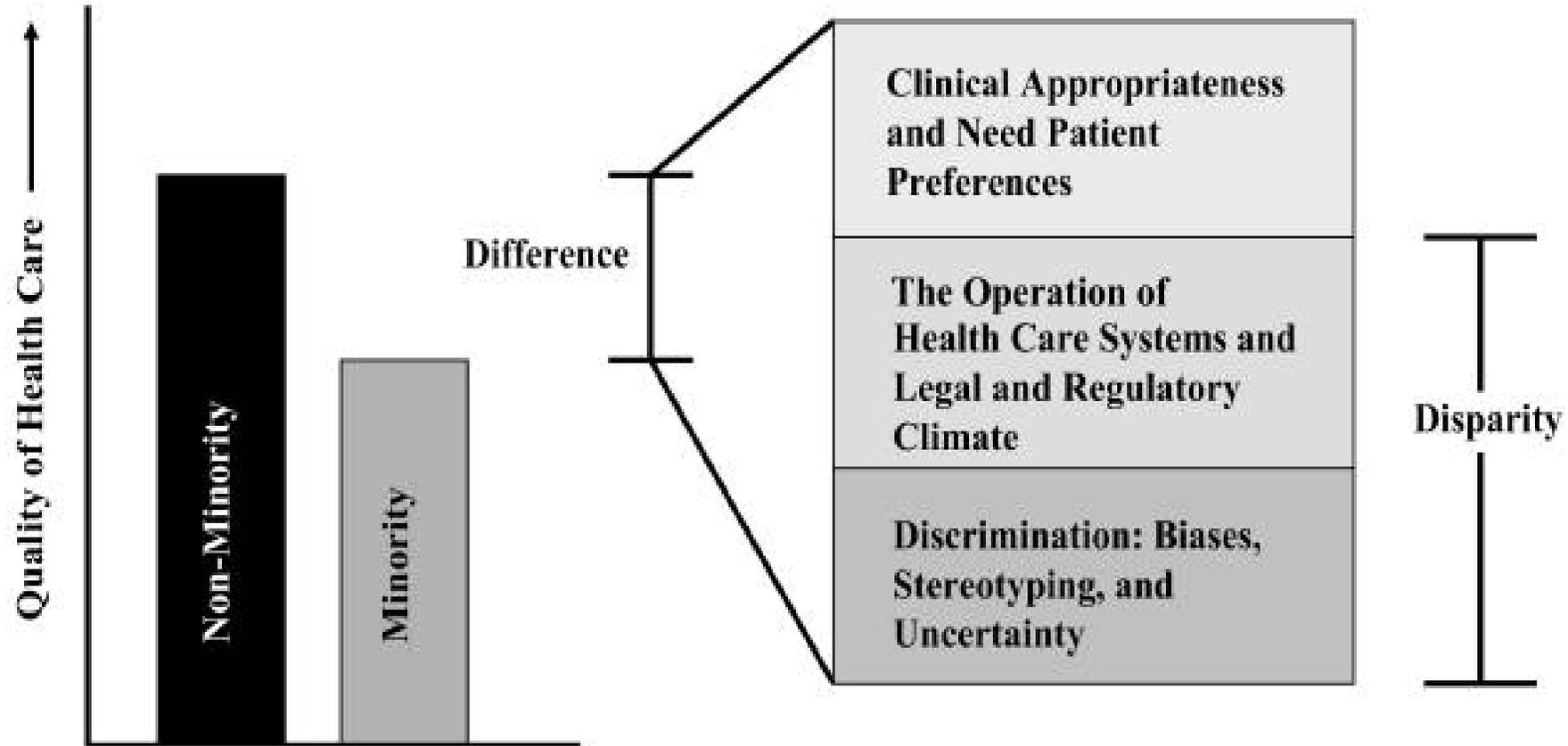
Defining disparities

- AHRQ: “Any difference among populations that are statistically significant and differ from the reference group by at least 10 percent”
- IOM: “the difference in treatment or access not justified by the differences in health status or preferences of the groups”
- WHO: “differences in health which are not only unnecessary and avoidable but, in addition, are considered unfair and unjust.”

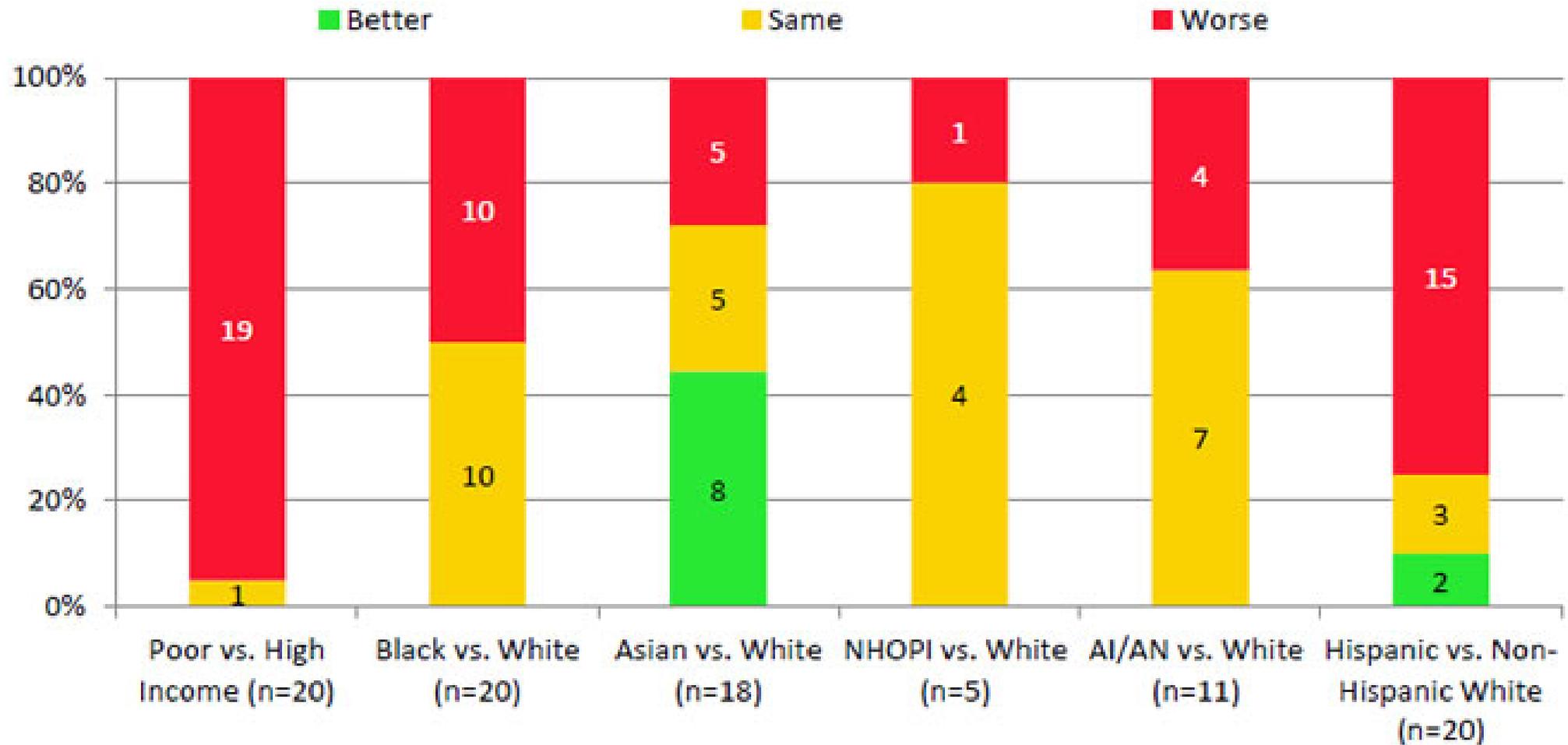
Defining disparities cont.

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Difference vs. Disparity



Access to Care, 2014



Unique Challenges to Genomic Medicine

- Ethical, Legal and Social
- (Genetic) literacy
- (Genetic risk) communication
- Rapidly-evolving knowledge
- Big Data
- Costs
- Many more...



Barriers to genomic medicine

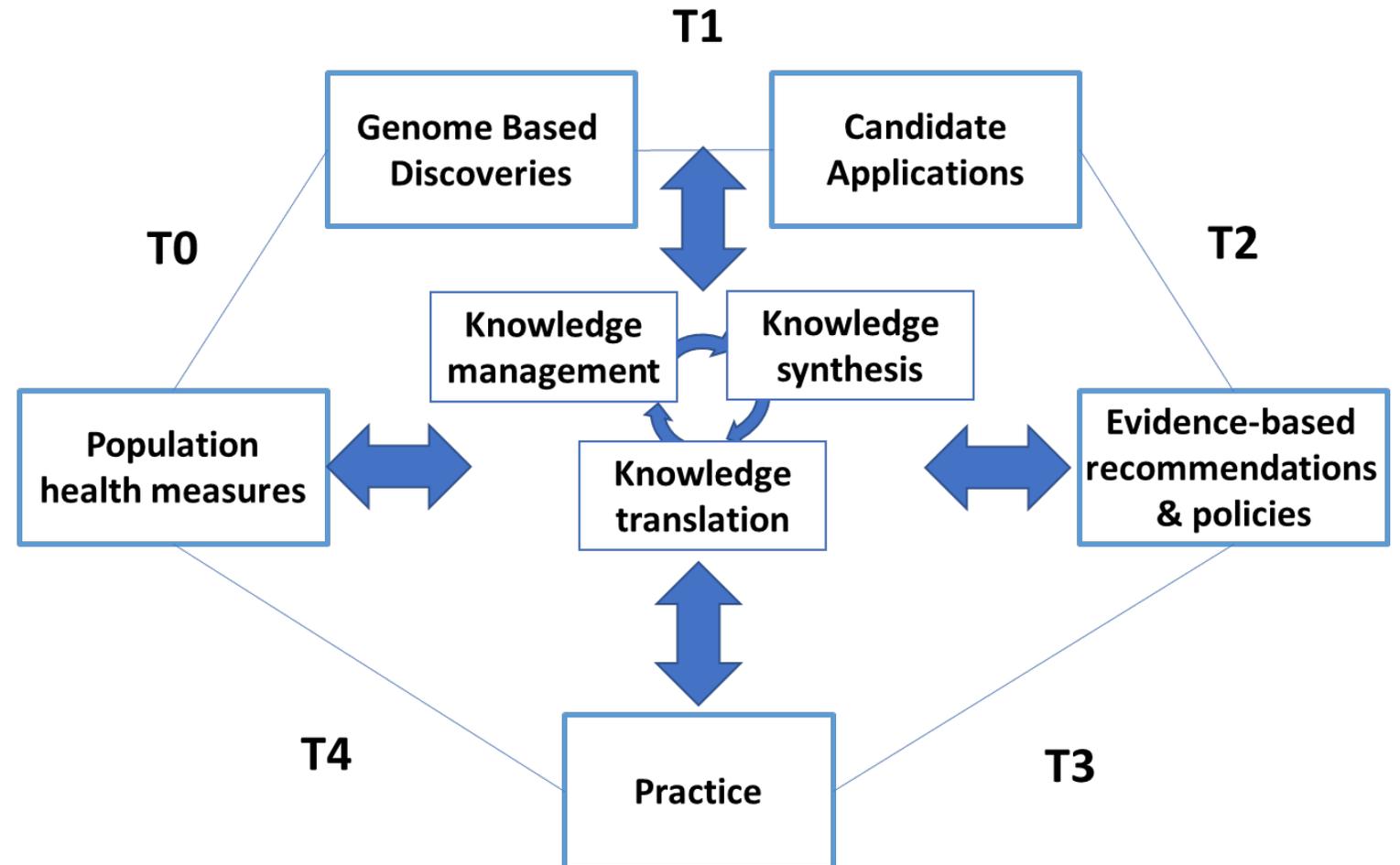
Level/Stakeholder	Example
Patients	Knowledge about genetic conditions and genetic testing
Relatives	Family dynamics
Providers	Communication about genetic conditions with patients and relatives
Laboratories	Different laboratory systems (e.g., centralized versus local) to undertake screening
Health-care organizations	Coordination between various specialties (e.g., primary care, cardiology, genetics); Electronic Health Records
Community/state leaders	State public health genomics programs to improve access to genetic testing
National health policymakers	Medicare and Medicaid benefits for genetic testing

Disparities in access to genomic medicine

- Barriers change over time
- Barriers vary between and within variation
 - Example:
 - Awareness of genetic testing for cancer risk varies by
 - Sub-ethnicity
 - Acculturation
 - Language level
 - Nativity
 - Racial and ethnic identity
- Barriers to cascade screening
 - State genetic privacy laws
 - Geography
 - Family Communication
 - Others

Disparities in quality care

- Lack of representation in research
 - Reliability of predictive models
 - Population prevalence
 - Testing benefits
- Access to risk management strategies



What tools do we have to
address complex disparities?

Implementation Science (IS)

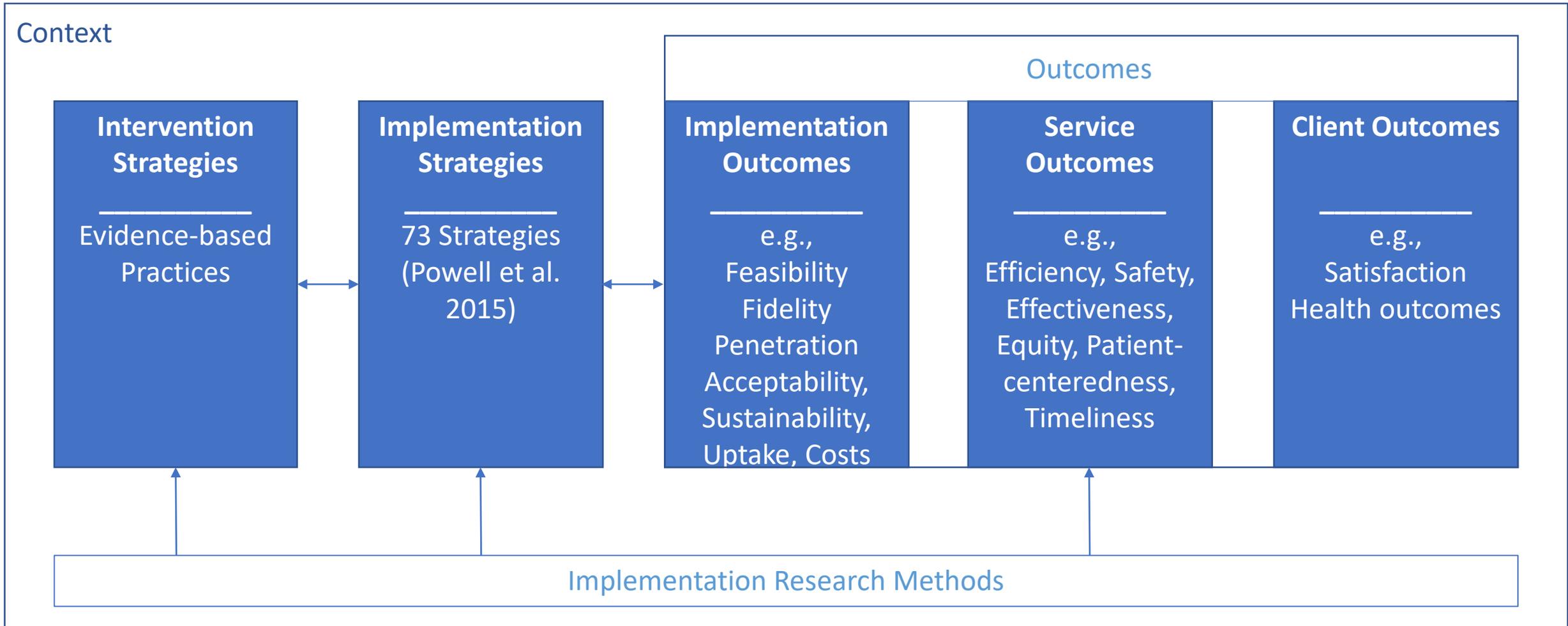
Study of methods to promote the adoption and integration of

- **evidence-based practices,**
- interventions, and
- policies

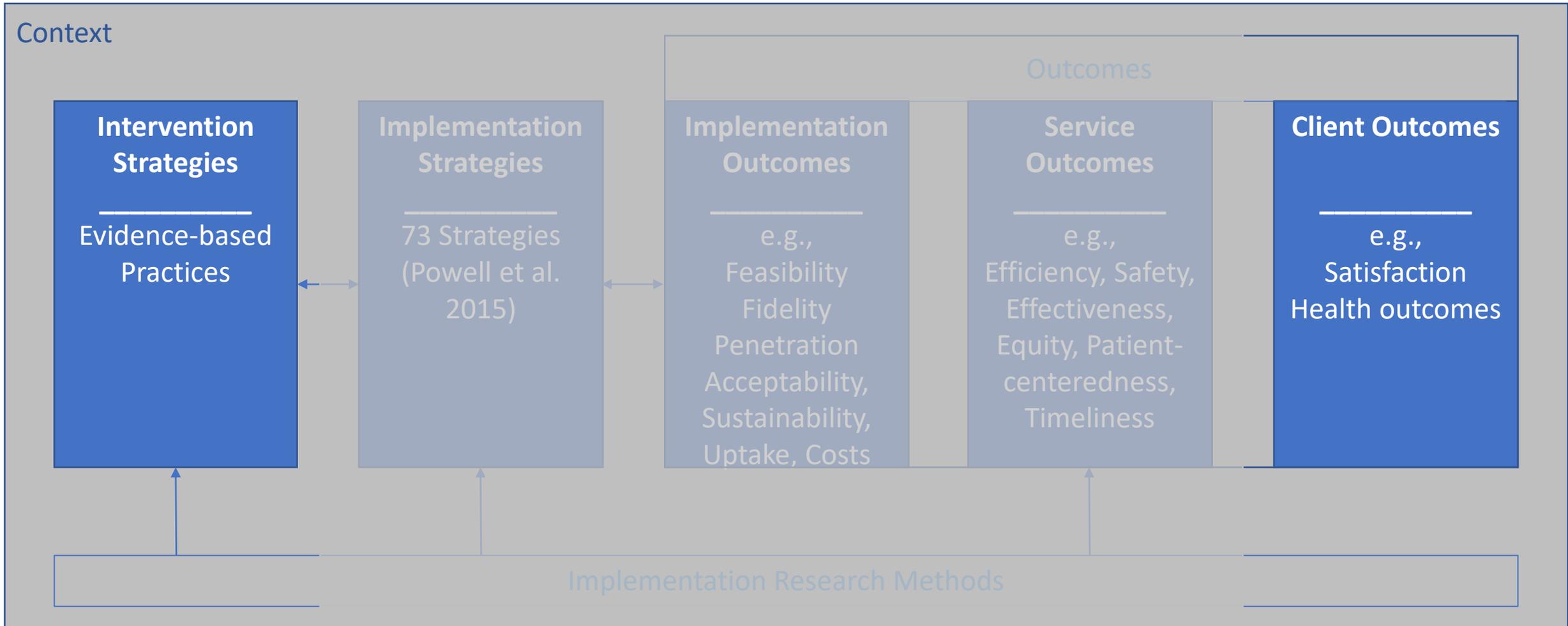
into routine health care and public health settings in order to improve our impact on **patient** and **population health.**

Implementation Science Research Methods

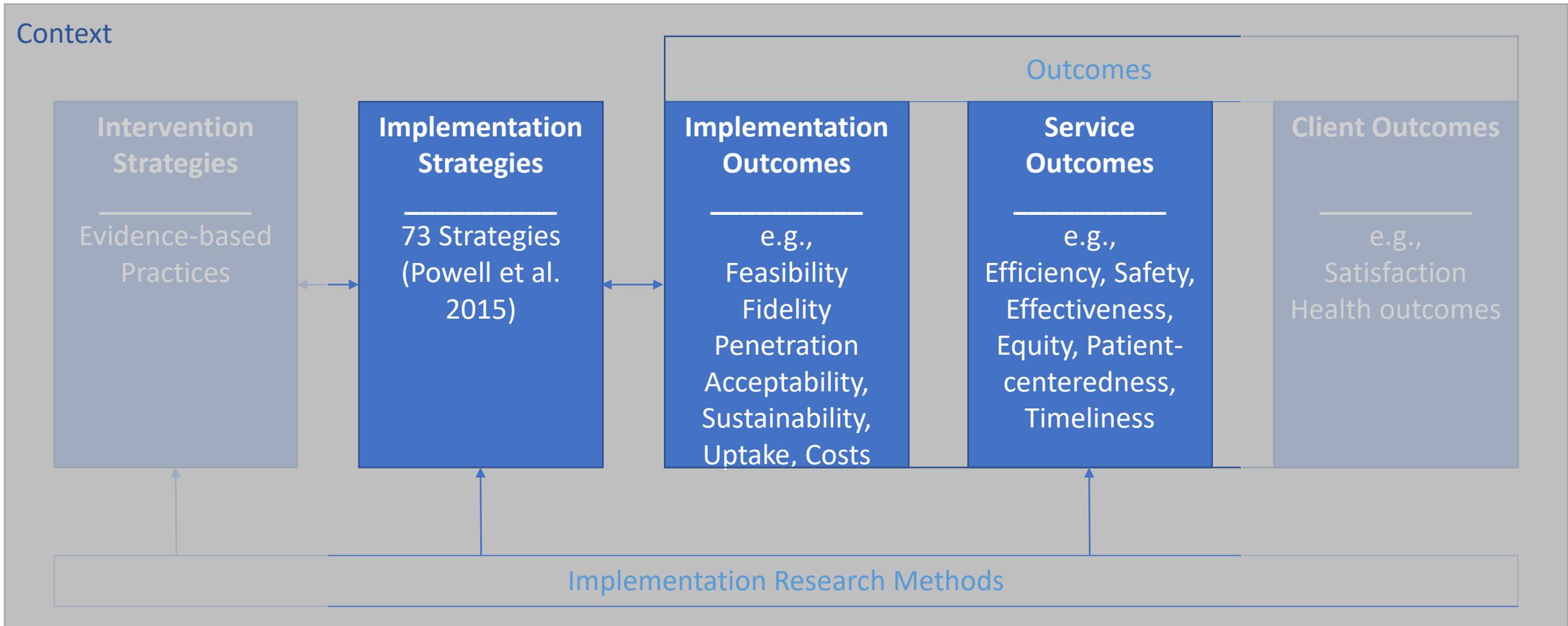
(Adapted from Proctor et al., *Adm Policy Ment Health*, 2009)



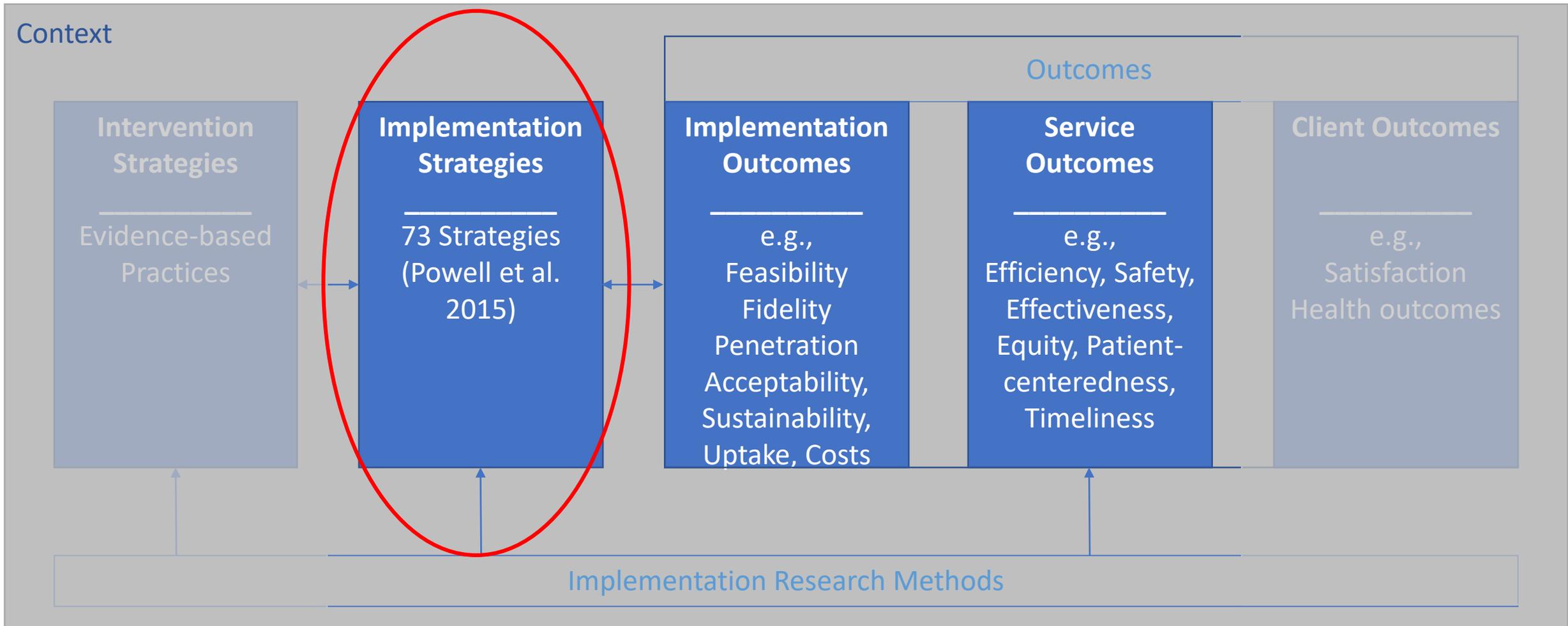
Usual Practice



Core of Implementation Science



Core of Implementation Science cont.

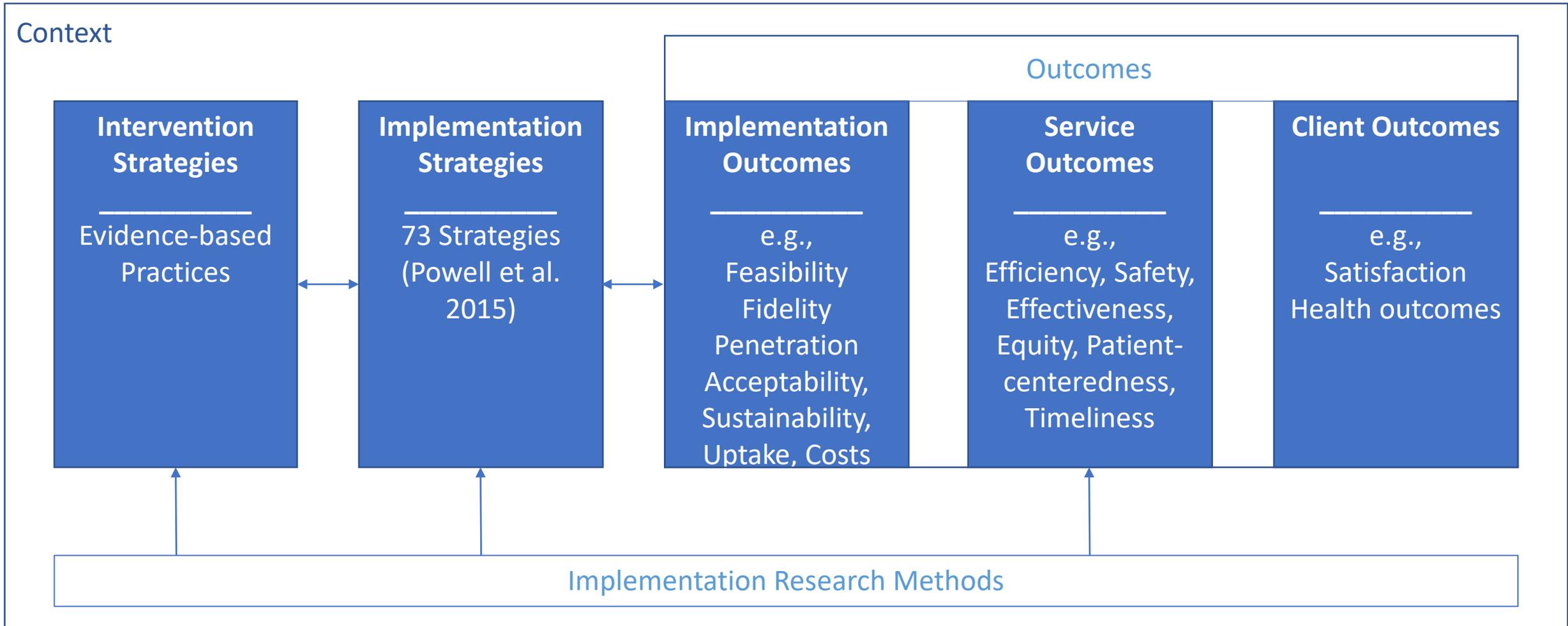


Implementation Strategies

Example Implementation Strategy	Definition
Build a coalition	Recruit and cultivate relationships with partners in the implementation effort
Conduct educational meetings	Hold meetings targeted toward different stakeholder groups (<i>e.g.</i> , providers, administrators, other organizational stakeholders, and community, patient/consumer, and family stakeholders) to teach them about the clinical innovation
Assess for readiness and identify barriers and facilitators	Assess various aspects of an organization to determine its degree of readiness to implement, barriers that may impede implementation, and strengths that can be used in the implementation effort
Conduct local needs assessment	Collect and analyze data related to the need for the innovation
Identify and prepare champions	Identify and prepare champions dedicated to supporting, marketing, and driving through an implementation, overcoming indifference or resistance that the intervention may provoke in an organization

Implementation Science Research Methods

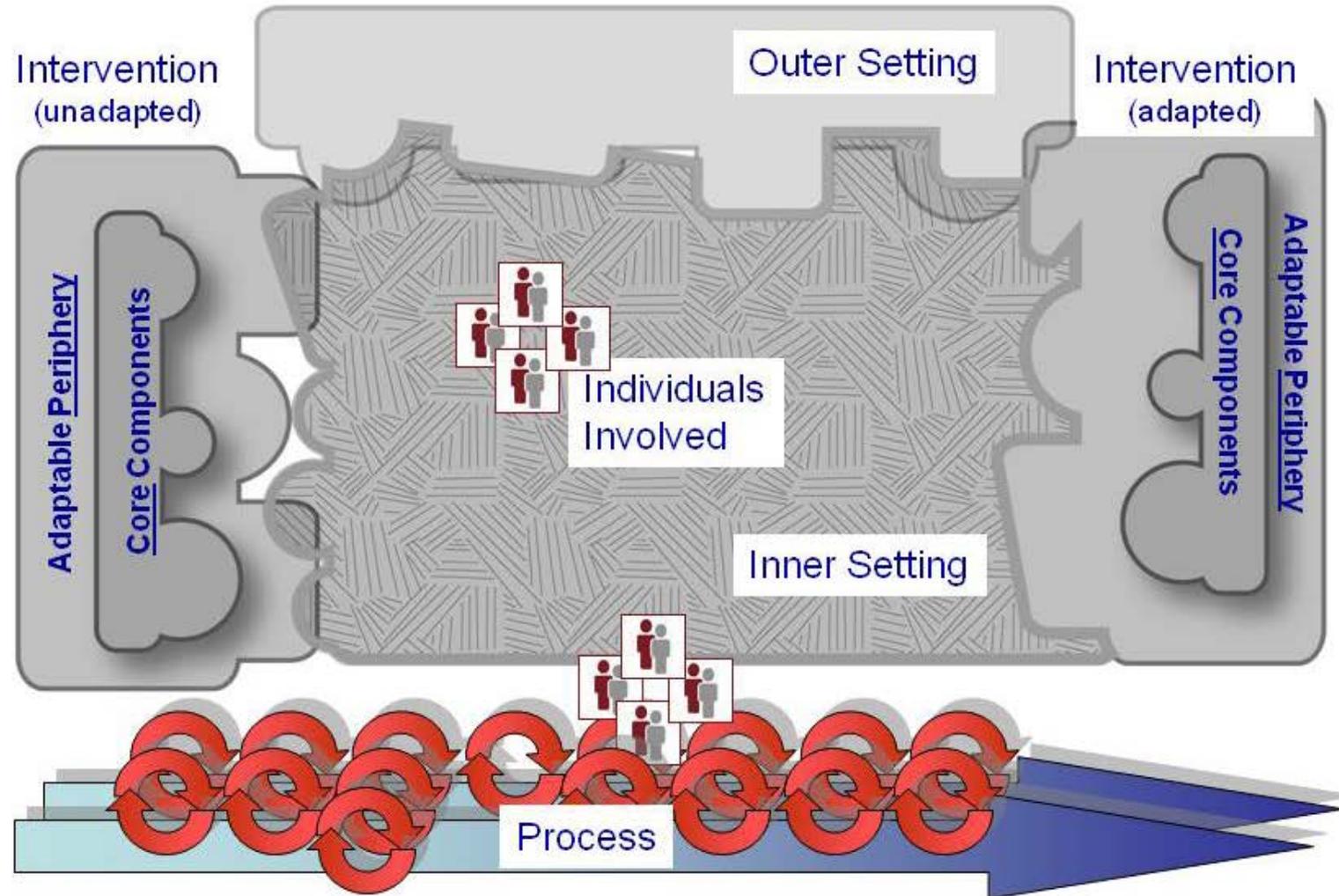
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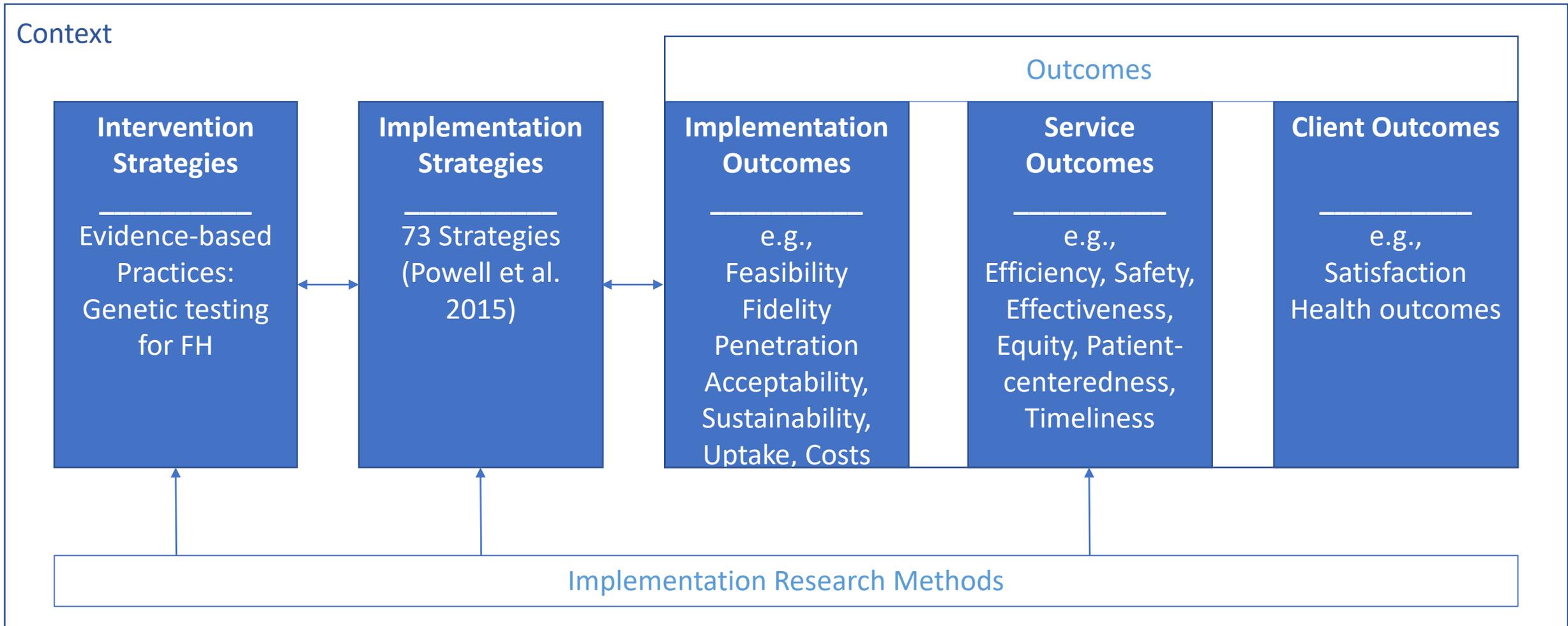
Implementation Frameworks

Category	Description	Example
Process frameworks	Specify stages, phases to describe/guide the process of translating research into practice	EPIS
Determinant frameworks	Specify types/classes or domains of determinants that can act as barriers and enablers (independent variables) that influence the implementation outcomes (dependent variables)	CFIR
Classic theories	Theories that originate from fields external to implementation science which can be applied to understand or explain aspects of implementation	Theory of planned behavior
Implementation theories	Theories developed by implementation researchers to provide understanding of aspects of implementation	ISF
Evaluation frameworks	Specify aspects of implementation that could be evaluated	RE-AIM

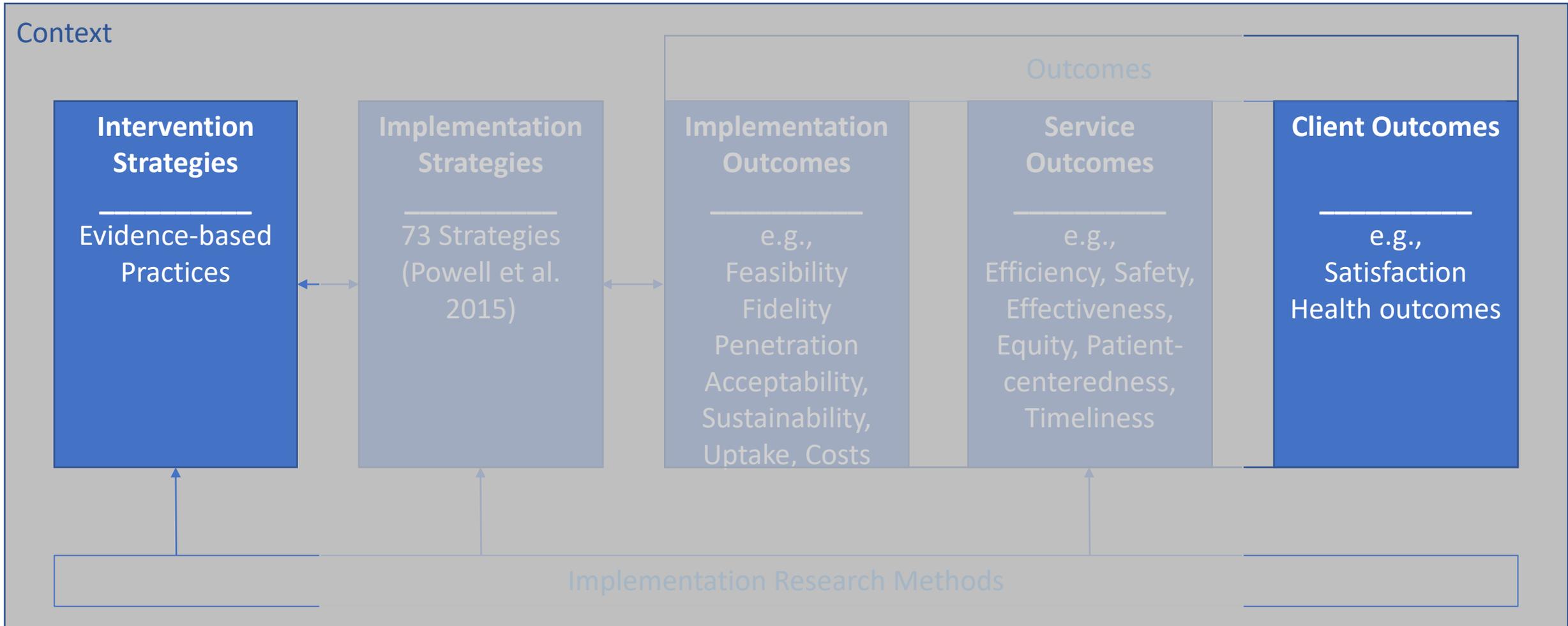
Example Framework: Consolidated Framework for Implementation Research (CFIR)



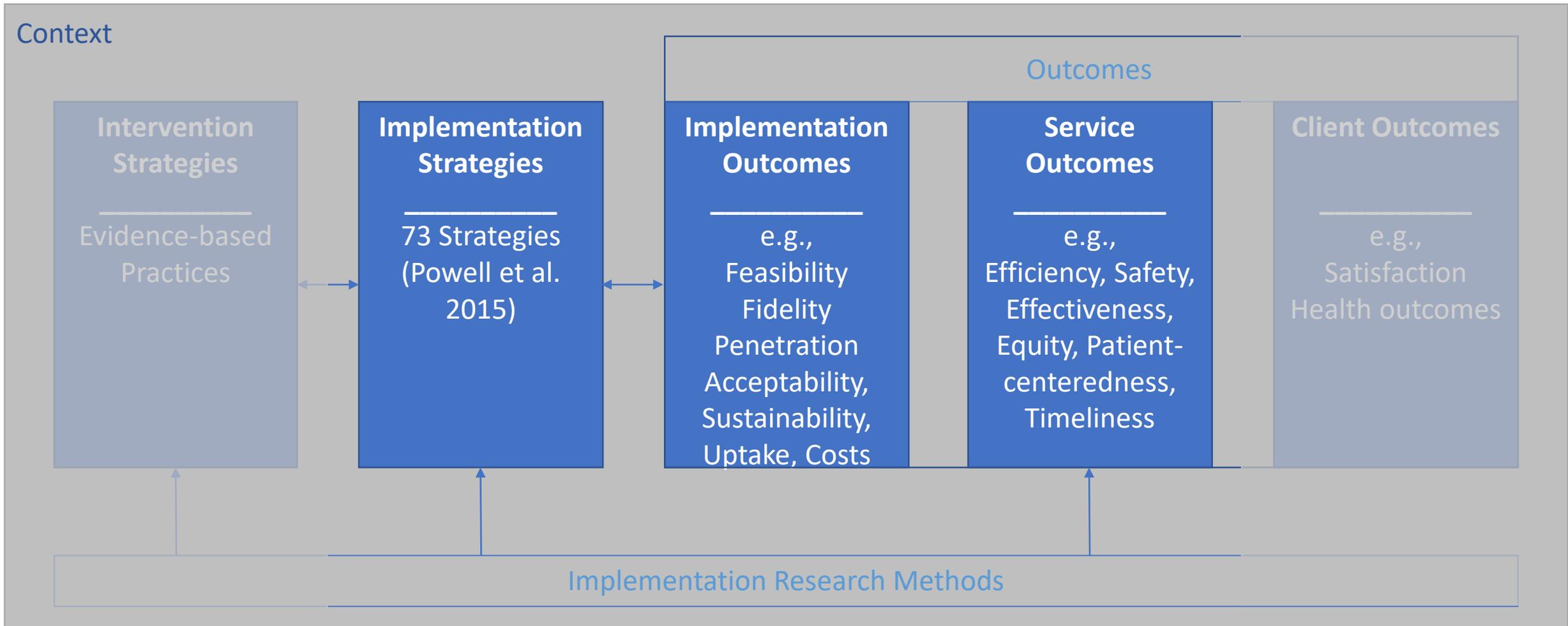
An example: Familial Hypercholesterolemia



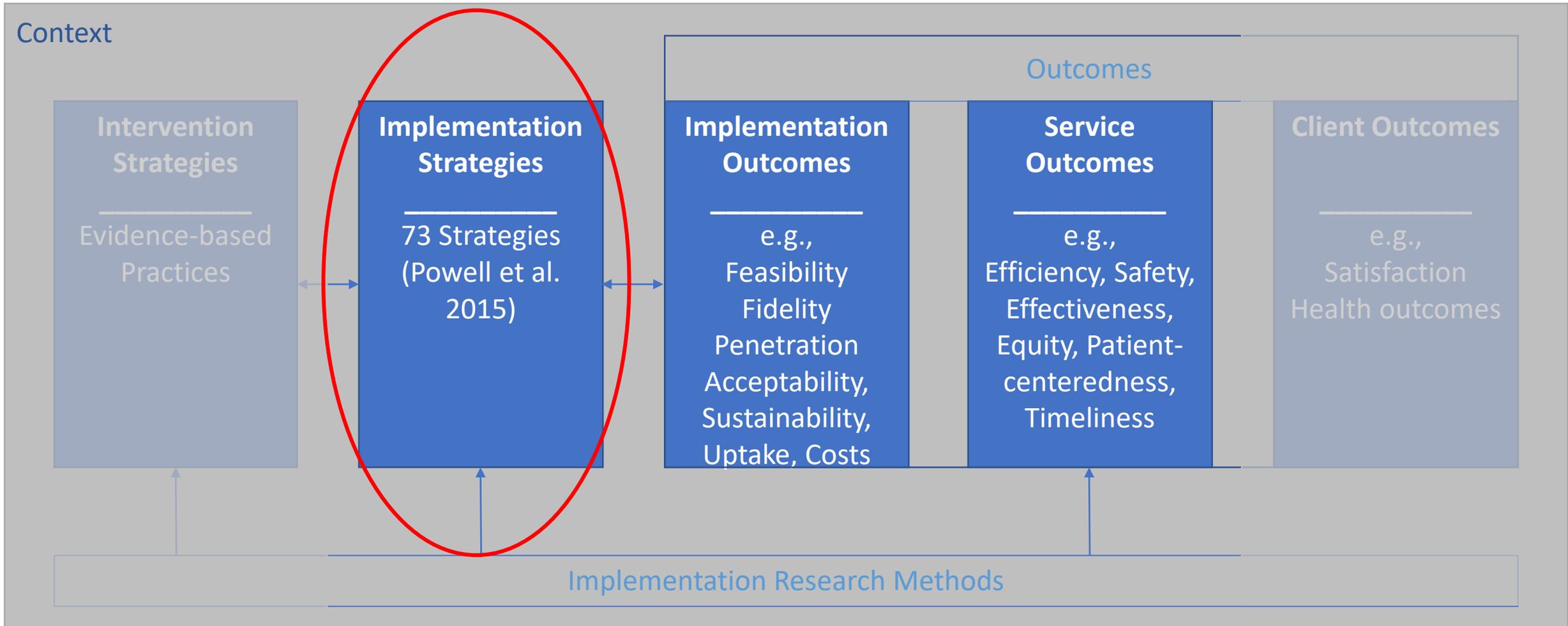
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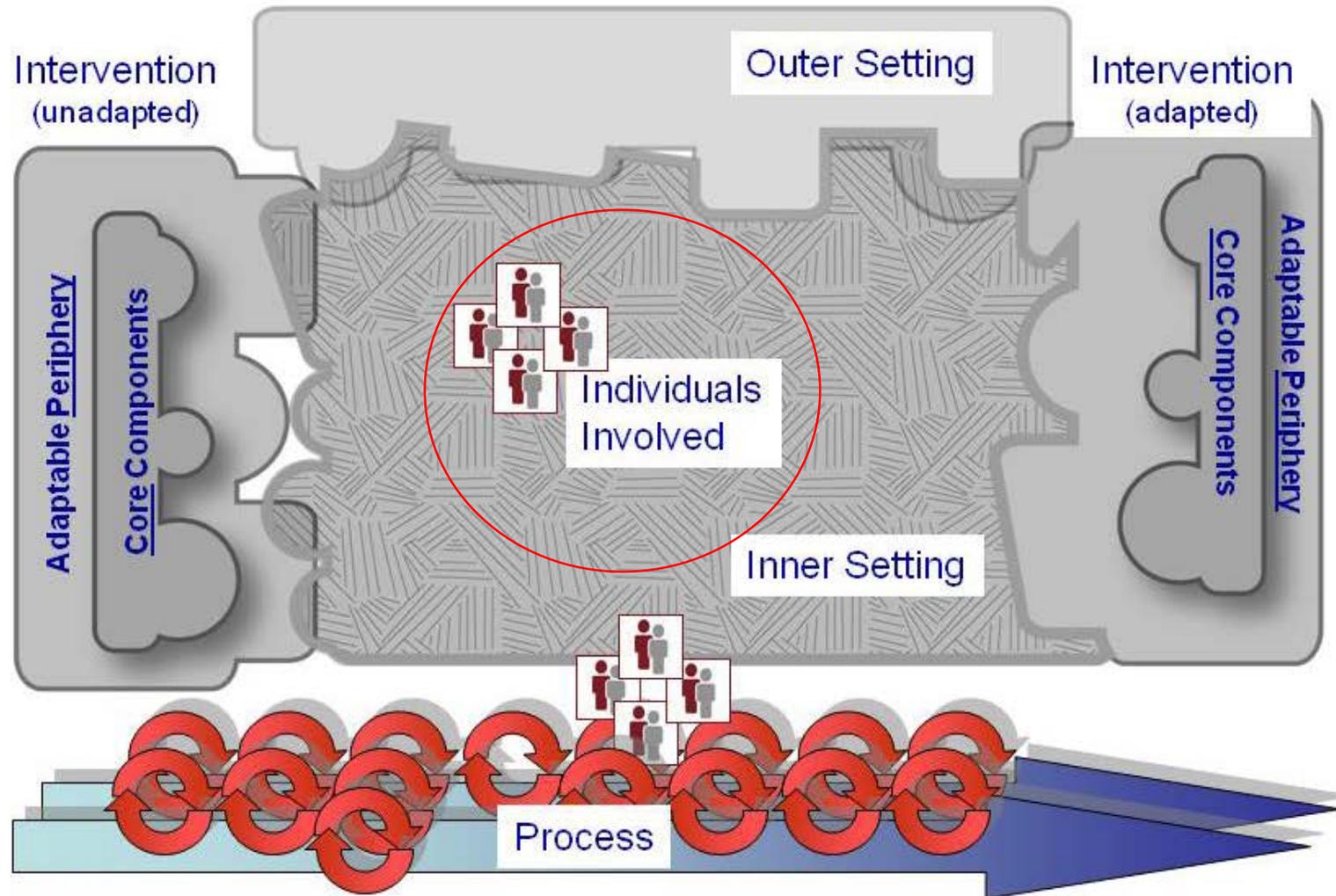
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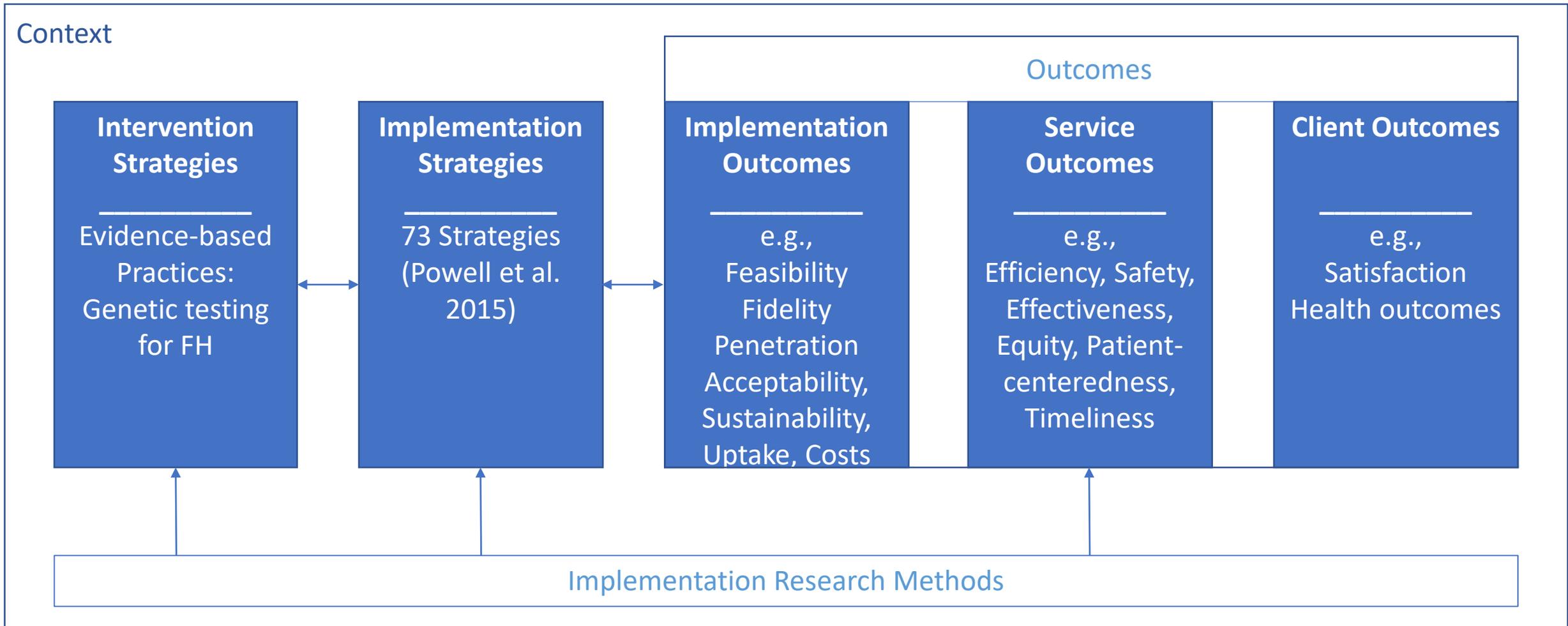
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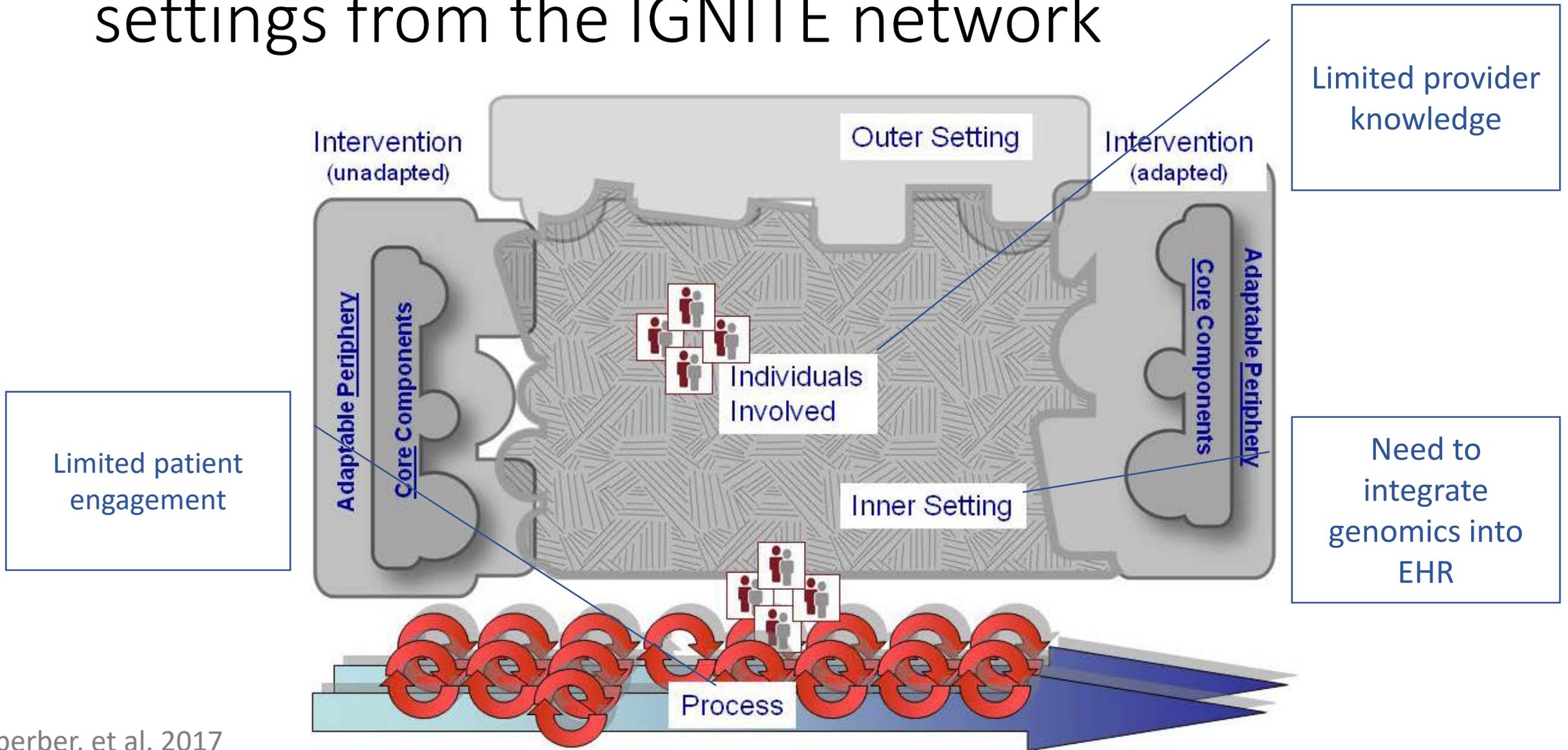


An example: Familial Hypercholesterolemia



Examples from the literature

Implementing genomic services in diverse settings from the IGNITE network



State-based public health genomics programs: An example of a multilevel approach

- Estimate burden of hereditary conditions
- Educate providers and public
- Promote policies to increase access to genetic services
- Build a coalition (collaborate with key stakeholders)
- Tailor programs to meet local needs

Implementation Science in Genomic Medicine

Genetics

Medicine

SYSTEMATIC REVIEW

Conclusion: Although genomic discovery provides the potential for population health benefit, the current knowledge base around implementation to turn this promise into a reality is severely limited. Current gaps in the literature demonstrate a need to apply implementation science principles to genomic medicine in order to deliver on the promise of precision medicine.

Purpose: The objective of this study was to identify trends and gaps in the field of implementation science in genomic medicine.

Methods: We conducted a literature review using the Centers for Disease Control and Prevention's Public Health Genomics Knowl-

particularly oncology (35%, $n = 99$). Key study design elements, such as racial/ethnic composition of study populations, were underreported in studies. Few studies incorporated implementation science theoretical frameworks, sustainability measures, or capacity building.

Future Directions

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Thank you.

Questions?