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

Cascade screening

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

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

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<tr>
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<th>Service Outcomes</th>
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Implementation Research Methods

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

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**Outcomes**

- e.g., Feasibility, Fidelity, Penetration, Acceptability, Sustainability, Uptake, Costs
- e.g., Efficiency, Safety, Effectiveness, Equity, Patient-centeredness, Timeliness
- e.g., Satisfaction, Health outcomes

**Implementation Research Methods**

Proctor et al. 2009
Reviews

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

- Reviewed literature 2018
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; cfarguide.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

Roberts et al *Health Affairs* 2018; Srinivasan et al. *EJHG* 2020
Enablers for cascade screening

Individuals Involved

Beliefs

Roberts et al. *Health Affairs* 2018; Srinivasan et al. *EJHG* 2020
CFIR
Barriers to Cascade Screening

Inner Setting

Compatibility
Relative priority
Available resources
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
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Thank You.