Health Disparities in Implementation of Genomic Medicine: Challenges and Opportunities

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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
Time Lags in Translational Research

- 55%
- 14%, 17 years

Balas and Boren 2000; Morris et al. 2011; Asch et al. 2003; Smedley, Stith, Nelson 2003
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.”

(Braveman, 2006)
Defining disparities cont.

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(Braveman, 2006)
Difference vs. Disparity

McGuire et al. 2006
Access to Care, 2014

AHRQ, 2016
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

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

Khoury et al. 2012
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

Landry, et al. 2018; Hall and Olopade 2006
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)

**Context**

- Intervention Strategies
  - Evidence-based Practices

- Implementation Strategies
  - 73 Strategies (Powell et al. 2015)

**Outcomes**

- Implementation Outcomes
  - e.g., Feasibility, Fidelity, Penetration, Acceptability, Sustainability, Uptake, Costs

- Service Outcomes
  - e.g., Efficiency, Safety, Effectiveness, Equity, Patient-centeredness, Timeliness

- Client Outcomes
  - e.g., Satisfaction, Health outcomes

Implementation Research Methods
Usual Practice

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Implementation Research Methods
Core of Implementation Science

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Implementation Research Methods
Core of Implementation Science cont.

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Implementation Research Methods
# Implementation Strategies

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<tr>
<th>Example Implementation Strategy</th>
<th>Definition</th>
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<tbody>
<tr>
<td>Build a coalition</td>
<td>Recruit and cultivate relationships with partners in the implementation effort</td>
</tr>
<tr>
<td>Conduct educational meetings</td>
<td>Hold meetings targeted toward different stakeholder groups (e.g., providers, administrators, other organizational stakeholders, and community, patient/consumer, and family stakeholders) to teach them about the clinical innovation</td>
</tr>
<tr>
<td>Assess for readiness and identify barriers and facilitators</td>
<td>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</td>
</tr>
<tr>
<td>Conduct local needs assessment</td>
<td>Collect and analyze data related to the need for the innovation</td>
</tr>
<tr>
<td>Identify and prepare champions</td>
<td>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</td>
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### Implementation Frameworks

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

Context

Intervention Strategies
- Evidence-based Practices: Genetic testing for FH

Implementation Strategies
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Implementation Research Methods
Examples from the literature
Implementing genomic services in diverse settings from the IGNITE network

- Limited patient engagement
- Limited provider knowledge
- Need to integrate genomics into EHR

Sperber, et al. 2017
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

Senier et al. 2018; Powell et al. 2015
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 Knowledge Portal, 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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    • Working group members

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Thank you.

Questions?