

The slide features decorative curved lines in shades of blue and green. One line starts at the top right and curves downwards. Another starts at the bottom left and curves upwards. A third line is positioned in the middle right, curving from the top towards the bottom.

Evaluating the Value Proposition for Population Genomic Screening

Marc S. Williams, MD, FAAP, FACMG, FACMI

Objectives

- Define value in the healthcare context
- Explore the concept of value from different stakeholder perspectives
- Propose areas to include in population genomic screening research that support value measurement

What is Value?

- Can be thought of as a relationship between outcomes and cost of care
- Definition “Value in health care is the measured improvement in a person’s health outcomes for the cost of achieving that improvement.”*

*Porter ME, Teisberg EO. Redefining Health Care: Creating Value-Based Competition on Results. 2006 Boston, MA: Harvard Business School Press.

Value Plot

Medical and/or Service Outcomes	Cost of care decreased	Cost of care unchanged	Cost of care increased
Improved	Immunizations		Molecularly-targeted cancer treatments
Unchanged			
Worsened			BMT for advanced breast cancer

The Perspective on Outcomes

Medical Outcomes

- Morbidity or Mortality
- Disease Free Survival
- Treatment effectiveness
- Treatment safety
- Preventive services (outcomes specific to specific intervention)

Patient-centered Outcomes

- Satisfaction
- Timeliness
- Access
- Health Behavior Change
- Empowerment/Engagement
- Knowledge
- Personal Utility

System Outcomes

- Costs incurred/avoided
- Utilization
- Visibility/reputation
- Patient experience

Cost

- Standard costs associated with the interventions and health states experienced by the patient. Cost/outcome (Cost/QALY)
- NB Hard to measure (at least in the US)

Process Outcomes

These measures are the specific steps in a process that lead — either positively or negatively — to a particular health outcome

Intermediate Outcomes

A biomarker associated — either positively or negatively — to a particular health outcome

Health Outcomes

Change in the health of an individual, group of people or population which is attributable to an intervention or series of interventions

Value-based Healthcare

- Goal of value-based care transformation is to enable the health care system to create more value for patients
- Focus on cost reduction without improvement in outcomes is incomplete
- Value-based health care is not the same as quality, although quality may be a component
- Patient satisfaction is not equivalent to value-based care, but outcomes must include the patient perspective
- Improving the patient experience associated with a value-based intervention enhances value from the patient perspective

Value-based Healthcare



Teisberg E, Wallace S, O'Hara S. Defining and Implementing Value-Based Health Care: A Strategic Framework. Acad Med. 2020 May;95(5):682-685
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Value-based Healthcare in Genomics

- Population Screening for CDC Tier 1 conditions
 - Outcomes from MyCode Community Health Initiative
 - Cost-effectiveness study Rational Integration of clinical SEquencing (RISE)

'Tier 1' Outcomes Paper

What is clinical utility of genomic screening program among MyCode patients with a 'CDC Tier 1' genomic condition?

- 350 patients with HBOC, Lynch, or FH result (May 2015-February 2018)
- Double-coded chart review performed by clinicians in June-Dec 2018
- Median follow-up window: 21.8 months (inter-quartile range 15-31 months)

Buchanan AH, et al. Clinical outcomes of a genomic screening program for actionable genetic conditions. Genet Med (2020). <https://doi.org/10.1038/s41436-020-0876-4>

'Tier 1' Outcomes Paper

Results, Conclusions

Majority of patients in genomic screening program:

Previously unaware of their Tier 1 variant (87%)	Eligible to perform risk management (86%)	Performed <i>some</i> management post-disclosure (68%)
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Ascertainment of genomic risk led to relevant disease diagnoses during follow-up period (13%)

Supports effectiveness of genomic screening programs in identifying previously undetected individuals at risk for preventable cancers and heart disease

Buchanan AH, et al. Clinical outcomes of a genomic screening program for actionable genetic conditions. Genet Med (2020). <https://doi.org/10.1038/s41436-020-0876-4>

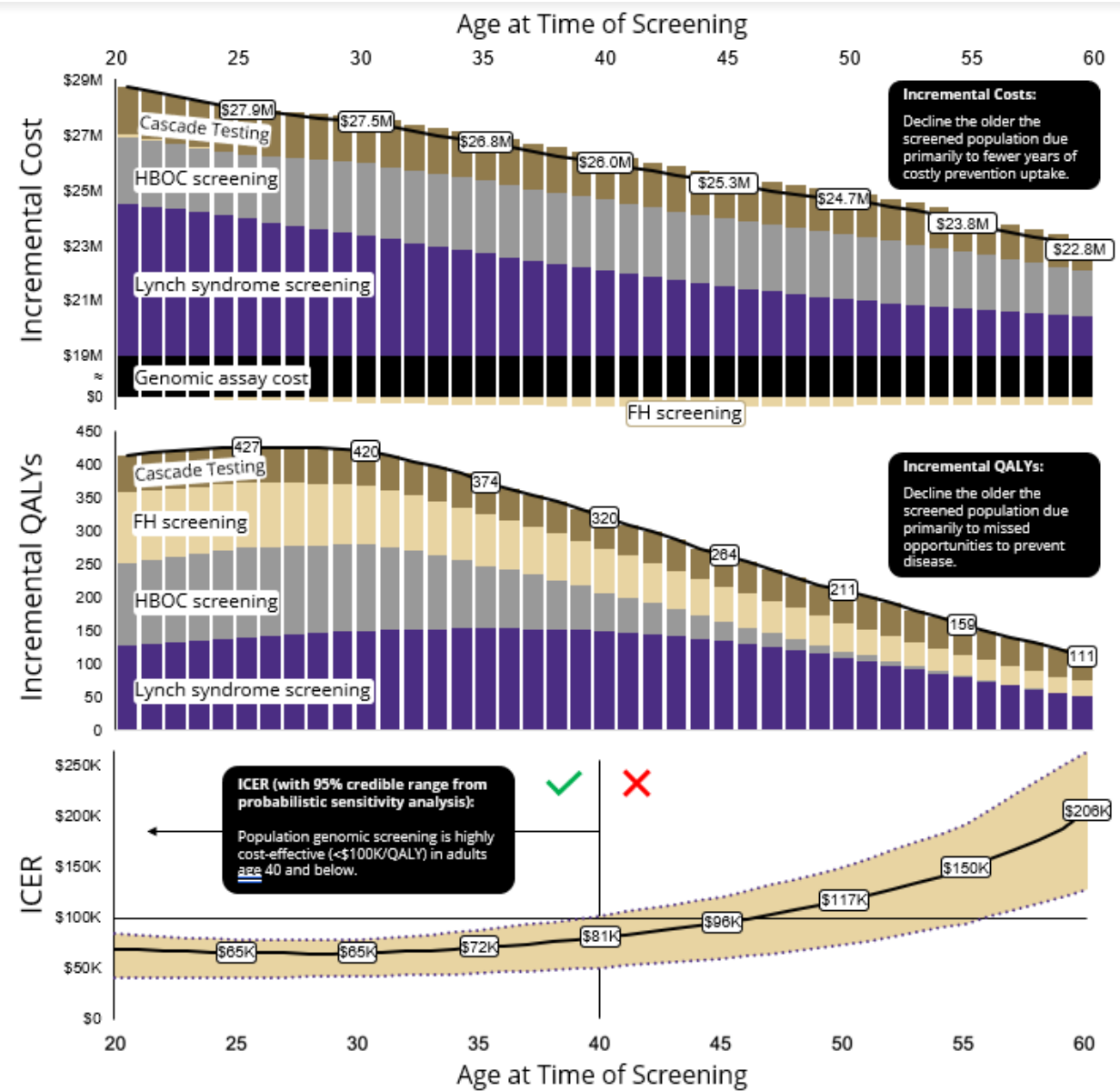
'Tier 1' Outcomes Paper

- Demonstrated some aspects of clinical utility
- Outcomes
 - Most outcomes were process (imaging, procedure, analyte analysis)
 - Some intermediate outcomes (LDL lowering, polyp removal)
 - No health outcomes
- Cost not measured

Buchanan AH, et al. Clinical outcomes of a genomic screening program for actionable genetic conditions. Genet Med (2020). <https://doi.org/10.1038/s41436-020-0876-4>

RISE Cost-effectiveness Paper

Single Con
Hereditary
Ovarian Ca
Syndrome
Lynch
Syndrome
Familial
hyperchole



ars old

Guzauskas GF, et al. Population Genomic Screening for Three Common Hereditary Conditions : A Cost-Effectiveness Analysis. Ann Intern Med. 2023 176:585-595. doi: 10.7326/M22-0846.

Value Plot for Tier 1 Population Genomic Screening

Medical and/or Service Outcomes	Cost of care decreased	Cost of care unchanged	Cost of care increased
Improved			??
Unchanged			
Worsened			

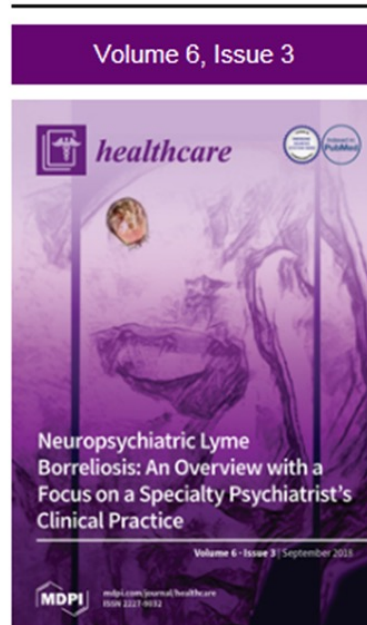
Value-based Healthcare in Genomics— Research Opportunities

- Outcomes Definition and Standardization

Statement of Problem

- Outcomes are critical to determination of the utility and value of any medical intervention
- Outcomes for genomic medicine are just beginning to be collected
- NIH- and NHGRI-funded projects are collecting outcomes
 - CSER
 - IGNITE
 - eMERGE
 - ClinSeq
 - UDN
 - Others
- Some institutions have projects for which outcomes are being collecting (PREDICT at Vanderbilt, Cardiovascular outcomes at Mayo, Cancer outcomes UW and Intermountain, exome/genome return of results Rady Children's, Geisinger MyCode CHI, and others)

Early Work on Harmonization



Volume 6, Issue 3

healthcare

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Healthcare 2018, 6(3), 83; <https://doi.org/10.3390/healthcare6030083>

Open Access Article

Harmonizing Outcomes for Genomic Medicine: Comparison of eMERGE Outcomes to ClinGen Outcome/Intervention Pairs

Janet L. Williams¹ ✉, Wendy K. Chung² ✉, Alex Fedotov³ ✉, Krzysztof Kiryluk⁴ ✉, Chunhua Weng⁵ ✉, John J. Connolly⁶ ✉, Margaret Harr⁶ ✉, Hakon Hakonarson^{6,7} ✉, Kathleen A. Leppig⁸ ✉, Eric B. Larson⁹ ✉, Gail P. Jarvik¹⁰ ✉, David L. Veenstra¹¹ ✉, Christin Hoell¹² ✉, Maureen E. Smith¹² ✉, Ingrid A. Holm¹³ ✉, Josh F. Peterson¹⁴ ✉ and Marc S. Williams^{1,*} ✉

¹ Genomic Medicine Institute, Geisinger, Danville, PA 17822, USA

² Departments of Pediatrics and Medicine, Columbia University, New York, NY 10025, USA

³ Irving Institute for Clinical and Translational Research, Columbia University, New York, NY 10025, USA

⁴ Department of Medicine, Division of Nephrology, Columbia University, New York, NY 10025, USA

⁵ Department of Biomedical Informatics, Columbia University, New York, NY 10025, USA

⁶ Children's Hospital of Philadelphia, Philadelphia, PA 19104, USA

⁷ Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA 19104, USA

⁸ Genetic Services, Kaiser Permanente of Washington, Seattle, WA 98101, USA

⁹ Kaiser Permanente Washington Health Research Institute, Seattle, WA 98101, USA

¹⁰ Departments of Medicine (Medical Genetics) and Genome Sciences, University of Washington, Seattle, WA 98195, USA

¹¹ Department Pharmacy, University of Washington, Seattle, WA 98195, USA

¹² Center for Genetic Medicine, Northwestern University, Chicago, IL 60611, USA

¹³ Division of Genetics and Genomics, Boston Children's Hospital, and Department of Pediatrics, Harvard Medical School, Boston, MA 02115, USA

¹⁴ Departments of Biomedical Informatics and Medicine, School of Medicine, Vanderbilt University, Nashville, TN 37232, USA

* Author to whom correspondence should be addressed.

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Full-Text | PDF [650 KB, uploaded 13 July 2018] | Figure

Early Work on Harmonization



BRIEF COMMUNICATION

Genomic medicine implementation protocols in the PhenX Toolkit: tools for standardized data collection

Wendy K. Chung¹, Kyle Brothers², Angela Bradbury^{3,4}, Sirisak Chanprasert⁵, Lori Orlando⁶, Ali Torkamani⁷, Heather Zierhut⁸, Marylyn D. Ritchie⁹, Michael Phillips^{10✉}, Jennifer Schoden¹⁰, Deborah Maiese¹⁰, Tabitha Hendershot¹⁰, Carol M. Hamilton¹⁰ and Erin M. Ramos¹¹

PURPOSE: The PhenX Toolkit (www.phenxtoolkit.org), an online catalog of recommended measurement protocols, facilitates cross-study analyses for research with human participants. The PhenX Steering Committee recommended genomic medicine implementation as a new research domain, with the following scope: genomic knowledge and education (both patients and providers); implementation science; changes in management and treatment; return of results; patient outcomes; and ethical, legal, and social issues (ELSI) associated with genomic research.

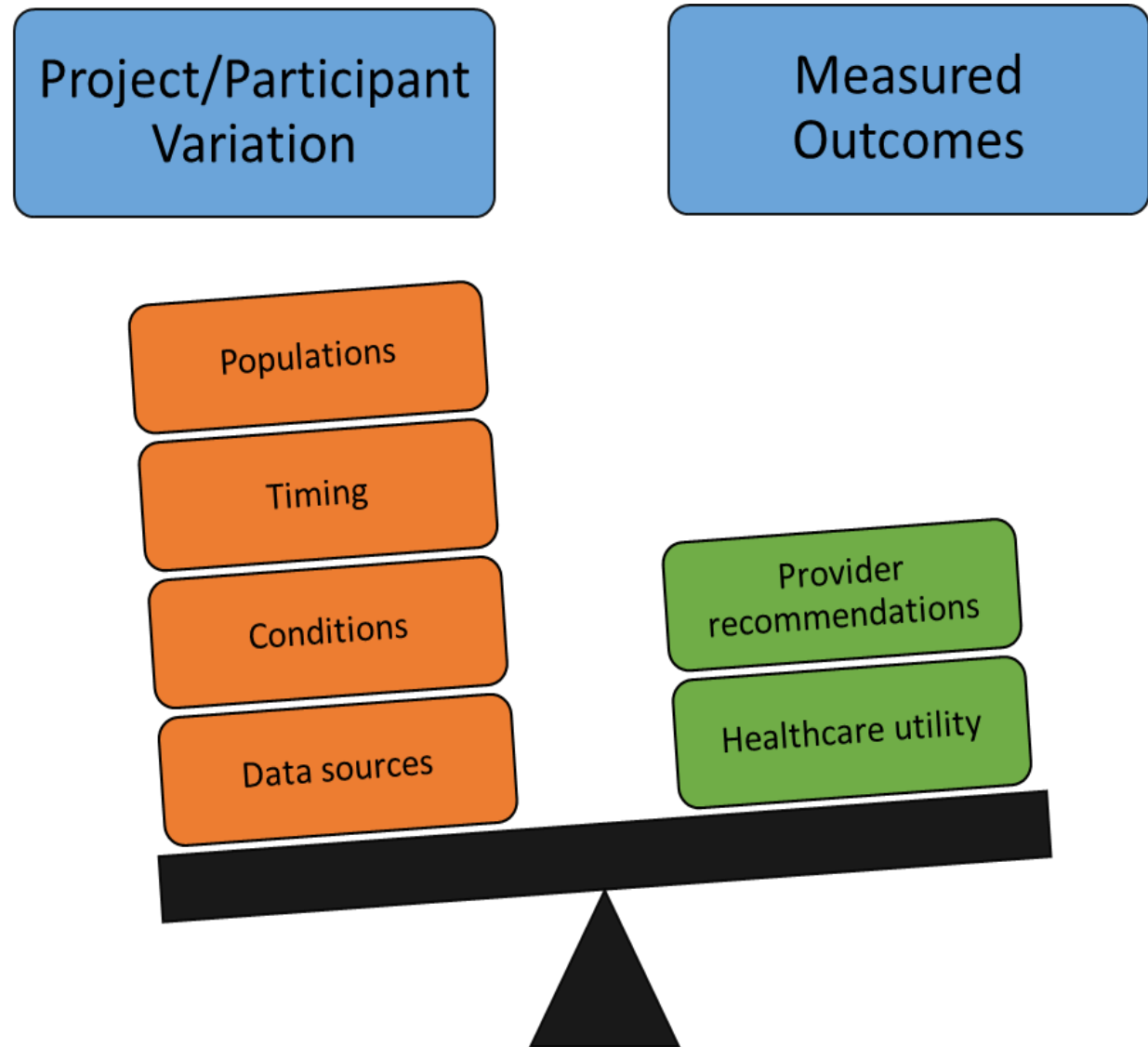
METHODS: A seven-member expert Working Group convened in October 2019 to identify well-established measurement protocols for a new genomic medicine implementation domain and used the established PhenX consensus process to select measurement protocols for inclusion in the PhenX Toolkit.

RESULTS: The Working Group recommended 15 measurement protocols for inclusion in the PhenX Toolkit, with priority given to those with empirical evidence supporting validity. Consortia funded by the National Institutes of Health, and particularly the National Human Genome Research Institute, proved critical in identifying protocols with established utility in this research domain, and identified protocols that were developed through a rigorous process for scope elements that lacked formally validated protocols.

CONCLUSION: Use of these protocols, which were released in September 2020, can facilitate standard data collection for genomic medicine implementation research.

Genetics in Medicine (2021) 23:1783–1788; <https://doi.org/10.1038/s41436-021-01183-0>

Early Work on Harmonization



Value-based Healthcare in Genomics— Research Opportunities

- Outcomes Definition and Standardization
- Engagement with broader stakeholder groups to expand outcomes that contribute to holistic consideration of value
 - Definition of health outcomes of most importance to patients essential
- Standardized cost definition and capture

Conclusion

“Value in health care is determined in addressing the patient’s particular medical condition over the full cycle of care, from monitoring and prevention to treatment to ongoing disease management.”

— Michael E. Porter, Redefining Health Care:
Creating Value-Based Competition on Results