Clinical Guidelines for Genomic Testing: Evidence Synthesis Considerations

Examining Clinical Guidelines for the Adoption of Genomic Testing: A Workshop

SESSION VI: Guideline Development in a Rapidly Evolving Field – A Look Ahead

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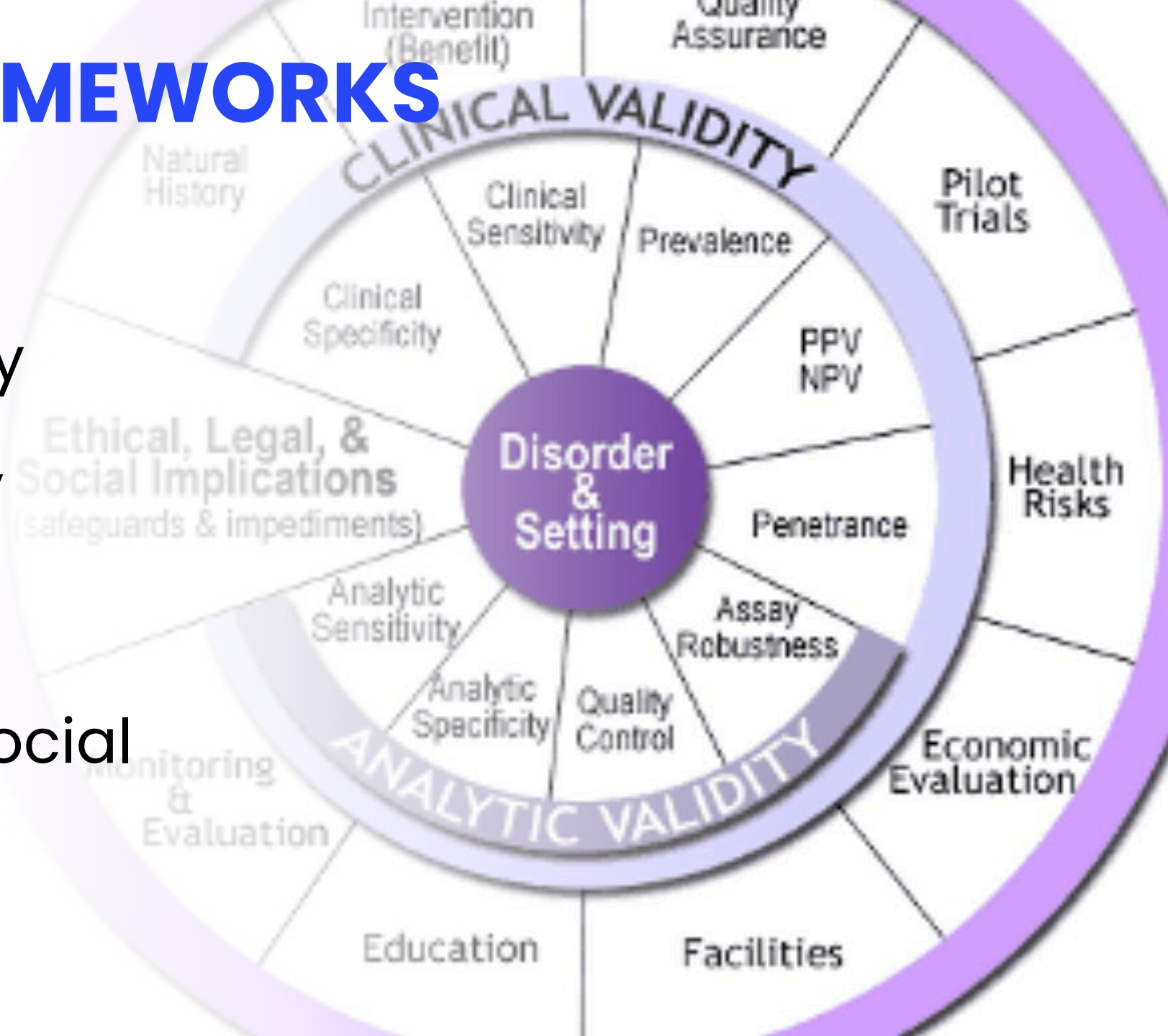
October 29, 2024

DECLARATIONS

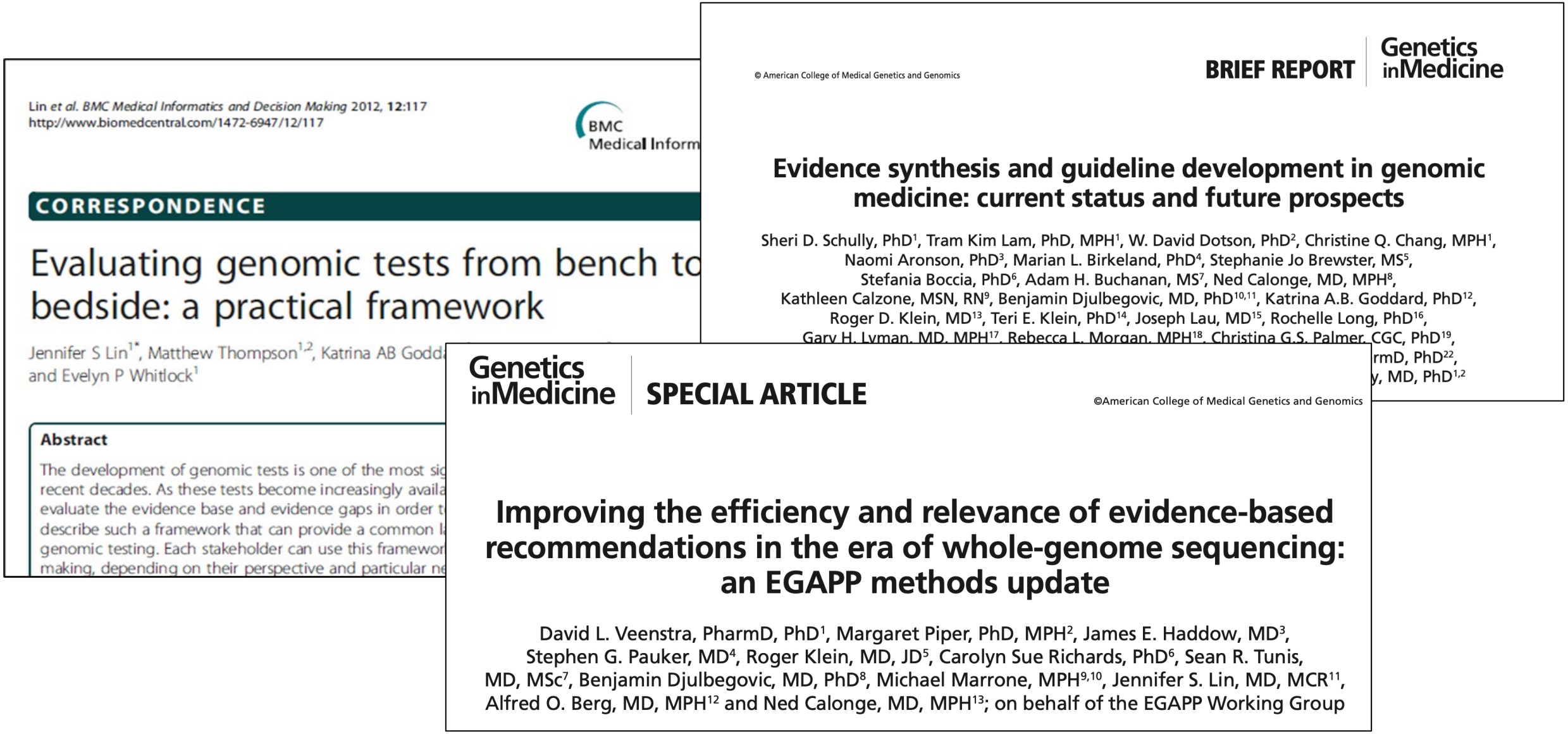
- No conflicting affiliations or financial interests

EVIDENCE FRAMEWORKS

- ACCE
  - Analytic validity
  - Clinical validity
  - Clinical utility
  - Ethical, legal, social issues



EVIDENCE FRAMEWORKS



Lin *et al. BMC Medical Informatics and Decision Making* 2012, **12**:117
http://www.biomedcentral.com/1472-6947/12/117

BMC Medical Inform

CORRESPONDENCE

Evaluating genomic tests from bench to bedside: a practical framework

Jennifer S Lin[1*], Matthew Thompson[1,2], Katrina AB Godd
and Evelyn P Whitlock[1]

Abstract

The development of genomic tests is one of the most sig
recent decades. As these tests become increasingly availa
evaluate the evidence base and evidence gaps in order t
describe such a framework that can provide a common l
genomic testing. Each stakeholder can use this framewor
making, depending on their perspective and particular ne

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BRIEF REPORT | Genetics inMedicine

Evidence synthesis and guideline development in genomic medicine: current status and future prospects

Sheri D. Schully, PhD[1], Tram Kim Lam, PhD, MPH[1], W. David Dotson, PhD[2], Christine Q. Chang, MPH[1], Naomi Aronson, PhD[3], Marian L. Birkeland, PhD[4], Stephanie Jo Brewster, MS[5], Stefania Boccia, PhD[6], Adam H. Buchanan, MS[7], Ned Calonge, MD, MPH[8], Kathleen Calzone, MSN, RN[9], Benjamin Djulbegovic, MD, PhD[10,11], Katrina A.B. Goddard, PhD[12], Roger D. Klein, MD[13], Teri E. Klein, PhD[14], Joseph Lau, MD[15], Rochelle Long, PhD[16], Gary H. Lyman, MD, MPH[17], Rebecca L. Morgan, MPH[18], Christina G.S. Palmer, CGC, PhD[19], rmD, PhD[22], y, MD, PhD[1,2]

Genetics inMedicine | SPECIAL ARTICLE

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Improving the efficiency and relevance of evidence-based recommendations in the era of whole-genome sequencing: an EGAPP methods update

David L. Veenstra, PharmD, PhD[1], Margaret Piper, PhD, MPH[2], James E. Haddow, MD[3], Stephen G. Pauker, MD[4], Roger Klein, MD, JD[5], Carolyn Sue Richards, PhD[6], Sean R. Tunis, MD, MSc[7], Benjamin Djulbegovic, MD, PhD[8], Michael Marrone, MPH[9,10], Jennifer S. Lin, MD, MCR[11], Alfred O. Berg, MD, MPH[12] and Ned Calonge, MD, MPH[13]; on behalf of the EGAPP Working Group

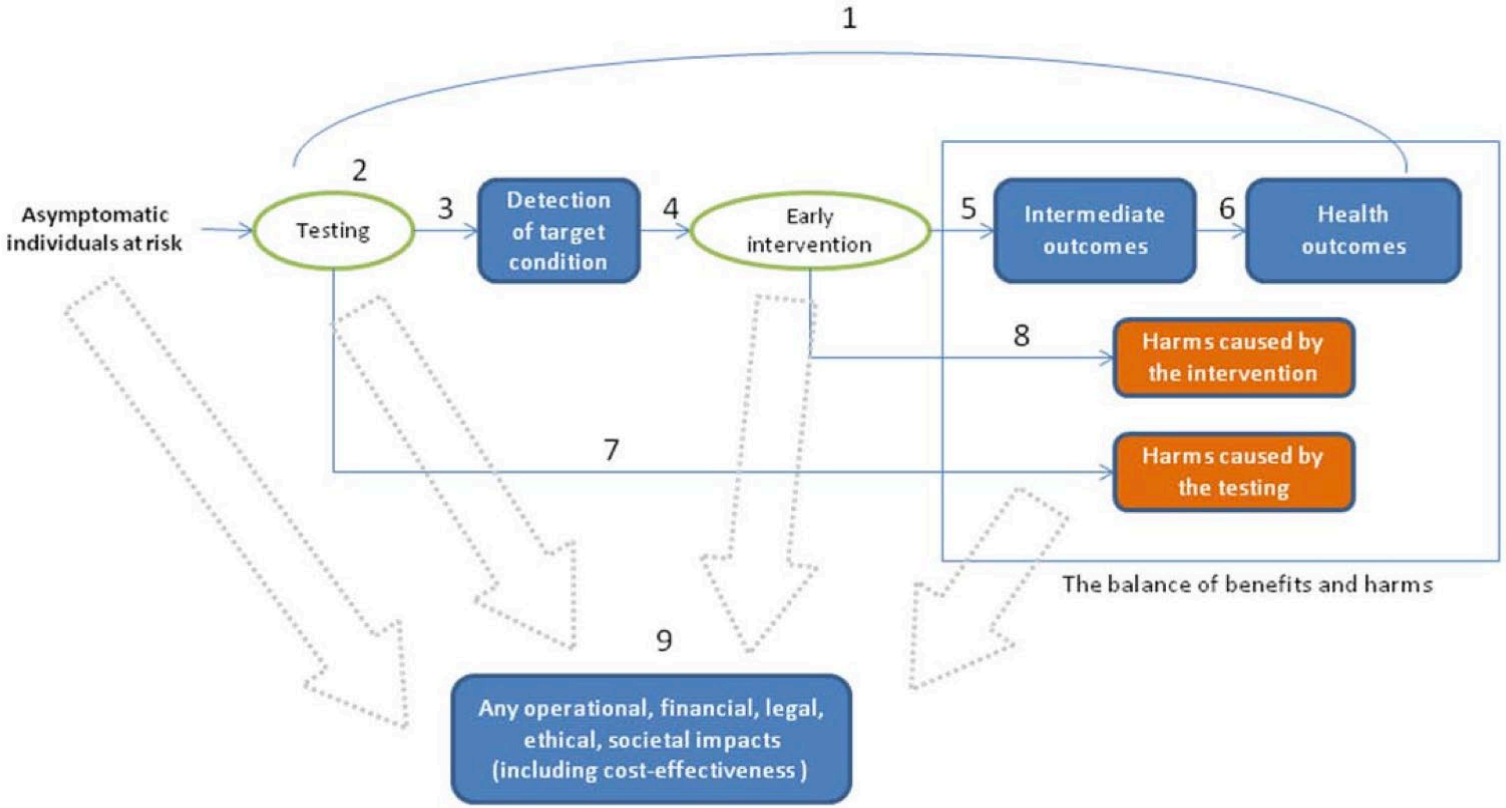
ANALYTIC FRAMEWORKS

Methods Research Report

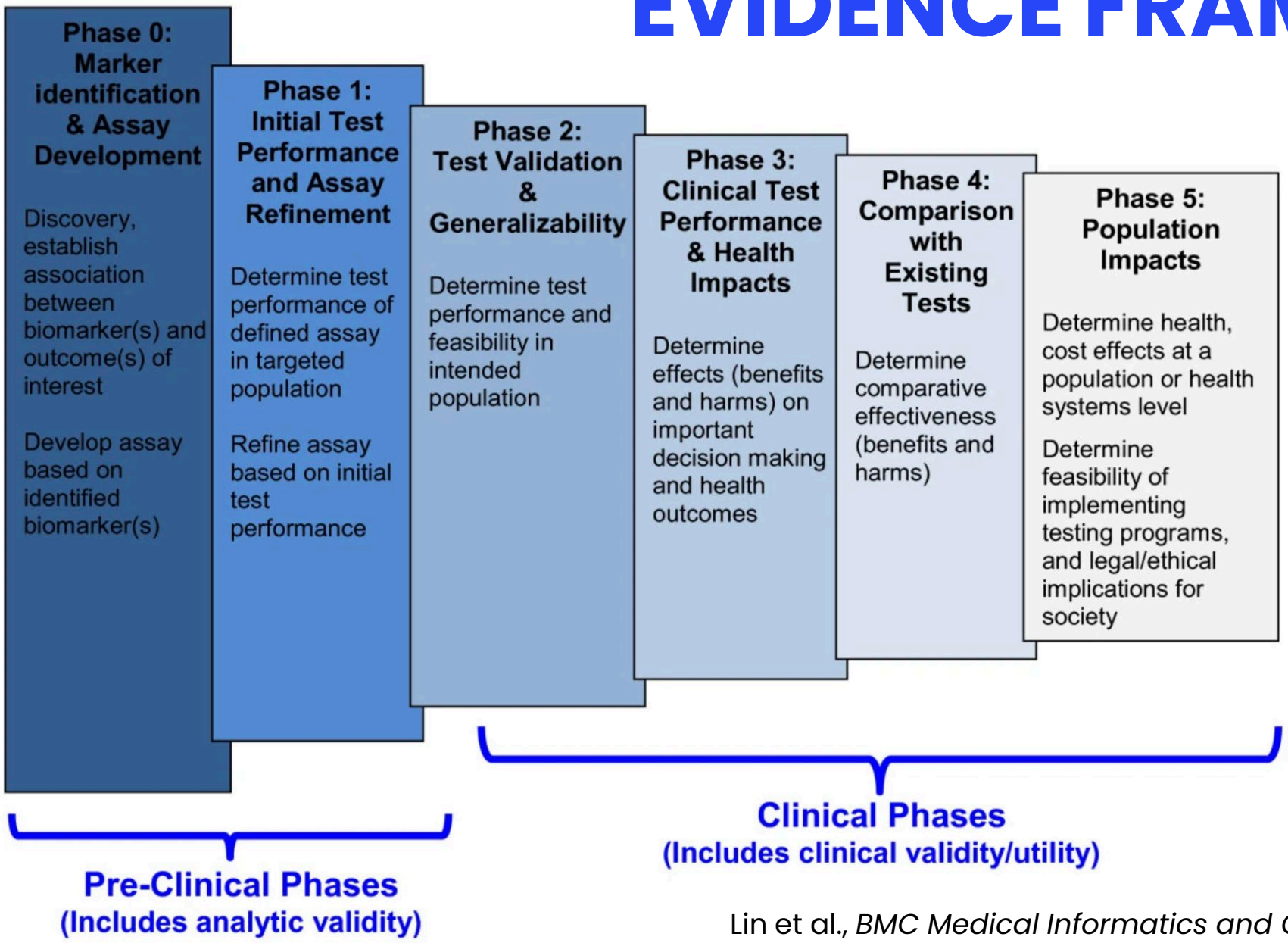
Addressing Challenges in Genetic Test Evaluation

Evaluation Frameworks and Assessment of Analytic Validity

Prepared for:
Agency for Healthcare Research and Quality
U.S. Department of Health and Human Services
540 Gaither Road
Rockville, MD 20850
www.ahrq.gov



EVIDENCE FRAMEWORKS

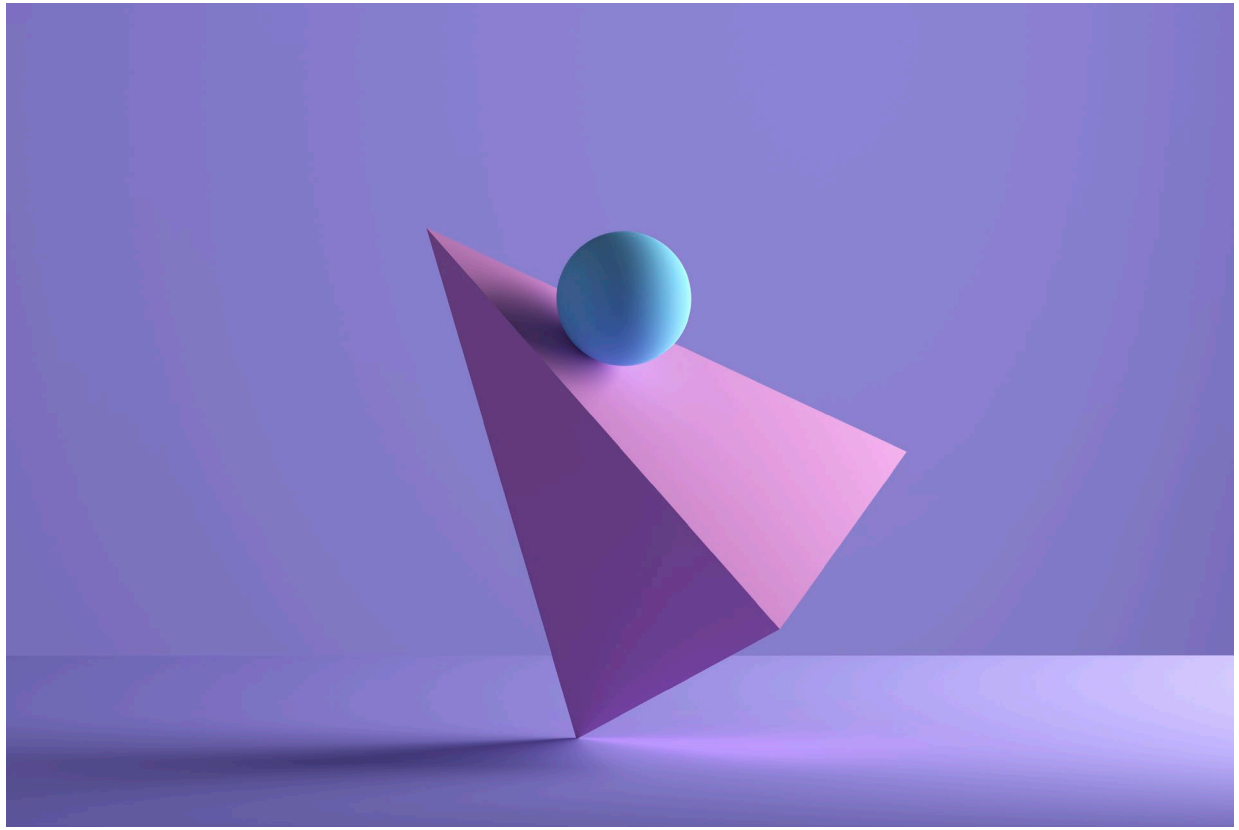


Lin et al., *BMC Medical Informatics and Clinical Decision Making*, 2012, 12:117

EVIDENCE CONSIDERATIONS

- Rapid development
  - Potential need to inform interest-holders with “less than ideal” evidence
    - Insufficient evidence due to:
      - Single studies
      - Small samples, etc.

EVIDENCE CONSIDERATIONS



Study Designs: “lower level”

- Observational, diagnostic accuracy
- Case series, case studies
- Grey literature

INFORMATION SOURCES

- Published literature
- Grey literature
  - Preprints
  - Media, newsletters
  - FDA 510k, device submissions
  - Other federal agencies
  - International sources

INFORMATION SOURCES

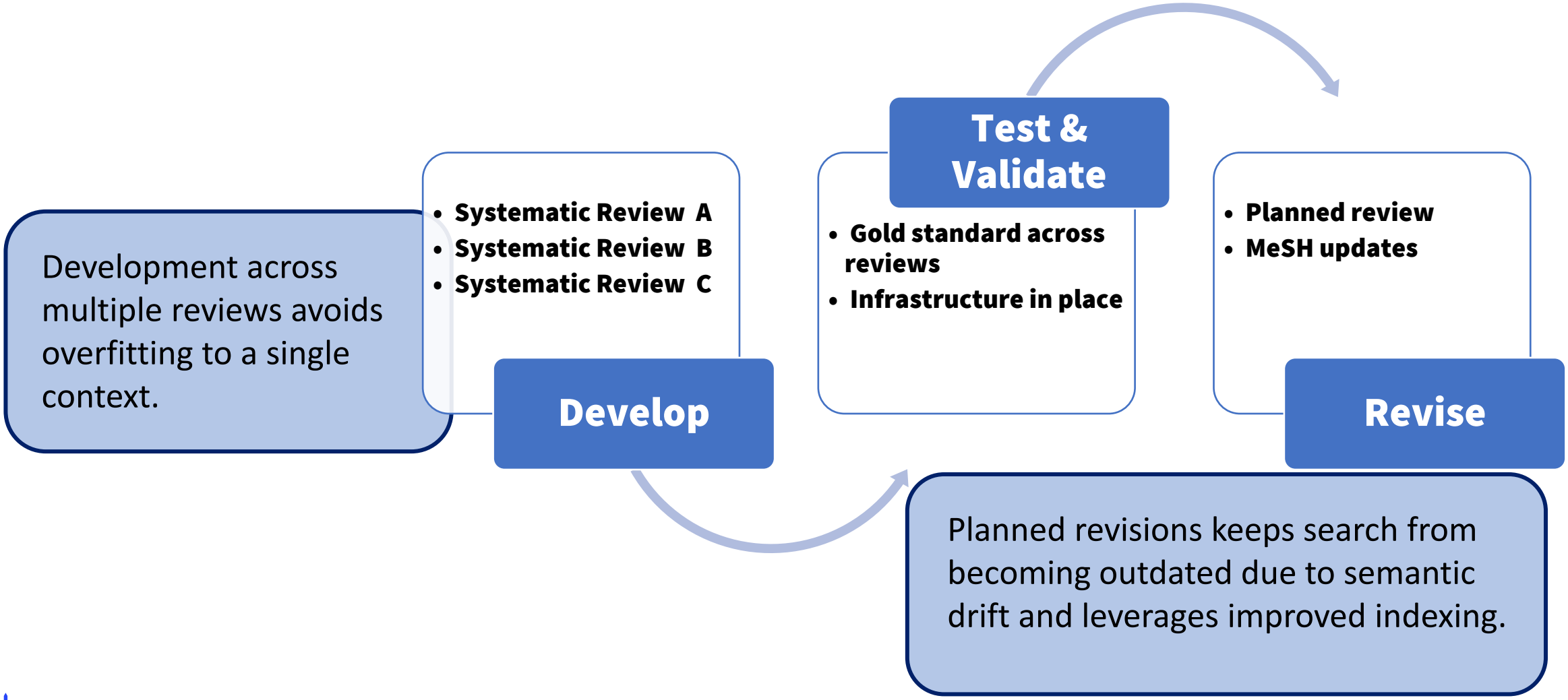
- Test developers, laboratories
  - Scientific Information Packets

- ClinicalTrials.gov
- Professional societies
- Interest holder review and feedback

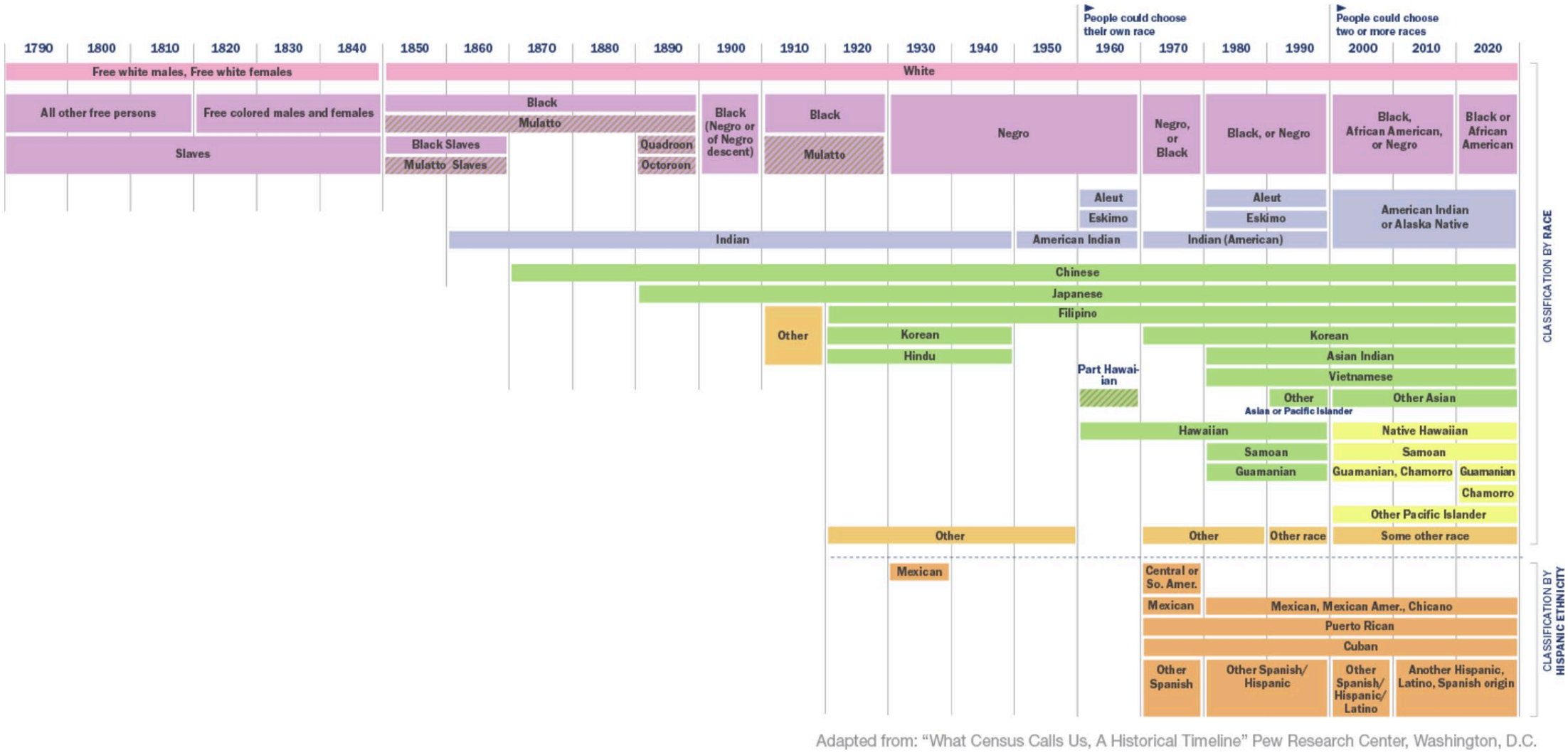
KEEPING IT CURRENT: LIVING REVIEWS

- Increasingly common
- Enables the currency of conclusions
  - Multiple data/evidence sources
  - Iterative process provides flexibility
  - Continual or regular surveillance
- Equity and implementation considerations

KEEPING IT CURRENT: HEALTH EQUITY SEARCH FILTER



KEEPING IT CURRENT: POPULATION DESCRIPTORS



Adapted from: "What Census Calls Us, A Historical Timeline" Pew Research Center, Washington, D.C.

Racial categories changed over time due to shifts in scientific, political and social thinking about race and ethnicity.

Source: Human Genome Research Institute/Use of Population Descriptors in Genomics

KEY POINTS

- Established frameworks exist: rapidly evolving field
- Use published & other evidence sources (FDA, SIP CT.gov, interest holders)
- Living reviews: continuous surveillance keeps conclusions up to date
- Importance of equity & implementation, & social, ethical, & legal implications

THANK YOU! For questions or further discussion: karli.kondo@cancer.org