Patient-centered research: from consent to outcomes

Steven Joffe, MD, MPH
Emanuel and Robert Hart Associate Professor
Department of Medical Ethics & Health Policy

CSER and Beyond
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Two paths to responsibly integrating sequencing into clinical practice

- Traditional approach
  - Demonstrate (narrowly defined) utility
  - Later, evaluate impact on psychosocial, economic and health-system outcomes
- CSER's approach
  - Define utility broadly
  - Integrate psychosocial, economic and health system outcomes into evaluation from the outset

What has CSER taught us about ELSI (so far)?

(With apologies to the many investigators whose work I don't mention)

Informed consent to sequencing

- Heterogeneity and best practices in consent forms (Henderson *J Law Med Ethics* 2014)
- Conceptual models of consent to return of incidental findings (Appelbaum *Hastings Cent Rep* 2014)
- Lessons from genetic counselors regarding consent to genomic sequencing (Bernhardt *Am J Med Genet A* 2015)

Patients' preferences for incidental & secondary results

- Most patients prefer to learn sequencing results, driven by interest in prevention and general desire to know (Facio *EJHG* 2013)

- Most families of children with cancer agree to tumor + germline sequencing; most opt to learn child's carrier status (Scollon *Genome Med* 2014)

Clinician and system challenges

- Genetic professionals' beliefs about return of incidental findings vary by patient population, age of onset and actionability (Yu *Am J Hum Genet* 2014)
- Primary care physicians, cardiologists and medical oncologists report being unprepared for sequencing (Christensen *Clin Genet* 2015; Gray *ASHG* 2014)

Electronic health record integration

- Little consistency to how genomic data are entered into EHR or where they are found; PDF-type documents most common (Tarczy-Hornoch *GIM* 2013)
- Acute need for clinical decision support integrated into EHR (Shirts *JAMIA* 2015)
- Alert systems based on pharmacogenomic results in the EHR are possible but resource-intensive, esp given evolving knowledge base (Nishimura *GIM* 2015)

Defining and measuring outcomes

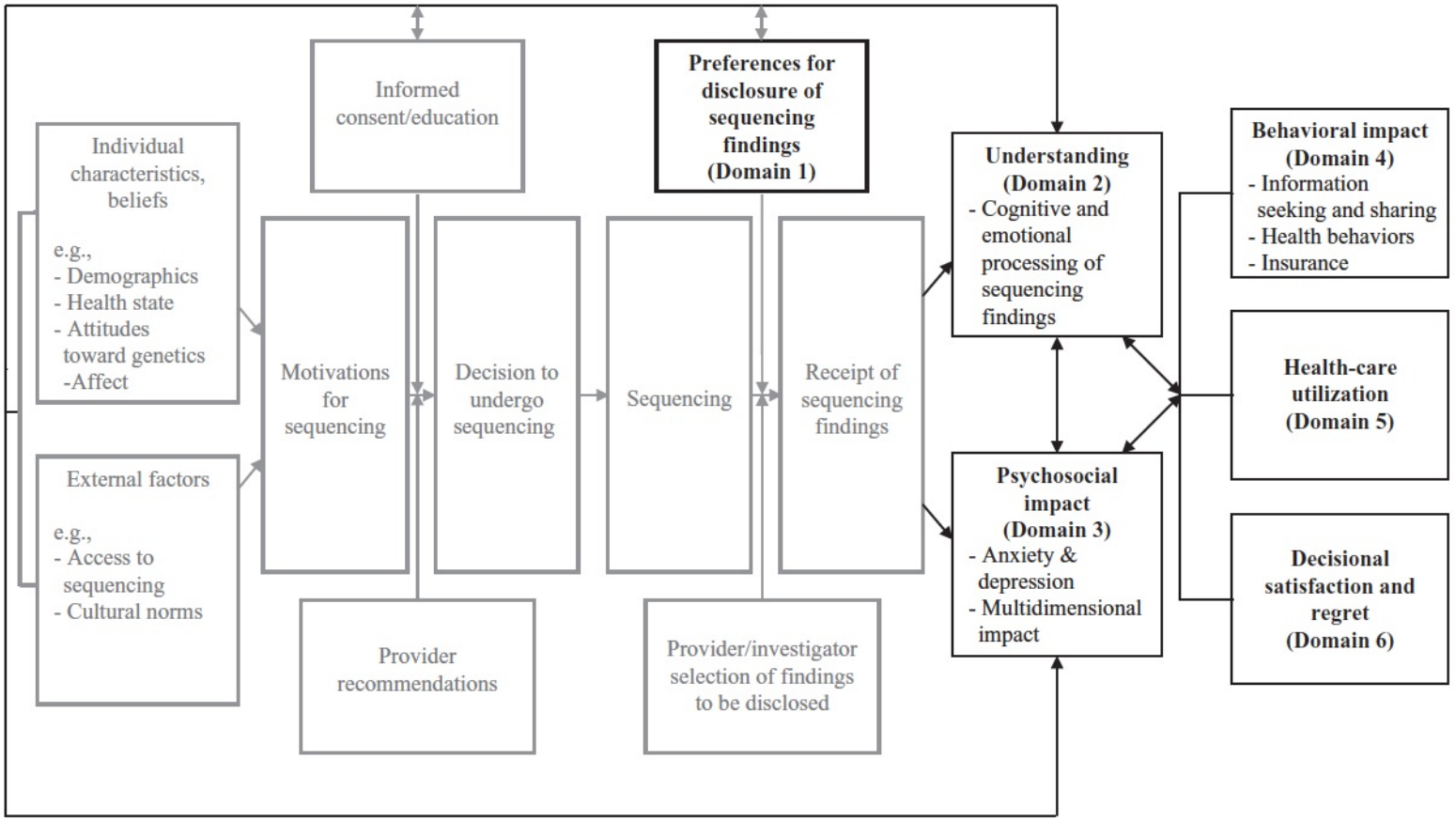
REVIEW | Genetics inMedicine

Social and behavioral research in genomic sequencing: approaches from the Clinical Sequencing Exploratory Research Consortium Outcomes and Measures Working Group

Stacy W. Gray, MD, AM[1–3], Yolanda Martins, PhD[1], Lindsay Z. Feuerman, BA[4], Barbara A. Bernhardt, MS, CGC [5,6], Barbara B. Biesecker, PhD, MS[7], Kurt D. Christensen, MPH, PhD[2,3], Steven Joffe, MD, MPH[8], Christine Rini, PhD[9,10], David Veenstra, Pharm. D, PhD[11], Amy L. McGuire, JD, PhD[4] and members of the CSER Consortium Outcomes and Measures Working Group

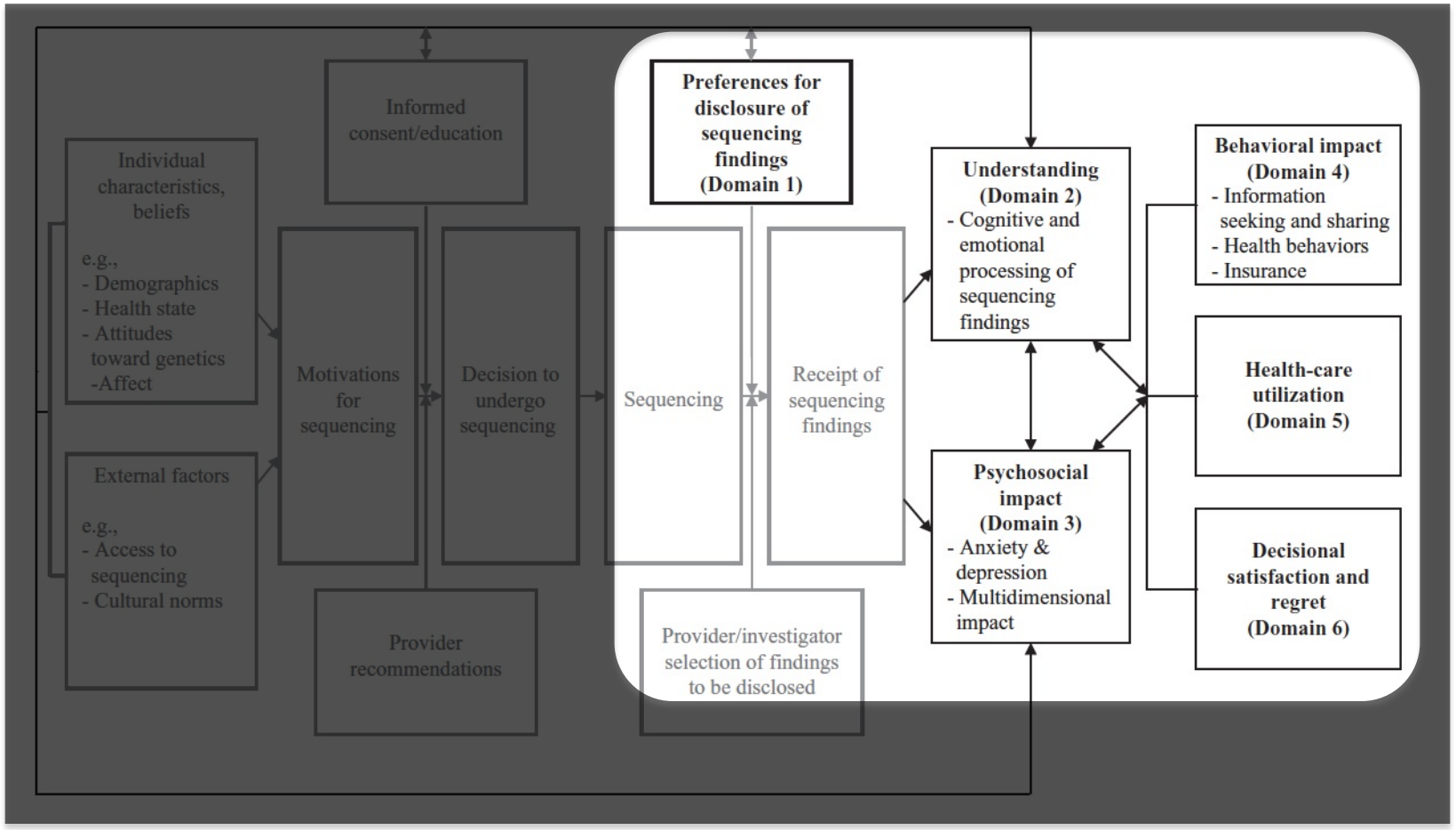
GIM 16:727, 2014

Defining and measuring outcomes



GIM 16:727, 2014

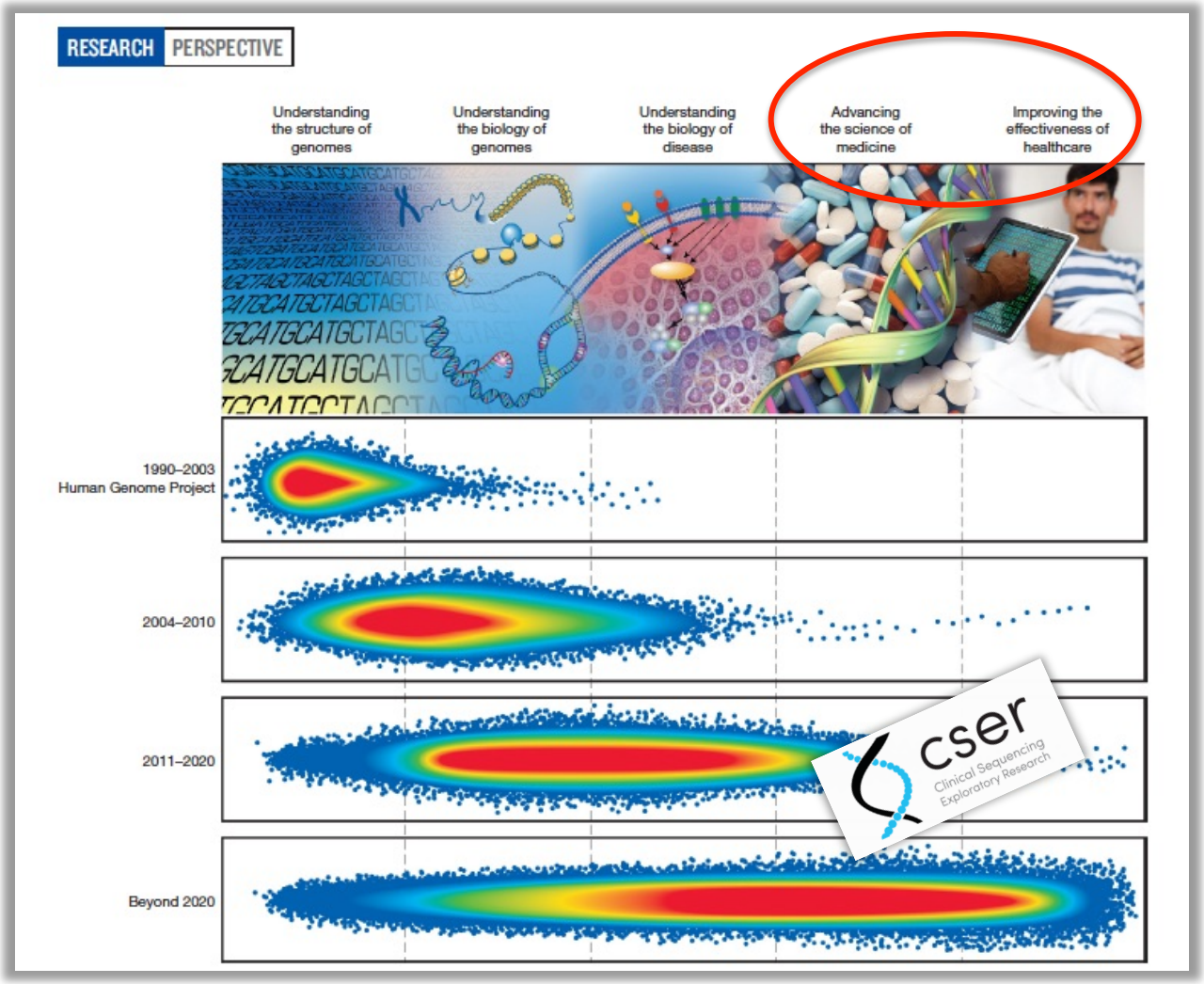
Defining and measuring outcomes



GIM 16:727, 2014

What do we still need to learn?

- Impact of return on patients and families
  - Defining patient- and family-centered measures of value of information
- Economic and health-utilization outcomes
- Extending sequencing to, & understanding outcomes in, community settings and diverse populations
- Better measures of key outcomes, with longitudinal followup



Nature 470:204, 2011

Insights from CSER will help move genomics from base pairs to bedside by…

- Advancing the science of medicine
  - Delivering genomic information to patients (and clinicians)
  - Addressing impact of genomics on health disparities
- Improving the effectiveness of healthcare
  - Integrating genomics into electronic health records
  - Demonstrating effectiveness
    - Defining metrics
    - Shaping & answering regulatory questions
  - Educating professionals, patients & the public
  - Increasing access to genomic medicine

Nature 470:204, 2011