

Personal Genomics Workshop

Panel Discussion

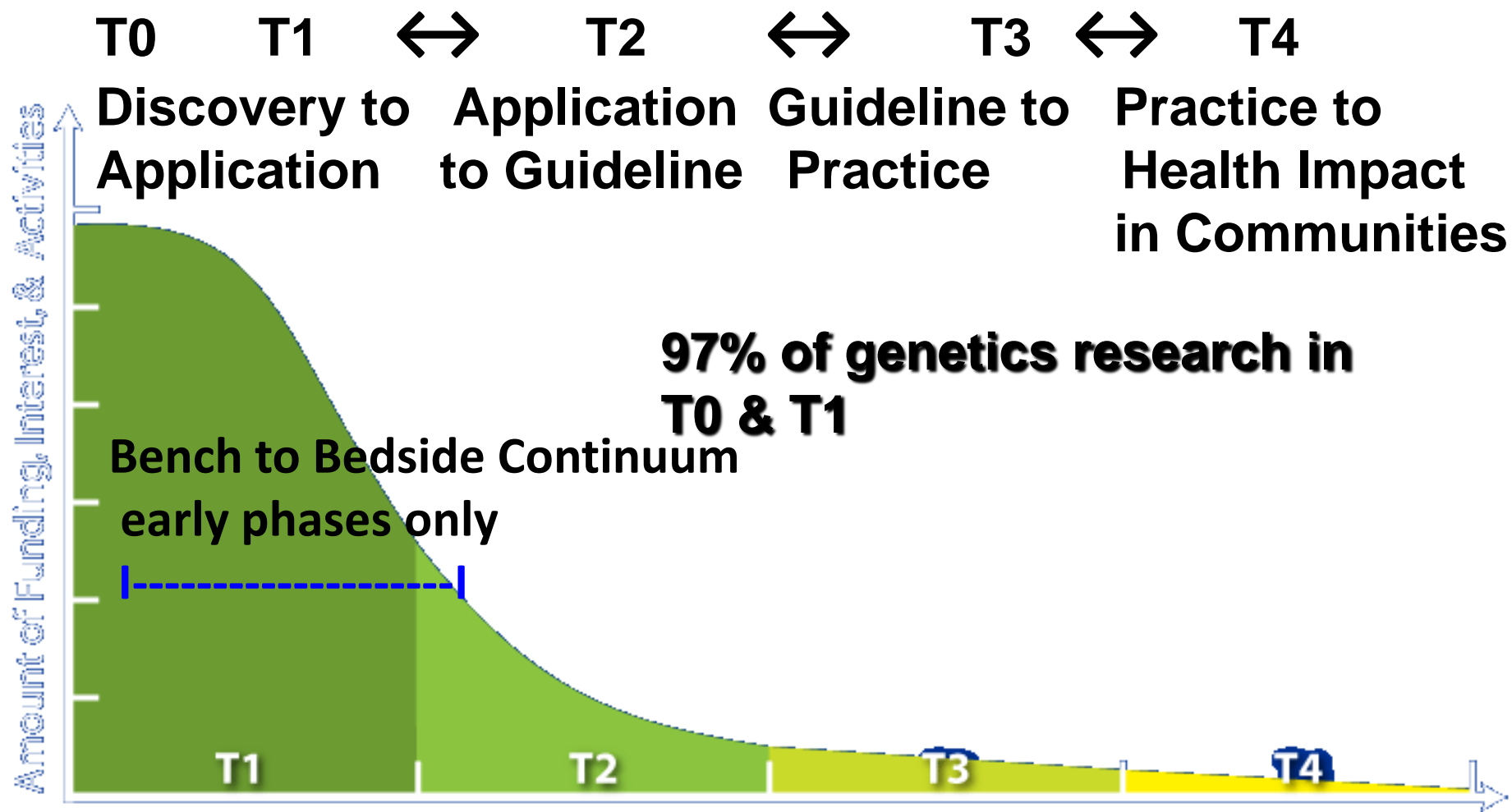
So What Should We Expect from this Meeting?

- Lots of Presentations, Discussions and Hopefully Lively Debate
- Structure
 - I. The Basics
 - II. Credibility of Genetic Associations
 - III, IV. Science of Clinical Validity and Utility
 - V. Case Studies
 - VI. Models for Translation Research
 - VII. Panel Discussion and Next Steps
- Products: A Multidisciplinary Research Agenda and a Scientific Report for Peer Reviewed Publication

Issues to consider

- What gene variants should be included?
- What research studies needed to establish clinical validity and clinical utility?
- Is there a public-private partnership model to conduct such research?
- What information and decision support tools are needed for consumers and providers are needed while research is done?
- What are the policy and oversight implications?

Limited Research on Either Public Health Challenge: Complete Evaluation, Implementation



Khoury Genet Med 2007;9:665; IOM The Future of the Public's Health in the 21st Century, 2003; Woolf JAMA 2008;299:211; IOM Clinical Research Roundtable- Sung JAMA 2003

Criteria for evaluation (short list)

0. Genetic associations → Janssens et al. *AJHG* 2008
1. Clinical validity : (is it a worthy test?)
 - Calibration and validation
 - Risk distribution / discriminative accuracy
2. Clinical utility : (is it worth testing?)
 - Benefits available (intervention or knowledge)
 - Change (clinical) decision → e.g risk difference and reclassification
3. Cost-effectiveness, etc
4. Feasibility, lab quality, counseling, etc.

Building the Infrastructure to Make this Work

- Biobanking
 - Coordinated efforts
 - Operational and informatics support
 - Standards
- Genomic Technologies
 - Core laboratories
 - Economies of scale
- Informatics
 - Reliable, interoperable EHRs
 - Integration of research, clinical, molecular data
- Biostatistics
 - Critical shortage must be addressed
 - Physician training in quantitative skills
- Decision Making
 - Understanding of human decision making
 - Biological, psychological and social factors
 - Education of health care professionals

Multidisciplinary Translation Sciences of Personal Genomics: Beyond Replication and Biology: Clinical Validity and Clinical Utility?

- Epidemiology
- Communication
- Behavioral/social
- Economics
- Health services research
- Outcomes research and surveillance
- Policy analysis
- Evidence guidelines
- ELSI..

RCTs?

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Stakeholder Driven Collaboration in Genomics Translation

