

Future Opportunities for Genome Sequencing and Beyond

A Planning Workshop for the National Human Genome Research Institute

July 28 and 29, 2014



National Human Genome
Research Institute

- 1. Welcome**
- 2. Agenda Rationale**
- 3. Agenda Walk-Through**
- 4. Acknowledgements**
- 5. Advice about Advice**
- 6. Logistics**

Rationale: Why now?

Genomic Science is Changing

- Technology
- Availability of “infrastructure” including data and analysis tools
- Community
- Clinical applications

Different era

- Federal budgets
- “Central planning” vs “Investigator initiated”

Rationale: How?

- **Involve the Community**
- **Acknowledge our history, but not be bound by it**
- **Agenda should not be about the existing program**
- **Agenda should include topics that are proximate to sequencing and could raise new scientific opportunities for exploring genomic problems at scale**

Rationale: What?

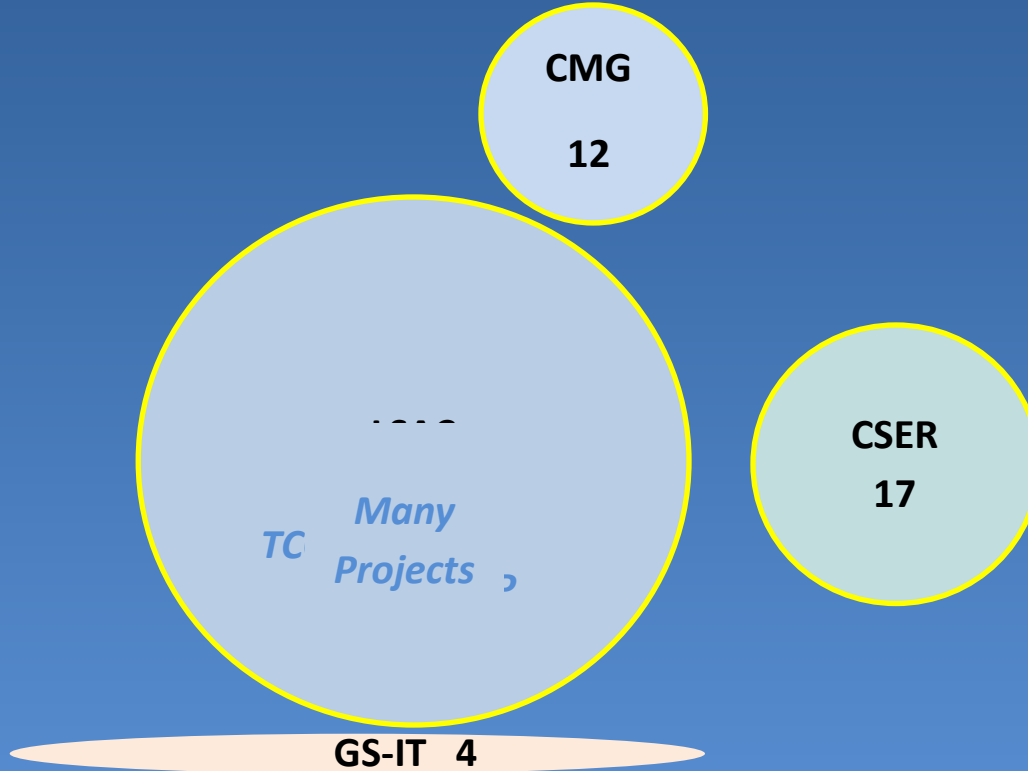
Structure of
Genomes

Biology of
Genomes

Biology of
Disease

Science of
Medicine

Effectiveness of
Healthcare



Rationale: What?

Structure of
Genomes

Biology of
Genomes

Biology of
Disease

Science of
Medicine

Effectiveness of
Healthcare

ENCODE
25

GGR 10

FunVar
5

CMG
12

Other
"science of
medicine"
with seq
30-40 next
year?

Tech Dev
15

LSAC
77

CSER
17

ClinGen 4

GS-IT 4

Other Seq Inf (non-
db) 24

Rationale: What?

Structure of
Genomes

Biology of
Genomes

Biology of
Disease

Science of
Medicine

Effectiveness of
Healthcare

**Genome
Function**

**(Related
Tech Dev)**

**Disease Gene
and Variant
Discovery**

**Clinical
Applications
of Sequencing**

**(Related
Informatics)**

Another “What”

What can (or should) be done “at scale”?

- **Explicit part of workshop: opportunities that can only (or best) be addressed at scale.**

Scaled efforts have two kinds of deliverables:

- **Answers to the scientific questions that require scale to address**
- **High-quality comprehensive resources; developed technologies, approaches, project designs, analysis methods, policies, file formats, etc.**

Agenda Walk-Through

Science

1. Talks: State of the science; Important questions
2. Breakouts: Four* topic discussions
3. “Challenge talks”: some examples of large scale projects that are not addressed in the current way NHGRI is thinking
4. Breakout reports

Implementation

5. “Program Elements”

Acknowledgements

National Advisory Council for Human Genome Research

Scientific Advisors to the Genome Sequencing Program

Workshop Agenda Committee: Ewan Birney, Eric Boerwinkle, Carlos Bustamante, Joe Ecker, Jim Evans, Bill Gelbart, Len Pennacchio

NHGRI Leadership: Larry Brody, Bettie Graham, Eric Green, Teri Manolio, Jeff Schloss, Mark Guyer*, Jane Peterson*

Shannon Biello, Deborah Colantuoni, Lucia Hindorff, Carolyn Hutter, Lu Wang, Elise Feingold, Mike Pazin, Mike Smith, Heidi Sofia, Chris Wellington, Kris Wetterstrand

About Advice



Logistics

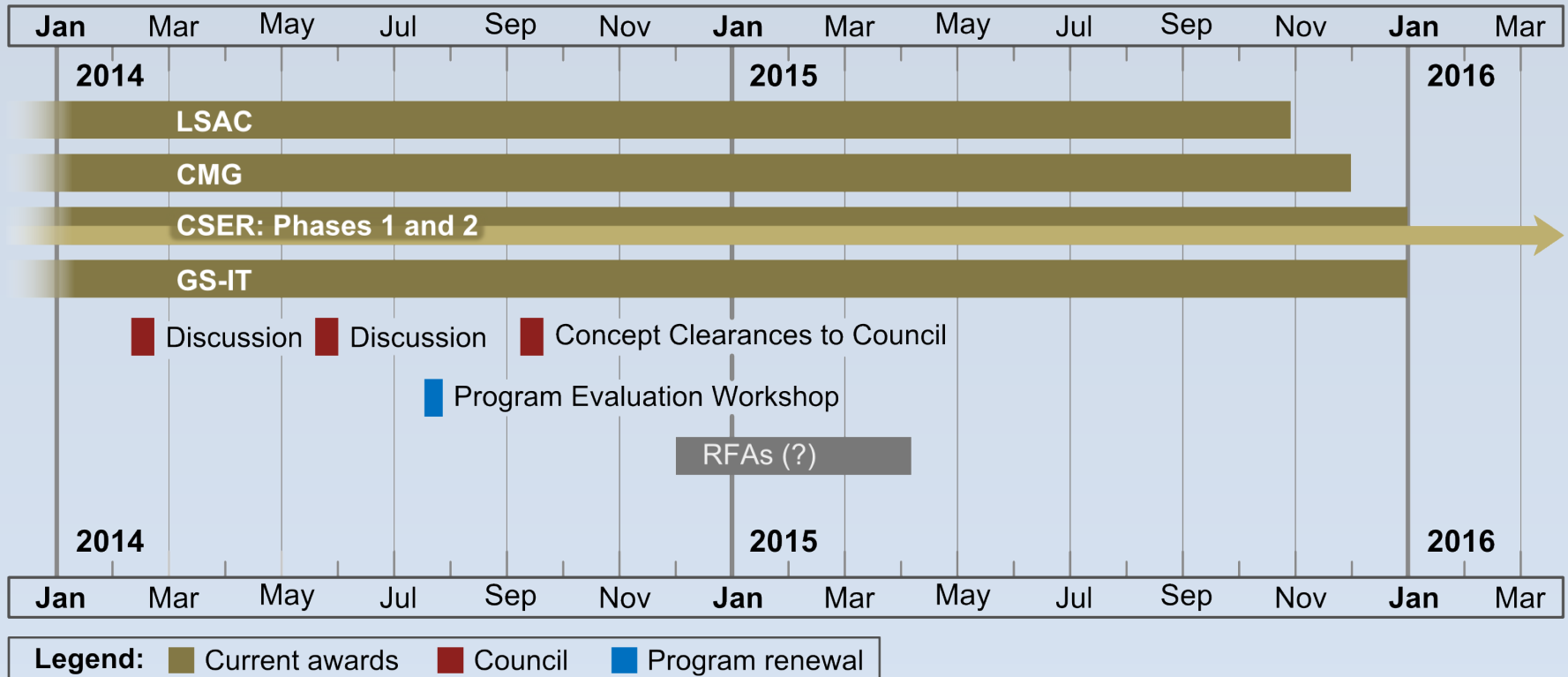
- Registration desk/Taxis/Re-imbusement vouchers
- Presentations
- Name badges/breakouts
- Food
- Hotel check out
- Free Wi-Fi
- Do not leave personal items/electronic devices in meeting room
- Workshop is being webcast (#GSPfuture)

END

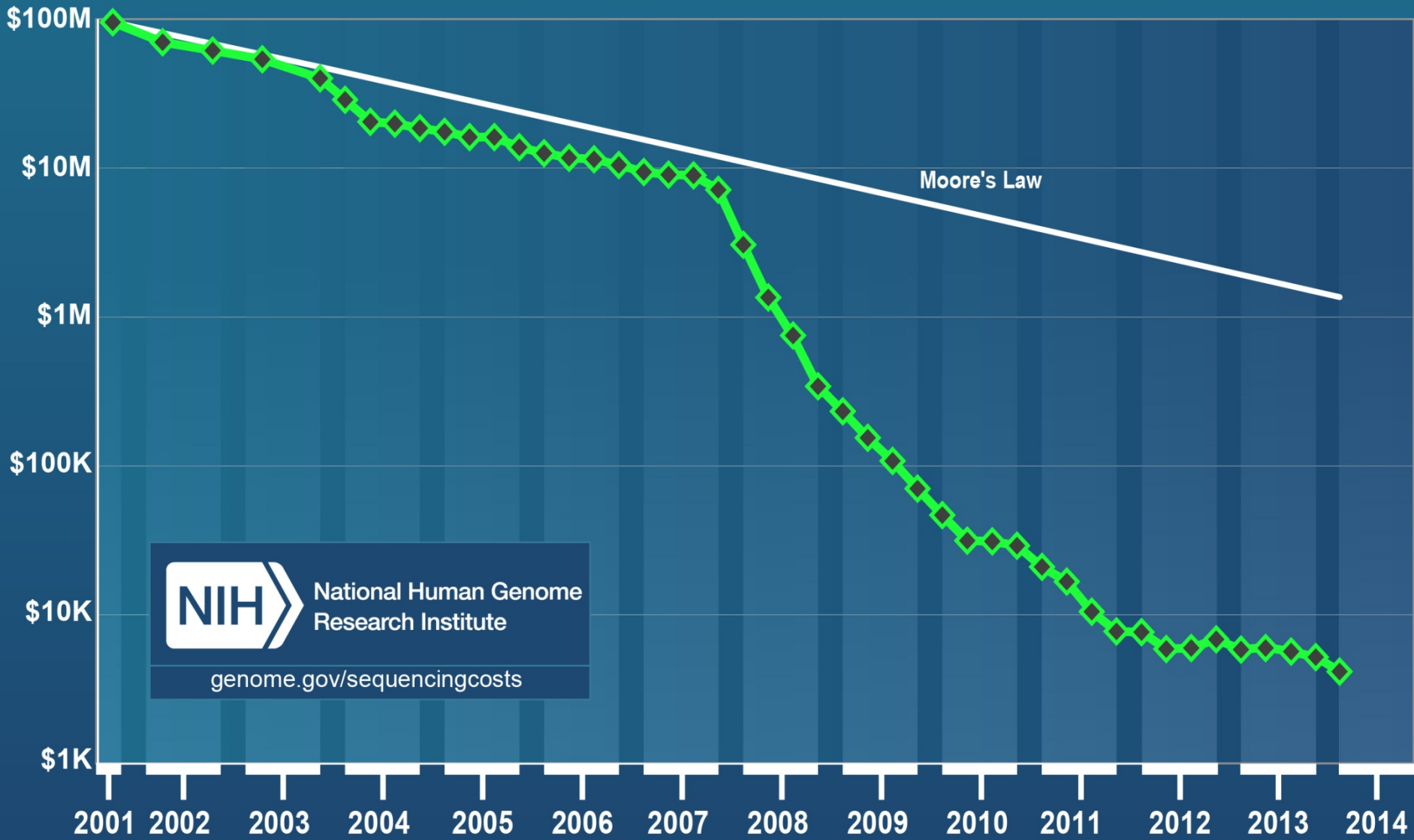
Morning Speakers

- **Michael Boehnke—University of Michigan**
- **Roderick McInnes—Lady Davis Institute for Medical Research**
- **Dan Roden—Vanderbilt University**
- **Joseph Ecker—HHMI and Salk Institute for Biological Studies**

Timeline



Cost per Genome



NIH National Human Genome Research Institute
genome.gov/sequencingcosts

Total Production (Gb) Y11Q2

