Concept Clearance

Center for Inherited Disease Research

Lawrence Brody, Ph.D.

National Advisory Council for Human Genome Research
September 19, 2022
1) Should NIH ICs support an integrated program to review and *carry out* large scale genomic research projects for investigators?

2) Should this facility retain flexibility with regard to what genomic services and in what scope are offered?

3) Should the program be structured so that it may be closed if its services are no longer required?
Center for Inherited Disease Research

• Founded in 1996

• Speed the discovery of genes that contribute to health and disease

• Supported by 10 NIH Institutes

CIDR – Muti-Institute Program

Shared

- Expertise
- Governance
- Resources
- Research projects
- Economies of scale
Shared Effort  Shared Benefits
Center for Inherited Disease Research

Governance - Review - Management

- CIDR Staff Meetings – every other week
- CIDR Access Committee – 6 times / year
- Board of Governors – 6 times / year

- NHGRI Budget Office
- NHLBI Contracting Office
CIDR Services

- Study Design Consultation

- SNP Genotyping
  - Human Genome Wide Association Studies
  - Custom Genotyping
  - Focused Content Panels
    - Methylation
    - Linkage
    - Consortium Arrays

- DNA Sequencing
  - Targeted
  - Exome
  - Whole genome
  - Low pass sequencing

- Statistical Analyses

- Data Wrangling
Is CIDR Relevant Today?

Commercial service suppliers

Academic service suppliers

Local academic based cores

Offshore service suppliers
CIDR vs Commodity Suppliers

CIDR Access Committee
CIDR ACCESS COMMITTEE (CAC)

Kathryn Lunetta, Ph.D, Chair  Boston University School of Public Health

John Belmont, MD, Ph.D.
  Baylor College of Medicine
Anne Bowcock, Ph.D.
  Mount Sinai School of Medicine
Karen Conneely, Ph.D.
  Emory University School of Medicine
Dana Crawford, Ph.D.
  Case Western University
Michael Hauser, Ph.D.
  Duke University Medical Center

Susan Neuhausen, Ph.D.
  City of Hope
Stephanie Santorico, Ph.D
  University of Colorado
Jinchuan Xing, Ph.D.
  Rutgers University
Sebastian Zöllner, Ph.D.
  University of Michigan
CIDR vs Commodity Suppliers

CIDR Access Committee

Data Quality
Study design
Plate map design
Sample pre-check

Sex checks
Call rate
Pedigree – known - hidden
Contamination/mixture flag

Study QC
Batch effects

Merge molecular and phenotype data
First pass association tests

Direct deposit to dbGAP for sharing
“Influenced” by CIDR
Completed CIDR Projects - 1997-2022

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<th>Projects</th>
<th>Samples</th>
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Projects 505
Samples 1,228,457
CIDR Publications

1,984 publications  133 dbGaP postings

Investigators - 173
Institutions - 93
Phenotypes - 122
Journals - 504
CIDR Program Renewal
Proposed Mechanism and Funds

Mechanism
• Investigators: X01 Application
• Research and Development Contract
  - ID IQ
  - 5-7 year term

Funding and Admin
• ~ Cap < $100M over 5-7 years depending on volume
• NHGRI support – in kind

Working Model - Open competition
NIH Concept /Operations Team

NHGRI
Jon Garvey
Mike Lacy
Marcia Morris
Barbara Thomas
(Rudy Pozzatti &
Ann Fitzpatrick)

NHLBI
Lynn Furtaw
Stacy Turner

CIDR Board of Governors
Concept Questions

1) Should NIH ICs support an integrated program to review and *carry out* large scale genomic research projects for investigators?

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The Future

Array Based

Samples per year

Sequenced Based

Samples per year