

Concept Clearance

**Center for Inherited Disease
Research**

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**National Advisory Council for Human Genome Research
September 19, 2022**

Concept Questions

- 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

***NHGRI – NCI – NEI – NIA – NIAAA – NICHD –
NIDCR – NIEHS – NINDS – OD***

CIDR – Multi-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?

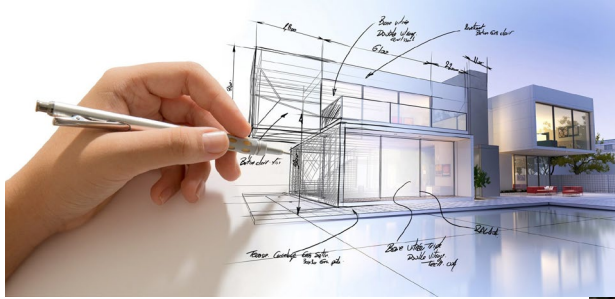
Commercial service suppliers

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

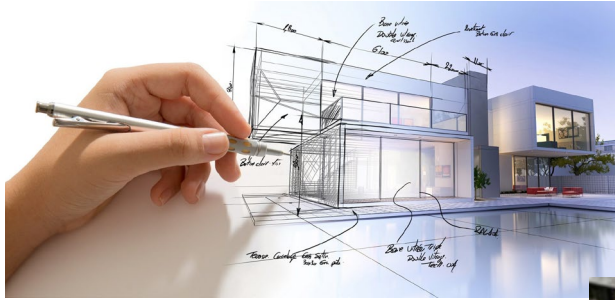
Jinchuan Xing, Ph.D.

Rutgers University

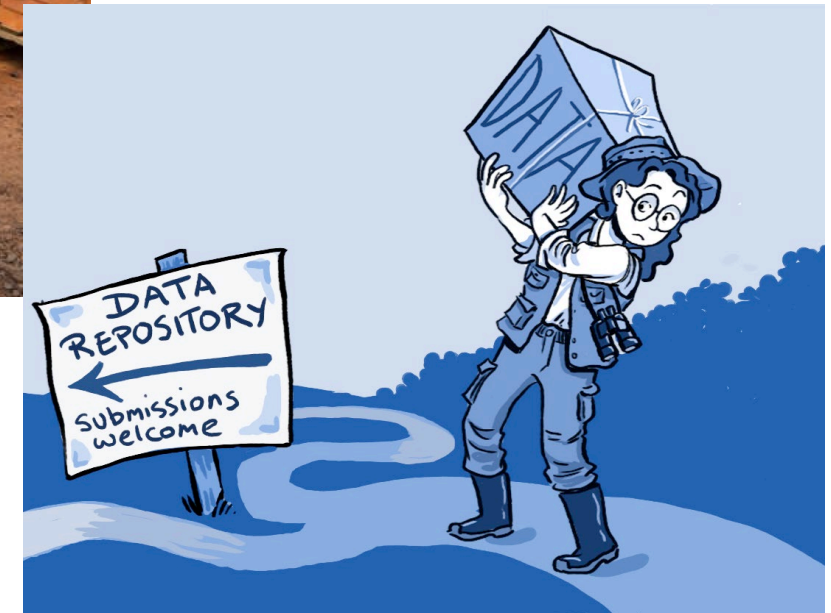
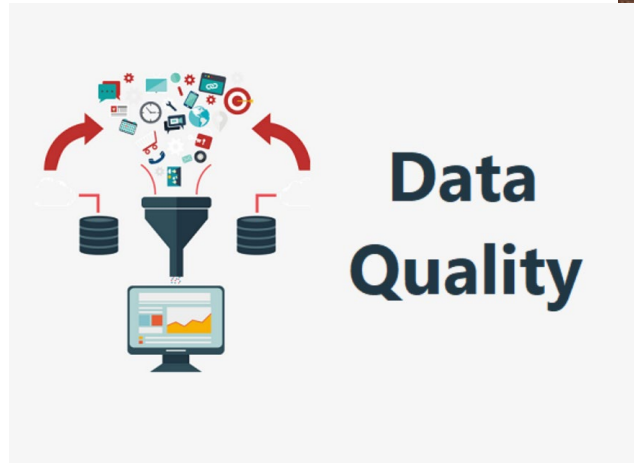
Sebastian Zöellner, Ph.D.

University of Michigan

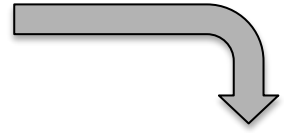
CIDR vs Commodity Suppliers



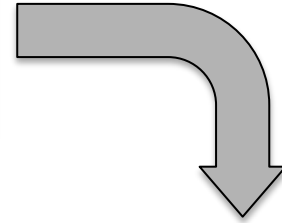
CIDR Access Committee



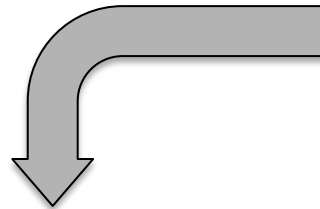
Study design
Plate map design
Sample pre-check



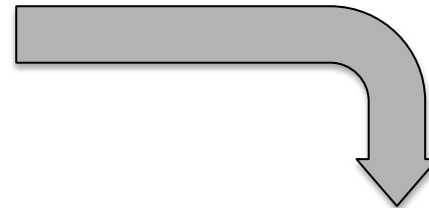
Sex checks
Call rate
Pedigree – known - hidden
Contamination/mixture flag



Study QC
Batch effects

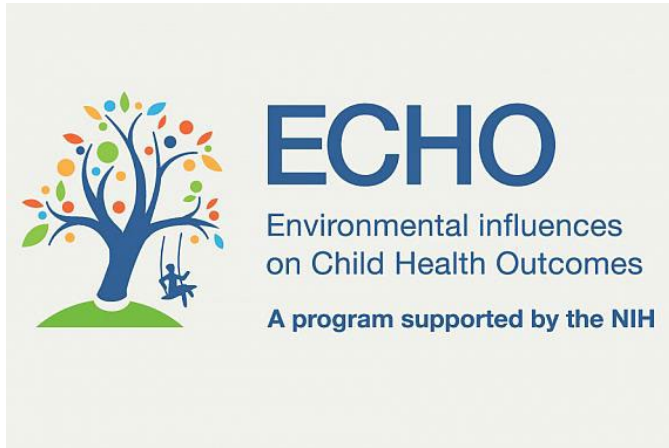


Merge molecular and phenotype data
First pass association tests



Direct deposit to dbGAP for sharing

“Influenced” by CIDR



Completed CIDR Projects - 1997-2022

	Projects	Samples
• NCI	87	470,738
• NEI	47	117,349
• NHGRI	31	107,868
• NIA	18	58,524
• NIAA	6	15,079
• NICHD	17	17,661
• NIDA	18	42,705
• NIDCD	39	10,886
• NIDCR	37	109,237
• NIDDK	59	105,121
• NIEHS	18	65,968
• NIMH	50	68,952
• NINDS	44	32,636
• NIAMS	2	5,501
• OD	1	5,744



Projects
505

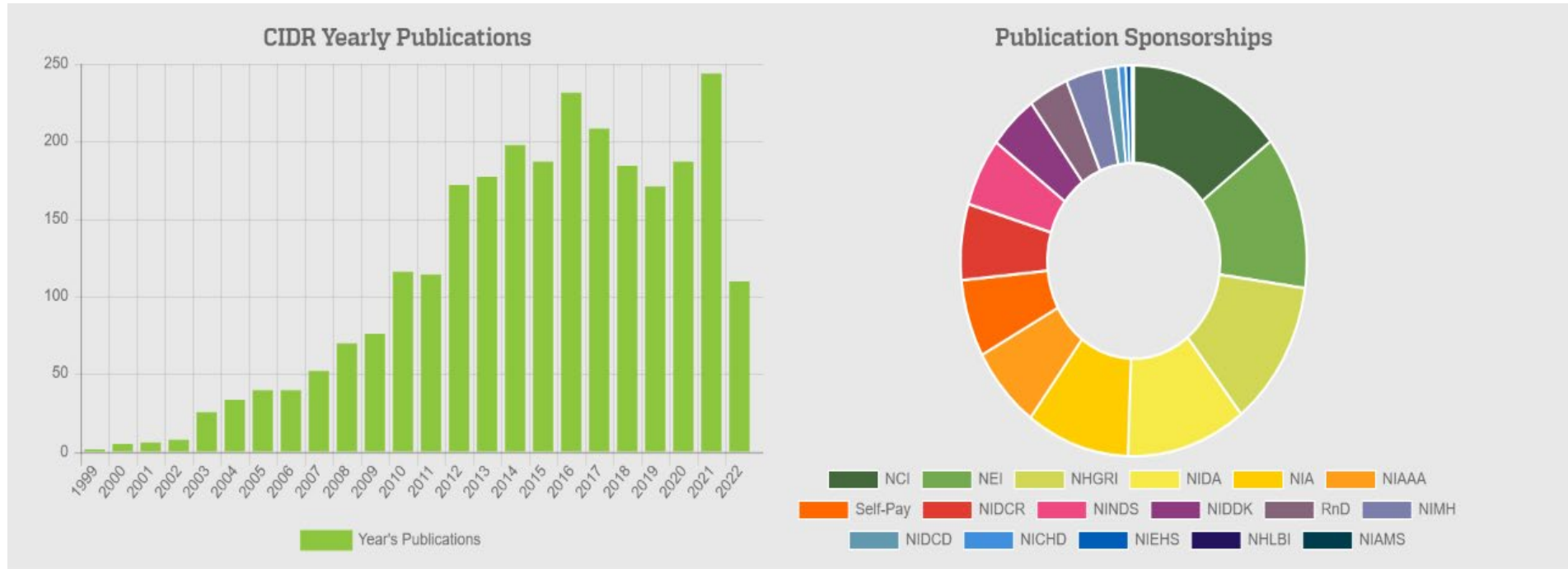


Samples
1,228,457

CIDR Publications

1,984 publications

133 dbGaP postings



Investigators - 173

Phenotypes - 122

Institutions - 93

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

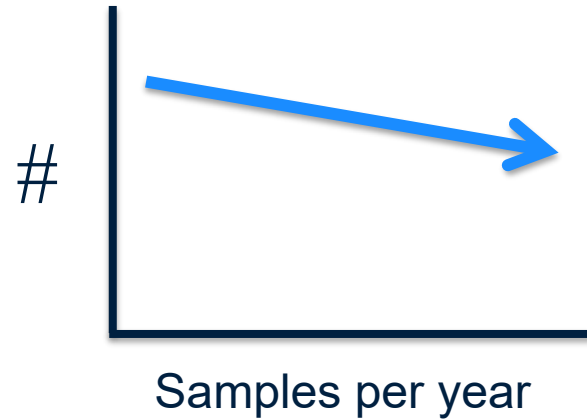
CIDR Board of Governors

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The Future

Array Based



Sequenced Based

