



National Human
Genome Research
Institute



National
Institutes of
Health



U.S. Department
of Health and
Human Services

LOGIN TO EMERGE

emerge network
ELECTRONIC MEDICAL RECORDS AND GENOMICS

451
Number of
network
publications

HOME ABOUT COLLABORATE PROJECTS TOOLS

<https://emerge.mc.vanderbilt.edu/>

eMERGE is a national network organized and funded by the National Human Genome Research Institute (NHGRI) that combines DNA biorepositories with electronic medical record (EMR) systems for large scale, high-throughput genetic research in support of implementing genomic medicine.

eMERGE and NHGRI's Genomic Medicine Portfolio

Teri Manolio, M.D., Ph.D.
eMERGE and Beyond Workshop
October 30, 2017

NHGRI's Division of Genomic Medicine

NHGRI defines genomic medicine as "*an emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the health outcomes and policy implications of that clinical use.*"

DGM plans, directs and facilitates multi-disciplinary research to identify genetic contributions to human health and to advance approaches for the use of genomic data to improve diagnosis, treatment, and prevention of disease.



NHGRI's Genomic Medicine Research Program

Program	Goal	Σ\$M	Years
UDN ¹	Diagnose rare and new diseases by expanding NIH's Undiagnosed Diseases Program	121	FY13-17
NSIGHT ²	Explore possible uses of genomic sequence information in the newborn period	25	FY13-17
CSER ³	Explore infrastructure, methods, and issues for integrating genomic sequence into clinical care	155	FY12-20
eMERGE ⁴	Use biorepositories with EMRs for genomics; assess penetrance of clinically relevant genes	135	FY07-18
IGNITE ³	Develop and disseminate methods for incorporating patients' genomic findings into their clinical care	28	FY13-17
ClinGen ⁴	Develop and disseminate consensus information on genes and variants relevant to clinical care	28	FY13-16
Investigator Initiated	Clinical sequencing research, HIV/AIDS drug response and co-morbidities, serious ADRs	~4	FY15-

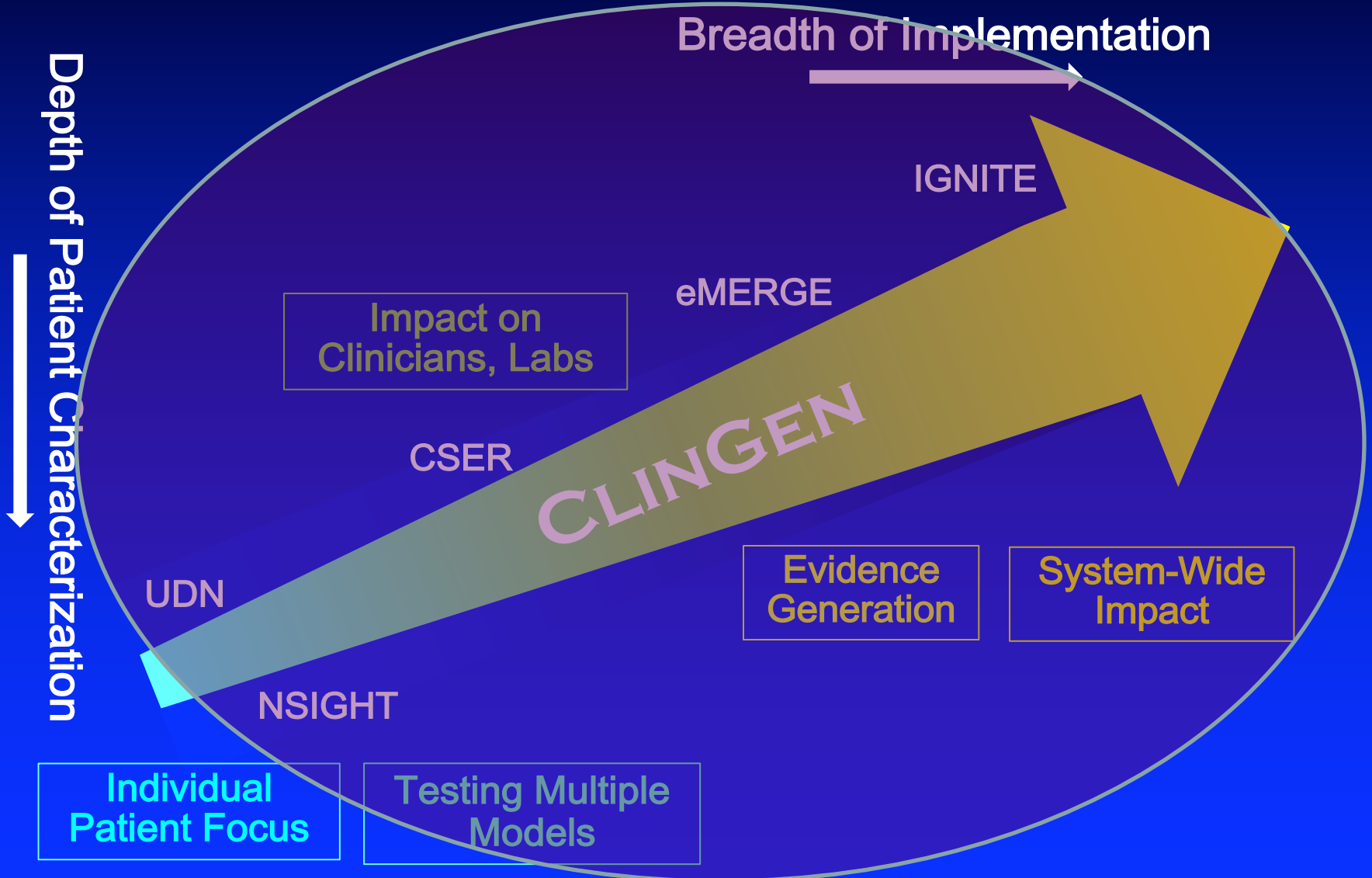
¹NIH Common Fund; ²Co-Funded by NICHD; ³Co-Funded by NCI; ⁴Co-Funded by OD.

NHGRI's Genomic Medicine Research Program

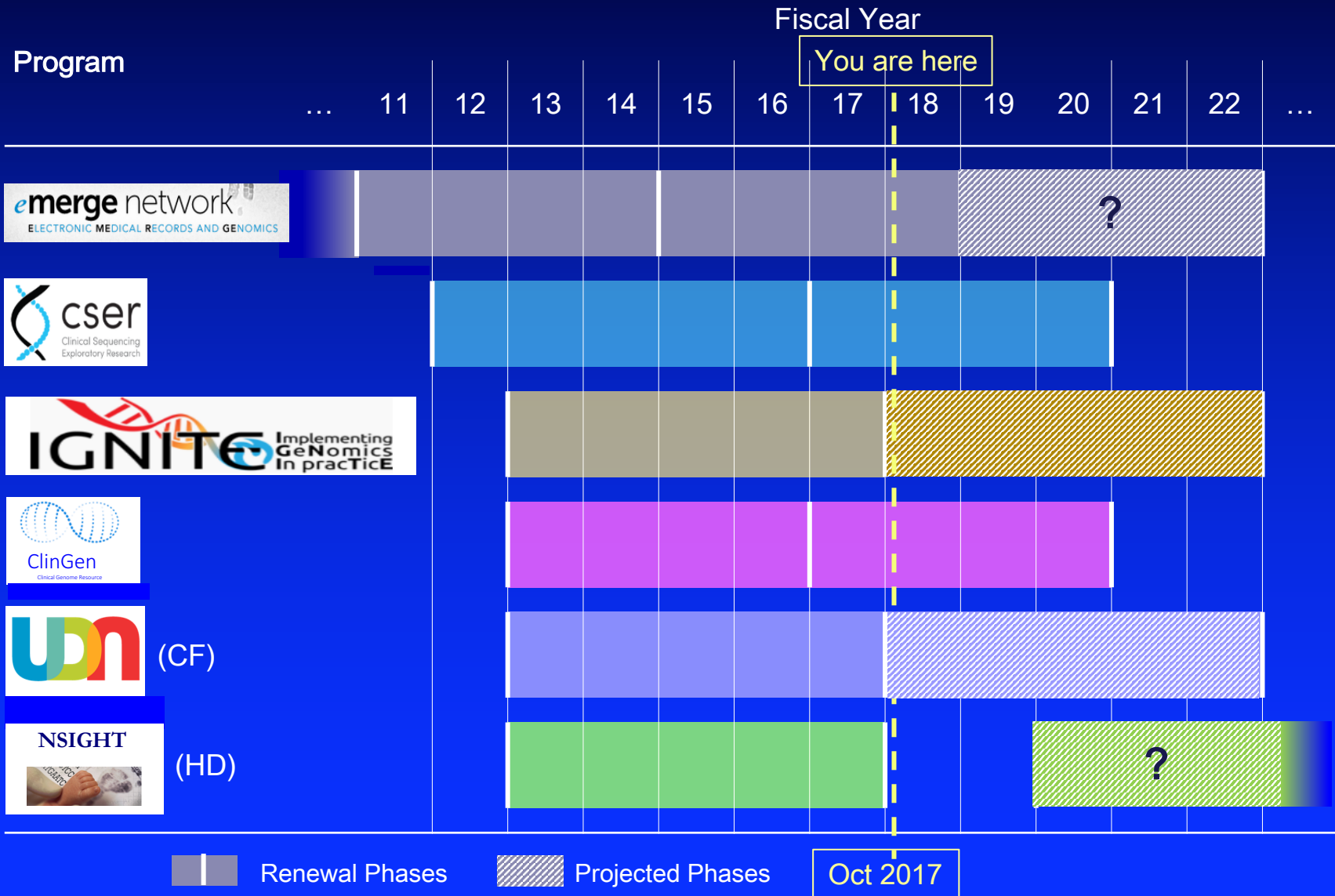
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Spectrum of Genomic Medicine Implementation: Intensity vs. Breadth



Timeline of NHGRI Genomic Medicine Programs



CSER, eMERGE, and IGNITE

Clinical
Sequencing
Evidence
Generating
Research
(FY2017-
2020)

Electronic
Medical
Records and
Genomics
(FY2015-
2018)

Implementing
Genomics in
Practice
(FY2018-
2022)

Commonalities and Complementarity of CSER and eMERGE

CSER (FY2017-2020)

- ~4,600 pts, 6 sites
- Community clinical scenarios
- Focus: clinical encounter
- Increased ethnic and socioeconomic diversity
- Evidence generation for clinical utility of genomic sequencing
- Real-world barriers to integrating genomic data for healthcare utilization

- EMR integration
- Clinical impact of RoR
- Data sharing concerns

eMERGE (FY2015-2018)

- 25K pts, 9 sites
- Electronic phenotyping
- Focus: system-wide
- Health outcomes of rare variants in ~100 clinically relevant genes
- System-wide impact of reporting actionable variants
- Improved e-phenotyping
- Novel variant discovery
- Electronic CDS

Commonalities and Complementarity of eMERGE and IGNITE

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- Novel variant discovery
- Electronic CDS

- EMR integration
- Cost-effectiveness
- Patient/clinician education

IGNITE (FY2018-2022)

- ~15K pts, 4-6 sites
- Diverse, real-world clinical settings
- Focus: pragmatic trials
- Clinical utility of established genomic medicine interventions
- Increased ethnic and socioeconomic diversity
- Generalizable knowledge on use of trials in genomic medicine interventions

Many Thanks...

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