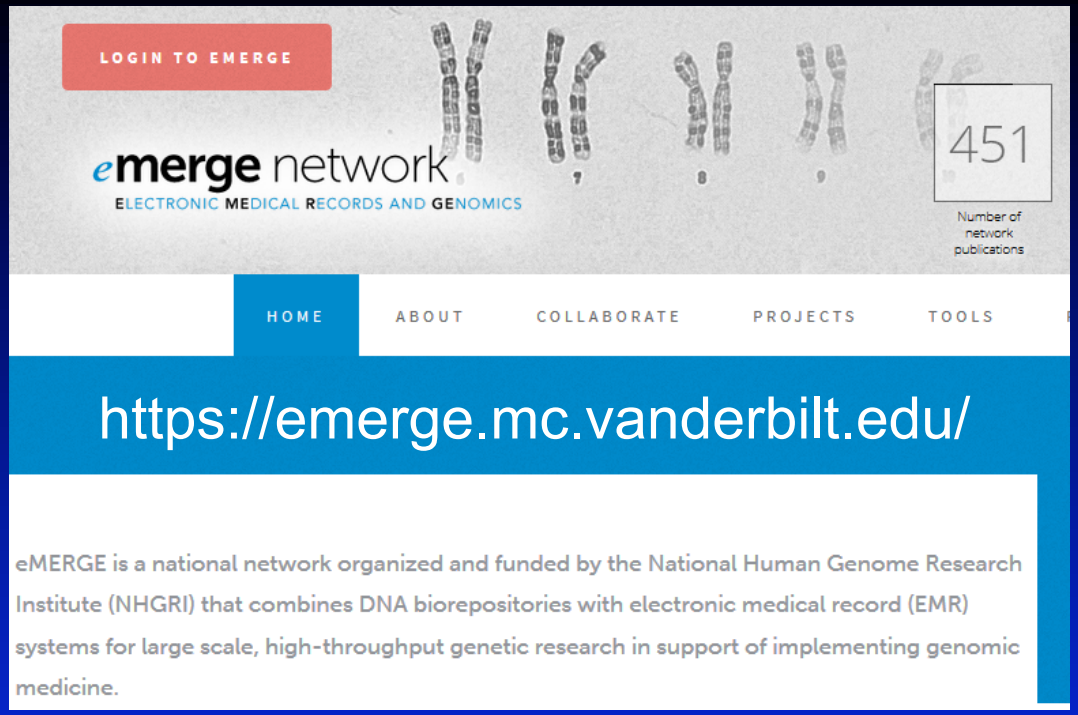
National Human Genome Research Institute

National Institutes of Health

U.S. Department of Health and Human Services



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-

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

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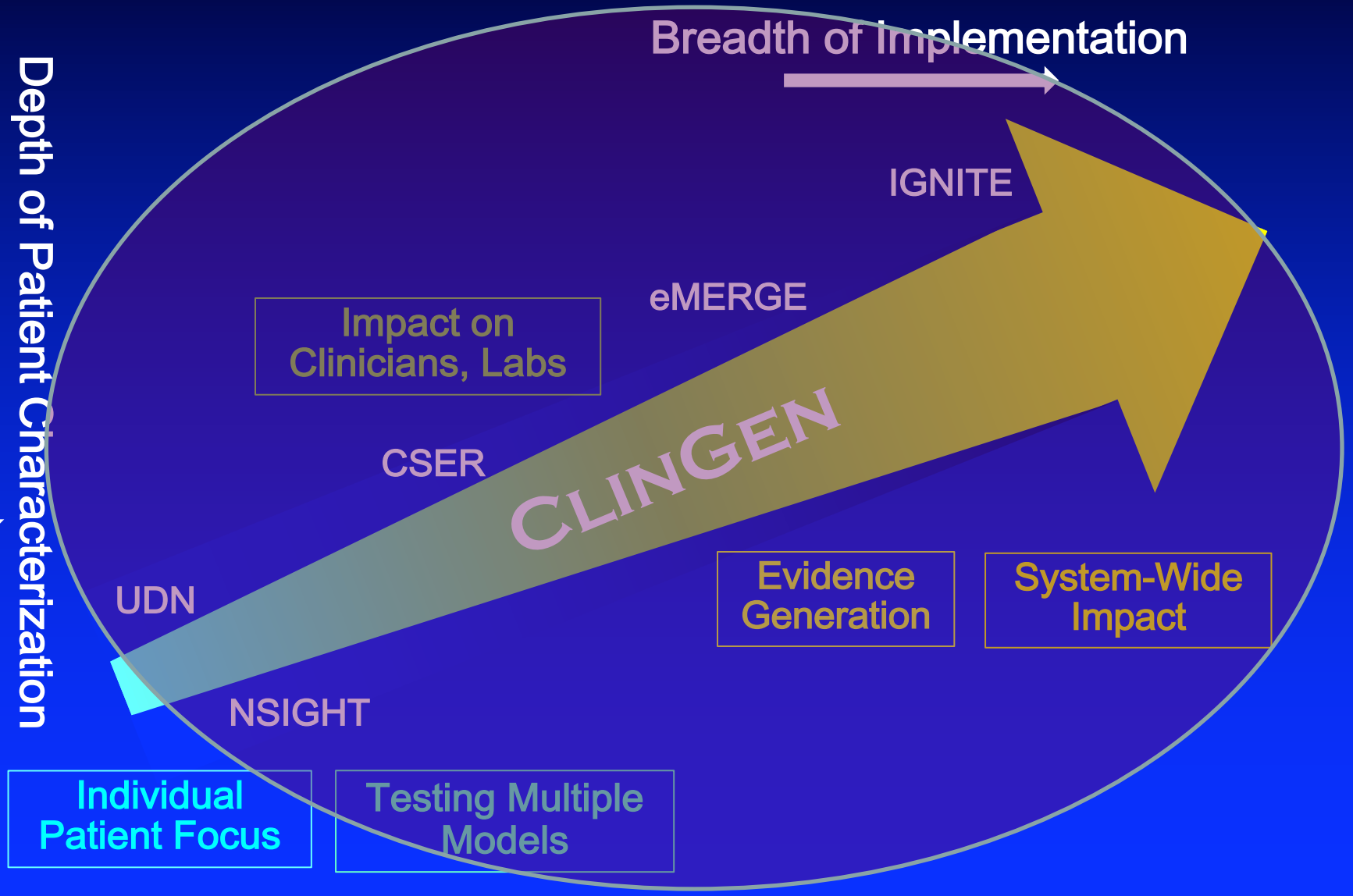
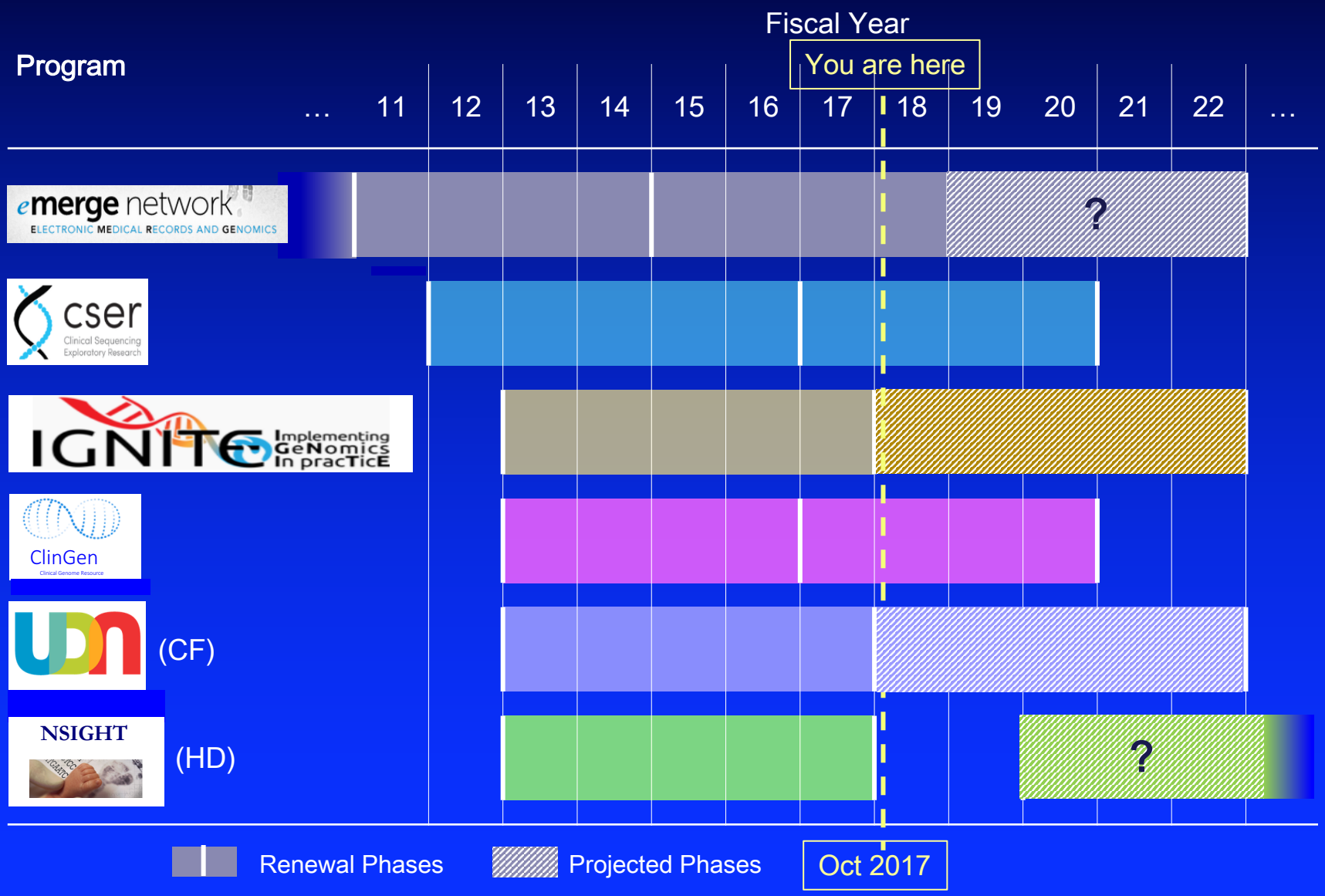
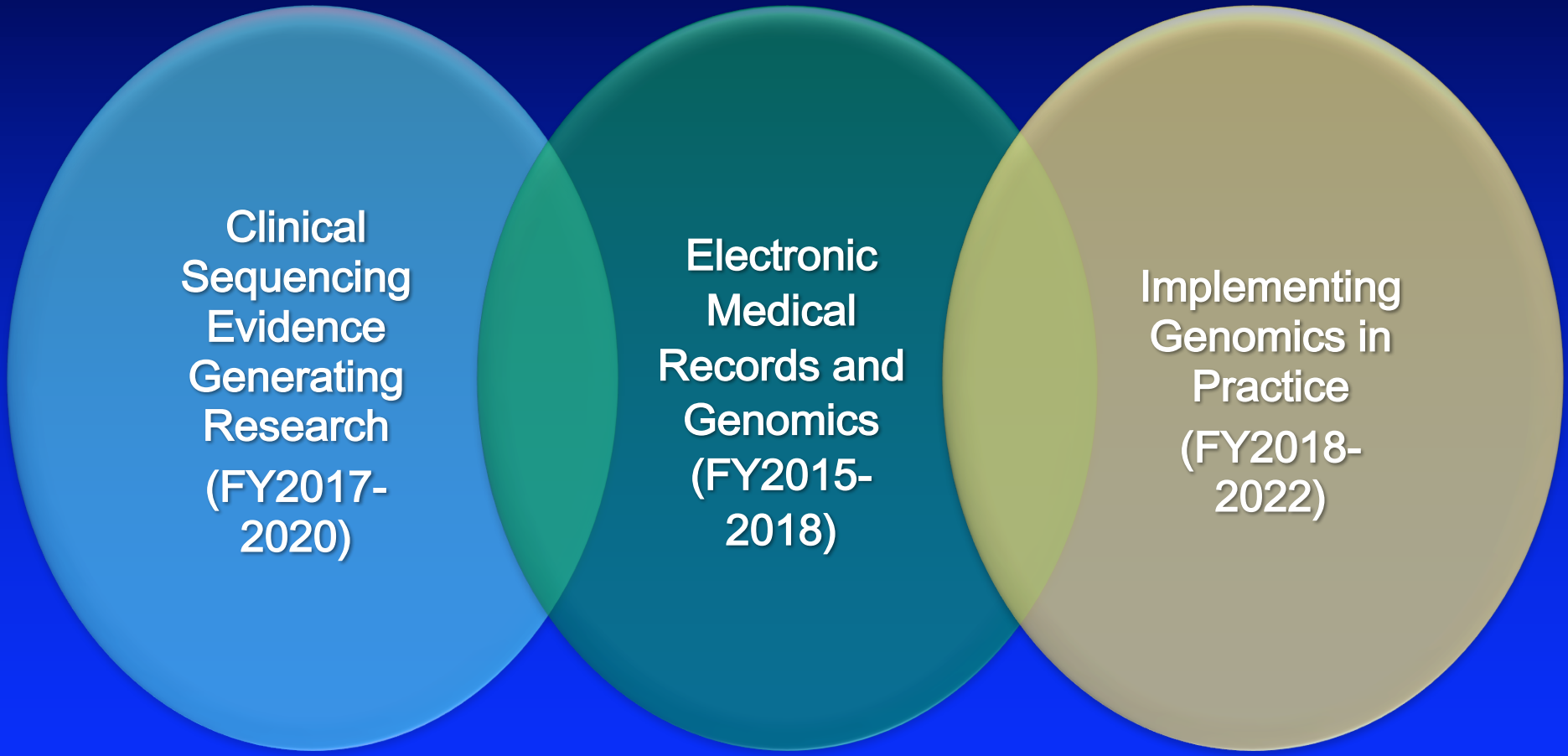
Spectrum of Genomic Medicine Implementation: Intensity vs. Breadth



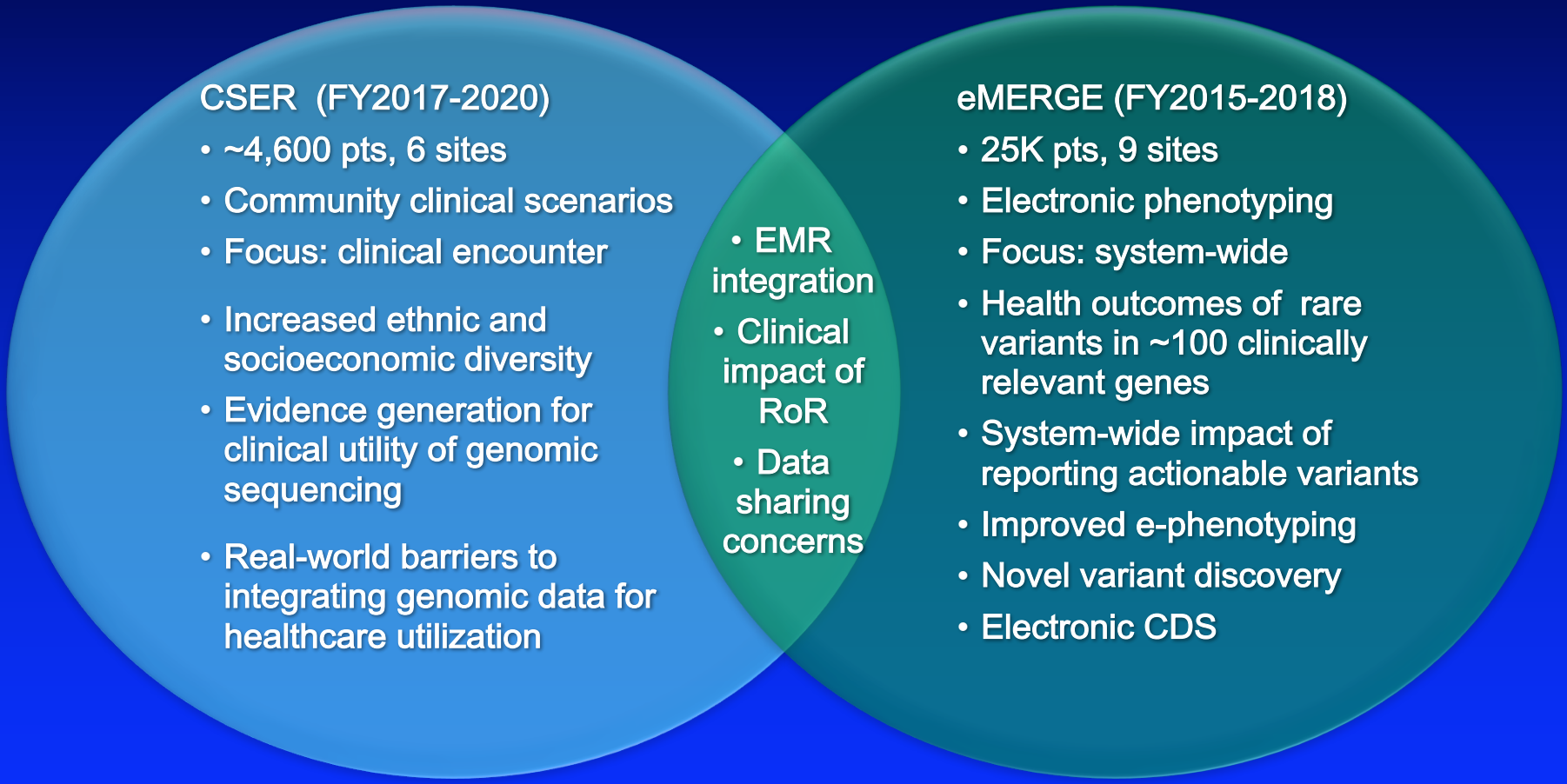
Timeline of NHGRI Genomic Medicine Programs



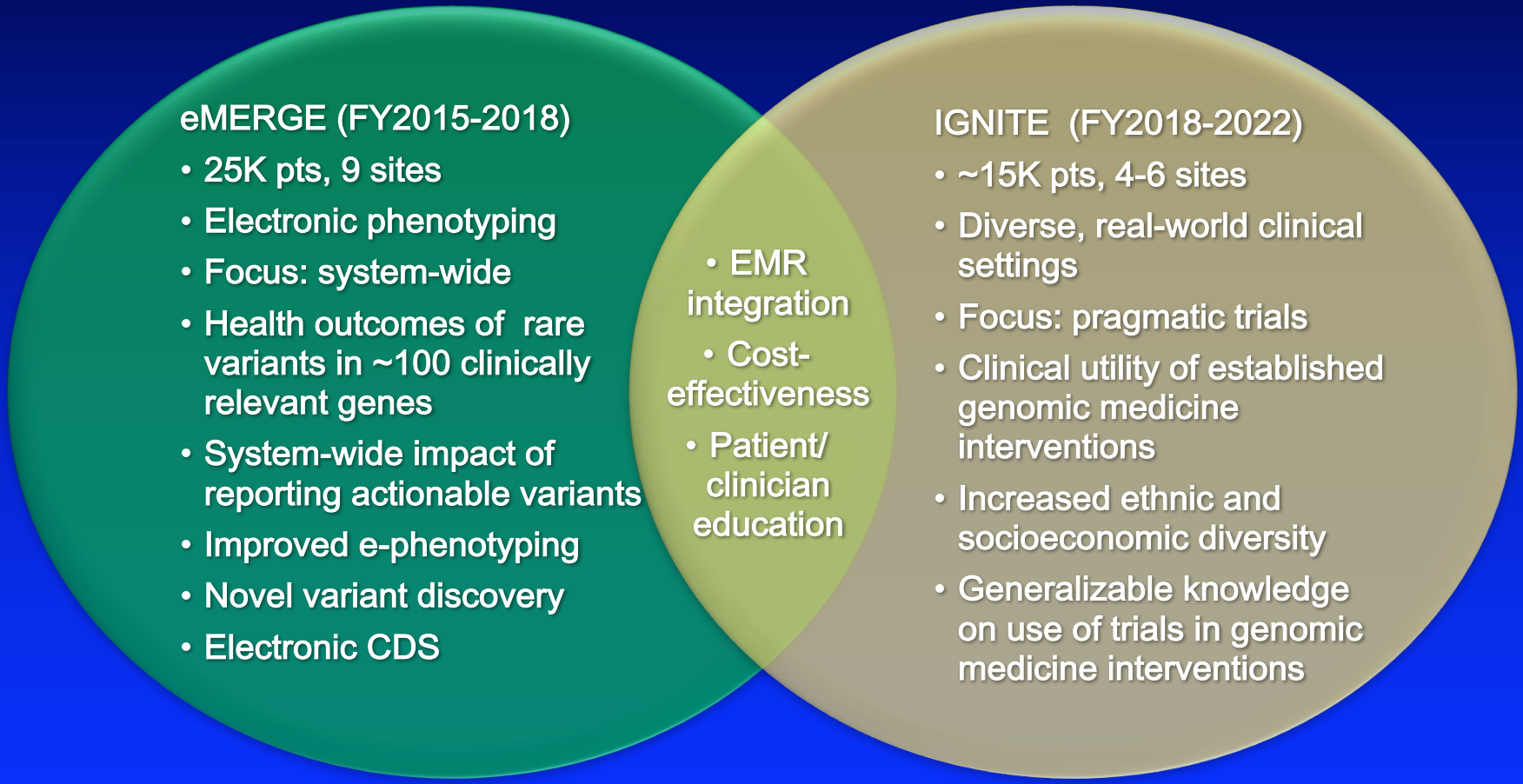
CSER, eMERGE, and IGNITE



Commonalities and Complementarity of CSER and eMERGE



Commonalities and Complementarity of eMERGE and IGNITE



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eMERGE network
ELECTRONIC MEDICAL RECORDS AND GENOMICS

IGNITE
Implementing GeNomics In practicE

NSIGHT

Newborn Sequencing

cser
Clinical Sequencing Exploratory Research

UDN
Undiagnosed Diseases Network

ClinGen
Clinical Genome Resource