National Human Genome Research Institute

National Institutes of Health

U.S. Department of Health and Human Services

Genomic Medicine Pilot Demonstration Projects

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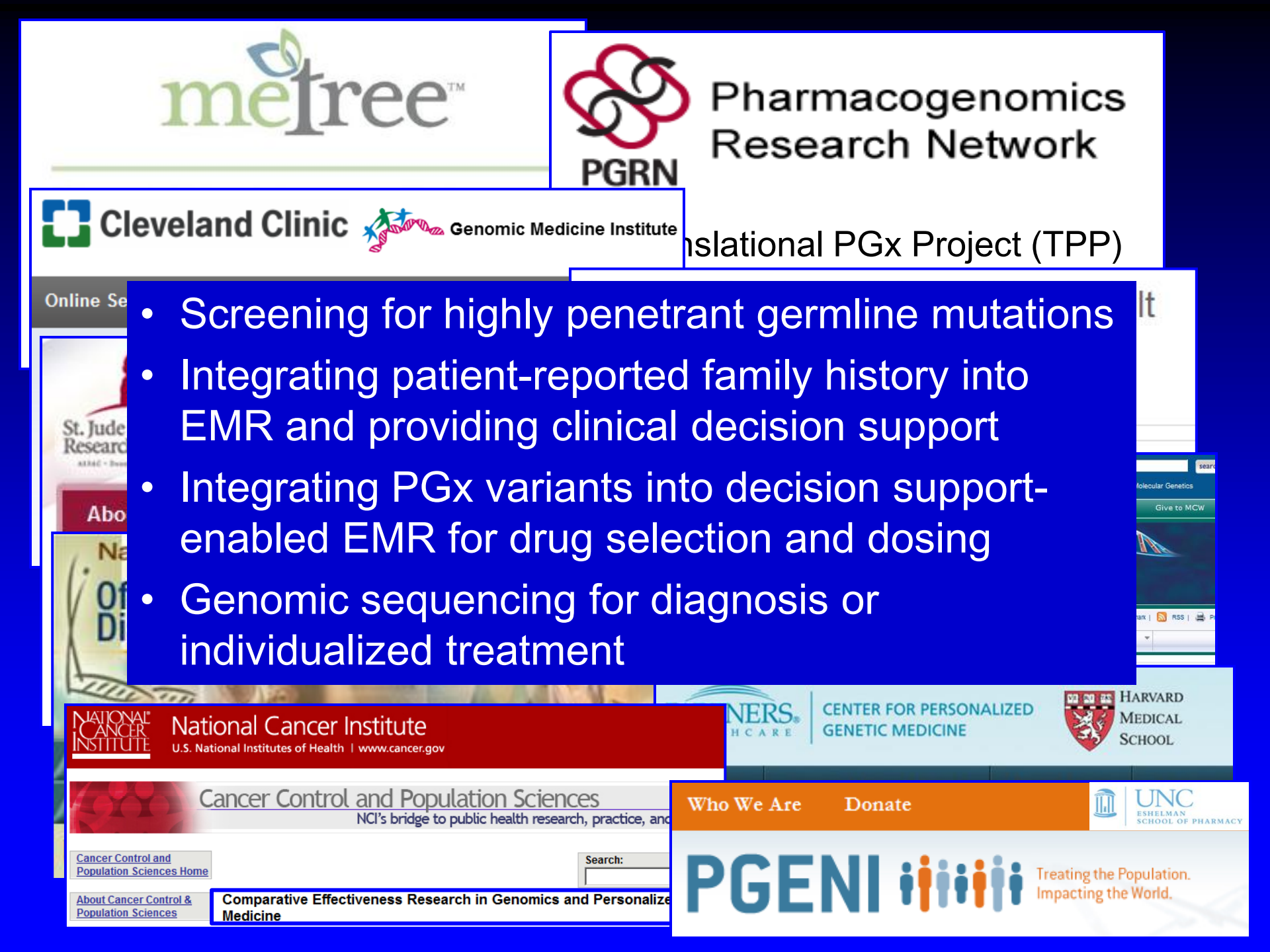
Genomic Medicine Demonstration Projects - Background

- Arises from strategic plan implementation efforts of Disease-Oriented Genomic Medicine WG, Council's Genomic Med WG
- Genomic Medicine Colloquium in June showed over 20 active GM centers at varying stages of implementation
- Supported through multiple institutional and NIH mechanisms (CTSAs, PGRN TPP)
- Numerous similar efforts, shared needs, common barriers



- Screening for highly penetrant germline mutations
- Integrating patient-reported family history into EMR and providing clinical decision support
- Integrating PGx variants into decision support-enabled EMR for drug selection and dosing
- Genomic sequencing for diagnosis or individualized treatment

Sources of Support for Ongoing Projects

- Institutional:
  - Mt. Sinai
  - Partners Healthcare
  - St. Jude
  - Vanderbilt
- Federal:
  - CDC's Genomic Applications in Practice and Prevention Network
  - Clinical Translational Science Awards (CTSA)
  - NCI Comparative Effectiveness Research for Genomics and Personalized Medicine
  - Pharmacogenetics Research Network (PGRN)

Common Barriers to Genomic Medicine Projects

- Skepticism, resistance, inertia
- Expectations for morbidity/mortality evidence
- Needs for CLIA certification, IRB approval
- Confusion over consent and counseling models
- Difficulty integrating results with existing EMR systems
- Burden of interpreting and following up massive numbers of results
- Reluctance to adopt novel patient care strategy

Genomic Medicine Demonstration Projects

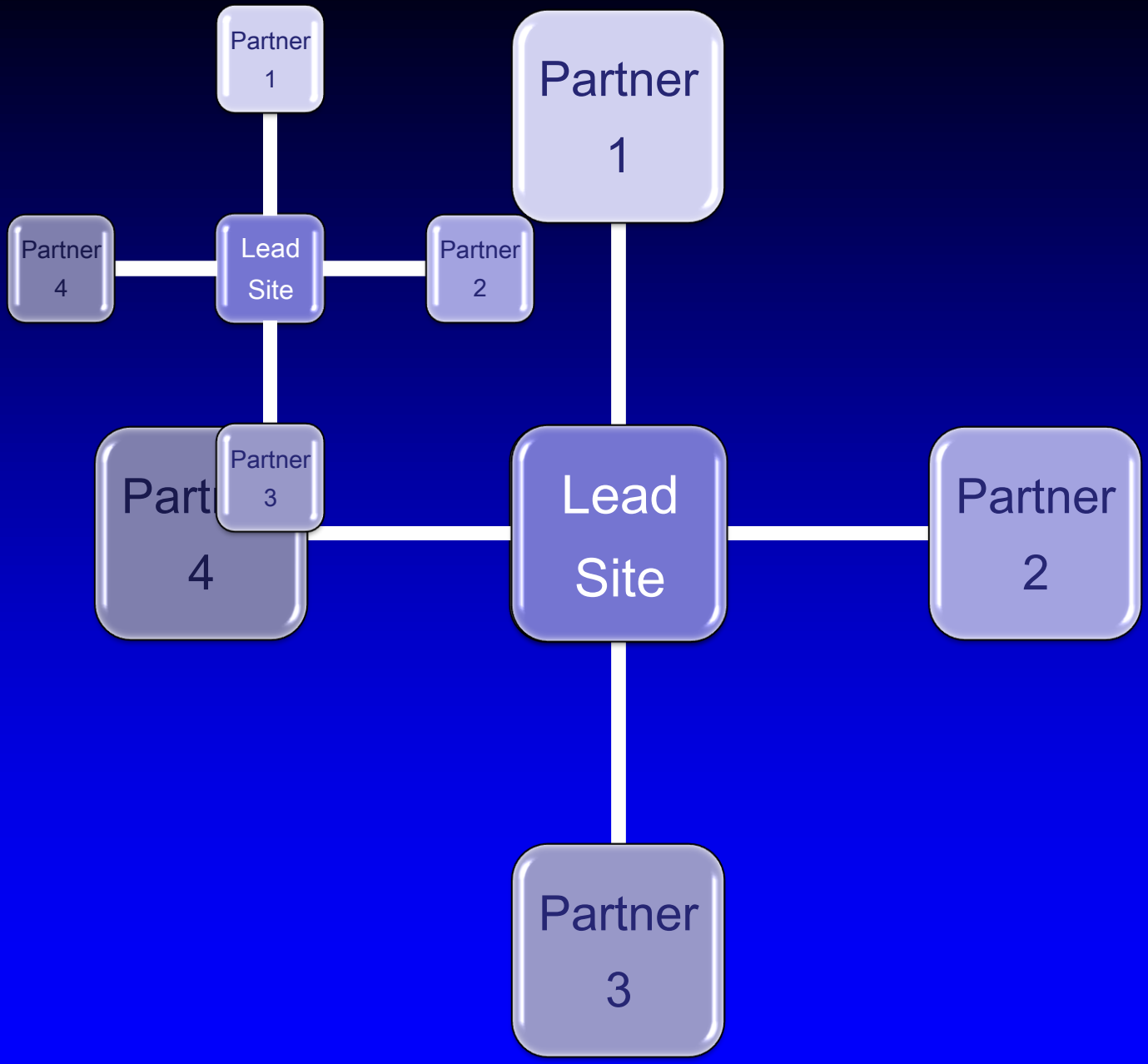
Proposal: Demonstrate feasibility of, and develop methods for, incorporating patients' genomic findings into their clinical care

Goals:

1. Expand existing GM efforts and develop new projects and methods, in diverse settings
2. Contribute to evidence base regarding outcomes of implementing GM
3. Define and disseminate processes of GM implementation, diffusion, and sustainability in diverse clinical settings

Proposed Approach

- Link early adopter sites to less experienced groups
- Expand number and types of sites at which implementation being done
  - Health maintenance organizations
  - Community hospitals and private practices
  - Military or veterans' care
  - Underserved and indigent populations
- Develop best practices
- Collect evidence of impact on outcomes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Partner 4
Lead Site
Partner 2
Partner 3
Partner 1
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Partner 2
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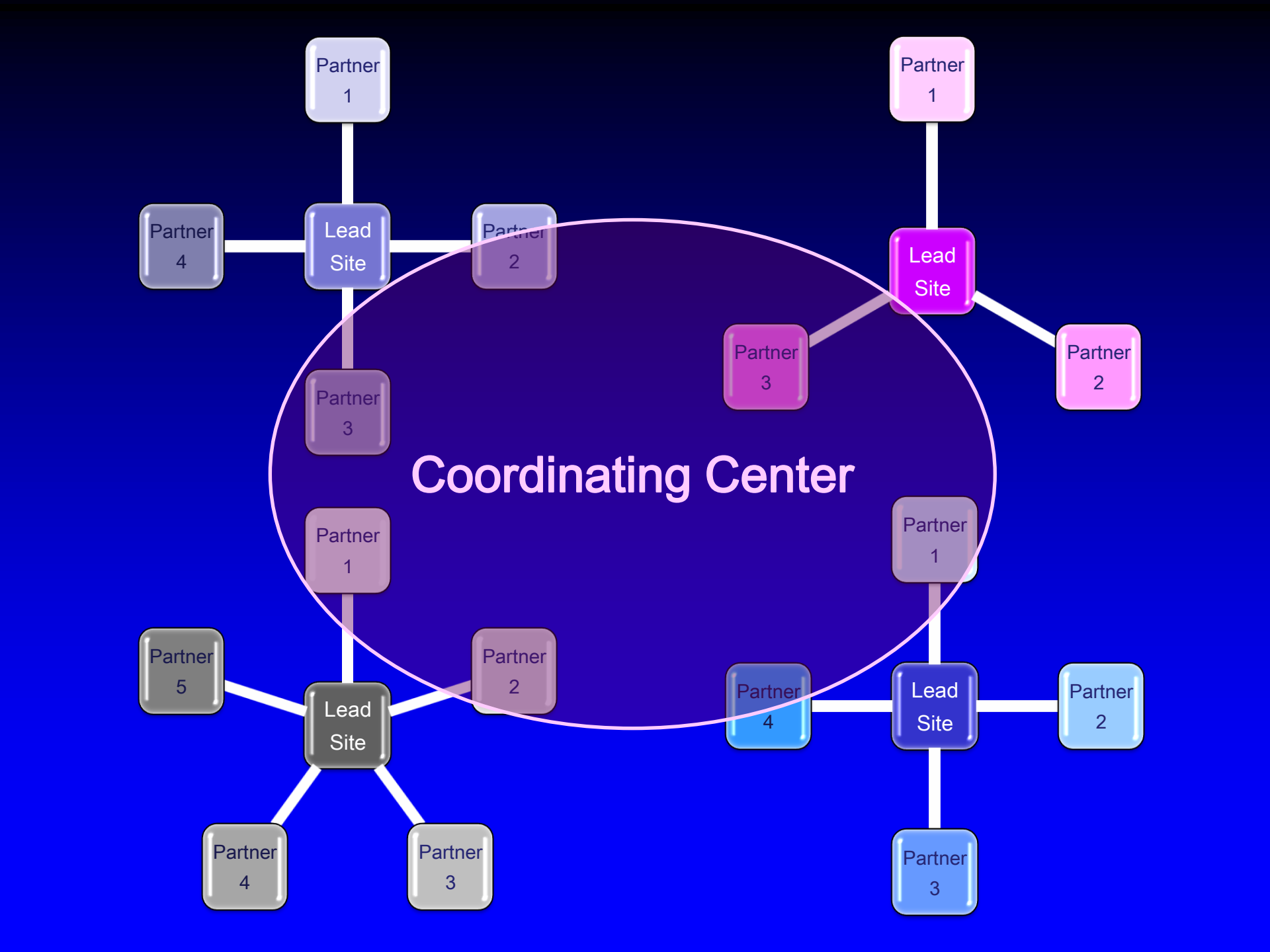
Partner 1
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Partner 2
Coordinating Center
Partner 1
Partner 1
Partner 5
Partner 2
Partner 4
Lead Site
Partner 2
Partner 4
Partner 3
Partner 3

Characteristics to be Encouraged

- Institutional endorsement, practitioner involvement, patient participation
- Identified group of clinicians willing to learn about, receive, and act upon genotyping results
- CLIA-certified genotyping environment and efficient workflow for assaying and reporting results
- Process for integrating genotype results into patients' EMRs and providing CDS
- Alternative non-computerized processes for settings without sophisticated EMRs

Characteristics to be Encouraged

- Defined outcomes such as satisfaction, uptake of recommended interventions, cost, morbidity
- Approaches for collecting and assessing outcomes
- Plan for sustaining, and possibly expanding, successful implementation projects
- Leveraging available institutional support and other resources and identifying clear path to sustainability of implementation project
- Ability to contribute objective evidence most likely to influence uptake and reimbursement of genomic medicine interventions

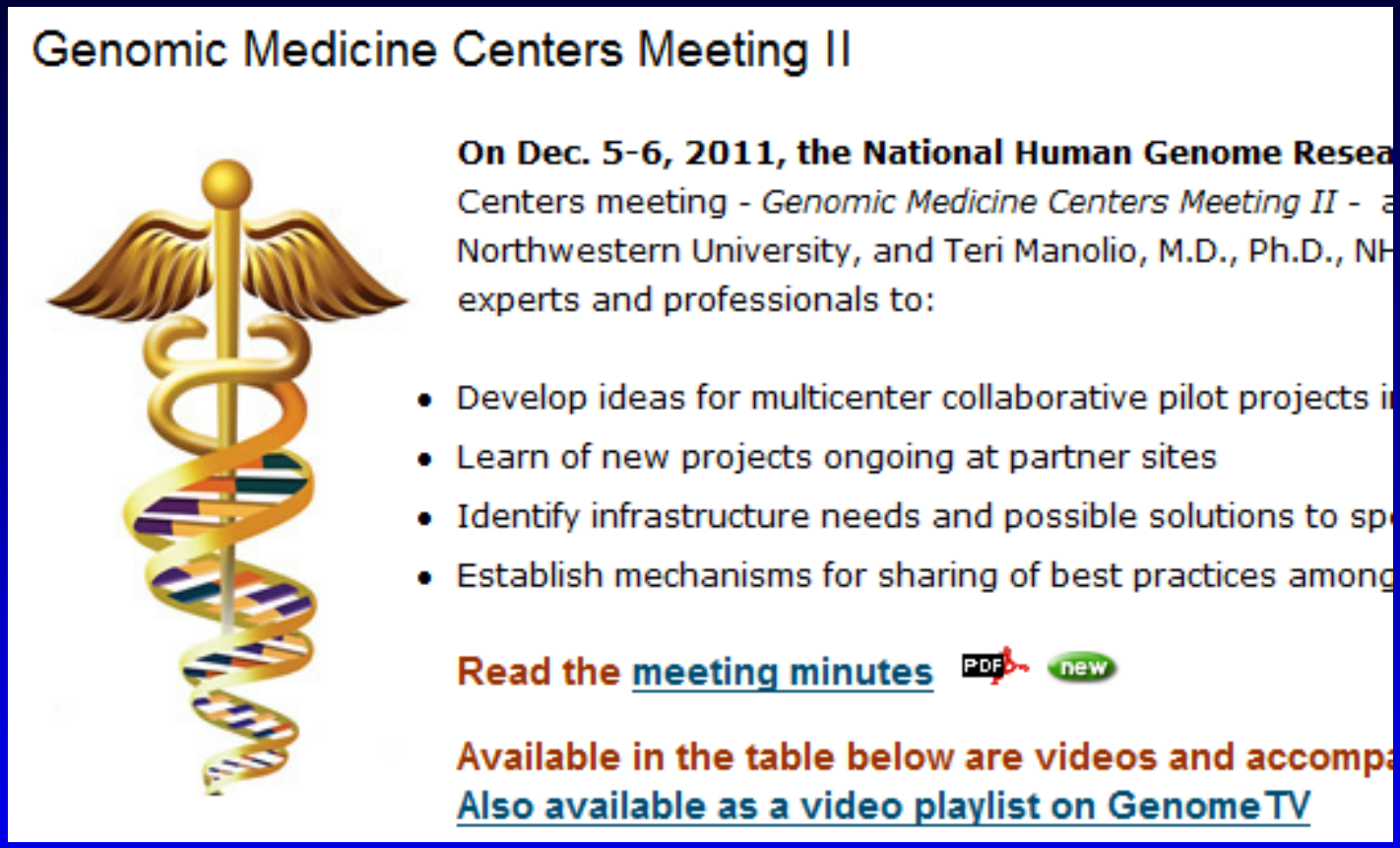
Role of Coordinating Center and Consortium

- Collect and disseminate protocols for successful implementation in variety of settings
- Organize broader, open meetings of genomic medicine community
- Coordinate with related NHGRI/NIH projects such as RoR Consortium, CSER, eMERGE, PGRN
- Explore potential to expand projects beyond initial lead-partner sites to other lead and partner sites
- Create cadre of institutions implementing and evaluating genomic medicine protocols, and collecting and disseminating successful approaches

Anticipated Funding

- $3M in FY13 and $4.4M per year in FY14-FY15
- Support 3-5 multisite demonstration projects each with one lead and 3-5 partner sites plus CC
- U01 (Cooperative Agreement) mechanism
- Seek support from other NIH Institutes, consider elevating priority for projects with proposed co-funding

Many thanks…



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