



# Clinical Sequencing Exploratory Research

*Moving the Genome Into the Clinic*

377 Researchers  
21 Institutions  
1 Consortium

## *Purpose & Structure*

- Explore, within an active clinical setting, the application of genomic sequence data to the care of patients.
  1. Generation of genomic sequence data,
  2. Interpretation and translation of data for the physician
  3. Communication to the patient
- **Three (3) Project Teams per Site**
  1. Practice: clinical setting being studied and what medical outcomes measured
  2. Lab: sequencing and reporting of genome-scale results to clinicians/EMR
  3. ELSI: ethical and psychosocial implications of bringing broad genomic data into the clinic

*Plan: Overview of CSER, Fall 2012*



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## *The Big Questions*

- What are the best practices in moving whole genome sequencing, using Next Generation technologies, from medical science to the clinical practice?
- What are the patient characteristics that signal potential utility (or lack thereof) for applying genome-scale sequencing?
- What are the best approaches to analyzing data?
- Are there special considerations in different populations?
- How should relevant information be best represented in the EMR?
- What criteria are most useful in guiding which results should be returned (and how) to the patient and physician?
- How should we deal with the plethora of highly heterogeneous “non-target” data generated when performing sequencing?

*Evans: Overview of CSER, Fall 2013 and NHGRI staff*



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## Working Groups & Steering Committees

CSER Steering Committee  
CSER Coordinating Center  
ELSI Steering Committee  
Actionable Variants & ROR  
Electronic Medical Records  
Genetic Counselors  
Informed Consent & Governance  
Pediatrics  
Outcomes & Measures  
Sequencing Standards  
Practitioner Education

## Leadership

Gail Jarvik  
Gail Jarvik  
Barbara Koenig  
Wendy Chung  
Brian Shirts  
Sarah Scollon  
Paul Appelbaum  
Kyle Brothers  
Stacy Gray  
Bradford Powell  
Sharon Plon  
Lucia Hindorff  
Gail Henderson  
Julia Wynn  
Joon Ho-Yu  
Ben Wilfond  
David Veenstra  
Will Salerno  
Kelly East

*As of 01/05/2017*



[www.genome.gov/CSER](http://www.genome.gov/CSER)  
[www.cser-consortium.org](http://www.cser-consortium.org)



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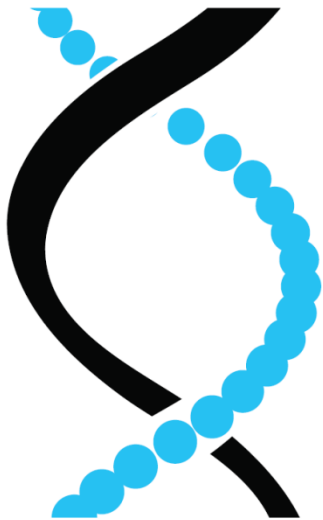


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## Full Logo



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Clinical Sequencing  
Exploratory Research

Logo Font:

Website Font:

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Open Sans

## Mark



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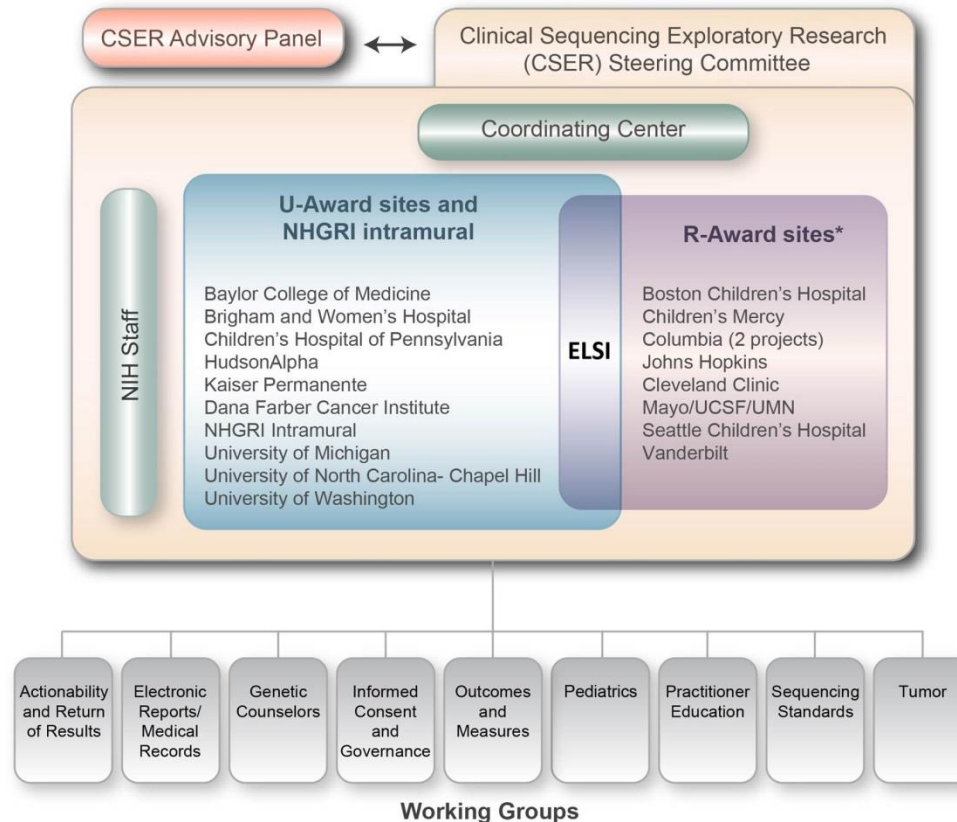
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\* Grants funded under RFA-HG-11-003 and RFA-HG-11-004 have ended but the investigators on those grants continue to participate in Consortium activities. Along with ELSI investigators on the U-grants, they meet regularly to discuss ELSI issues relevant to CSER.



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## CSER Site (U-Award)

Baylor College of Medicine  
Brigham & Women's Hospital  
Children's Hospital of Philadelphia  
Dana-Farber Cancer Institute  
HudsonAlpha Institute

Kaiser Permanente  
University of Michigan  
University of North Carolina  
University of Washington  
ClinSeq (NHGRI Intramural)

## Patient Focus

WES in tumor/germline childhood cancers  
WGS in adults, healthy and w. cardiomyopathy  
WES/WGS in children w. pediatric disease  
WES in tumor/germline in lung, colorectal cancers  
WES in children with intellectual, developmental disability  
WGS in preconception carrier screening  
WES in adults & children with advanced rare cancer  
WES in adults & children with genetic disease  
WES in adults with colorectal cancer  
WGS in healthy adults and adults with heart disease

*Evans: Overview of CSER, Fall 2013*



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## CSER Site (U-Award)

## PI(s)

Baylor College of Medicine	Sharon Plon & Will Parsons
Brigham & Women's Hospital	Robert Green
Children's Hospital of Philadelphia	Ian Krantz
Dana-Farber Cancer Institute	Pasi Janne & Eliezer van Allen
HudsonAlpha Institute	Greg Cooper
Kaiser Permanente	Katrina Goddard & Ben Wilfond
University of Michigan	Arul Chinnaiyan, Kenneth Pienta, Scott Roberts
University of North Carolina	Jim Evans, Jonathan Berg & Gail Henderson
University of Washington	Gail Jarvik
ClinSeq (NHGRI Intramural)	Leslie Biesecker
Coordinating Center (UW)	Gail Jarvik, Wylie Burke, Debbie Nickerson, & Peter Tarczy-Hornoch





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## CSER Site (R-Award)

## PI(s)

Boston Children's Hospital

Ingrid Holm

Columbia University

Paul Appelbaum

Columbia University

Wendy Chung

Children's Mercy Hospital

Jeremy Garrett

Johns Hopkins University

Michelle Lewis

Mayo Clinic

Rich Sharp

Seattle Children's Hospital

Holly Tabor

UC - San Francisco,  
Mayo College of Medicine, &  
University of Minnesota

Barbara Koenig, Gloria Peterson, & Susan Wolf

Vanderbilt University &  
McGill University

Ellen Clayton & Bartha Knoppers

Coordinating Center (UW)

Gail Jarvik, Wylie Burke, Debbie Nickerson, & Peter Tarczy-Hornoch



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