Introductory Remarks

Dr. Ben Heavner
GREGoR Data Coordinating Center
(with slides courtesy of other GREGoR members)

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Genomics Research to Elucidate the Genetics of Rare Diseases

@GREGoR_research    www.gregorconsortium.org
Exome sequencing ushered in a paradigm shift in Mendelian gene discovery

chart recreated from Chong et al., 2015 and Bamshad et al., 2019
How do we increase the solve rate?
What will catalyze the next paradigm shift?
The GREGoR Consortium

University of Washington Center for Rare Disease Research
Seattle, WA

University of Washington Data Coordinating Center
Seattle, WA

Broad Institute
Cambridge, MA

GREGoR Stanford Site
Stanford, CA

Baylor College of Medicine - GREGoR
Houston, Texas

Children's National Hospital / Invitae Pediatric Mendelian Genomics
Washington, DC
**Mission Statement**

To significantly increase the proportion of Mendelian conditions with an identified genetic cause. A key objective of the GREGoR Consortium is to develop and apply approaches to discover causal genes underlying Mendelian conditions for which a candidate gene was not identified using whole exome sequencing alone.

**Scientific Goals**

- Shift the Paradigm
- Increase diagnostic yield
- Develop best practices
- Create a dataset with broad utility
- Make RD diagnosis applicable to all
Today’s Session

- **Unraveling the Complex Connections between Genomic Variation and Disease Trait Manifestation**
  
  *Dr. Jennifer Posey*
  
  Baylor College of Medicine Research Center

- **Expanding our understanding of human genetic variation through long-read sequencing of 1000 Genomes Project samples**
  
  *Dr. Danny Miller*
  
  University of Washington Center for Rare Disease Research

- **Getting to know GREGoR Data in AnVIL**
  
  *Dr. Ben Heavner*
  
  GREGoR Data Coordinating Center
Housekeeping notes

- Please register your attendance at this session on the sign-in sheets on the back table.
- Learn more about GREGoR at https://gregorconsortium.org/.
- Visit GREGoR Posters at ASHG!
- Contact the GREGoR Data Coordinating Center via email at gregorconsortium@uw.edu.

Session Feedback and Interest Survey
https://tinyurl.com/gregor-post-session-survey
Thank you