Introductory Remarks

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

November 2, 2023

GREGoR consortium

***G**enomics **R**esearch to **E**lucidate the **G**enetics **o**f **R**are Diseases*

@GREGoR_research www.gregorconsortium.org

Exome sequencing ushered in a paradigm shift in Mendelian gene discovery

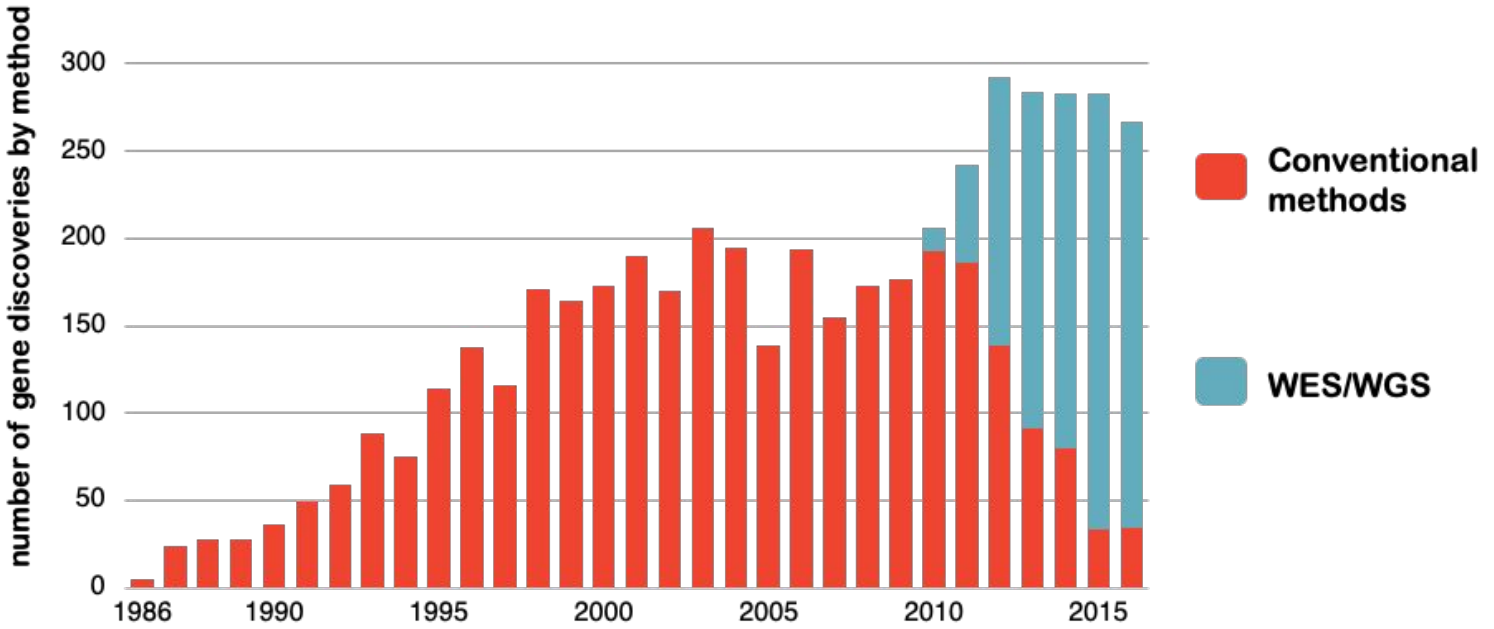
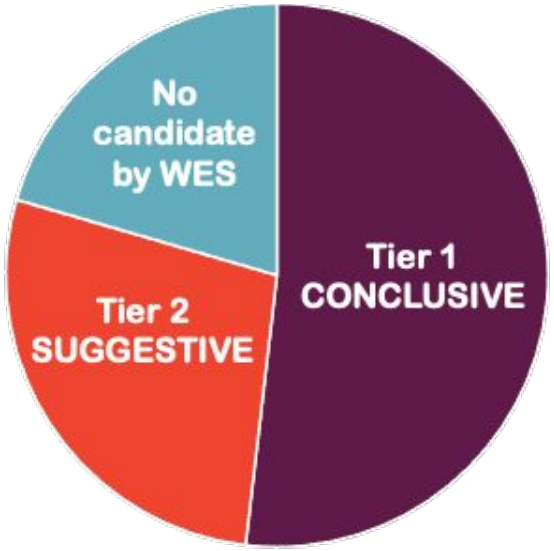


chart recreated from Chong et al., 2015 and Bamshad et al., 2019

How do we increase the solve rate?
What will catalyze the next paradigm shift?



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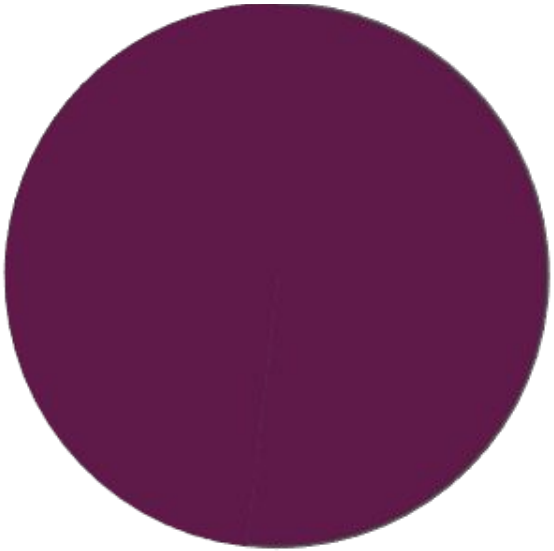



University of Washington Center for Rare Disease Research
Seattle, WA
University of Washington Data Coordinating Center
Seattle, WA
Broad Institute
Cambridge, MA
The GREGoR Consortium
GREGoR Stanford Site
Stanford, CA
Baylor College of Medicine - GREGoR
Houston, Texas
Children's National Hospital / Invitae
Pediatric Mendelian Genomics
Washington, DC

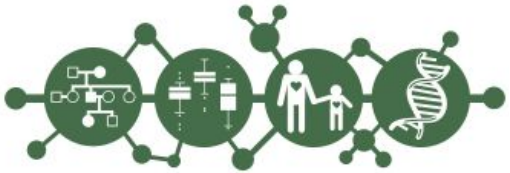
GREGoR

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

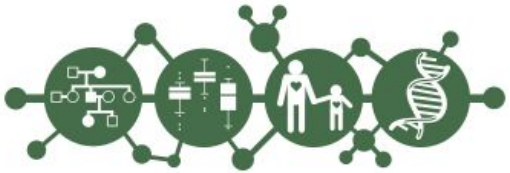


Today's Session

- **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

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

- 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

Thank you

GREGoR consortium

New GREGoR Grant Opportunities

September 2023: The GREGoR Consortium announces Grant Opportunities to bring in needed expertise not currently represented within the Consortium in order to expand the impact that GREGoR provides (the **Research Grant awards**), and to increase workforce diversity in genomic research (the **Grants to Support Workforce Diversity**).

Visit our Grant Opportunities web page for information including how to apply.

Visit GREGoR's Learning Hub

This new section of the GREGoR website provides information and resources for families and patients impacted by rare disease and addresses:

- What is rare disease?
- What methods and new technologies is GREGoR using?
- How does research participation work?

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