GREGoR consortium

Genomics Research to Elucidate the Genetics of Rare Diseases

GREGoR Funding Opportunities

The GREGoR Consortium announces Grant Opportunities to bring in needed expertise not currently represented within the Consortium in order to expand GREGoR's impact:

- **Research Grants** - Letters of Intent due Nov. 13
- **Grants to Support Workforce Diversity in Genomics Research** - applications due Jan. 12

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?

Connect with GREGoR online

Visit us on the web

Follow us on Twitter/X

Follow us on LinkedIn

Email us

GREGoR at ASHG 2023

- **Dr. Lisa Chadwick, NHGRI, "Everything You Ever Wanted to Know About Applying for NIH Grants", Nov. 2nd 7 - 8:30 am ET**
- **GREGoR Ancillary Session Nov. 2nd, 12:15 - 1:45 pm**
  Agenda:
  1. **12:15** Gathering and Conversation
  2. **12:25 Dr. Ben Heavner** Introductory Remarks
  3. **12:30 Dr. Jennifer Posey** "Unraveling the Complex Connections between Genomic Variation and Disease Trait Manifestation"
  4. **12:55 Dr. Danny Miller** "Expanding our understanding of human genetic variation through long-read sequencing of 1000 Genomes Project samples"
  5. **1:20 Dr. Ben Heavner** "Getting to know GREGoR Data in AnVIL"
  6. **1:45** Adjourn

See our GREGoR at ASHG web page

Visit the GREGoR Poster: Poster Hall AB, Board Number: PB4711
Presentation Date/Time: Nov. 3rd 3-5 pm ET:
"Data sharing in the GREGoR Consortium to support rare genetic disease research"

- See all GREGoR abstracts on our GREGoR at ASHG webpage -

GREGoR Data

Researchers can now apply for access to the GREGoR Consortium Dataset (R01) via dbGaP study phs003047

See the Data page on GREGoR's website

GREGoR release version	phs003047.v1.p1
Release Date	Sep 1, 2023
Number of participants[1] Short read WGS Short read WES RNA-seq	2512 2438 997 183
Number of families	990
Consent groups[2]	GRU, HMB
Genome build	GRCh38
GREGoR Data Model Version	1.1
Size (TB)	72.9

Most frequent HPO Terms in GREGoR Dataset:

- Global developmental delay
- Seizure
- Hypotonia
- Intellectual disability
- Muscle weakness
- Morphological central nervous system abnormality
- Bicuspid aortic valve
- Thoracic aortic aneurysm
- Microcephaly
- Abnormal cerebral cortex morphology