Release Notes

Release 2 (R02):

To see what's new in a Release, please refer to the Release Notes below and <u>GREGOR Dataset</u> characteristics by release version. Additionally, the GREGOR Consortium Data Coordinating Center maintains a list of errata for Release 2 at

https://gregorconsortium.org/data/release-notes/r02-errata

Release Date: November, 2024

AnVIL Workspaces:

- AnVIL GREGOR R02 GRU
- AnVIL GREGOR R02 HMB

Summary of changes since previous release:

- 1. R02 adds data for an additional 4882 participants as well as long-read data (Nanopore and PacBio) to the GREGoR Dataset.
- 2. The R02 version of GREGoR Data Model includes data tables that support long-read data (Nanopore and PacBio) and genetic findings reported by GREGoR Research Centers.
- 3. A subset of aligned DNA short-read files were uniformly processed by the GREGoR Data Coordinating Center (DCC). The R02 dataset contains these reprocessed, aligned DNA short-read files instead of the aligned DNA short-read files separately processed and uploaded by GREGoR Research Centers. The aligned_dna_short_read_id for each reprocessed file begins with "GREGoR DCC A1".
- 4. Multi-sample VCFs (split by chromosome) and single-sample genomic VCFs (gVCFs) derived from DCC reprocessed aligned DNA short-read files are available in the R02 release. These VCFs contain genotype data for single-nucleotide variants and short-indels and can be found in the called variants dna short read table.
- 5. The following participants were present in R01 and removed in the R02 dataset due to sex and ped check failures:
 - BCM Subject BH15330 2
 - BCM Subject BH16155 1
 - BCM Subject BH16330 4
 - GSS795357