

## Release Notes

## Release 3 (R03):

To see what's new in this Release, please refer to the Release Notes below and <u>GREGOR Dataset characteristics by release version</u>. Additionally, the GREGOR Consortium Data Coordinating Center maintains a list of errata for Release 3 at <a href="https://gregorconsortium.org/data/release-notes/r03-errata">https://gregorconsortium.org/data/release-notes/r03-errata</a>

Release Date: July 2025

## **AnVIL Workspaces:**

- AnVIL GREGOR R03 GRU
- AnVIL GREGOR R03 HMB

## Summary of changes since previous release:

- R03 adds an additional 1,446 participants as well as short-read ATAC-seq data to the GREGoR Dataset. In total, R03 includes a total of 8,840 participants.
- The R03 version of GREGoR Data Model includes data tables that support short-read ATAC-seq data.
- A subset of aligned DNA short-read files were uniformly processed by the GREGOR
  Data Coordinating Center (DCC). The R03 dataset contains these reprocessed, aligned
  DNA short-read files instead of the aligned DNA short-read files separately processed
  by GREGOR Research Centers. The aligned\_dna\_short\_read\_id for each
  reprocessed file begins with "GREGOR DCC A1".
- Jointly called, multisample VCFs (split by chromosome) and single-sample genomic VCFs (gVCFs) are available for DCC reprocessed aligned DNA short-read files. These VCFs contain genotype data for single-nucleotide variants and short indels and can be found in the called variants dna short read table.
- R03 fixes the following errors identified in R02:
  - The subject UW\_CRDR-sub-1443008 has been removed from the R03 dataset as it was inappropriately included in the R02 GRU release consent group.
  - R03 corrects metadata for family PMGRC-1067.
  - A small subset of participants were present in the dataset R02 with multiple, unique participant\_ids. These were entered erroneously because the participant has multiple tissue (analyte) or experiment types. We identified this issue in R02 release for the following families:
    - Broad BON B09 24

- Broad\_BON\_B11-09
- Broad\_BON\_B16-87
- Broad\_BON\_B17-01
- Broad\_BON\_B17-54
- Broad\_BON\_B17-77
- Broad\_BON\_B17-97
- Broad\_BON\_B17\_15
- Broad\_BON\_B18-37
- Broad\_BON\_B18-97
- Broad\_BON\_B19-52
- Broad\_BON\_B20\_32
- Broad\_BON\_UC35
- Broad\_BON\_UC355
- Broad\_BON\_UC522
- Broad HK079
- Broad\_HK080
- Broad\_HK081
- Broad HK085
- UW\_CRDR-fam-1590041
- UW\_CRDR-fam-1590067

R03 corrects this issue by retaining one unique participant\_id for each individual and mapping both analyte\_ids to this participant\_id. The file R03 remapped participant ids.tsv has a table with the mapping of participant\_ids retired in R03 to new participant ids.