



TOPMed Genomic Summary Results

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Background

Genomic Summary Results (GSR) are defined by the [National Institutes of Health Office of Science Policy](#) [6] as “the output of analyses of genomic data across the many individuals included within a specific study’s dataset.” Association analysis statistics and allele frequency information are two common types of GSR, though GSR may encompass other metrics or summary data calculated from individual-level data.

Individual-level genomic and phenotypic data are typically available through controlled-access. In contrast, access controls for GSR have varied over time (see [NOT-OD-19-023](#) [7] for a historical overview) and can also vary by study. Specifically, GSR are made available through controlled access when a study designates as “sensitive,” a designation that may be motivated by the study of potentially stigmatizing conditions or identifiable or vulnerable study populations. GSR from studies not designated as sensitive can be made publicly available.

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GSR in TOPMed

The TOPMed program comprises over 80 studies, including many designated as “sensitive” for the purposes of GSR sharing. To accommodate this heterogeneity of study designations while promoting broad access and use of TOPMed GSR by the scientific community, the TOPMed DCC collaborated with the NHLBI Data Access Committee, NHLBI TOPMed program staff, the University of Washington IRB, and TOPMed study PIs to establish a dbGaP accession for the controlled-access sharing of TOPMed GSR: [phs001974](#) [8], “NHLBI TOPMed: Genomic Summary Results for the Trans-Omics for Precision Medicine Program.”

GSR from TOPMed manuscripts and other Working Group analyses may be posted to this accession and made accessible, via controlled access and a General Research Use (GRU) consent designation, to applicants via the [normative dbGaP application process](#) [9].

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How to access TOPMed GSR

Full TOPMed GSR are available via controlled access, but a subset of the association test results for each released GSR are publicly available.

To apply for access to the full TOPMed GSR in phs001974, investigators should follow [dbGaP instructions](#) [9] to initiate a project request and submit a Data Access Request (DAR) for phs001974. Investigators with an active dbGaP project may request phs001974 as an additional DAR. Requests for access to phs001974 will be reviewed by the NHLBI Data Access Committee. Please note that an applicants' proposed Research Use Statement will be evaluated against a GRU consent designation.

A subset of records from each released GSR are also publicly available via the dbGaP Genome Browser and the [dbGaP FTP site](#) [10]. The publicly-available results include the GSR results with low p-values. They also do not include allele frequencies and direction of effect. To access the Genome Browser, click on the "Analyses" tab on the [main page for phs001974](#) [11], click on the analysis of interest in the sidebar on the right, and then click on "View association results in Genome Browser".

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References

NIH, NOT-OD-19-023: "Update to NIH Management of Genomic Summary Results Access" <https://grants.nih.gov/grants/guide/notice-files/NOT-OD-19-023.html> [7], November 01, 2018.

NIH, Genomic Data Sharing, <https://osp.od.nih.gov/scientific-sharing/genomic-data-sharing/> [12]

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Source URL (modified on 08/02/2021 -

1:50pm):<https://topmed.nhlbi.nih.gov/topmed-genomic-summary-results-public>

Links

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