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gtexr: A convenient R interface to the Genotype-Tissue Expression (GTEx) Portal API

Alasdair Neil Warwick [1,2], **Benjamin Zuckerman** [3], **Chuin Ying Ung** [4], **Robert Luben** [2,5], and **Abraham Olvera-Barrios** [2,5]

1 University College London Institute of Cardiovascular Science, London, UK **2** NIHR Biomedical Research Centre, Moorfields Eye Hospital NHS Foundation Trust **3** Centre for Rheumatic Diseases, King's College London, London, UK **4** Guy's and St Thomas' NHS Foundation Trust **5** University College London Institute of Ophthalmology, London, UK

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Software

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Editor: Arfon Smith

Reviewers:

- @ernestguevarra
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Summary

The Genotype-Tissue Expression (GTEx) project (GTEx Consortium, 2013, 2020) is a comprehensive public resource for studying tissue-specific gene expression and regulation in human tissues. Through systematic analysis of RNA sequencing data from 54 non-diseased tissue sites across nearly 1000 individuals, GTEx provides crucial insights into the relationship between genetic variation and gene expression. This data is accessible through the GTEx Portal API (GTEx Consortium, 2023), enabling programmatic access to human gene expression data.

The `gtexr` package (Warwick et al., 2024) provides a convenient R interface to the GTEx Portal API (v2), making this valuable resource more accessible to researchers. It offers a suite of R functions with intuitive naming conventions that mirror the complete set of API endpoints. The package includes comprehensive documentation with working examples for all functions, and is complemented by an interactive web application that allows users to explore the API functionality through a graphical interface. Common use cases include retrieving quantitative trait loci data for specific genes or variants, accessing tissue-specific expression data, and converting between different identifier systems (e.g., gene symbols to GENCODE IDs). Results are returned in tabular format, facilitating integration with downstream analysis pipelines and allowing researchers to focus on their analyses rather than API implementation details.

Statement of need

GTEx data is extensively used across diverse areas of biomedical research, from basic regulatory genomics to clinical applications. For example, researchers use GTEx to investigate tissue-specific gene expression patterns, interpret genome-wide association study findings, and infer causal relationships between gene expression and disease. While the GTEx Portal provides a web interface for data access, many analyses require programmatic access to query multiple genes, variants, or tissues systematically.

Current tools for programmatic GTEx access are limited to either downloading bulk data releases or require significant programming expertise to construct API queries. `gtexr` (Warwick et al., 2024) fills this gap by providing researchers with a straightforward interface to query GTEx data programmatically.

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