

Battle, A. et al. Characterizing the genetic basis of transcriptome diversity through RNA-sequencing of 922 individuals. *Genome Res.* 24, 14–24 (2014).

“Here, we provide a direct window into the regulatory consequences of genetic variation by sequencing RNA from 922 genotyped individuals... We detect variants influencing expression of over ten thousand genes, and through the enhanced resolution offered by RNA-sequencing... We have sequenced RNA from whole blood of 922 genotyped individuals from the Depression Genes and Networks cohort (Methods), all of European ancestry”

Depression Genes and Networks study (National Institute of Mental Health Grant 5RC2MH089916) was designed to collect samples of individuals with and without major depressive disorder, ages 21–60, from a survey research panel that was broadly representative of the United States population.

Data

Genotype, raw RNA-seq, quantified expression, and covariate data are available by application through the NIMH Center for Collaborative Genomic Studies on Mental Disorders. Instructions for requesting access to data can be found at:

https://www.nimhgenetics.org/access_data_biomaterial.php,

Inquiries should reference the “Depression Genes and Networks study (D. Levinson, PI)”.

For convenience, significant QTLs (FDR 0.05) are available for download here.

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Zhernakova, Daria V., et al. "Identification of context-dependent expression quantitative trait loci in whole blood." *Nature genetics* 49.1 (2017): 139-145.

Here we generated peripheral blood RNA-seq data from 2,116 unrelated individuals and systematically identified context-dependent eQTLs using a hypothesis-free strategy that does not require previous knowledge of the identity of the modifiers. Of the 23,060 significant cis-regulated genes (false discovery rate (FDR) ≤ 0.05), 2,743 (12%) showed context-dependent eQTL effects. The majority of these effects were influenced by cell type composition... We created the Biobank-Based Integrative Omics Study (BIOS) data set by sequencing whole peripheral blood mRNA in 2,116 healthy adults from four Dutch cohorts.

Data availability:

Raw RNA-seq data can be obtained from the European Genome-phenome Archive (EGA; accession EGAD00001001623).

No access to download

Please log in before attempting to download data from the EGA. If you do not have an EGA account and want to request access, contact information for the DAC responsible for access to this data is on the right under the heading 'Who controls access to this dataset'.

eQTL results can be accessed via our web browser (<http://genenetwork.nl/biosqtlbrowser/>). -- dc2all: the server is down

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Mostafavi, Sara, et al. "Type I interferon signaling genes in recurrent major depression: increased expression detected by whole-blood RNA sequencing." *Molecular psychiatry* 19.12 (2014): 1267-1274.

"We recruited a European ancestry sample of 463 individuals with recurrent major depressive disorder and 459 controls, and ... sequenced RNA from whole blood ..."

"We present here the largest whole-transcriptome study of MDD to date and the first using RNA-seq. After collecting psychiatric, demographic, environmental and medical information, we studied 922 European ancestry individuals (463 cases and 459 controls) with RNA-seq of whole-blood RNA and with a GWAS assay... Complete genotype and RNA-seq data are available form <https://nimhgenetics.org>."

processing:

"Only uniquely aligned reads were used to quantify gene expression levels. Reads were assigned to 21 578 of the 22 339 annotated protein-coding genes (NCBI v37). Analyses included 13 857 autosomal genes with ≥ 100 individuals with ≥ 10 reads (in total from anywhere across the transcript). Effects of technical covariates (for example, per individual 5' bias, GC bias, sequencing depth, and percent globin reads) and biological covariates (for example, estimates of blood cell-type proportions and time of day of blood draw; see Supplementary Table S1 for complete list) were removed by ridge regression of logarithm-transformed read counts. Cell-type proportions were inferred using a method based on non-negative least squares,¹⁷ making use of external microarray data on cell-type specific expression signatures¹⁸ (Supplementary Methods)."

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Zhernakova, Daria V., et al. "DeepSAGE reveals genetic variants associated with alternative polyadenylation and expression of coding and non-coding transcripts." *PLoS genetics* 9.6 (2013): e1003594.

Zhernakova ref in Westra paper:

"... investigated the 3' UTR using Deep Serial Analysis of Gene Expression (DeepSAGE) next-generation RNA sequencing data from 94 peripheral blood samples."

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Jansen, R., et al. "Gene expression in major depressive disorder." *Molecular psychiatry* 21.3 (2016): 339-347.

"We measured gene expression in peripheral blood from 1848 subjects from The Netherlands Study of Depression and Anxiety. Subjects were divided into current MDD (N=882), remitted MDD (N=635) and control (N=331) groups."

"Gene expression in peripheral venous blood was assayed at RUCDR Infinite Biologics (<http://www.rucdr.org/>). Our prior paper contains full details. ... Gene expression profiles were determined using Affymetrix U219 arrays ..."

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Zhang, Xiaoling, et al. "Identification of common genetic variants controlling transcript isoform variation in human whole blood." *Nature genetics* 47.4 (2015): 345-352.

We performed genome-wide screening to identify SNPs that might control mRNA splicing in whole blood collected from 5,257 Framingham Heart Study participants.

Others (less useful):

Shin, Heesun, et al. "Variation in RNA-Seq transcriptome profiles of peripheral whole blood from healthy individuals with and without globin depletion." *PLoS One* 9.3 (2014): e91041.

"...we designed a study of a small collection of biological replicates (6 healthy individuals; 3 males, 3 females), as well as 6 technical replicates created from pooled total RNA extracted from peripheral whole blood across all 6 biological samples"

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Dorr, Casey, et al. "Differentially expressed gene transcripts using RNA sequencing from the blood of immunosuppressed kidney allograft recipients." *PloS one* 10.5 (2015): e0125045.

"...DEGs were determined in 32 adult kidney recipients"

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Wright, Fred A., et al. "Heritability and genomics of gene expression in peripheral blood." *Nature genetics* 46.5 (2014): 430-437.

Uses array-based gene expression data