

GWAS resources

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Learning outcomes

- Get acquainted with databases that store GWAS summary statistics
 - Disease traits
 - Quantitative traits
 - Ancestry specific
 - Metabolomics

GWAS Catalogue



GWAS Catalog

The NHGRI-EBI Catalog of human genome-wide association studies



Examples: [breast carcinoma](#), [rs7329174](#), [Yao](#), [2q37.1](#), [HBS1L](#), [6:160000000-250000000](#)

Coming soon! New format for GWAS summary statistics.
Read all about it in our [blog](#) and [preprint](#).

Download

Download a full copy of the GWAS Catalog in spreadsheet format as well as current and older versions of the GWAS diagram in SVG format.

Summary statistics

Documentation and access to full summary statistics for GWAS Catalog studies where available.

Submit

Submit summary statistics to GWAS Catalog.

<https://www.ebi.ac.uk/gwas/>

Meta-analysis of sub-Saharan African studies provides insights into genetic architecture of lipid traits

Ananyo Choudhury ^{# 1}, Jean-Tristan Brandenburg ^{# 2}, Tinashe Chikowore ^{2 3}, Dhriti Sengupta ², Palwende Romuald Boua ^{2 4}, Nigel J Crowther ⁵, Godfred Agongo ^{6 7}, Gershim Asiki ⁸, F Xavier Gómez-Olivé ⁹, Isaac Kisiangani ⁸, Eric Maimela ¹⁰, Matshane Masemola-Maphutha ¹¹, Lisa K Micklesfield ³, Engelbert A Nonterah ⁶, Shane A Norris ³, Hermann Sorgho ⁴, Halidou Tinto ⁴, Stephen Tollman ⁹, Sarah E Graham ¹², Cristen J Willer ^{12 13 14}; AWI-Gen study; H3Africa Consortium; Scott Hazelhurst ^{2 15}, Michèle Ramsay ^{16 17}

Data availability

Go to: ▶

The full dataset generated in this study is in the EGA [<https://ega-archive.org/>] database under the study accession code [EGAS00001002482](#). This includes the phenotype dataset [EGAD00001006425](#) and the genotype dataset [EGAD00010001996](#). These datasets are available subject to controlled access through the Data and Biospecimen Access Committee of the H3Africa Consortium. The processed data generated in this study are provided in [Supplementary Information](#) and Supplementary Data. Summary statistics reported in the paper are accessible on GWAS Catalog (<https://www.ebi.ac.uk/gwas/>) at the accession numbers: [GCST90101741](#), GCST90101742, GCST90101743, GCST9010174, GCST90101745, GCST90101746, GCST90101747, GCST90101748. All data that support the findings of this study are available from the corresponding authors on request. Publicly available datasets included in the study are the following: 1000 Genomes Project Phase 3 (<ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp>), UGR meta-analysis summary statistics, GLGC summary Statistics, PAGE consortium summary statistics available at GWAS Catalog (<https://www.ebi.ac.uk/gwas/>) and gnomAD (<https://gnomad.broadinstitute.org/>).

Query output

Search results for *GCST90101741*

GCST90101741

Meta-analysis of sub-Saharan African studies provides insights into genetic architecture of lipid traits.

Associations 



Meta-analysis of sub-Saharan African studies provides insights into genetic architecture of lipid traits.

Choudhury A et al. 2022 Nat Commun PMID:35546142

Associations  Studies 



Study description

Study information

Reported trait	Low density lipoprotein cholesterol levels	Trait(s)	low density lipoprotein cholesterol measurement
Genotyping technology	Genome-wide genotyping array	Background Trait(s) ⓘ	-
Discovery sample description	10,389 Sub-Saharan African ancestry individuals	Platform [SNPs passing QC]	Illumina [13900000] (imputed)
Discovery ancestry (country of recruitment)	10389 Sub-Saharan African (Burkina Faso, South Africa, Ghana, Kenya)	Replication sample description	NA
PubMed ID	35546142	Replication ancestry (country of recruitment)	NR
First author	Choudhury A ⓘ	Journal	Nat Commun
		Publication date ⓘ	2022-05-11

Full Summary Statistics

[FTP Download](#) 

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[CC0](#)

Title Meta-analysis of sub-Saharan African studies provides insights into genetic architecture of lipid traits.

Authors Choudhury A ⓘ , Brandenburg JT ⓘ , Chikowore T, Sengupta D, Boua PR... [Show more >](#)

Download link

Index of /pub/databases/gwas/summary_statistics/GCST90101001-GCST90102000/GCST90101741

<u>Name</u>	<u>Last modified</u>	<u>Size</u>	<u>Description</u>
 Parent Directory		-	
 GCST90101741_buildGRCh37.tsv.gz	2022-05-14 00:04	323M	
 README.txt	2022-05-14 00:04	1.4K	
 harmonised/	2022-07-11 23:14	-	
 md5sum.txt	2022-05-14 00:04	109	

Downloading the data

```
wget http://ftp.ebi.ac.uk/pub/databases/gwas/summary_statistics/GCST90101001-  
GCST90102000/GCST90101741/GCST90101741_buildGRCh37.tsv.gz
```

```
[tinash@cream-ce CHAIR]$ zcat GCST90101741_buildGRCh37.tsv.gz | head
```

variant_id	p_value	chromosome	base_pair_location	beta	standard_error	effect_allele	other_allele	effect_allele_frequency
rs144434834	0.844322	1	723918 -0.00561981	0.0221663	G	0.890893		
rs111533735	0.928312	1	724103 0.00240934	0.0175528	A	0.809232		
rs1222283314	0.989279	1	724169 0.000988917	0.0390022	A	0.9630523		
rs12069907	0.315583	1	724300 -0.0170174	0.0154994	C	0.711316		
rs28692873	0.219178	1	724324 -0.0607959	0.0672038	G	0.9838896		
rs578177050	0.283721	1	725211 0.0638079	0.0546656	C	0.981092		
rs111203397	0.0819076	1	725286 0.108617	0.0702693	G	0.9820085		
rs1408251607	0.128085	1	725499 -0.05659	0.0455097	A	0.9571677		
rs28454925	0.375904	1	726794 -0.0450522	0.0635831	C	0.9838896		

```
[tinash@cream-ce CHAIR]$
```


Metabolomics and GWAS

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Human Metabolic Individuality

A blog about metabolomics, genomics, and where the two fields meet.

A Table of all published GWAS with metabolomics

by karsten | posted in: Resources

Here you find references to all published mGWAS in humans, ordered by publication date. If a study is missing from this list, please let me know. Please note that I do not apply a strict inclusion criterion for what I would call a GWAS with metabolomics, or rather a GWAS with biochemical traits (such as a GWAS with cholesterol levels). The latter are well covered by the [GWAS catalogue](#).

This table was initially published in [Kastenmüller et al., Genetics of human metabolism: an update. Hum. Mol. Genet. 2015](#) and has been **updated as of 20 January 2023**. If you are interested in associations at specific loci you may also use the [SNIPA](#) block annotation tool [[read this post](#)], and if you are looking for associations specific to certain metabolites, try the [Metabolomics GWAS Server](#) [[read this post](#)].

<http://www.metabolomix.com/list-of-all-published-gwas-with-metabolomics/>

UK Biobank



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Pan-UK Biobank

Pan-ancestry genetic analysis of the UK Biobank

The UK Biobank is a collection of a half million individuals with paired genetic and phenotype information that has been enormously valuable in studies of genetic etiology for common diseases and traits. However, most genome-wide analyses of this dataset use only the European ancestry individuals. Analyzing a more inclusive and diverse dataset increases power and improves the potential for discovery. Here, we present a multi-ancestry analysis of 7,228 phenotypes, across 6 continental ancestry groups, for a total of 16,131 genome-wide association studies. We release these summary statistics freely to the community ahead of publication.

<https://pan.ukbb.broadinstitute.org/>

GLGC



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STATISTICAL GENETICS

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Global Lipids Genetics Consortium Results

Result Files

[Ancestry-specific GWAS summary statistics for HDL-C, LDL-C, nonHDL-C, TC and TG \(90 files plus README\)](#)

[Trans-ancestry GWAS summary statistics for HDL-C, LDL-C, nonHDL-C, TC and C \(20 files plus README\)](#)

[LDL-C polygenic score weights for ALL, Admixed African, East Asian, European, Hispanic and South Asian ancestries \(12 files plus README\)](#)

[Trans-ancestry credible sets for HDL-C, LDL-C, nonHDL-C, TC and TG \(5 files plus README\)](#)

[Noncoding variant prioritization at lipid loci](#)

[ChrX GWAS summary statistics for HDL-C, LDL-C, nonHDL-C, TC and TG \(180 files\)](#)

[Sex-specific GWAS summary statistics for HDL-C, LDL-C, nonHDL-C, TC and TG \(20 files\)](#)

Reference

Graham et al. (2021) The power of genetic diversity in genome-wide association studies of lipids. In press at Nature.

Kanoni et al. medRxiv (2021) Implicating genes, pleiotropy and sexual dimorphism at blood lipid loci through multi-ancestry meta-analysis.

Ramdas et al. bioRxiv (2021) A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids.

Description

The Global Lipids Genetics Consortium aggregated GWAS results from 1,654,960 individuals from 201 primary studies representing five genetic ancestry groups: Admixed African or African (AdmAFR, N=99.4k, 6.0% of sample), East Asian (EAS, N=146.5k, 8.9%), European (EUR, N=1.32m, 79.8%), Hispanic (HIS, N=48.1k, 2.9%), and South Asian (SAS, N=41.0k, 2.5%). We performed GWAS for five blood lipid traits: low-density lipoprotein cholesterol (LDL-C), high-density lipoprotein cholesterol (HDL-C), triglycerides (TG), total cholesterol (TC), and non-high-density lipoprotein cholesterol (nonHDL-C). We performed meta-analysis within each ancestry using RAREMETAL. For the meta-analysis of all ancestries we performed meta-analysis of single cohort files using MR-MEGA to account for heterogeneity in variant effect sizes on lipids between ancestry groups. For the all ancestry analysis, fixed-effects meta-analysis was carried out with METAL to estimate effect sizes and se. The meta-analysis summary statistics

<https://csg.sph.umich.edu/willer/public/glglc-lipids2021/>

GIANT



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GIANT consortium

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GIANT: Genetic Investigation of ANthropometric Traits

The **Genetic Investigation of ANthropometric Traits (GIANT)** consortium is an international collaboration that seeks to identify genetic loci that modulate human body size and shape, including height and measures of obesity. The GIANT consortium is a collaboration between investigators from many different groups, institutions, countries, and studies, and the results represent their combined efforts. The primary approach has been meta-analysis of genome-wide association data and other large-scale genetic data sets. Anthropometric traits that have been studied by GIANT include body mass index (BMI), height, and traits related to waist circumference (such as waist-hip ratio adjusted for BMI, or WHRadjBMI). Thus far, the GIANT consortium has identified common genetic variants at hundreds of loci that are associated with anthropometric traits.

Data Release

We are releasing the summary data from our meta-analyses of GWAS data, in order to enable other researchers to examine particular variants or loci for their evidence of association with anthropometric traits. The files include p-values and direction of effect at over 2 million directly genotyped or imputed single nucleotide polymorphisms (SNPs). To prevent the possibility of identification of individuals from these summary results, we are not releasing allele frequency data from our samples.

[Click here to access the Summarized Genome-Wide Meta-analysis data files](#)

[Click here to create regional association plots from GIANT data using LocusZoom](#)

Then select *"Plot Using Published GWAS Results"*

https://portals.broadinstitute.org/collaboration/giant/index.php/GIANT_consortium_data_files

MAGIC



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Ongoing projects

MAGIC (the Meta-Analyses of Glucose and Insulin-related traits Consortium)

MAGIC (the Meta-Analyses of Glucose and Insulin-related traits Consortium) represents a collaborative effort to combine data from multiple GWAS to identify additional loci that impact on glycemic and metabolic traits.

MAGIC investigators have initially studied fasting glucose, fasting insulin, 2h glucose and HBA1c, as well as performed meta-analysis of more sophisticated measures of insulin secretion and sensitivity. Through these efforts, dozens of loci influencing these traits have been identified, a subset of which also influence risk of type 2 diabetes.

We are releasing results from our meta-analyses and these can be accessed through the data downloads pages.

<https://magicinvestigators.org/>

Next steps

