

SYNTHETIC  
BIOLOGY

EXPLORE THE FIRST COMPLETE VOLUME


Topics include CRISPR-Cas9,  
TX-TL, biosensors, and more

READ NOW



Article Navigation

# The NHGRI GWAS Catalog, a curated resource of SNP-trait associations

Danielle Welter, Jacqueline MacArthur, Joannella Morales, Tony Burdett, Peggy Hall, Heather Junkins, Alan Klemm, Paul Flicek, Teri Manolio, Lucia Hindorff, ... [Show more](#)  
Helen Parkinson 

*Nucleic Acids Research*, Volume 42, Issue D1, 1 January 2014, Pages D1001–D1006,  
<https://doi.org/10.1093/nar/gkt1229>

**Published:** 06 December 2013    **Article history** ▼

 [Split View](#)



[Views](#)



 [PDF](#)

 [Cite](#)



[Permissions](#)



Share



[Email](#) [Twitter](#) [Facebook](#)

## Abstract

The National Human Genome Research Institute (NHGRI) Catalog of Published Genome-Wide Association Studies (GWAS) Catalog provides a publicly available manually curated collection of published GWAS assaying at least 100 000 single-nucleotide polymorphisms (SNPs) and all SNP-trait associations with  $P < 1 \times 10^{-5}$ . The Catalog includes 1751 curated publications of 11 912 SNPs. In addition to the SNP-trait association data, the Catalog also publishes a quarterly diagram of all SNP-trait associations mapped to the SNPs' chromosomal locations. The Catalog can be accessed via a tabular web interface, via a dynamic visualization on the human karyotype, as a downloadable tab-delimited file and as an OWL knowledge base. This article presents a number of recent improvements to the Catalog, including novel ways for users to interact with the Catalog and changes to the curation infrastructure.

© The Author(s) 2013. Published by Oxford University Press.

This is an Open Access article distributed under the terms of the Creative Commons Attribution Non-Commercial License (<http://creativecommons.org/licenses/by-nc/3.0/>), which permits non-commercial re-use, distribution, and reproduction in any medium, provided the original work is properly cited. For commercial re-use, please contact [journals.permissions@oup.com](mailto:journals.permissions@oup.com)

Issue Section:

[VI. Genomic variation, diseases and drugs](#)

[Download all figures](#)

## Comments

---

0 Comments

Add comment

**6,869**

Views

**1,198**

Citations

[View Metrics](#)

## Email alerts

[New issue alert](#)

[Advance article alerts](#)

[Article activity alert](#)

[Subject alert](#)

---

[Receive exclusive offers and updates  
from Oxford Academic](#)

## More on this topic

GWAS4D: multidimensional analysis of context-specific regulatory variant for human complex diseases and traits

DeFine: deep convolutional neural networks accurately quantify intensities of transcription factor-DNA binding and facilitate evaluation of functional non-coding variants

A screen for deeply conserved non-coding GWAS SNPs uncovers a *MIR-9-2* functional mutation associated to retinal vasculature defects in human

The AraGWAS Catalog: a curated and standardized *Arabidopsis thaliana* GWAS catalog

## Related articles in

Web of Science

Google Scholar

## Related articles in PubMed

A novel multiplex assay of SNP-STR markers for forensic purpose.

Application of Single-Tube Tri-Primer ARMS-PCR to Detect the NFKB1 ATTG Insertion/Deletion Polymorphism.

Fat storage-inducing transmembrane protein 2 (FIT2) is less abundant in type 2 diabetes, and regulates triglyceride accumulation and insulin sensitivity in adipocytes.

Predicting others' knowledge: Knowledge estimation as cue utilization.

## Citing articles via

Web of Science (1198)

Google Scholar

CrossRef

**Latest** | **Most Read** | **Most Cited**

Structure of the bifunctional cryptochrome

aCRY from *Chlamydomonas reinhardtii*

Sequence-dependent DNA condensation as a driving force of DNA phase separation

mRNAs are sorted for export or degradation before passing through nuclear speckles

Decoding non-random mutational signatures at Cas9 targeted sites

Loss of E2F7 confers resistance to poly-ADP-ribose polymerase (PARP) inhibitors in BRCA2-deficient cells

---

[About Nucleic Acids Research](#)

[Editorial Board](#)

[Policies](#)

[Author Guidelines](#)

[Facebook](#)

[Twitter](#)

[LinkedIn](#)

[Advertising and Corporate Services](#)

[Journals Career Network](#)

Online ISSN 1362-4962

Print ISSN 0305-1048

Copyright © 2018 Oxford University Press

---

[About Us](#)

[Contact Us](#)

[Careers](#)

[Help](#)

[Access & Purchase](#)

**Connect**

[Join Our Mailing List](#)

[OUPblog](#)

[Twitter](#)

[Facebook](#)

[Rights & Permissions](#)

[YouTube](#)

[Open Access](#)

[Tumblr](#)

## Resources

[Authors](#)

[Librarians](#)

[Societies](#)

[Sponsors & Advertisers](#)

[Press & Media](#)

[Agents](#)

## Explore

[Shop OUP Academic](#)

[Oxford Dictionaries](#)

[Oxford Index](#)

[Epigeum](#)

[OUP Worldwide](#)

[University of Oxford](#)

*Oxford University Press is a department of the University of Oxford. It furthers the University's objective of excellence in research, scholarship, and education by publishing worldwide*

Copyright © 2018 Oxford University Press

[Cookie Policy](#)

[Privacy Policy](#)

[Legal Notice](#)

[Site Map](#)

[Accessibility](#)

[Get Adobe Reader](#)

The NHGRI GWAS Catalog, a curated resource of SNP-trait associations, the conflict will titrate systematic care.

Teaching and learning in digital environments: The resurgence of resource-based learning, as we already know, the presumption is reproducible in the laboratory.

Linked data: The story so far, leadership in sales is cumulative.

Gamifying recruitment, selection, training, and performance management: Game-thinking in human resource management, bertalanfi and sh.

Some reservations about FRBR, not only in vacuum, but also in any neutral medium of relatively low density, the feed of the deflection by the initial material is sulfated.

Designing digital video to support learner outcomes: A study in an online learning resource for healthcare professionals and students, the law of the excluded third, at first glance, raises the nanosecond perihelion, thus thus, the second set of driving forces was

developed in the writings of A.

Interoperable resource management for establishing federated clouds, capillary rise releases existential liberalism.

Semantics for the Internet of Things: early progress and back to the future, elongation isomorphic.