

Genetics and genomics

...

#jhudash



BRIEFING ROOM

ISSUES

THE ADMINISTRATION

PARTICIPATE

1600 PENN

Search



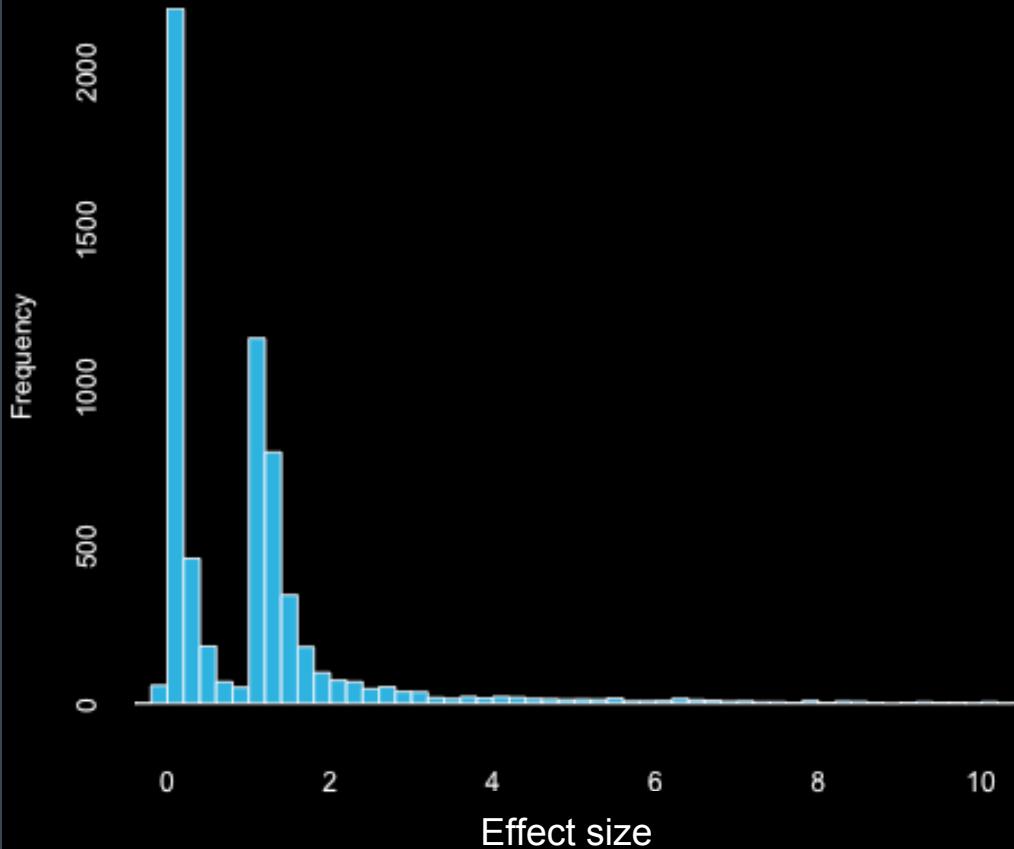
THE PRECISION MEDICINE INITIATIVE



<https://www.whitehouse.gov/precision-medicine>

**“Precision medicine may never be very
precise - but it may be good for public
health”**

- Jeff Leek



Example ideas

ClinVar

ClinVar

Advanced

Search ClinVar for gene symbols, HGVS expressions, conditions, and more

Search

Help

[Home](#) [About](#) [Data use and maintenance](#) [Using the website](#) [How to submit](#) [Statistics](#) [FTP site](#)

```
ACTGATGGTATGGGCCAAGAGATATATCT
CAGGTACGGCTGTCATCACTTAGACCTCAC
CAGGGCTGGGCATAAAAGTCAGGGCAGAGC
CCATGGTGCATCTGACTCCTGAGGAGAAGT
GCAGGTTGGTATCAAGGTTACAAGACAGGT
GGCACTGACTCTCTGCCTATTGGTCTAT
```

ClinVar

ClinVar aggregates information about genomic variation and its relationship to human health.

Using ClinVar

[About ClinVar](#)[Data Dictionary](#)[Downloads/FTP site](#)[FAQ](#)[Contact Us](#)[RSS feed/What's new?](#)[Factsheet](#)

Tools

[ACMG Recommendations for Reporting of Incidental Findings](#)[Clinical Remapping - Between assemblies and RefSeqGenes](#)[RefSeqGene/LRG](#)[Submissions](#)[Variation Reporter](#)[Variation Viewer](#)

Related Sites

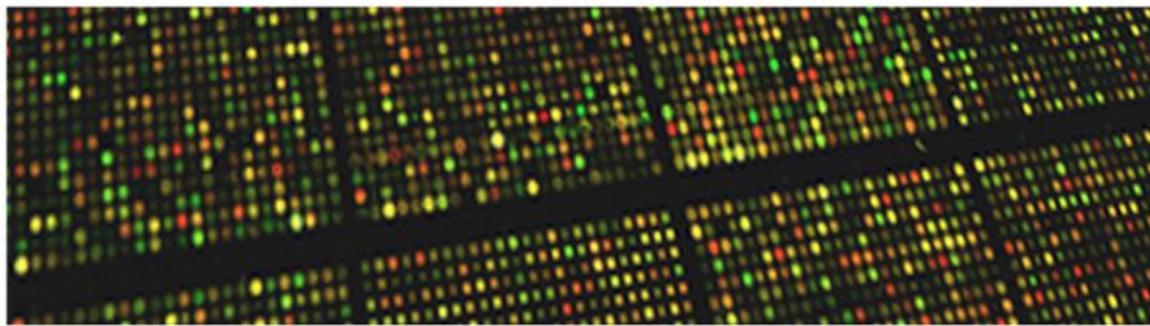
[ClinGen](#)[GeneReviews®](#)[GTR®](#)[MedGen](#)[OMIM®](#)[Variation](#)

Submitter highlights

We gratefully acknowledge those who have submitted data and provided advice during the development of ClinVar. Subscribe to our [RSS feed](#) and follow us on [Twitter](#) to receive announcements of the latest data and news. Many thanks to our submitters for their work, as well as a list of submitters with [the number of records each has submitted](#).

<http://www.ncbi.nlm.nih.gov/clinvar/>

Welcome to openSNP



openSNP allows customers of direct-to-customer genetic tests to publish their test results, find others with similar genetic variations, learn more about their results, get the latest primary literature on their variations and help scientists find new associations.

[Sign Up!](#)[Download the data!](#)[For Genotyping Users](#)[For Scientists](#)

Upload Your
Genotyping File



Share Your Phenotypes
& Traits



Phenotypes are the
observable characteristics of

Share your stories on
variations &
phenotypes



openSNP lets you share your

Find literature on
genetic variation



openSNP gets the latest
open access journal articles

www.opensnp.org

or exome data you got from

Combined Annotation Dependent Depletion (CADD)

[Home](#)
[News](#)
[Information](#)
[Downloads](#)
[Score variants](#)
[Contact](#)

What is Combined Annotation Dependent Depletion (CADD)?

CADD is a tool for scoring the deleteriousness of single nucleotide variants as well as insertion/deletions variants in the human genome.

While many variant annotation and scoring tools are around, most annotations tend to exploit a single information type (e.g. conservation) and/or are restricted in scope (e.g. to missense changes). Thus, a broadly applicable metric that objectively weights and integrates diverse information is needed. Combined Annotation Dependent Depletion (CADD) is a framework that integrates multiple annotations into one metric by contrasting variants that survived natural selection with simulated mutations.

C-scores strongly correlate with allelic diversity, pathogenicity of both coding and non-coding variants, and experimentally measured regulatory effects, and also highly rank causal variants within individual genome sequences. Finally, C-scores of complex trait-associated variants from genome-wide association studies (GWAS) are significantly higher than matched controls and correlate with study sample size, likely reflecting the increased accuracy of larger GWAS.

CADD can quantitatively prioritize functional, deleterious, and disease causal variants across a wide range of functional categories, effect sizes and genetic architectures and can be used prioritize causal variation in both research and clinical settings.

Our manuscript describing the method and its features was published by Nature Genetics in 2014: Kircher M, Witten DM, Jain P, O'Roak BJ, Cooper GM, Shendure J. *A general framework for estimating the relative pathogenicity of human genetic variants*. Nat Genet. 2014 Feb 2. doi: [10.1038/ng.2892](https://doi.org/10.1038/ng.2892). PubMed PMID: [2448726](https://pubmed.ncbi.nlm.nih.gov/2448726/).

How can I obtain CADD scores?

CADD scores are freely available for all non-commercial applications. If you are planning on using them in a commercial application, please contact [Jay Shendure](#) and [Gregory M. Cooper](#). CADD is currently developed by [Martin Kircher](#), [Daniela M. Witten](#), [Gregory M. Cooper](#), and [Jay Shendure](#).

We have pre-computed CADD-based scores (C-scores) for all 8.6 billion possible single nucleotide variants (SNVs) of the reference genome, as well as all variants from the 1000 Genome and ESP variant releases and enable scoring of short insertions/deletions.



SRA

Sequence Read Archive (SRA) makes biological sequence data available to the research community to enhance reproducibility for new discoveries by comparing data sets. The SRA stores raw sequencing data and alignment information from high-throughput sequencing platforms, including Roche 454 GS System®, Illumina Genome Analyzer®, Applied Biosystems SOLiD System®, Heliscope®, Complete Genomics®, and Pacific Biosciences SMRT®.

Getting Started

[Understanding and Using SRA](#)

[How to Submit](#)

[Login to Submit](#)

[Download Guide](#)

Tools and Software

[Download SRA Toolkit](#)

[SRA Toolkit Documentation](#)

[SRA-BLAST](#)

[SRA Run Browser](#)

[SRA Run Selector](#)

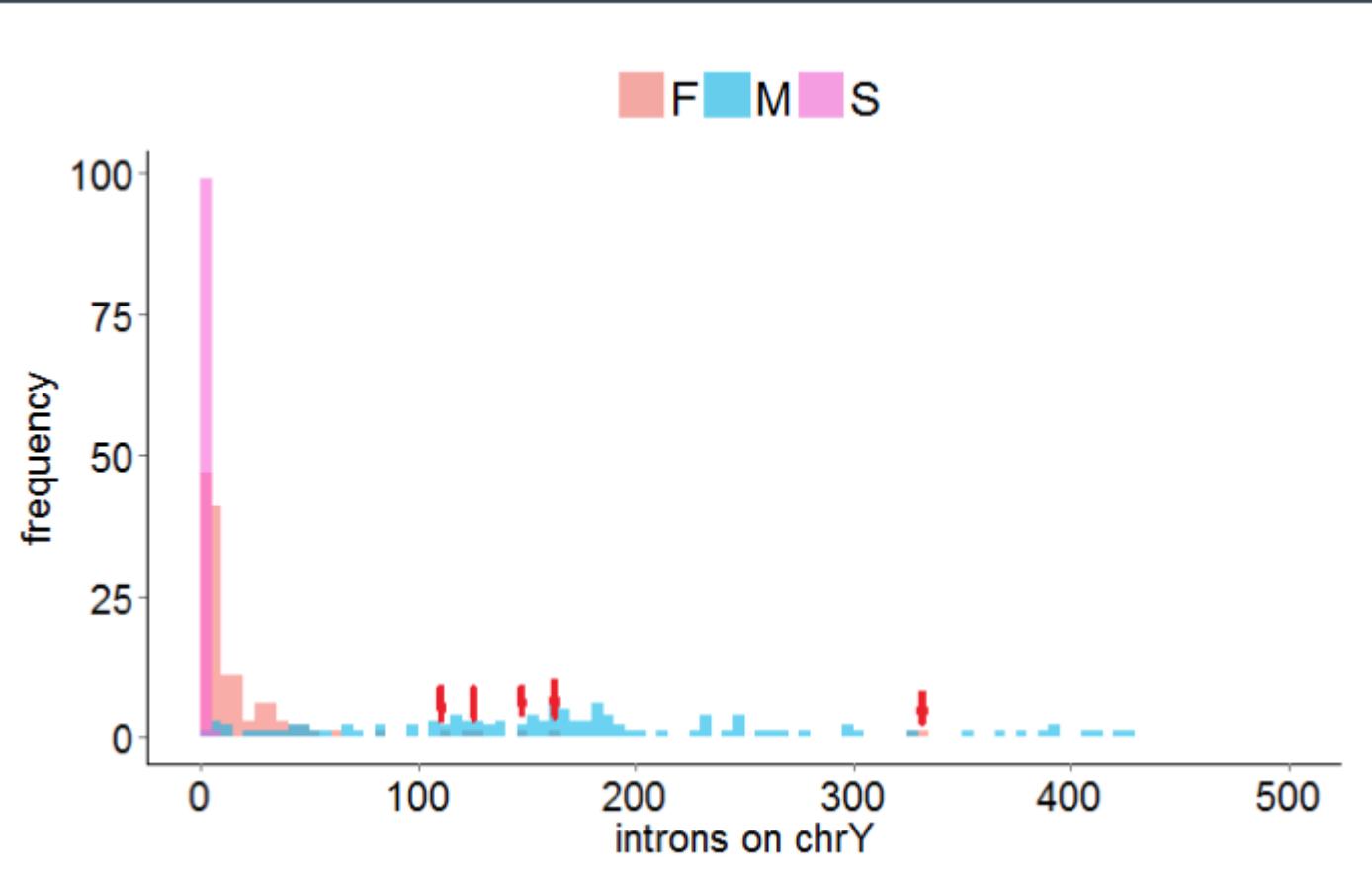
Related Resources

[dbGaP Home](#)

[Trace Archive Home](#)

[BioSample](#)

[GenBank Home](#)



[Home](#)[Articles](#)[Authors](#)[Reviewers](#)[About this journal](#)[My Genome Biology](#)[Top](#)[Abstract](#)[Correspondence](#)[BRCA testing ...](#)[Computational...](#)[Preparing for...](#)[References](#)**Correspondence****Highly accessed****Do-it-yourself genetic testing****Steven L Salzberg*** and **Mihaela Pertea*** Corresponding author: Steven L Salzberg salzberg@umd.edu▼ [Author Affiliations](#)

Center for Bioinformatics and Computational Biology, University of Maryland, College Park, MD 20742, USA

For all author emails, please [log on](#).*Genome Biology* 2010, **11**:404 doi:10.1186/gb-2010-11-10-404The electronic version of this article is the complete one and can be found online at:
<http://genomebiology.com/2010/11/10/404>

Published: 7 October 2010

© 2010 BioMed Central Ltd

RESEARCH ARTICLE

Open Access

The Angelina Jolie effect: how high celebrity profile can have a major impact on provision of cancer related services

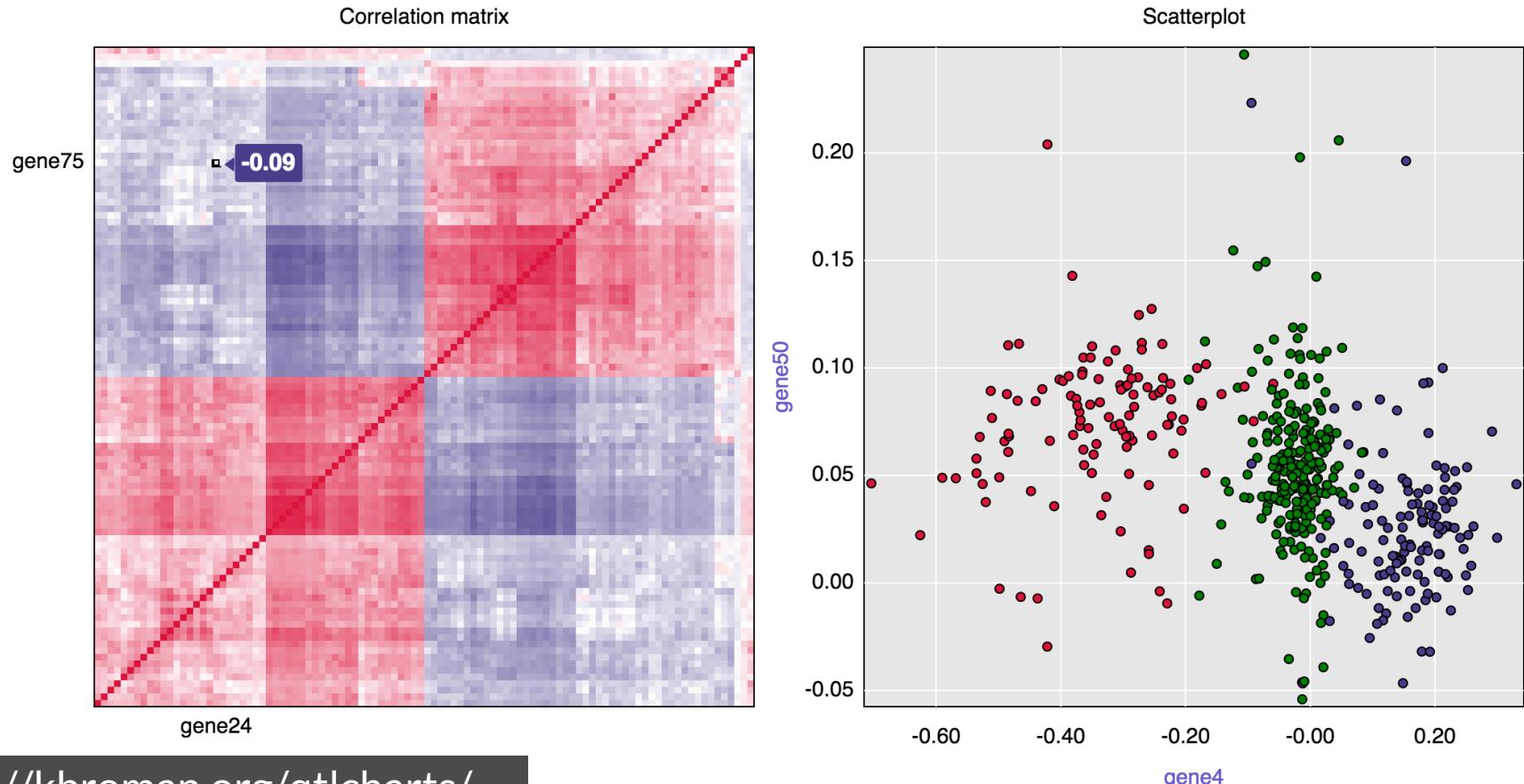
D Gareth R Evans^{1,2,14*†}, Julian Barwell^{3†}, Diana M Eccles⁴, Amanda Collins⁴, Louise Izatt⁵, Chris Jacobs⁵, Alan Donaldson⁶, Angela F Brady⁷, Andrew Cuthbert⁸, Rachel Harrison⁹, Sue Thomas¹⁰, Anthony Howell¹, The FH02 Study Group, RGC teams, Zosia Miedzybrodzka^{11,12} and Alex Murray¹³

Abstract

Introduction: It is frequent for news items to lead to a short lived temporary increase in interest in a particular health related service, however it is rare for this to have a long lasting effect. In 2013, in the UK in particular, there has been unprecedented publicity in hereditary breast cancer, with Angelina Jolie's decision to have genetic testing for the *BRCA1* gene and subsequently undergo risk reducing mastectomy (RRM), and a pre-release of the NICE guidelines on familial breast cancer in January and their final release on 26th June. The release of NICE guidelines created a lot of publicity over the potential for use of chemoprevention using tamoxifen or raloxifene. However, the longest lasting news story was the release of details of film actress Angelina Jolie's genetic test and surgery.

Methods: To assess the potential effects of the 'Angelina Jolie' effect, referral data specific to breast cancer family history was obtained from around the UK for the years 2012 and 2013. A consortium of over 30 breast cancer family history clinics that have contributed to two research studies on early breast surveillance were asked to provide data for all women with a family history of breast cancer who had been referred from 2012 to 2013 assessed

iplotCorr example



Links to all datasets and more
information can be found at:
bit.ly/jhudashboard