



Two GWAS Databases

CGEMS at NCI

dbGaP at NCBI



CGEMS

Cancer Genetic Markers of Susceptibility

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Cancer Genetic Markers of Susceptibility Project

The Cancer Genetic Markers of Susceptibility (CGEMS) is a three-year, \$14 million initiative that will identify genetic alterations that make people susceptible to prostate and breast cancer. Scientists involved will use DNA available from five large studies of prostate cancer and five large studies of breast cancer to “scan” the genome for common genetic variations between patients who have these cancers and controls who do not have cancer.

[Learn more >>](#)

[Background](#)

Spotlight

[Cancer Genetic Markers of Susceptibility \(CGEMS\)](#)

February 13, 2006

NCI begins studies to identify genetic risk factors for prostate and breast cancer. [more](#)

[DCEG and CGF Collaborate on CGEMS Initiative](#)

CGEMS: caBIG Posting Pre-Computed Analysis



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 **CGEMS**
Cancer Genetic Markers of Susceptibility

  Division of Cancer Epidemiology and Genetics

Home | Browse Data | Bulk Data Download | Feedback



This is the home page of the Cancer Genetic Markers of Susceptibility (CGEMS) data access. The following links provide information on the [project](#) and [background](#). The CGEMS study design uses cases and controls drawn from well designed epidemiological studies of prostate and breast cancer. DNA from these subjects is being used to generate genotypes to perform a Genome-Wide Association Study (GWAS) on over 500,000 genetic variants to determine their role in cancer susceptibility.

CGEMS Prostate Scan Phase 1

A GWAS has been conducted in a large, national study in the U.S.A., the Prostate, Lung, Colorectal, and Ovary study ([PLCO](#)). The analysis includes 1,177 subjects who developed prostate cancer during the observational period and 1,105 individuals who did not develop prostate cancer during the same time period. The prostate scan is being conducted in two parts, Phase 1A and Phase 1B

The data generated from these scans can be accessed through this portal. The first posting includes data from Phase 1A of the prostate cancer scan and includes:


- Association test results for over 300,000 SNPs
- Frequency and descriptive statistics on these SNPs
- Individual phenotypic and genotypic data for the study participants and control samples. Note that these data can only be made available to eligible investigators after a registration process (link).

The results of Phase 1B will be available in February 2007.

[Browse Data](#) [Bulk Data Download](#)

For more information on:

- [About CGEMS Study](#)
- [How to use the CGEMS data portal](#)
- [Register to access raw data](#)

 Click the question mark icon for context sensitive help throughout the application.


CGEMS updates:


- This release, Version 1.0, was deployed on Oct 10, 2006.
- The current dataset in use was deployed on Oct 10, 2006


**Pre-computed Analysis
No Restrictions for
Publication**


**Raw Genotype
Case/control
Age (in 5 yrs)
Registered Access**

<http://cgems.cancer.gov/data>

 National Cancer Institute U.S. National Institutes of Health | www.cancer.gov

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Cancer Genetic Markers of Susceptibility


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Welcome Browse Data Bulk Data Download Support Feedback Login/Register

Browse Data:

Select a Study: 


- CGEMS Breast Cancer WGAS Phase 1
- CGEMS Prostate Cancer WGAS Phase 1
- CGEMS Prostate Cancer WGAS Phase 1A

Study Description:

There are multiple studies available. Please review the context-sensitive description when you use the list above to select a specific study.

Select a Dataset:

- Association Finding
- Population Frequency
- Subjects Data (login required)

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Study: CGEMS Breast Cancer WGAS Phase 1

Search Association Findings by:

Analysis Name:



Analysis Abstract: [\[Analysis Description \(PDF\)\]](#)

There are multiple analyses of the data in this study. Please review the context-sensitive abstract when you use the dropdown list above to select a specific analysis.

AND

Panel:

Genomic Location:

Chromosome

Based on NCBI Genome build 35

From bp

To bp

AND

HUGO Gene Symbol List

Fill in list of HUGO symbols (ex. MET) or upload a new line separated list file with max. 100 gene symbols via Browse... button

Enter Symbols

Upload File no file selected

AND

dbSNP Identifier List

Fill in list of dbSNP IDs (ex. rs38840) or upload a new line separated list file with max. 1000 dbSNP ids via Browse... button

Enter IDs

Upload File: no file selected

AND

p-value

<=

Whole Genome Rank

<=





National Cancer Institute

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Cancer Genetic Markers of Susceptibility



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Study: CGEMS Prostate Cancer WGAS Phase 1A

SNP Association Finding Report - (19 results)

| dbSNP ID | Chromosome | Physical Position (bp) | Associated Genes | Analysis Name | p-value | Whole Genome Rank |
|----------------------------|------------|------------------------|------------------|---|----------|-------------------|
| rs12334695 | 8 | 128523110 | | Incidence density sampling, Adjusted score test | 0.025361 | 7583 |
| rs7012462 | 8 | 128526872 | | Incidence density sampling, Adjusted score test | 0.61895 | 187681 |
| rs4871791 | 8 | 128527826 | | Incidence density sampling, Adjusted score test | 0.569441 | 172475 |
| rs6470517 | 8 | 128529586 | | Incidence density sampling, Adjusted score test | 0.353344 | 106901 |
| rs7841228 | 8 | 128530060 | | Incidence density sampling, Adjusted score test | 0.753514 | 228046 |
| rs7841264 | 8 | 128535996 | | Incidence density sampling, Adjusted score test | 0.101898 | 30853 |
| rs1447293 | 8 | 128541502 | | Incidence density sampling, Adjusted score test | 0.026153 | 7829 |
| rs921146 | 8 | 128544367 | | Incidence density sampling, Adjusted score test | 0.109914 | 33365 |
| rs4871799 | 8 | 128551824 | | Incidence density sampling, Adjusted score test | 0.069611 | 21001 |
| rs1447295 | 8 | 128554220 | | Incidence density sampling, Adjusted score test | 4.16E-4 | 149 |
| rs9297758 | 8 | 128555770 | | Incidence density sampling, Adjusted score test | 0.572839 | 173461 |
| rs6985504 | 8 | 128565958 | | Incidence density sampling, Adjusted score test | 0.281571 | 85131 |
| rs12155672 | 8 | 128576206 | | Incidence density sampling, Adjusted score test | 0.282398 | 85399 |
| rs1562432 | 8 | 128576784 | | Incidence density sampling, Adjusted score test | 0.285649 | 86401 |
| rs4242382 | 8 | 128586755 | | Incidence density sampling, Adjusted score test | 9.6E-5 | 38 |
| rs7017300 | 8 | 128594450 | | Incidence density sampling, Adjusted score test | 1.58E-4 | 67 |
| rs7837688 | 8 | 128608542 | | Incidence density sampling, Adjusted score test | 3.8E-5 | 19 |
| rs6991990 | 8 | 128614565 | | Incidence density sampling, Adjusted score test | 0.106728 | 32421 |
| rs4407842 | 8 | 128619305 | | Incidence density sampling, Adjusted score test | 0.854811 | 258529 |

Association Tests

8q24

Scan 1A

<http://cgems.cancer.gov>

NCBI



dbGaP Current Activities

- Genetic Association Information Network (GAIN)
- Genetics and Environment Initiative (GEI)
- Framingham Genetic Study
- National Institute for Neurological Disease and Stroke (NINDS)
- NHGRI Medical Resequencing
- NEI Macular Degeneration
- Control sets

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Finding dbGaP

<http://view.ncbi.nlm.nih.gov/dbGaP>

Entrez PubMed - Windows Internet Explorer
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By Studies By Diseases Advanced Search

| Disease | Studies | Variables | Documents | Participants | Type of Study | Status |
|---|---------|-----------|-----------|--------------|---------------|-----------|
| + Cataract | 1 | - | - | - | | |
| + Macular Degeneration | 1 | - | - | - | | |
| - Parkinson Disease | 3 | - | - | - | | |
| NINDS Parkinsonism Study | - | 100 | 5 | 2573 | case-control | Completed |
| NINDS Parkinsonism Study - Cases | - | 40 | 3 | 1498 | case-control | Completed |
| NINDS Parkinsonism Study - Controls | - | 60 | 1 | 1075 | case-control | Completed |

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Study: National Eye Institute (NEI) Age-Related Eye ...

Conerger analysis of the in clinic and telephone batteries from the AREDS cognitive function ancillary study. AREDS Report No. 15. Ophthalmic epidemiology 2005 Aug;12(4):

- [Yaffe K, Clemons TE, McBee WL, Lindblad AS, Age-Related Eye Disease Study Research Group](#)
Impact of antioxidants, zinc, and copper on cognition in the elderly: a randomized, controlled trial. Neurology 2004 Nov;63(9):

Diseases Related to This Study (MESH terms)

- [Macular Degeneration](#)
- [Cataract](#)

Principal Investigators

- Susan B. Bressler, Wilmer Eye Institute, The Johns Hopkins Medical Institutions, Baltimore, MD
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- Hemin Chin, The National Eye Institute
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Access Restrictions

Study: National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS) - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/study.cgi?id=phs000001

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Study: National Eye Institute (NEI) Age-Related Eye ...

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National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS)

ID: phs000001
Version: 1

National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS)

Description

The Age-Related Eye Disease Study (AREDS) was initially conceived as a long-term multicenter, prospective study of the clinical course of age-related macular degeneration (AMD) and age-related cataract. In addition to collecting natural history data, AREDS included a clinical trial of high-dose vitamin and mineral supplements for AMD and a clinical trial of high-dose vitamin supplements for cataract. Results from these clinical trials have been published. The two clinical trials generally shared 1 pool of participants (Figure 1). The clinical trials were initiated largely because of the widespread public use in the United States of commercially available pharmacologic doses of vitamins and minerals to treat these two eye conditions and the absence of definitive studies on the safety and efficacy of their use.

Eligible AREDS participants were age 55 to 80 years of age at enrollment and had to be free of any illness or condition that would make long-term follow-up or compliance with study medications unlikely or difficult. On the basis of fundus photographs graded by a central reading center, best-corrected visual acuity and ophthalmologic evaluations, participants were enrolled in one of several AMD.

It is hoped that this resource will help researchers better understand two important diseases that affect an aging population. The AREDS Research Group hopes that data from AREDS on progression rates and risk factors for AMD and cataract will further understanding of the clinical course of both conditions, generate hypotheses about etiology and aid in the design of clinical trials of potential interventions.

[AREDS, The National Eye Institute](#)

[AREDS, The EMMES Corporation](#)

- Subjects: 600
- Type: case-control
- Status: Completed

Inclusion/Exclusion Criteria

- [Inclusion/exclusion criteria](#)

Search Within This Study

Search for:

Associated Analyses

- NEI Age-Related Eye Disease Study (AREDS)
 - All [Genome-Wide Allelic Associatio...](#)

Associated Variables

- + NEI Age-Related Eye Disease Study (AREDS)

Associated Documents

- NEI Age-Related Eye Disease Study (AREDS)
 - + All
 - Manual of Operations
 - [Chapter 1: Background and rati...](#)
 - [Chapter 2: Literature review](#)
 - [Chapter 3: Study design](#)
 - [Chapter 4: Study rationalization](#)
 - [Chapter 5: Study policies](#)
 - [Chapter 6: Examination schedule](#)
 - [Chapter 7: Examination procedures](#)
 - [Chapter 8: Photographic proced...](#)
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 - [Chapter 12: Quality enhancement](#)
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NCBI dbGaP Document NEI Age-Related Eye Disease Study

Chapter 7 EXAMINATION PROCEDURES

7.1 INTRODUCTION

The procedures for carrying out the examinations required in the study are described in this chapter. Required ocular examinations include refraction and visual acuity measurements, intraocular pressure measurement, and ophthalmoscopic examination. General characteristic assessments include measurement of height, weight, and blood pressure and determination of past medical history. Risk factor assessments will require the administration of the food frequency and sunlight exposure questionnaires as well as collection of blood specimens. Procedures for participant identification, masking, distribution and management of the supplementation, adherence assessment, and home visit examination are also described. Procedures for taking photographs of the lens and fundus are described in detail in Chapter 8. The schedule and description of participant visits in Chapter 6 outline the examinations required during each visit.

7.2 REFRACTION AND VISUAL ACUITY

A manifest refraction and visual acuity measurement according to the detailed study protocol must be performed during (a) the Qualifying Visit when the visual acuity score using Chart R is 73 letters or less in at least one eye, (b) the Randomization Visit, (c) Annual Visits, and (d) any Nonannual Visit when the visual acuity score using Chart R has dropped by 10 letters or more compared to the Randomization Visit score for the first time. Participants' pupils should not be dilated at the time of visual acuity testing at any study visit; except they may be dilated during the Qualifying Visit. Pinhole acuity will not be tested as part of AREDS. At the Qualifying Visit, visual acuity may be initially assessed utilizing the participant's current distance glasses. At the Nonannual Visits, visual acuity is initially assessed utilizing the previously obtained manifest refraction. Participants will be asked to read the letters on Chart R only (not Charts 1 or 2), using the equipment described in Section 7.2.1. They will start reading from the top left-most letters--first with the right eye and then with the left eye. A visual acuity score will be calculated as described in Section 7.2.3.3. If at the Qualifying Visit

Pages

AREDS: NEI Age-Related Eye Disease Study

Comments Attachments

AREDS: NEI Ag

1 of 36

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Genotype and Phenotype - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=search&db=gap&term=systolic%20blood%20pressure&doptcmdl=SVaria

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Genotype and Phenotype

| | | |
|--|--|---------|
| dias04 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: dias04 | Sitting diastolic blood pressure (at follow-up year 4) | unknown |
| dias03 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: dias03 | Sitting diastolic blood pressure (at follow-up year 3) | unknown |
| dias00 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: dias00 | Sitting diastolic blood pressure (at follow-up year 0) | integer |
| syst13 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst13 | Sitting systolic blood pressure (at follow-up year 13) | unknown |
| syst12 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst12 | Sitting systolic blood pressure (at follow-up year 12) | integer |
| syst11 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst11 | Sitting systolic blood pressure (at follow-up year 11) | integer |
| syst10 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst10 | Sitting systolic blood pressure (at follow-up year 10) | integer |
| syst09 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst09 | Sitting systolic blood pressure (at follow-up year 9) | integer |
| syst08 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst08 | Sitting systolic blood pressure (at follow-up year 8) | integer |
| syst07 NEI Aqe-Related Eye Disease Study (AREDS) >> Variable: syst07 | Sitting systolic blood pressure (at follow-up year 7) | integer |

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Variable: syst07 - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/GetDocument.cgi?id=phd000007#Sec7.6

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Variable: syst07

"160"
"132"
"134"
"142"

Document Parts Related to Variable

AREDS: Chapter 7. EXAMINATION PROCEDURES - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/GetDocument.cgi?id=phd000007#Sec7.6

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AREDS: Chapter 7. EXAMINATION PROCEDURES

7.6: BLOOD PRESSURE MEASUREMENT

Variable(s) associated with this section: syst00 syst03 syst04 syst05 syst06 syst07 syst08 syst09 syst10 syst11 syst12 syst13 dias00 dias03 dias04 dias05 dias06 dias07 dias08 dias09 dias10 dias11 dias12 dias13

Blood pressure measurements will be taken by a certified examiner using a standard mercury sphygmomanometer. Instructions for preparing the participant, using the proper techniques, utilizing equipment, and measuring and recording the blood pressure are provided below. Some institutions have installed electronic automated sphygmomanometers. In the interest of data consistency, choice; however it is recognized that staff at those centers may have no alternative.

AREDS: BASELINE INTERVIEW — PHASE II - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/GetDocument.cgi?id=phd000020#V11

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AREDS: BASELINE INTERVIEW — PHASE II

of the Manual of Operations.):

a. Systolic (mmHg)

a. Diastolic (mmHg)

b. Certification number of blood pressure examiner:

9. Have you ever smoked cigarettes for a total of 6 months or more?

no
 yes

If no, skip to 10

- a. How old were you when you first started smoking?
- b. Over your lifetime of smoking, on the average, how many packs per day have you smoked?
- ≤ ½ pack
 > ½, ≤ 1 pack
 > 1, ≤ 2 packs
 > 2 packs
- c. Do you smoke cigarettes at present?
- no
 yes

If no, skip to e

- d. If you currently smoke, how many cigarettes a day do you smoke? Skip to 10
- e. If you do not smoke currently, how old were you when you last quit smoking?
10. Have you ever smoked cigars, a pipe, or chewed tobacco for a total of 6 months or more?
- no
 yes

If no, skip to 11

- a. How old were you when you first started smoking cigars, a pipe, or chewing tobacco?
- b. Do you now smoke cigars, a pipe, or chew tobacco?
- no
 yes

If yes skip to 11

- c. How old were you when you last quit smoking cigars, a pipe, or chewing tobacco?

th feet flat and on the floor and legs uncrossed, with the right arm bared, supported, could not have smoked, eaten, ingested caffeine or been exposed to exertion or cold for surement. The participant should be seated and quiet for at least 5 minutes prior to the talk while blood pressure is being taken.

to be tested. The rubber bladder should encircle at least two-thirds of the arm. If the re reading will be erroneously high; if it is too wide, the reading may be low. A cuff that the average adult arm.

nometer to measure the blood pressure. The mercury manometer must be handled he level of mercury in the tube should be observed with no pressure applied to the cuff. led to the reservoir to bring the edge of the mercury meniscus exactly to the zero mark. l manometer must be vertical for correct reading. Some mobile or floor-based mercury d at a reclined angle and the gradations are adjusted accordingly. It is important that e and its scale in the correct position. The tube of the mercury manometer should be f oxidation. Clogging in the air vent or filter at the top of the manometer tube will cause gishly to declining pressure in the bladder and will cause an erroneous reading. The filter least annually to ensure continued accuracy.

ing connections approximately 1 inch above natural crease of the inner aspect of elbow

latable inner bladder centered over area of brachial artery (medial surface of arm).

tached to the mercury column is away from the participant's body and that the tube e to the participant's body. Secure the wrapped cuff firmly by applying pressure on the e where it is applied to the cuff.

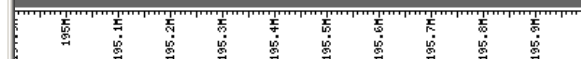
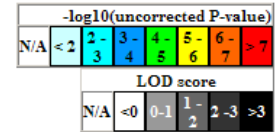
dbGaP study [phs000001](#): rs203674 Genotype Summary

| phv00000173 | Genotype | | | Genotype Frequency | | |
|-----------------------------|----------|-----|-----|--------------------|-----|-------|
| | AA | AC | CC | 25% | 50% | 75% |
| Case | 51 | 157 | 186 | 39.8% | | 47.2% |
| Control | 82 | 88 | 23 | 42.5% | | 45.6% |

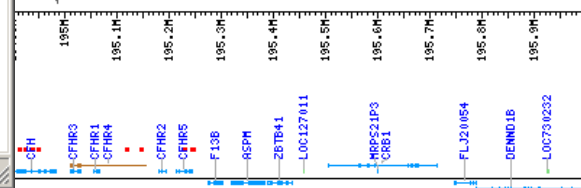
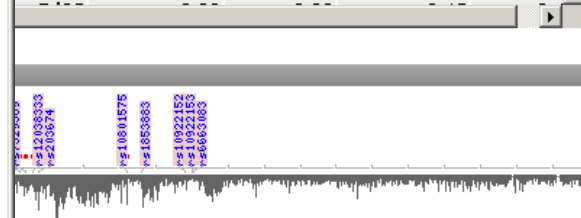
| phv00000173 | Allele | | Allele Frequency | | |
|-----------------------------|--------|-----|------------------|-----|-------|
| | A | C | 25% | 50% | 75% |
| Case | 259 | 529 | 32.9% | | 67.1% |
| Control | 252 | 134 | 65.3% | | 34.7% |

| phv00000173 | Number of Samples | | Success Rate | | |
|-----------------------------|-------------------|-------|--------------|-----|-----|
| | Genotyped | Total | 25% | 50% | 75% |
| Case | 394 | 395 | 99.7% | | |
| Control | 193 | 198 | 97.5% | | |

Case pHWE: 0.053
 Control pHWE: 1.000
 Odds ratio of minor allele 'A': 0.260
 Chi-square: 109.447
 p-value of Chi-square test: 1.29e-25



| Rank | MAF (control) | MAF (case) | HWE(control) | HWE(case) |
|-------|---------------|------------|--------------|-----------|
| 4/25 | 0.34 | 0.37 | 0.64 | 0.0 |
| 1/25 | 0.35 | 0.33 | 1.00 | 0.0 |
| 6/25 | 0.40 | 0.20 | 0.46 | 1.0 |
| 2/25 | 0.38 | 0.31 | 0.36 | 0.0 |
| 9/25 | 0.22 | 0.11 | 0.13 | 0.3 |
| 10/25 | 0.21 | 0.11 | 0.19 | 0.1 |



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GaP Chromosome Browser - Windows Internet Explorer

http://www.ncbi.nlm.nih.gov/SNP/GaP.cgi

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GaP Chromosome Browser

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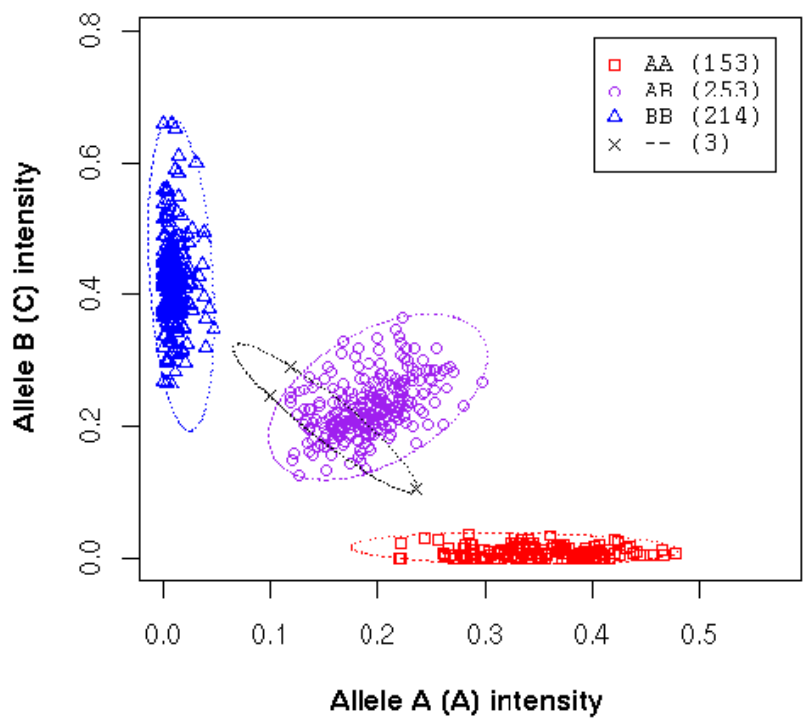
dbGaP GENOTYPE and PHENOTYPE

AREDS

| -log ₁₀ (uncorrected P-value) | | | | | | |
|--|----|-----|-----|-----|----|--|
| N/A | <2 | 2-3 | 4-5 | 6-7 | >7 | |
| LOD score | | | | | | |
| N/A | <0 | 0-1 | 1-2 | 2-3 | >3 | |

http://www.ncbi.nlm.nih.gov/projects/SNP/fetch_xy_density_plot.cgi?snp_id=203...
 http://www.ncbi.nlm.nih.gov/projects/SNP/fetch_xy_density_plot.cgi?snp_id=203674&display_id=1

AREDS (Illumina 100K) Normalized Allele Intensity for SNP rs203674 (623 individual samples)



reset Go

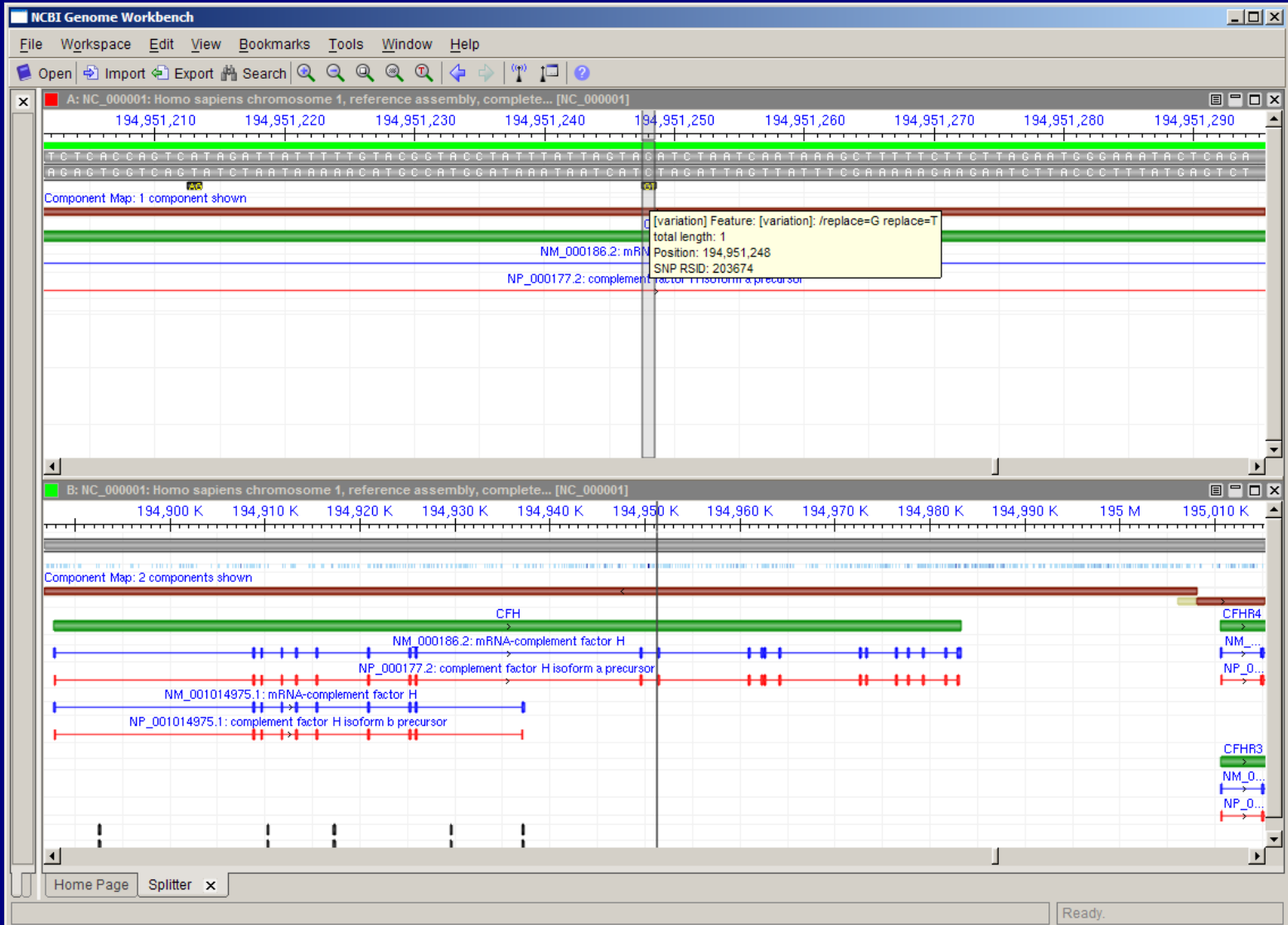
Status in Illumina 100k Chip | Univariate SNP Allelic Association Method

| Position | P-value | P-value Rank | MAF (control) | MAF (case) | HWE(control) | HWE(case) |
|----------|----------|--------------|---------------|------------|--------------|-----------|
| 9077 | 2.77e-20 | 4/25 | 0.34 | 0.37 | 0.64 | 0.0 |
| 1248 | 1.29e-25 | 1/25 | 0.35 | 0.33 | 1.00 | 0.0 |
| 9404 | 8.31e-14 | 6/25 | 0.40 | 0.20 | 0.46 | 1.0 |
| 3223 | 1.08e-22 | 2/25 | 0.38 | 0.31 | 0.36 | 0.0 |
| 5922 | 2.51e-06 | 9/25 | 0.22 | 0.11 | 0.13 | 0.3 |
| 5933 | 4.87e-06 | 10/25 | 0.21 | 0.11 | 0.19 | 0.1 |

Gene track showing SNPs: rs579515, rs7523689, rs2108393, rs2108374, rs1863883, rs1863883, rs10922153, rs10321153, rs66633083.

Gene track showing genes: KONT2, CFH, CFHR3, CFHR1, CFHR4, CFHR2, CFHR5, F13B, HSPH, ZBTB41, LOC127011, RPS21P3, CNG1, FLJ121054, DENND1B, LOC730322.

Associations to the Basepair



http://dbGaP.nih.gov



dbGaP Authorized Access is the management portal for individual-level data.

Authorized users can use this site to submit a data access request, manage access requests, and download approved data sets.

[Log in](#) to the Authorized access system.

Help

In order to apply for authorized access to dbGaP studies you must have one of the following accounts:

- ♦ [eRA Commons](#) (for NIH Extramural principal investigators, grantees, or other extramural investigators)
- ♦ [NIH Login](#) (for intramural NIH scientists and staff)

Who can apply? NIH is committed to respecting the privacy and intentions of research participants with regard to how data pertaining to their individual information is used. Data access is therefore intended only for scientific investigators pursuing research questions that are consistent with the informed consent agreements provided by individual research participants. Furthermore, investigators provided access will be expected to utilize appropriate data security measures (<http://www.ncbi.nlm.nih.gov/projects/gap/pdf/dbGaPLevel2SecurityProcedures.pdf>)

What is an authorized user within the data access request system? Authorized users are the Principal Investigators who may request data sets for specific research uses, the Institutional Signing Officials from the PI's home organization who certify and submit such requests, and the NIH staff who will review and process requests (e.g., members of the Data Access Committees).

dbGaP also maintains a help desk to assist investigators, institutional signing officials and NIH staff with authorized access management, and answer any questions related to the application process. Contact the help desk with your queries.

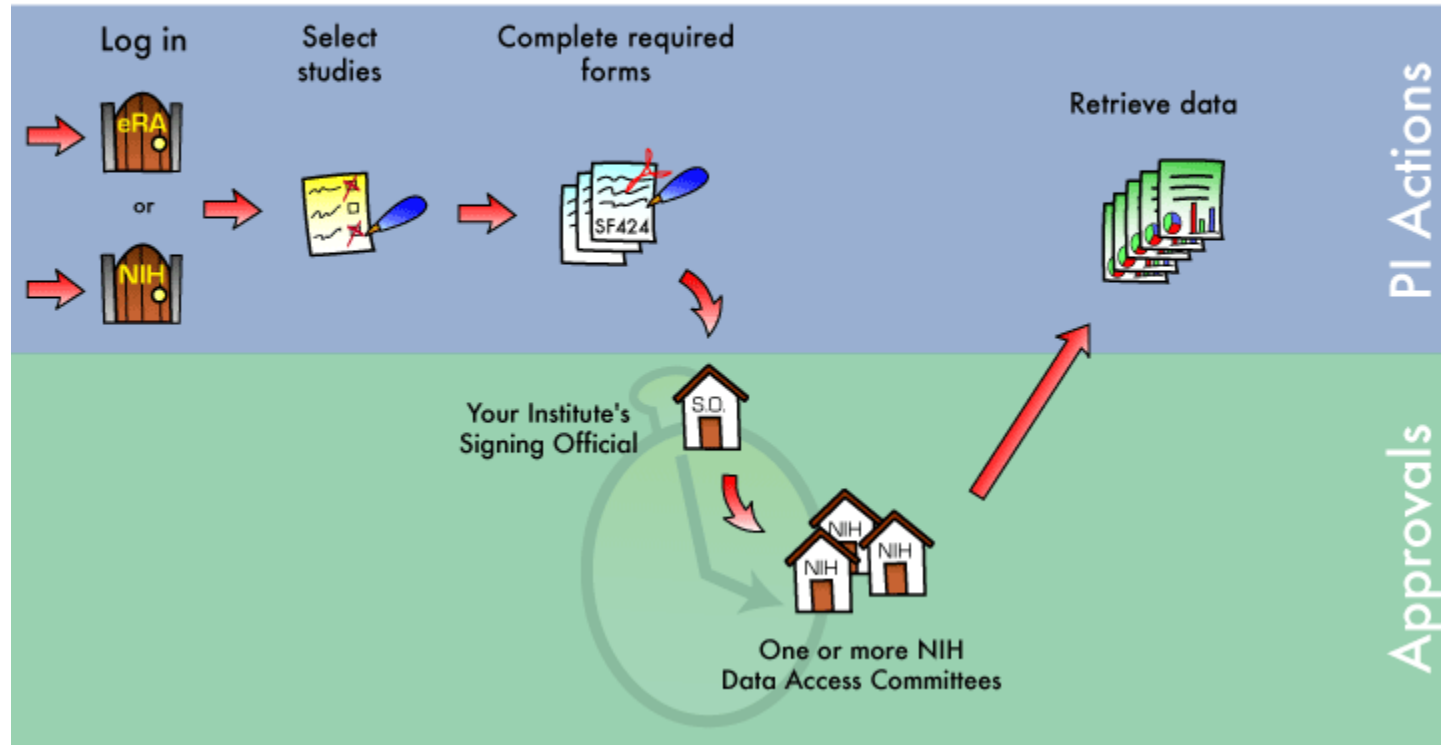
The system is a service of NCBI. Please [contact us](#) with any questions.

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[Privacy Notice](#) | [Disclaimer](#) | [Accessibility](#)

[National Institute of Health](#) | [United States Department of Health and Human Services](#) | [FirstGov.gov: The U.S. Government's Official Web Portal](#)

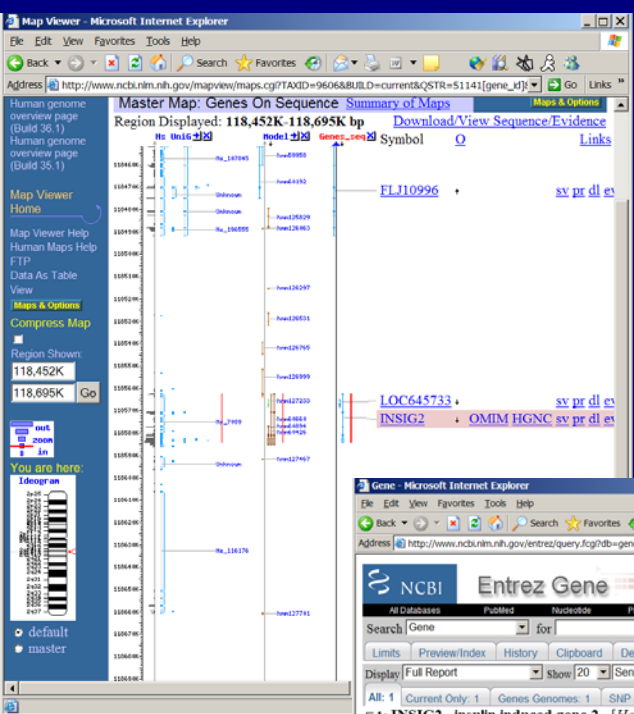
The dbGaP data request process



- Data access requires request from PI and approval by institutional Signing Official (SD for NIH) – Requester must have PI role.
- Uses NIH login for authentication
- Data Access Request uses SF424 R&R form with Co-PI form and Statement of Intent
- Data is downloaded via Web with Commons account controlling access to data.



Closing the Loop



Entrez GEO - Gene Expression Omnibus - Microsoft Internet Explorer
Address: http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene&cmd=Display&dopt=gene_geo&from_uid=51141
Search: GEO Profiles for
Display: Summary Show 20
Items 1 - 20 of 221
1: GDS501 record | GPL371 27849 [Homo Annotation: INSIG2: insulin induced gene 2 (Reporter: AL080184) Experiment: Inflammatory cytokine effect on gene expression array-based log
2: GDS1048 record | GPL564 1001267094 [Homo Annotation: INSIG2: insulin induced gene 2 (Reporter: AL080184) Experiment: Lymphoblastoid cell lines from C

Entrez PubMed - Microsoft Internet Explorer
Address: http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?cmd=Retrieve&db=pubmed&dopt=Abstract&list_uids=1
Search: PubMed for
Display: Abstract Show 20 Sort by
All: 1 Review: 0
1: Science. 2006 Apr 14;312(5771):279-83. Related Articles, Links

Entrez Gene - Microsoft Internet Explorer
Address: http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene&cmd=Retrieve&dopt=full_report&list_uids=51141
Search: Gene for
Display: Full Report Show 20 Send to
All: 1 Current Only: 1 Genes Genomes: 1 SNP GeneView: 1
1: INSIG2 insulin induced gene 2 [Homo sapiens] updated 19-Apr-2006
GenID: 51141 Primary source: HGNC:20452
Summary
Official Symbol: INSIG2 and Name: insulin induced gene 2 provided by HUGO Gene Nomenclature Committee
See related: HPRD-16361, MIM-608660
Gene type: protein coding
Gene name: INSIG2
Gene description: insulin induced gene 2
RefSeq status: Reviewed
Organism: Homo sapiens
Lineage: Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorhini; Catarrhini; Hominoidea; Homo
Gene aliases: MGC26273
Summary: The protein encoded by this gene is highly similar to the protein product encoded by gene INSIG1. Both INSIG1 protein and this protein are endoplasmic reticulum proteins that block the processing of sterol regulatory element binding proteins (SREBPs) by binding to SREBP cleavage-activating protein (SCAP), and thus prevent SCAP from escorting SREBPs to the Golgi.
Genomic regions, transcripts, and products
RefSeq below

A common genetic variant is associated with adult and childhood obesity.

Herbert A, Gerry NP, McQueen MB, Heid IM, Pfeuffer A, Illig T, Wichmann HE, Meitinger T, Hunter D, Hu FB, Colditz G, Hinney A, Hebebrand J, Koberwitz K, Zhu X, Cooper R, Ardlie K, Lyon H, Hirschhorn JN, Laird NM, Lenburg ME, Lange C, Christman MF.

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Obesity is a heritable trait and a risk factor for many common diseases such as type 2 diabetes, heart disease, and hypertension. We used a dense whole-genome scan of DNA samples from the Framingham Heart Study participants to identify a common genetic variant near the INSIG2 gene associated with obesity. We have replicated the finding in four separate samples composed of individuals of Western European ancestry, African Americans, and children. The obesity-predisposing genotype is present in 10% of individuals. Our study suggests that common genetic polymorphisms are important determinants of obesity.