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Critical Database Support

- DAC and more for Orphan Studies
- Consents and pro-active Policy
- Sequencing, Epigenetics, CNV
- ? Facilitate Structured Review of Clinical Significance of variant alleles
- Registration of Genetic Test Reagents

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Building on Data in dbGaP

- Collect on investments made through new associations on deposited data.
- Provide a stable, public base for a functional exploration of emerging phenotype standards.
- Provide new approaches to allele calling, population stratification, imputation.
- Increase power by combining controls or less well characterized phenotypes.

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dbGaP Captures Existing Phenotype Investment As Is

The screenshot displays a web browser window with the URL <http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/study.cgi?id=phs000001>. The page features the NCBI logo, the dbGaP logo (Genotype and Phenotype), and the AREDS logo. The main heading is "National Eye Institute (NEI) Age-Related Eye Disease Study (AREDS)" with the accession number "phs000001.v1.p1".

Description

The Age-Related Eye Disease Study (AREDS) was initially designed as a long-term multi-center, 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. AREDS participants were 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, over 4,700 participants were enrolled in one of several AMD categories, including persons with no AMD.

The clinical trials for AMD and cataract were conducted concurrently. AREDS participants were followed on the clinical trial for a median time of 6.5 years. Subsequent to the conclusion of the clinical trial, participants were followed for an additional 5 years and natural history data were collected. The AREDS research design is detailed in AREDS Report 1. AREDS Report 8 contains the mainline results from the AMD trial; AREDS Report 9 contains the results of the cataract trial. Blood samples were also collected for genetic research. Genetic samples from 600 AREDS participants were evaluated with a genome-wide scan for inclusion in the dbGaP.

It is hoped that this resource will better help researchers understand two important diseases that affect an aging population. These data may be applied to examination and inference on genetic and genetic-environmental bases for age-related diseases of public health significance and may also help elucidate the clinical course of both conditions, generate hypotheses, and aid in the design of clinical trials of preventive interventions.

[AREDS, The National Eye Institute](#)

[AREDS, The EMMES Corporation](#)

• Subjects: 600

Search Within This Study

Search for: Go

Associated Analyses

- NEI Age-Related Eye Disease Study (AREDS)
- AMD status

Associated Variables

- Physical Observations
 - Clinical Examination
 - Organ Systems
 - Eye
- rpscscore
- rpscbase
- rpsc
- rpscbase

Associated Documents

- NEI Age-Related Eye Disease Study

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Original Semantics in Protocols and Questionnaires

The image shows a screenshot of a web browser displaying the NCBI dbGaP document for the NEI Age-Related Eye Disease Study, Chapter 7: Examination Procedures. The browser window is titled "Chapter 7. EXAMINATION PROCEDURES (dbGaP ID: phd000007) - Windows Internet Explorer". The address bar shows the URL "http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/GetDocument.cgi?id=phd000007". The document content is displayed in a PDF viewer, showing the title "NCBI dbGaP Document NEI Age-Related Eye Disease Study" and the chapter title "Chapter 7 EXAMINATION PROCEDURES". The document is divided into sections, with "7.1 INTRODUCTION" and "7.2 REFRACTION AND VISUAL ACUITY" visible. The text in the introduction describes the procedures for carrying out the examinations required in the study, including required ocular examinations, general characteristic assessments, and risk factor assessments. The text in section 7.2 describes the procedure for manifest refraction and visual acuity measurement.

Chapter 7. EXAMINATION PROCEDURES (dbGaP ID: phd000007) - Windows Internet Explorer

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

Chapter 7. EXAMINATION PROCEDURES (dbGaP I...

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NEI Age-Related Eye Disease Study

Chapter 7: EXAMINATION PROCEDURES

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- 7.1 INTRODUCTION
- 7.2 REFRACTION AND VISUAL ACUITY
- 7.3 INTRAOCULAR PRESSURE MEASUREMENT
- 7.4 PUPIL DILATION
- 7.5 HEIGHT AND WEIGHT MEASUREMENT
- 7.6 BLOOD PRESSURE MEASUREMENT
- 7.7 NUTRITION AND SUNLIGHT EXPOSURE QUESTIONNAIRES
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- 7.9 BLOOD SPECIMEN COLLECTION
- 7.10 PARTICIPANT IDENTIFICATION, MASKING, DISTRIBUTION AND MANAGEMENT OF THE SUPPLEMENTATION, ADHERENCE ASSESSMENT, AND HOME VISIT EXAMINATION
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- 7.18 REPORTING
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- 7.22 END-OF-STUDY
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7.1. INTRODUCTION

7.2 REFRACTION AND VISUAL ACUITY

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.

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

commented that 2 sets each of 2.5% Neoination, the ophthalmologist examines the fundus, slit-lamp biomicroscopy. Ocular photography should face and impair the quality of photographs.

procedures are set forth below.

recorded with indoor clothing only.

g weight balances are set at "zero" and the scale

m the scale.

ght and hands relaxed and at your side, and your

ds).

Done

Unknown Zone | Protected Mode: On

Internet | Protected Mode: On

100%

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Stable Public IDs Provide A Base of Real Data For Standardized Measures

Chapter 7. EXAMINATION PROCEDURES (dbGaP ID: phd000007) - Windows Internet Explorer

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

7.6. BLOOD PRESSURE MEASUREMENT

Blood pressure measurements will be taken by a certified examiner preparing the participant, using the proper techniques, utilizing equipment provided below. Some institutions have installed electronic automated standard mercury units are the instruments of choice; however it is

7.6.1. Participant Preparation

1. The participant should be seated with feet flat and on the floor positioned at heart level and should not have smoked, eaten, or drunk 30 minutes prior to the measurement. The participant should be requested not to talk while blood pressure measurement is being taken.
2. Choose appropriate cuff size for arm to be tested. The rubber cuff is too narrow, the blood pressure reading will be erroneously high. A 12 cm wide is satisfactory for the average adult arm.

7.6.2. Technique

1. Use a standard mercury sphygmomanometer to measure the blood pressure. To avoid loss of mercury. The level of mercury in the tube should be necessary, mercury should be added to the reservoir to bring the column of the usual desk or wall manometer must be vertical. Manometers are designed to be read at a reclined angle and the instrument be used with the tube and its scale in the correct position. Inspected regularly for dirt or sign of oxidation. Clogging in the mercury column to respond sluggishly to declining pressure in the vent should be serviced at least annually to ensure continued accuracy.
2. Place lower edge of cuff with its tubing connections approximately 2.5 cm above antecubital space.
3. Wrap cuff snugly about arm with inflatable inner bladder centered over the brachial artery.
4. Be sure that the connecting tube attached to the mercury column is close to the participant's body and the locking fabric fastener over the area where it is applied to the cuff.
5. Attach the cuff connection and inflate the cuff while palpating the radial artery. Inflate the cuff to a pressure 20 mmHg above the systolic pressure. Deflate the cuff slowly (2-3 mmHg per second) and note the pressure at which the sound disappears. This is the diastolic pressure. Repeat the procedure for the other arm.

Variable: syst12 - Windows Internet Explorer

BASELINE INTERVIEW — PHASE II (dbGaP ID: phd000020) - Windows Internet Explorer

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

I would like to take your blood pressure now and again later during this interview.

8. Sitting blood pressure. (Participant must have been seated and quiet for at least 5 minutes prior to the measurement. See Section 7.6 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

Internet | Protected Mode: On 100%

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Standards Emerge from Utility

Variable: syst12 - Windows Internet Explorer
http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/variable.cgi?id=phv00000094

NCBI dbGaP GENOTYPE and PHENOTYPE AREDS

syst12
Accession: phv00000094.v1.p1
>> [NEI Age-Related Eye Disease Study \(AREDS\)](#) >> syst12

Description
Sitting systolic blood pressure (at follow-up year 12)

Variable: L_ANKLE - Windows Internet Explorer
http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/variable.cgi?id=phv00021576

NCBI dbGaP GENOTYPE and PHENOTYPE BOSTON UNIVERSITY National Heart Lung and Blood Institute People Science Health

L_ANKLE
Accession: phv00021576.v1.p1
>> [Framingham SHARE](#) >> [Framingham SHARE Ankle Arm BP](#) >> L_ANKLE

Description
SYSTOLIC BLOOD PRESSURE BY DOPPLER IN LEFT ANKLE

Variable: SYSBP - Windows Internet Explorer
http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/variable.cgi?id=phv00019997

NCBI dbGaP GENOTYPE and PHENOTYPE GENETIC ASSOCIATION INFORMATION NETWORK GoKID Genetics of Kidneys in Diabetes Study

SYSBP
Accession: phv00019997.v1.p1
>> [Search for Susceptibility Genes for Diabetic Nephropathy in Type 1 Diabetes](#) >> SYSBP

Description
Systolic blood pressure (mmHg)

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Standards Emerge from Utility

Recommendations for Blood Pressure Measurement in Humans and Experimental Animals: Part 1: Blood Pressure Measurement in Humans: A Statement for Professionals From the Subcommittee of Professional and Clinical Practitioners of the American Heart Association. *Hypertension*. 2005;45:142-161. doi: 10.1161/01.HYP.0000150859.47929.8e

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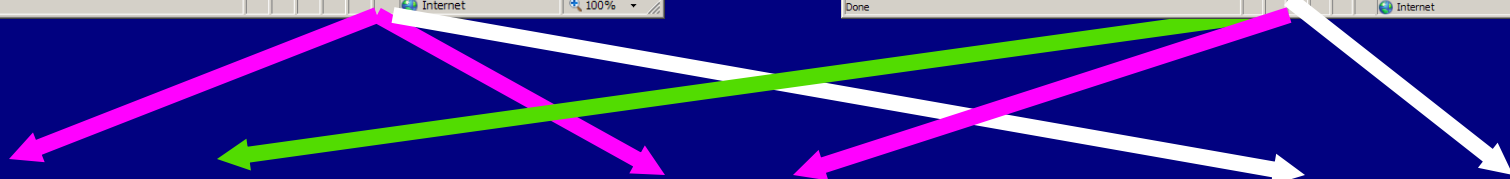
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Google Scholar

- Articles by Ward, M.
- Articles by Langton, J. A.

PubMed



sys12
Accession: phv000000094.v1.p1
>> NEI Age-Related Eye Disease Study (AREDS) -> sys12

Description
Sitting systolic blood pressure (at follow-up year 12)

L_ANKLE
Accession: phv00021576.v1.p1
>> Erasmigham SHARe -> Erasmigham SHARe Arkle Arm BP -> L_ANKLE

Description
SYSTOLIC BLOOD PRESSURE BY DOPPLER IN LEFT ANKLE

SYSBP
Accession: phv00019997.v1.p1
>> Search for Susceptibility Genes for Diabetic Nephropathy in Type 1 Diabetes -> SYSBP

Description
Systolic blood pressure (mmHg)

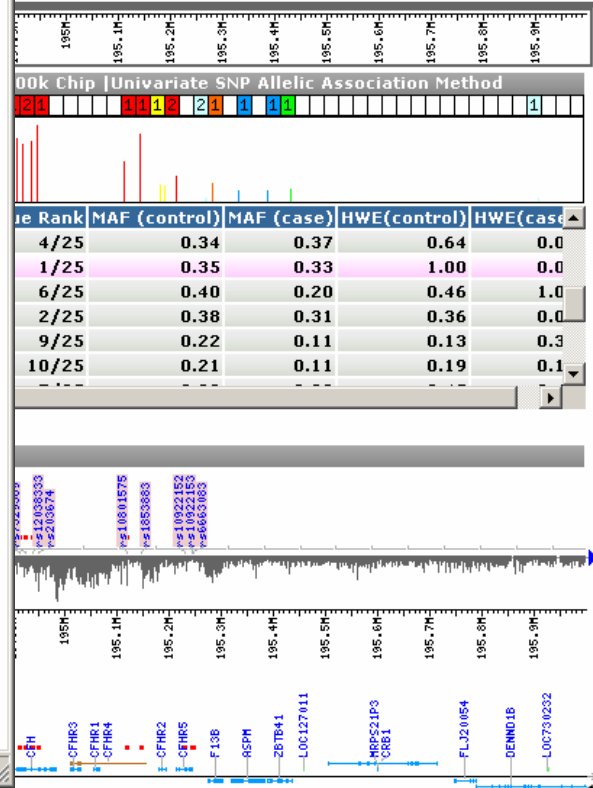
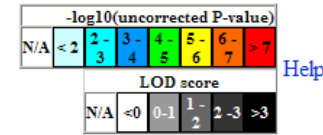
dbGaP study [phs000001](#): rs203674 Genotype Summary

phv00000173	Genotype			Genotype Frequency		
	AA	AC	CC	25%	50%	75%
Case	51	157	186			
Control	82	88	23			

phv00000173	Allele		Allele Frequency		
	A	C	25%	50%	75%
Case	259	529			
Control	252	134			

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

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



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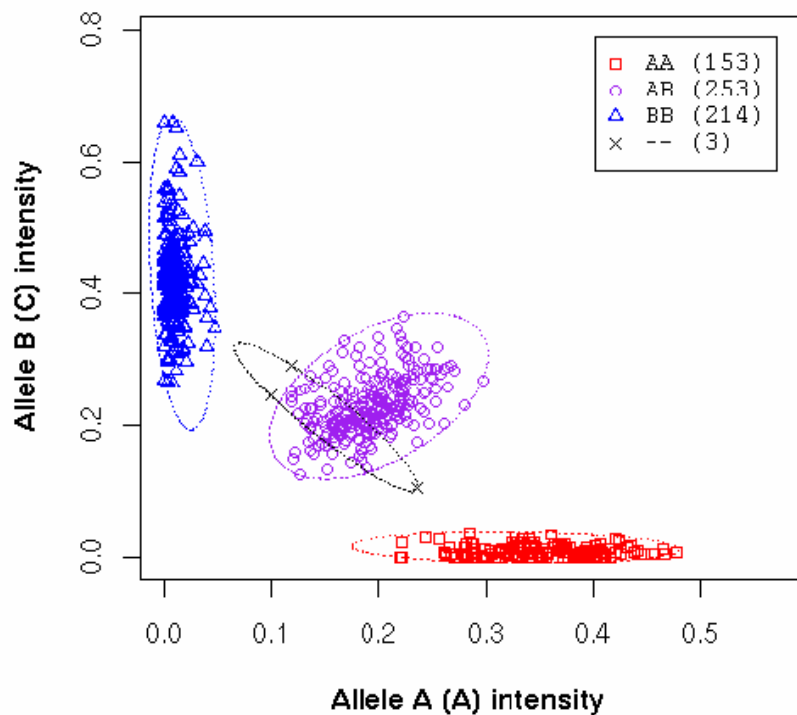
110101



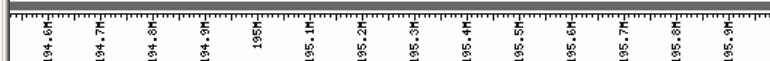
-log ₁₀ (uncorrected P-value)						
N/A	<2	2-3	3-4	4-5	5-6	>6
Lod score						
N/A	<0	0-1	1-2	2-3	>3	

Help

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



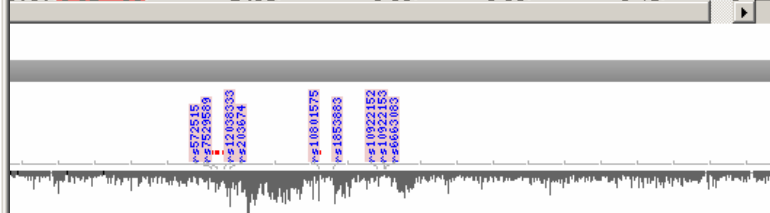
reset Go



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



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