

NIAGADS Update

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NIAGADS

Datasets at NIAGADS

41 datasets | 54,000 samples | 24 billion genotypes

- ▣ 18 GWAS
- ▣ 1 Expression
- ▣ 1 Linkage
- ▣ 9 Genotyping (<100k)
- ▣ 11 Summary Statistics
- ▣ 1 Whole Exome Sequencing
- ▣ 16 GWAS datasets with Imputation

In Progress:

- ▣ 1 Whole Exome Sequencing (PSP- after pub.)

Incoming Datasets:

- ▣ 2 Targeted Sequencing
- ▣ 2 GWAS
- ▣ 2 Linkage
- ▣ ADGC Exome Chip (after pub.)

ADSP Data Update

Phase	Discovery Phase		Discovery Extension Phase
Data Summary	Whole Genome Family Based	Whole Exome Case/Control	Whole Genome Family Based
Subjects	578 Subjects in 111 families	10,913 Subjects	428 Subjects
DNA-Seq (BAM)	Released Nov, 2013 & March, 2014	Released Nov 2014	Anticipate Release Fall 2016
Phenotypes	Updated Annually		Anticipate Release Fall 2016
Pedigrees	ADSP Portal	Enriched Case Families under QC	Anticipate Release Fall 2016
Genotype Calls	Released SNV VCF files April 2016		Anticipate Release Spring 2017
	Anticipate Indel VCF files Spring 2017		

New NIAGADS Website

NIAGADS DATA ▾ RESOURCES ▾ NEWS ▾ GENOMICS DB ABOUT ▾ CONTACT LOGIN/REGISTER 🔍

The National Institute on Aging Genetics of Alzheimer's Disease Data Storage Site

NIAGADS is the National Institute on Aging Genetics of Alzheimer's Disease Data Storage Site. NIAGADS is a national genetics repository created by NIA to facilitate access by qualified investigators to genotypic data for the study of genetics of late-onset Alzheimer's disease.

BROWSE DATASETS

DISEASE ▾

DATA TYPE ▾

SEARCH

AVAILABLE DATA

41	53,826	24,582,130,282
DATASETS	SAMPLES	GENOTYPES

RESEARCHERS

Submit a Data Access Request, provide documents for a dataset submission, report any publications, or view NIAGADS

TOOLS AND SOFTWARE

Software and other tools developed for analysis of data in NIAGADS and other databases.

NIAGADS GenomicsDB Version 2.1

NIAGADS GenomicsDB










Search for a gene, variant, or region

SEARCH

Examples - Gene: [APOE](#) - Variant: [rs6656401](#) - Region: [chr19:45309039-45512650](#)


A searchable annotation resource that provides access to publicly available NIAGADS summary statistics datasets for Alzheimer's Disease and related neuropathologies.


What would you like to do?

-  Explore the region around a gene or SNP on the genome browser.
-  Perform [pathway or functional enrichment analysis](#) on a list of genes.
-  Find SNPs with genome-wide significance in NIAGADS datasets. →
-  Get a list of [beta-amyloid binding genes](#).
-  Explore [gene-pathway memberships](#).
-  Find SNPs associated with [Alzheimer's Disease](#) in the NHGRI GWAS Catalog.
-  Upload genomic locations from a [BED file](#) to compare against curated features.
-  Build your own query.
-  Explore example [search strategies](#).

Get SNPs with genome-wide significance in a NIAGADS dataset

Retrieve a list of SNPs based on summary statistics from NIAGADS Genome Wide Association data.

ACCESSION 

DATASET  Select...

Select...

Cerebral amyloid angiopathy (presence vs. absence)

Hippocampal sclerosis of the elderly (presence vs. absence)

Lewy body disease (3 category ranking)

Lewy body disease (5 category ranking)

Lewy body disease (presence vs. absence)

Neuritic plaques (presence vs. absence)

Neuritic plaques (presence vs. absence); conservative

Neuritic plaques (presence vs. absence); relaxed

Neuritic plaques (ranked by CERAD score)


Neurofibrillary tangles (ranked by Braak groups)

Neurofibrillary tangles (ranked by Braak stages)

Neuropathologic features of AD and related dementias (case/control)

Vascular brain injury (3 category ranking)

Vascular brain injury (presence vs. absence); relaxed

P-VALUE 

Search

This search

- [NG00041](#) Summary statistics from a multi-ethnic exome array study to identify low-frequency coding variants that affect susceptibility to late-onset Alzheimer disease
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NIAGADS GenomicsDB Version 2.1

Variants, gene annotations, AD-relevant functional genomics datasets allow AD researchers to easily identify and interpret genomic regions

- Table of Contents
 - Genomic Context
 - Functional Genomics
 - Co-located SNPs
 - Functional Annotation
- Related Resources
 - NCBI: 348
 - HUGO: HGNC:613
 - ENSEMBL: ENSG00000130203
 - VEGA: OTTHUMG00000128901
 - OMIM: 107741
- Explore Further
 - PubMed
 - StringDB
 - ExAC Browser
 - UCSC Genome Browser
- Expression
 - GTEx Portal
 - Human Protein Atlas
 - EMBL-EBI Expression Atlas

APOE
apolipoprotein E
Also known as: AD2, LDL2Q5, LPG, APO-E

Genomic Context
chr19, (45409039..45412650)

Variant Effect Impact (snpEff)

My Strategies: New Opened (1) All (1) Basket Public Strategies (11)

Strategy: SNPs with genome-wide significance for established CSF biomarkers and Alzheimer's disease (IGAP)*

Expanded View of Step CSF biomarkers

16 SNPs from Step 2
Strategy: SNPs with genome-wide significance for established CSF biomarkers and Alzheimer's disease (IGAP)

About the Result Table

Filter results by putative impact of SnpEff-inferred effects


SNP Results

SNP	Reference Allele	Minor Allele/MAF	Strongest Effect Impact	M
rs11566005	C	T/0.114	LOW	synor
rs12972155	C	G/0.08806	LOW	seq
rs12972970	G	A/0.08806	LOW	seq
rs1305082	G	C/0.3425	MODIFIER	3 pin



Preview: Genotype-Phenotype Portal

- ❑ Collaboration with Penn ADC (InQuery)
- ❑ Available to approved investigators
- ❑ Query subjects by genotypes and basic phenotypes
- ❑ Planned feature: link with NCRAD sample availability
- ❑ To be released in Spring 2017


University of Pennsylvania
 Neurodegenerative Diseases

MAIN ▾

Reset
Commit
Export
Search

Query By By SNP By Position

Markers

NIAGADS_ID	subject_id	niagads_data...	ncrad_id	cohort	dx	sex	apoe	age_at_onset...	age_last_exa...	rs760893	rs4930128
			NA	ADC3	2	1	33	78	83	CT	CC
			NA	ADC3	2	2	33	78	83	CC	TC
			NA	ADC3	NA	2	33	78	83	CT	CC
			NA	ADC3	NA	1	33	78	83	TT	TC
			NA	ADC1	2	1	34	78	83	CC	CC
			NA	ADC1	2	2	33	78	82	CT	CC
			NA	ADC1	2	1	33	78	82	CT	CC
			NA	ADC2	2	1	33	78	82	CT	CC
			NA	ADC3	2	2	34	78	82	TT	CC
			NA	ADC1	2	1	34	78	82	CT	CC
			NA	ADC1	2	1	34	78	82	CT	CC
			NA	ADC1	2	2	24	78	82	CT	CC
			NA	ADC1	2	1	33	78	82	CT	TC
			NA	ADC1	2	1	34	78	82	TT	CC
			NA	ADC1	2	1	33	78	82	TT	TC
			NA	ADC2	2	1	34	78	81	TT	CC
			NA	ADC2	2	2	33	78	81	TT	CC
			NA	ADC2	2	2	33	78	81	TT	TC
			NA	ADC2	2	2	34	78	81	TT	CC
			NA	ADC2	2	1	23	78	81	CC	TC
			NA	ADC2	NA	1	33	78	81	TT	CC
			NA	ADC3	2	1	34	78	81	TT	CC
			NA	ADC3	2	2	24	78	81	TT	CC
			NA	ADC3	2	2	34	78	81	TT	TC
			NA	ADC3	2	2	44	78	81	CC	TC
			NA	ADC3	2	1	33	78	81	TT	CC

Total Patients: 5033 Total Rows: 5033

Stage 1 >> Stage 2 >>

Acknowledgements

Many thanks to

- Patients and families
- NIA ADRC program and centers
- NACC and NCRAD
- ADSP
- IGAP/ADGC/CHARGE/EADI/GERAD
- Support from NIA/NHGRI/NIH

NIAGADS Team:

- | | |
|-------------------------|-------------------|
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EAB:

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- Carole Ober
- Eric Schadt
- Brad Hyman
- Mark Daly

DUC:

- Tatiana Foroud (Chair)
- Steve Estus
- Mel Feany
- Todd Golde
- Leonard Petrucelli

NIA Program Officer:

- Marilyn Miller

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