



# NCRAD

January 2016

## Genomic Data Sharing Policy Documentation

The Alzheimer's Disease Sequencing Project (ADSP) is continuing to plan for the replication phase. In order for your Centers' samples to be included in this phase, the NIH Genomic Data Sharing Policy (<https://gds.nih.gov/index.html>) documentation must be updated. This documentation certifies that the samples you have provided to NCRAD are allowed to be shared and how they can be shared.

NCRAD has been contacting every ADC to update the required paperwork for the Genomic Data Sharing (GDS) Policy to ensure ADC samples can be included. Please note, that this document was updated in the fall of 2015. If you completed the documentation in the spring of 2015 or earlier, the form will need to be updated to the latest version.

The ADSP replication phase is already in the process of selecting samples. We appreciate your help in providing the required documentation as quickly as possible. If you are unsure if you have completed this or not, please contact Kelley Faber at [kelfaber@iu.edu](mailto:kelfaber@iu.edu).

A huge THANK YOU to those who have already completed this updated form!

**A Central Repository with DNA available to match the rich dataset collected for all subjects seen in the ADCs is a very valuable resource for the field of AD research. We hope you will continue to support this effort!**

## Conference Calls

Thank you to everyone who participated in a conference call with us last year. We appreciated the opportunity to discuss blood sample collection strategies with each Center and new initiatives. It is time again to touch base with all Centers. Kelley will begin to contact each Center soon to schedule a call for late February or March.

We look forward to speaking with you soon!

## Annual Sample Distribution Reports

It is time again for NCRAD's annual Sample Distribution Reports. This is sent out every January to the Center Director. Please contact Kelley Faber ([kelfaber@iu.edu](mailto:kelfaber@iu.edu)) if you would like a copy sent to you directly as well. As a reminder, this report is intended to assist your Center in demonstrating your contribution to sample banking efforts encouraged by NIA.

The report summarizes the number of samples provided by your site as part of initiatives banking samples at NCRAD. For example, a Center could be sending samples as part of the ADGC, ADNI, and LOAD initiatives. Please note that the number of samples provided on the report is a count of unique individuals within a study. Some studies are collecting longitudinal samples or multiple types of samples and these are not distinguished.



# NCRAD

The annual report also summarizes how frequently samples contributed by your site are requested by researchers. The report shows the total number of samples contributed by your site that were ever distributed to researchers as well as the number that were distributed in the past year. This is a total count and is not restricted to unique subjects. Samples from the same subject may be requested by more than one investigator. This count does not reflect the samples that were returned to the contributing site as their one free aliquot.

Our summary report also provides the number of unique investigators that have requested samples contributed by your Center. We provide this information for the past year and also in a cumulative form across all years. Finally, all NIH grants that were supported by the samples contributed by your site are listed.

We always welcome suggestions for how to improve our report. We are also glad to answer any questions you have after you review the report.

## New NCRAD Catalogs

As part of the new NCRAD website ([www.ncrad.org](http://www.ncrad.org)), we have built a catalog request system. This catalog system is designed for researchers interested in accessing samples to determine which sample collections best fit their research needs before applying for the samples.

In the Accessing Biospecimens and Data section of the website, researchers can complete a web-based Data Agreement to obtain a username and password to gain access to the restricted catalogs. Once in the catalog, a data dictionary is available to describe each variable. While some columns are shown on the screen by default, additional columns may be added according to the needs of each investigator. One of the columns that can be added in is a "NIAGADS Data" variable. This variable provides a link to the relevant NIAGADS dataset information page if the subject has genomic data available for request through the NIAGADS site.

After selecting which columns to display, each one can be filtered to further select samples. After selecting samples, the list of subjects and data can be downloaded into an Excel or CSV file. This file can be used by a statistician to determine feasibility of the sample set and to submit to NCRAD as part of the request.

Catalogs are currently available for the NIA-LOAD and Indianapolis-Ibadan studies with more to come. Some screen shots are shown on the following page.

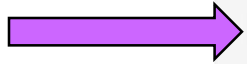


# NCRAD



Biospecimens and Data section of NCRAD website

The variables to be displayed in the catalog can be selected individually.



Each column can be filtered to select samples that fit research criteria.

**Catalog Selections**

Please follow the steps below to select the criteria necessary for your research.

1. View the study **Data Dictionary** to see all available catalog fields' and their descriptions.
 

[Dictionary](#)

\*Additional data were collected for this study. To download a list of additional variables click here. Contact us to request more information about data that are not available in this catalog: [alzstudy@iupui.edu](mailto:alzstudy@iupui.edu) or 800-526-2839.
2. Click on **Select Columns** below to choose the fields you would like to display in the catalog.
 

[Select Columns](#)
3. Click on **Filter Columns** below to filter the catalog by applying criteria to individual fields.
 

[Filter Columns](#)
4. Please **Download** the records you have selected from the catalog. The downloaded CSV file will include all of the columns you have selected and the subjects that met your filtering criteria. Please use this list to work with your statistician to determine feasibility for your research project.
 

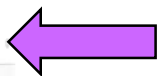
[Download](#)
5. **Contact** us. Please use your downloaded file to discuss with your statistician to determine feasibility for your research project. When you are comfortable with the list of samples you have selected, please contact NCRAD to begin the request process. You can reach us at: [alzstudy@iupui.edu](mailto:alzstudy@iupui.edu) or 800-526-2839.

**Late-Onset Alzheimer's Disease Family Study**

Welcome and thank you for visiting the NCRAD catalog for the NIA-LOAD study! Please use this tool to help you determine feasibility of this collection for your request. Located on the left hand side of the screen, you will find expandable data mining tools for personalized sample selection. Use these tools to narrow our catalog to only those samples applicable for your research. This application is designed to help you find a list of subjects that can meet your research needs. Please do not hesitate to contact us at any point in the process to ask any questions, provide comments, or talk through your sample needs. You can reach us at [alzstudy@iupui.edu](mailto:alzstudy@iupui.edu) or 800-526-2839.

Showing 1 to 25 of 7,126 entries

Subject ID	Family ID	Sex	Autopsy	Dementia Status	Age at Onset	NIAGADS Data
100001	1000	Female	Missing/Unknown	Probable AD	72	NG00020, NG00032
100101	1001	Female	Missing/Unknown	Probable AD	78	NG00020, NG00032
100102	1001	Male	Missing/Unknown	Probable AD	65	NG00020, NG00032
100103	1001	Female	Missing/Unknown	Not demented, no neurological disorder		NG00020, NG00032
100104	1001	Female	Missing/Unknown	Not demented, no neurological disorder		NG00020, NG00032
100201	1002	Female	Missing/Unknown	Probable AD	73	NG00020, NG00032
100301	1003	Male	Missing/Unknown	Probable AD	69	NG00020, NG00032
521641	1003	Female	Missing/Unknown	Probable AD	71	NG00020, NG00032
100401	1004	Female	Missing/Unknown	Probable AD	63	NG00020, NG00032
521642	1004	Male	Missing/Unknown	Questionable dementia or cognitive impairment		
521643	1004	Female	Missing/Unknown	Dementia by family report	70	
521644	1004	Male	Missing/Unknown	Not demented, no neurological disorder		
100501	1005	Female	Missing/Unknown	Probable AD	76	NG00020, NG00032
521645	1005	Male	Missing/Unknown	Other		
100601	1006	Female	Missing/Unknown	Probable AD	81	NG00020, NG00032
100604	1006	Male	Missing/Unknown	Not demented, no neurological disorder		NG00020, NG00032
100605	1006	Female	Missing/Unknown	Possible AD	65	NG00020, NG00032
100701	1007	Male	Missing/Unknown	Probable AD	60	NG00020,



Samples with genetic data at NIAGADS are hyperlinked directly to the information page for the dataset.

The selected dataset can be downloaded and sent to NCRAD as part of the request process.



## NIAGADS/Alzheimer's Disease Sequencing Project (ADSP) Update

The ADSP has released high-quality datasets of single nucleotide variant (SNV) and insertion-deletion polymorphism (indel) genotypes for both the whole-genome sequencing (WGS) and whole-exome sequencing (WES) data, including 578 subjects from 111 families and the WES sequenced subjects include ~11,000 case/control subjects. To date, whole-exome and whole-genome SNV concordant genotypes in PLINK file format have been released through dbGaP, phs000572. The next wave of release (expected February 2016) will provide additional files including WES and WGS SNV consensus genotypes in VCF format and concordant indel calls in PLINK file format.



## SWAN (Statistical Structural Variant Analysis)

The SWAN toolkit is now available for download for detecting CNVs and INDELS in WGS data here: <https://www.niagads.org/content/swan-sv>. SWAN uses a multifaceted approach to improve sensitivity in detecting a variety of events, including insertions, deletions, duplications, inversions and translocations. SWAN has been tested and benchmarked with various WGS datasets on multiple environments, including AWS and HPC clusters (manuscripts are pending). SWAN is easy to install, provides genotyping information and outputs results in the BED format, making it easy to upload tracks in your favorite visualization tool. SWAN has been developed in collaboration with Nancy Zhang, Charlie Xia and NIAGADS.

## New Dataset Available

[NG00045](#): Progressive Supranuclear Palsy (PSP) Summary Statistics

Progressive Supranuclear Palsy (PSP) is a movement disorder with prominent tau neuropathology. A genome wide association study of PSP was performed to identify genes that modify risk for this primary tauopathy ([GWAS data available](#)). A two-stage analysis was performed to maximize efficiency while maintaining power. Stage 1 is comprised of autopsied cases and stage 2 contains clinically diagnosed PSP cases. Available in this dataset are the summary statistics described in [Hoglinger et al.](#) The p-value data is generally available to all users using the link below; however, gaining access to the allele frequencies requires a formal data request.

[Download P-value only data here.](#)

## Beta-release for an enhanced GenomicsDB is now available!

Visit the GenomicsDB to access a beta-version of the upcoming, enhanced GenomicsDB to explore new datasets, experience an enhanced search interface, and try out new tools for data analysis.

### New NIAGADS Datasets

Data from NG00040 (GWAS summary statistics for a multi-ethnic exome array study of AD, FTD, and PSP) and NG00041 (GWAS summary statistics for Neuropathologic Features of AD and Related Dementias) now available for search or exploration via the beta-release of the GenomicsDB and associated Genome Browser.

### New Tools for Data Analysis

Use new graphical query toolkits to discover SNPs or Genes co-located within sequence feature annotations, such as:

- Expressed enhancers (FANTOM5)
- Histone modifications (ENCODE)
- DNase Hypersensitivity Regions (ENCODE)
- Transcription Factor Binding Sites (ENCODE)
- Disease-Trait Associations (NHGRI GWAS Catalog)
- AD-relevant GWAS Significance (NIAGADS)
- User uploaded annotations
- Perform functional or pathway enrichment analysis on results from gene searches or uploaded gene lists.
- View the distribution of search results across the genome.

## The NIAGADS Genomics Database

Advanced Search ?

Q Search

### 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 GWAS 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 feature annotations.