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.

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) 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.

Please contact us with any questions or concerns about NCRAD at 800-526-2839/317-274-7360, by email at kelfaber@iu.edu or visit our web-site: www.ncrad.org Thanks!!



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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), 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.

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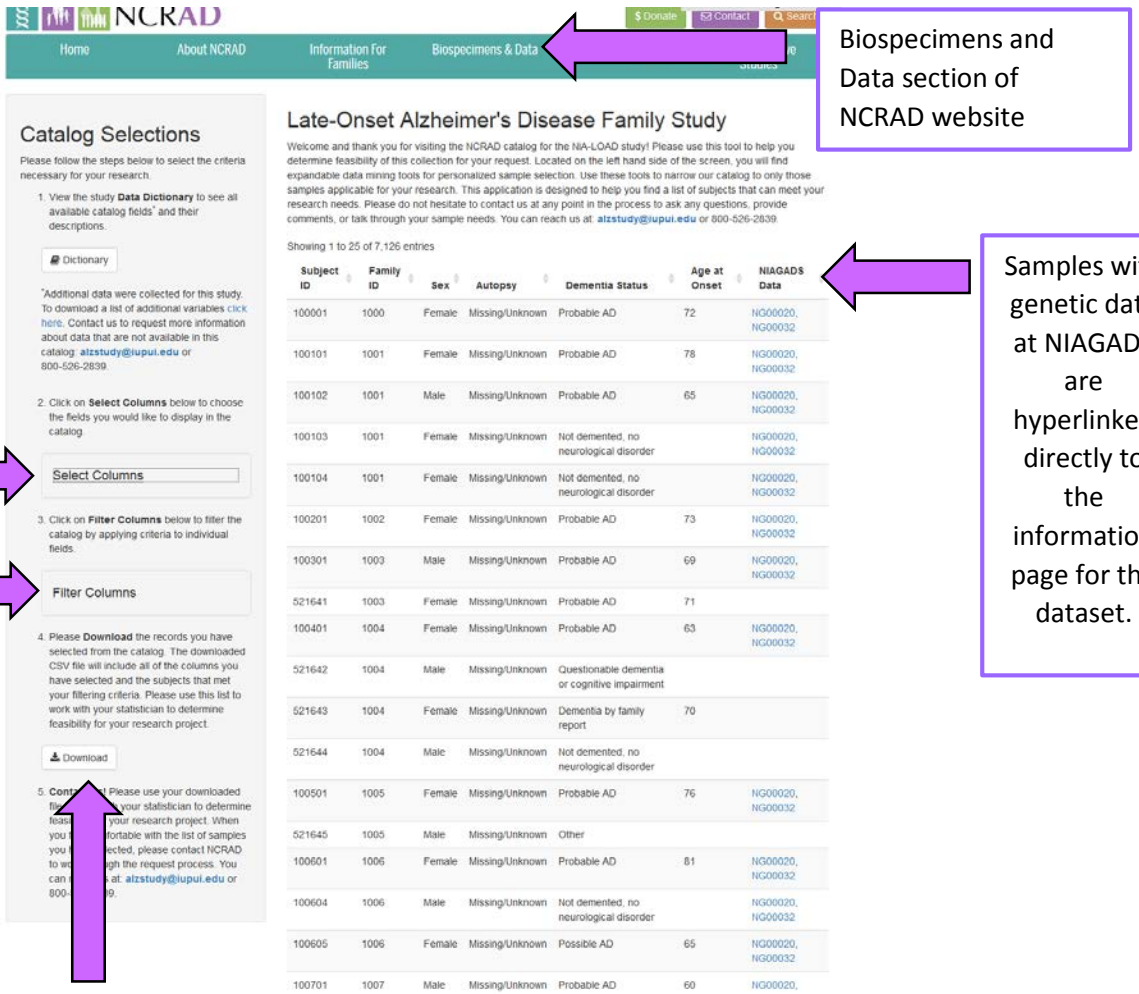

Biospecimens and Data section of NCRAD website

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

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

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

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

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NIAGADS

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

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 form 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

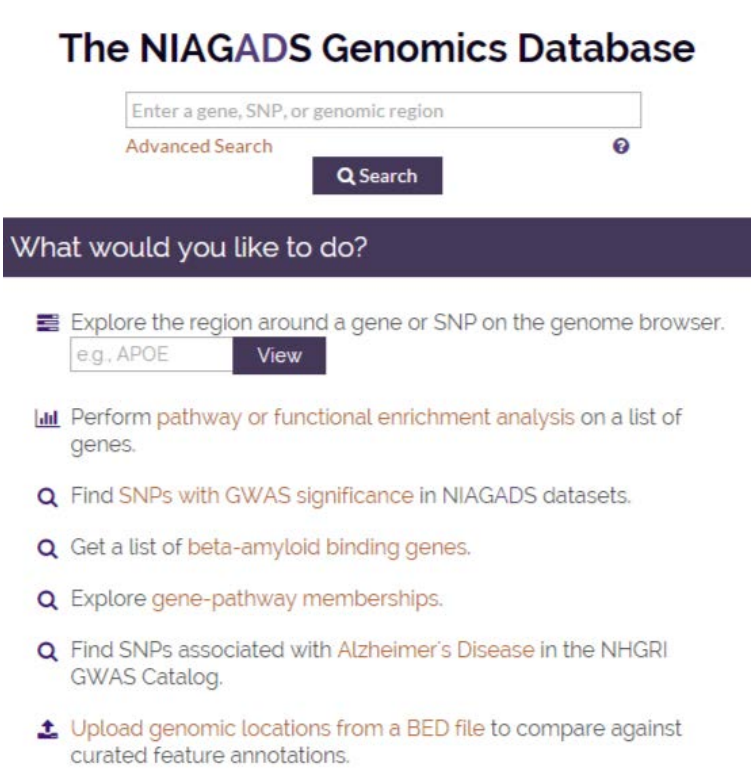
Progressive Supranuclear Palsy (PSP) is a movement disorder with prominent tau neuropathology. A genome wide association study of PSP was performed to identify genes that modify risk for this primary tauopothy (GWAS data available). A two-stage analysis was performed to maximize efficiency while maintaining 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.

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

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.

Visit the NIAGADS website at http://www.niagads.org.
Contact us with questions at support@niagads.org.