# NCRAD

## January 2017

**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.

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.

**APOE Genotyping**

As part of NCRAD's continuing effort to provide *APOE* genotypes on all ADC samples provided to NCRAD, additional data were posted in December. There is a single Excel file with all *APOE* genotypes included.

To obtain these *APOE* data, visit the NACC website at: https://www.alz.washington.edu/adgc.html, select your Center and choose the option to download *APOE* data from NCRAD. As always, please compare these data with any internal *APOE* genotype data you have generated. Please notify Kelley Faber (kelfaber@iu.edu) if you have any questions or find any discrepancies. We want to work with you in order to resolve these.

**Key for *APOE* results on NACC site**

| | | |
|---|---|---|
| 1=e3/e3 | 4=e4/e4 | |
| 2=e3/e4 | 5=e2/e4 | 9=missing/unknown/ |
| 3=e2/e3 | 6=e2/e2 | not assessed |

# NCRAD

## Genomic Data Sharing Policy and Alzheimer's Disease Related Dementias

First, a huge THANK YOU to those who have already completed this updated Genomic Data Sharing (GDS) Institutional Certification form!

In order for your Centers' samples to be included in sequencing projects going forward, the NIH Genomic Data Sharing Policy (https://gds.nih.gov/index.html) documentation must be completed. This document certifies that the samples you have provided to NCRAD are allowed to be shared and how they can be shared.

Under "The National Alzheimer's Project Act (NAPA)", Alzheimer's Disease is defined to include the Alzheimer's Disease Related Dementias (ADRDs) in research and other activities delineated to decrease the growing public health burden due to dementia in our aging population. An email describing this definition has been drafted by the NIH and will be distributed to all ADCs in January.

Since this update to disease definition will also apply to studies already submitted to the Database for Genotypes and Phenotypes (dbGaP), NIH requests that you notify the official(s) who originally signed your Institutional Certification submitted about this update, and about the implication for any "Alzheimer's Disease" Data Use Limitations relevant to your study.

*Please watch your inboxes closely for this email and contact NIH as instructed in the email with any concerns. If there is no response from you or your site, we will presume your Institution's concurrence with the update.*

## ADC Samples to NCRAD

NCRAD continues to accept samples from all subjects with an MDS or UDS at NACC. NACC has updated the lists of samples for submission to NCRAD as of the December 2016 NACC data. You can find them in the Phase 2 section on the NACC website: https://www.alz.washington.edu/GWASPHASE2/gwasphase2.html.

While fresh whole blood samples are preferred, NCRAD also accepts frozen buffy coats, transferred DNA and brain tissue samples. Our goal is to have a DNA sample banked and available from all MDS and UDS subjects. When sending blood, NCRAD will cover the cost of shipping, provide a blood kit, and return 25ug of DNA for free. In addition, APOE genotyping is done on all samples submitted once they match to the NACC lists. More details about sample requirements, kit requests, and forms for submitting samples to NCRAD can be found on our NCRAD website here: https://www.ncrad.org/sample_requirements.html.

Please continue to send samples for this very valuable project!

## Annual ADC Conference Calls

We appreciated the opportunity to review progress and discuss new initiatives with ADCs during our annual conference call. It is time to schedule our spring calls 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!

**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!**

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!!

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

## We're Published!

In the November 2016 issue of *Alzheimer's & Dementia: The Journal of the Alzheimer's Association*, an article has been published that explains how the database works to advance the analysis of Alzheimer's disease genetic and genomic studies. "NIAGADS: The NIA Genetics of Alzheimer's Disease Data Storage Site" is in the print edition of *Alzheimer's & Dementia*, and available online at http://dx.doi.org/10.1016/j.jalz.2016.08.018.

## Introducing GCAD

The Genome Center for Alzheimer's Disease (GCAD) is funded under the NIA cooperative agreement/specialized center award U54 AG052427 to identify genetic variants that cause, influence risk, or protect against AD, and to identify the underlying genes affected by these variants.

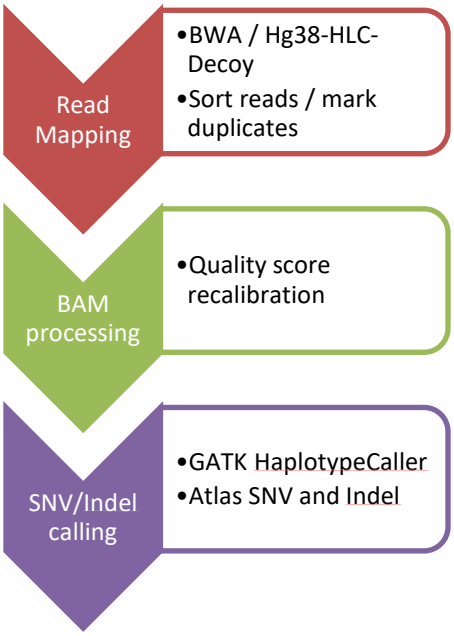
The role of GCAD is to coordinate the integration and meta-analysis of all available AD relevant genetic data with the goal of identifying AD risk/causative/protective genetic variants and eventual therapeutic targets.

In keeping with NIH policy, the GCAD will broadly disseminate all results and derivative data [e.g. imputed genotypes, variant call format (VCF) files recalled using ADSP protocols, etc.]. All GCAD results and derivative data will be distributed to ADSP and other AD investigators, particularly those working on functional analysis of AD-associated genetic variants.

Visit the GCAD website at http://www.adgenomics.org.

## GCAD/ADSP Production Workflow

GCAD/ADSP production workflow is developed by the GCAD team and will be used to apply to all ADSP Discovery, Discovery Extension, and Follow-up study WGS and WES data. The workflow takes as input BAMs and re-maps to the most recent human genome build hg38. It performs marking duplicated reads, then individual-level single nucleotide variant (SNV) and insertion-deletion polymorphism (Indel) calling using GATK-Haplotypecaller (Broad Institute) and Atlas2 (Baylor College of Medicine). Outputs of this workflow include re-calibrated BAMs, individual-level VCFs, and project-level VCFs (pVCF). These pVCFs are then analyzed by the ADSP Quality Control workflow to generate QCed pVCFs. The GCAD/ADSP production workflow will be available soon via Bitbucket.




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