

April 2014

Conference Calls

Thank you to everyone who participated in our annual NCRAD-ADC conference calls this year. We spoke with the Center Director and/or Clinical Core Director at each ADC. We appreciate having the opportunity to speak with each center and learn about ways we can help facilitate sample collection and transfer to our central repository. We hope you find the calls informative and please don't hesitate to contact us if you have any further questions!

Alzheimer's Disease Sequencing Project (ADSP)

The ADSP project includes an initial discovery phase which explores the role of rare variants in late onset AD (LOAD). The discovery phase included whole genome sequencing in 578 individuals from 111 families having multiple LOAD members. The discovery phase will also generate whole exome sequencing in 11,000 individuals, approximately half LOAD cases (LOAD) and half controls.

The sequencing work is well underway and we are now preparing for the replication phase. The target for the replication project is at least 40,000 samples, half of which are LOAD cases and half are controls. Racial and ethnic diversity is a priority. ADC samples contributed to NCRAD will be used once again play a major role in this effort. We have several ongoing initiatives which are designed to maximize the number of samples available at NCRAD from the ADCs for genetic studies.

Replenishing Samples

Because samples provided by the ADCs have been widely used for genetic studies, NCRAD has depleted or nearly depleted many of our stocks. In order to continue to provide ADC samples for additional projects, it is vital for NCRAD to obtain additional DNA for many subjects. Lists with samples that have less than 10ug of DNA at NCRAD have been sent to centers. By sending in additional

Minimum Data Set (MDS) Initiative

NACC is working closely with NCRAD to identify subjects who might be eligible for the replication phase. NACC has generated a list of subjects at each site that are reported to have DNA but that were never part of the ADGC Phase 1 or Phase 2 lists. These lists can be found at: <https://www.alz.washington.edu/NONMEMBER/ncradmds.html>.

We are asking for ADCs to review the lists and help us identify any individuals with DNA samples who are either LOAD cases (possible, probable or autopsy confirmed AD) or controls. We ask that sites please use the most current diagnosis information known to the center when reporting counts. We are focused on cases and controls and would appreciate a count of how many are available. Please note that the lists are posted with the last diagnostic data provided to NACC. They also included diagnoses other than AD and normal subjects. This is because we realize that centers may have updated data available. NACC appreciates receiving these updates as well. Samples must have at least 5ug of DNA which could be shared with NCRAD.

Phase I and Phase II Lists

The reimbursement phase of the ADGC initiative ended March 14, 2014. However, NCRAD continues to accept samples from all subjects on the NACC Phase 1 and 2 lists. While NCRAD is not able to provide monetary compensation, we will cover the cost of shipping. When sending blood, NCRAD will provide a blood kit and return 25ug of DNA for free. In addition, APOE genotyping will be done on all samples submitted. Please continue to send samples for this very valuable project!

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!

New Look of NIAGADS

The NIAGADS (NIA Genetics of Alzheimer's Disease Data Storage Site) website (<https://www.niagads.org/>) has been overhauled to enhance the user experience. NIAGADS is a national repository that facilitates access to genetic, genomic, and related data to qualified investigators for the study of Alzheimer's disease (AD).

Second Batch of ADSP Data Released

Whole genome sequencing data has been generated for 584 subjects from 111 families. This was released in two phases. The first release in December 2013 included 410 individuals. The most recent release includes the remaining 168 subjects. Subsequent releases will contain whole exome sequence data from ~11,000 additional subjects. Approved investigators can browse and download available data through the [ADSP Data Portal](#) or through the [dbGaP Study page](#).

NIAGADS Genomics Database

The NIAGADS Genomics Database provides a simple, but powerful, workspace for searching and identifying genes, SNPs, and genomics locations of interest or with special relevance to Alzheimer's Disease. Register with NIAGADS to bookmark favorite search results or save and share search results and workflows (strategies). The resource also supports a genome browser, allowing users to browse tracks generated from NIAGADS datasets and relevant functional genomics tracks.

<https://www.niagads.org/genomics/>

New Datasets Available Through NIAGADS

NG00030

- [WASHU1](#)
- A case control dataset collected by the Knight ADRC at Washington University.

NG00031

- [MIRAGE CAUCASIAN](#)
- MIRAGE (Multi Institutional Research on Alzheimer Genetics Epidemiology) is a cross-sectional family-based study of genetic and environmental risk factors for AD.

NG00032

- [NIA-LOAD \(ADGC SUBSET\)](#)
- GWAS to identify putative genetic loci related to the risk of late-onset Alzheimer disease (LOAD). This dataset is a subset of NG00020 and was analysed as part of the ADGC project.

NG00033

- [IDENTIFYING RARE VARIANTS THAT INCREASE RISK FOR ALZHEIMER'S DISEASE](#)
- Exome sequencing data for families with Late-Onset AD.

NG00034

- [ACT AND GENETIC DIFFERENCES GWAS](#)
- The Adult Changes in Thought (ACT) study is a longitudinal prospective cohort study and the Genetic Differences study was an epidemiologic case control study.

To access NIAGADS data, please submit an account request through the [NIAGADS website](#). If you would like further information about the data available through NIAGADS or inquire about submitting your AD genetics data to NIAGADS, please contact support@niagads.org.

