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](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.


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.

Contact us with questions at niagads@upenn.edu.