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.

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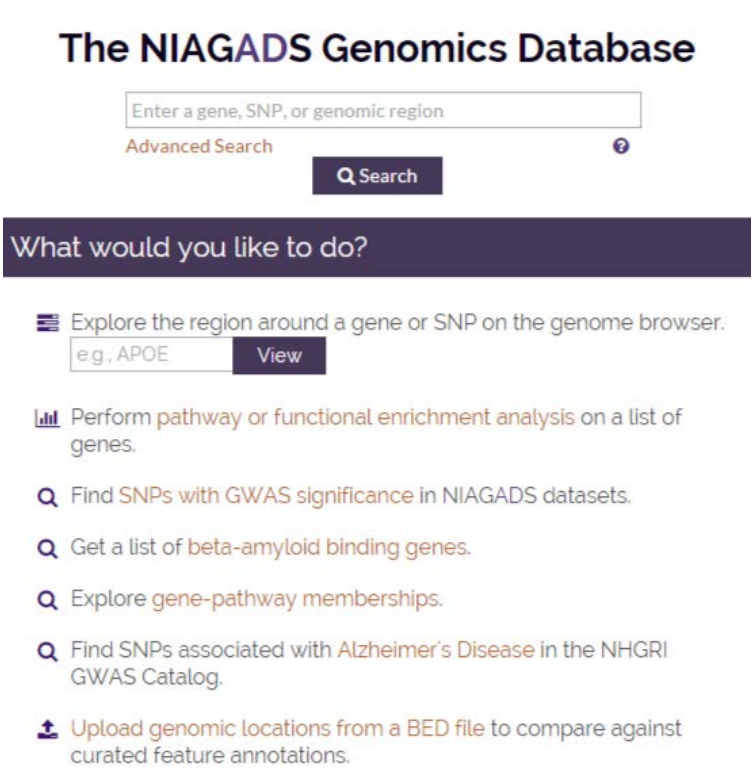
Beta-release for an enhanced GenomicsDB is now available!

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.