

Spotlight: Coming Soon to the GenomicsDB

Functional Enrichment Analysis

Perform enrichment analysis (Gene Ontology, Pathways) on uploaded gene lists or gene search results.

More powerful search tools

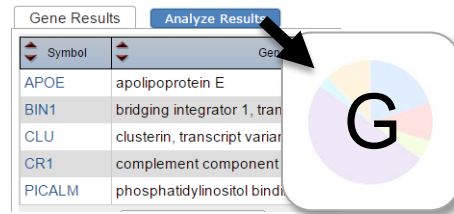
Use a flexible query toolkit to discover SNPs or Genes co-located with sequence feature annotations, including:

- 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

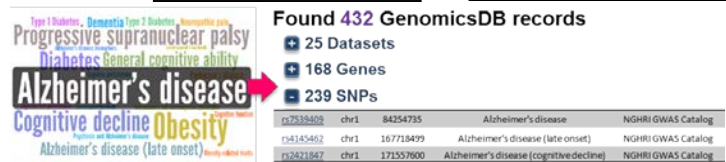
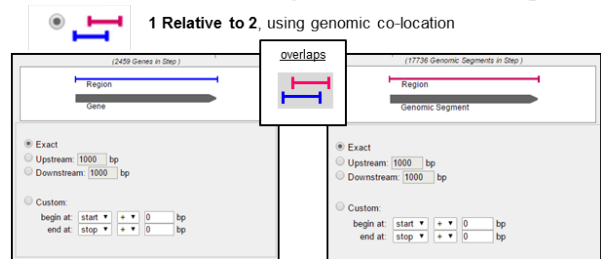
Graphical search tools

Use interactive visualizations to quickly discover relevant datasets, sequence features, and annotations

To volunteer as a beta-tester for the new GenomicsDB, email support@niagads.org



Combine results from Step 1 with Annotated Regions



NIAGADS/Alzheimer's Disease Sequencing Project (ADSP) Update

The ADSP Quality Control (QC) Work Group has worked to provide 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. The QC process integrated genotype data from samples sequenced at three NHGRI funded large scale sequencing centers. Data were pre-processed by two genotype calling pipelines to produce a single high-quality set of "consensus" genotypes. The first WGS consensus genotype calls on 578 individuals from 111 families was released in June 2015. WES SNV QC is underway and

genotypes will be available as release 2 in September; a third data release with additional QC steps is expected in mid-November 2015. The QC team is reviewing WGS and WES indel data with an anticipated public release of quality-checked data during late winter/early spring 2016.

NIAGADS is currently developing a software pipeline to mirror the genotype calling and QC process developed by ADSP. This tool will allow investigators to prepare their WGS or WES genotype calling data for comparison with ADSP data. This pipeline is currently under development and should be publicly available later this year. We will announce and release the software and documentation through the NIAGADS website.

Two New Caucasian Datasets Available

NG00042 – Miami and Medical School of Mount Sinai

913 Cases and 896 Controls
GWAS and Imputation data available

NG00043 – Mayo

844 Cases and 1255 Controls
GWAS data available

For all available datasets, visit:
<https://www.niagads.org/available-data>

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