New data at NIAGADS

ADSP Data and how to link to your ADC information

NIAGADS Genomics DB
Datasets at NIAGADS

34 datasets | 49,000 subjects | 30 billion genotypes

- **16 GWAS**
- 1 eGWAS
- 1 Expression
- 1 Linkage
- 9 Genotyping (<100K)
- 6 Summary Statistics
- 1 Whole Exome Sequencing
- 11 GWAS datasets with Imputation

**Incoming Datasets:**
- 2 Targeted Sequencing
- 3 GWAS
- 3 Summary Statistics
- 1 Exome Chip
- 1 PSP WES
- 2 Linkage
ADSP Data

- Raw data are available
  - WGS: 578 individuals / 111 multiplex families
  - WES: 10,939 samples/10,959 individuals
  - 3,264 WES are ADC subjects

- Genotypes
  - WGS: July
  - WES: October/November

- See ADSP website (www.niagads.org/adsp) for more information on
  - Study design
  - Apply for data access
Apply for ADSP data access

Go to [www.niagads.org/adsp](http://www.niagads.org/adsp), click Apply for Data

- All applications submitted to dbGaP, reviewed by NIH
- ADSP requires:
  - **Secondary/derived data return plan** that will be generated by your study and deposited into NIAGADS
  - Signed **NIA Data Sharing Plan** and **NIAGADS Data Distribution Agreement** for ADSP
- Contact NIAGADS ([data@niagads.org](mailto:data@niagads.org)) for data application, we’d be glad to help step-by-step
Linking ADSP WES ADC samples

- ADSP WES data are labeled using ADSP IDs
  
es.g. A-ADC-AD003257-BR-NCR-08AD11443

- ADCs can link with their own phenotypic data with these steps:

  - Contact NIAGADS (data@niagads.org) if you plan to link the samples and we’ll walk you through the process
NIAGADS Genomics Database

Welcome to the NIAGADS Genomics Database

A simple, but powerful, workspace for browsing and identifying genes, SNPs, and genomic regions with special relevance to Alzheimer's Disease.

- Search the GenomicsDB
- View a genomic region
Quickly access curated annotations for known genes, SNPs, and genomics regions.

With a single-click, **discover** SNPs with GWAS significance in published NIAGADs datasets.
Bookmark records or add to a basket

Link to other databases with a single click.

Quickly find significant SNPs
Additional Search Functions

GENES

FIND A SNP
Enter a dbSNP rs identifier to view details and curated annotations for a specific SNP.

SNPs

rs6656401
Go
More

GENOMIC REGIONS

Enter a genomic region
Go

Sample Strategies
Upload a list of locations

Expressed Enhancers

Brain-specific expression?
chr19:35409039..65412650

FIND EXPRESSED ENHANCERS WITHIN A REGION OF INTEREST
Search pre-defined enhancer sets for brain-relevant tissues and cells from PANTOM5.
Enter a genomic location in the format chrH:start-end (e.g., chr19:35409039..65412650) to find all expressed enhancers within that region.

Explore further to make novel discoveries
Explore and Analyze Gene Functions

**My Strategies:**
- New
- Opened (2)
- All (2)
- Basket
- Public Strategies (0)

**Strategy:** Genes associated with SNPs with GWAS Significance in IGGAP 2013

**59 Genes from Step 2**

**Strategy:** Genes associated with SNPs with GWAS Significance in IGGAP 2013

**Genome overview**

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<th>Location</th>
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Showing 1 to 8 of 8 entries (filtered from 25 total entries)
New Result Pages: Explore and Analyze

Functional / pathway analysis

Gene Ontology Enrichment

Pathway Enrichment
Genome Browser Tracks: GWAS + ENCODE + FANTOM5
October Release

- Currently contains the following data
  - **Genes** and pathway annotations
  - **SNP** information (dbSNP v142) with population allele frequency and SNPEff impact predictions
  - Published **AD GWAS** summary statistics
  - NHGRI GWAS Catalog
  - **Functional genomics** data relevant to brain
    - ENCODE: DNasel HS / Histone modification / TF Binding ChIP-Seq
    - FANTOM5: Enhancer-TSS associations / Cell-expressed enhancers

- Planned data
  - Gene-gene interactions
  - Gene expression tissue specificity
  - Cross-species conservation (PhastCons)
  - Genome interaction database
  - Structural variant calls (dbVar/DGV/CNVD)
  - GTex express QTL
Take-home message
-- please share with your ADC

- New datasets at NIAGADS

- **ADSP genotypes** for WGS (available now) and WES (October ~ November)

- **Linking ADSP WES samples** with ADC’s own phenotype data – contact NIAGADS so we can help you

- Improved **NIAGADS Genomics Database** will be available in October

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