

# NIAGADS Update

April 2016 ADC Director's Meeting

Li-San Wang

University of Pennsylvania

[lswang@upenn.edu](mailto:lswang@upenn.edu)

[data@niagads.org](mailto:data@niagads.org)

**NIAGADS**

# Datasets at NIAGADS

34 datasets | 54,000 samples | 30 billion genotypes

- ▣ 15 GWAS
- ▣ 1 eGWAS
- ▣ 1 Expression
- ▣ 1 Linkage
- ▣ 9 Genotyping (<100k)
- ▣ 6 Summary Statistics
- ▣ 1 Whole Exome Sequencing
- ▣ 12 GWAS datasets with Imputation

## In Progress:

- ▣ 1 Whole Exome Sequencing (PSP)
- ▣ 1 GWAS

## Incoming Datasets:

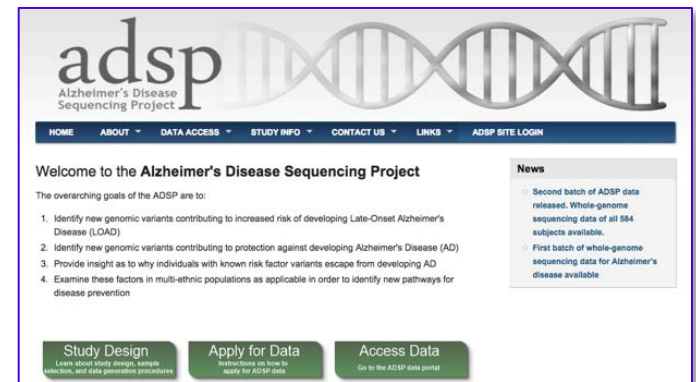
- ▣ 2 Targeted Sequencing
- ▣ 2 GWAS
- ▣ 2 Linkage
- ▣ 2 Summary Statistics

# ADSP Data Update

Data Summary	Family Based Whole Genome Sequenced	Case/Control & Enriched Case Whole Exome Sequenced
Subject Count	578 Subjects in 111 families	10,913 Subjects
DNA-Seq (BAM)	Released Nov, 2013 & March, 2014	Released Nov 2014
Concordant Genotypes (PLINK format)	Released SNV files July 2015 <b>Released Indel files April 2016</b>	Released files Dec 2015 <b>Released Indel files April 2016</b>
Consensus Genotypes (VCF format)	<b>Released SNV VCF files April 2016</b> Anticipate Indel release June 2016	
Phenotypes	Annual Updates	
Pedigrees	ADSP Portal	Enriched Case Families under QC

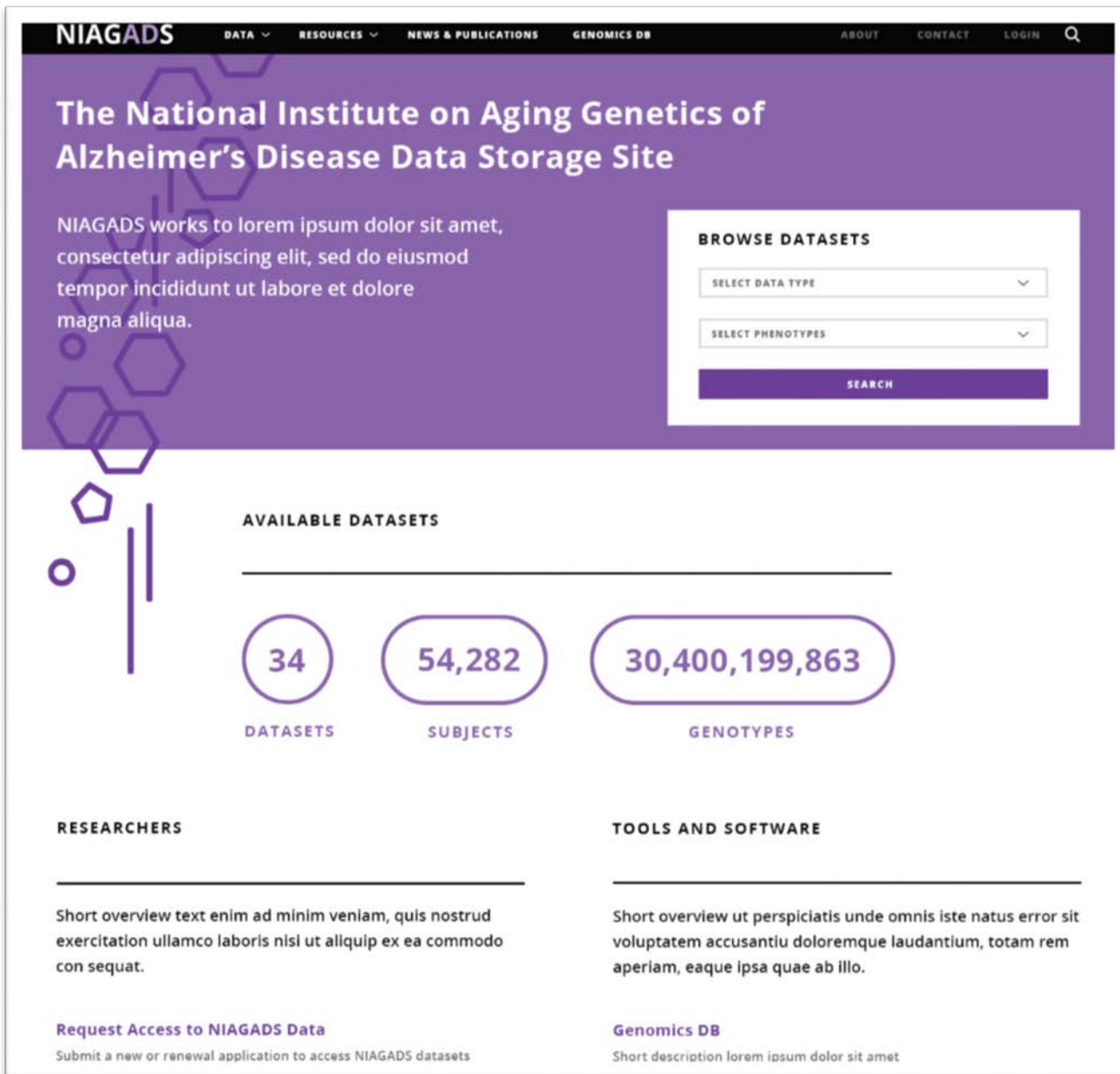
# ADSP dbGaP v7 release April 2016

- ▣ 578 subjects with whole-genome data
  - ▣ Consensus SNV VCF files
  - ▣ Concordant Indel PLINK files
  
- ▣ 10913 subjects with whole-exome data
  - ▣ Consensus SNV PLINK and VCF files
  - ▣ Concordant Indel PLINK files
  
- ▣ See ADSP website  
[www.niagads.org/adsp](http://www.niagads.org/adsp)  
for more information on
  - ▣ Study design
  - ▣ Apply for data access



# ADSP SNV calling / QC Pipeline

- ▣ Combine your data with ADSP data
- ▣ **Release 1: Concordant Calling**
  - ▣ Early access available as of today
  - ▣ Email [data@niagads.org](mailto:data@niagads.org) to get help
  - ▣ Or use the following preview URL:  
[https://github.com/jmal0403/ADSP\\_QC\\_Beta](https://github.com/jmal0403/ADSP_QC_Beta)
- ▣ **Release 2: Consensus Calling**
  - ▣ Available in summer 2016



NIAGADS  
Website  
Update

July 2016

# Acknowledgements

## Many thanks to

- Patients and families
- NIA ADRC program and centers
- NACC and NCRAD
- ADSP
- IGAP/ADGC/CHARGE/EADI/GERAD
- Support from NIA/NHGRI/NIH

## NIAGADS Team:

- |                         |                   |
|-------------------------|-------------------|
| • Gerard Schellenberg   | • Fanny Leung     |
| • Chris Stoeckert       | • Micah Childress |
| • Adam Naj              | • Rebecca Cweibel |
| • Emily Greenfest-Allen | • Prabhakaran K.  |
| • Amanda Partch         | • Han Jen Lin     |
| • Otto Valladares       | • Zhao Yi         |
| • John Malamon          | • Liming Qu       |

## EAB:

- Matthew Farrer
- Barry Greenberg
- Carole Ober
- Eric Schadt
- Brad Hyman
- Mark Daly

## DUC:

- Tatiana Foroud (Chair)
- Steve Estus
- Mel Feany
- Todd Golde
- Leonard Petrucelli

## NIA Program Officer:

- Marilyn Miller

NIAGADS is funded by NIA  
U24-AG041689