NIAGADS Update
October 2016 ADC Director's Meeting

Li-San Wang

University of Pennsylvania

lswang@upenn.edu

niagads@upenn.edu

NIAGADS

NIAGADS

Datasets at NIAGADS

41 datasets | *54,000* samples | 24 *billion* genotypes

- 18 GWAS
- 1 Expression
- 1 Linkage
- 9 Genotyping (<100k)
- 11 Summary Statistics
- 1 Whole Exome Sequencing
- 16 GWAS datasets with Imputation

In Progress:

- 1 Whole Exome Sequencing (PSP- after pub.)

Incoming Datasets:

- 2 Targeted Sequencing
- 2 GWAS
- 2 Linkage
- ADGC Exome Chip (after pub.)

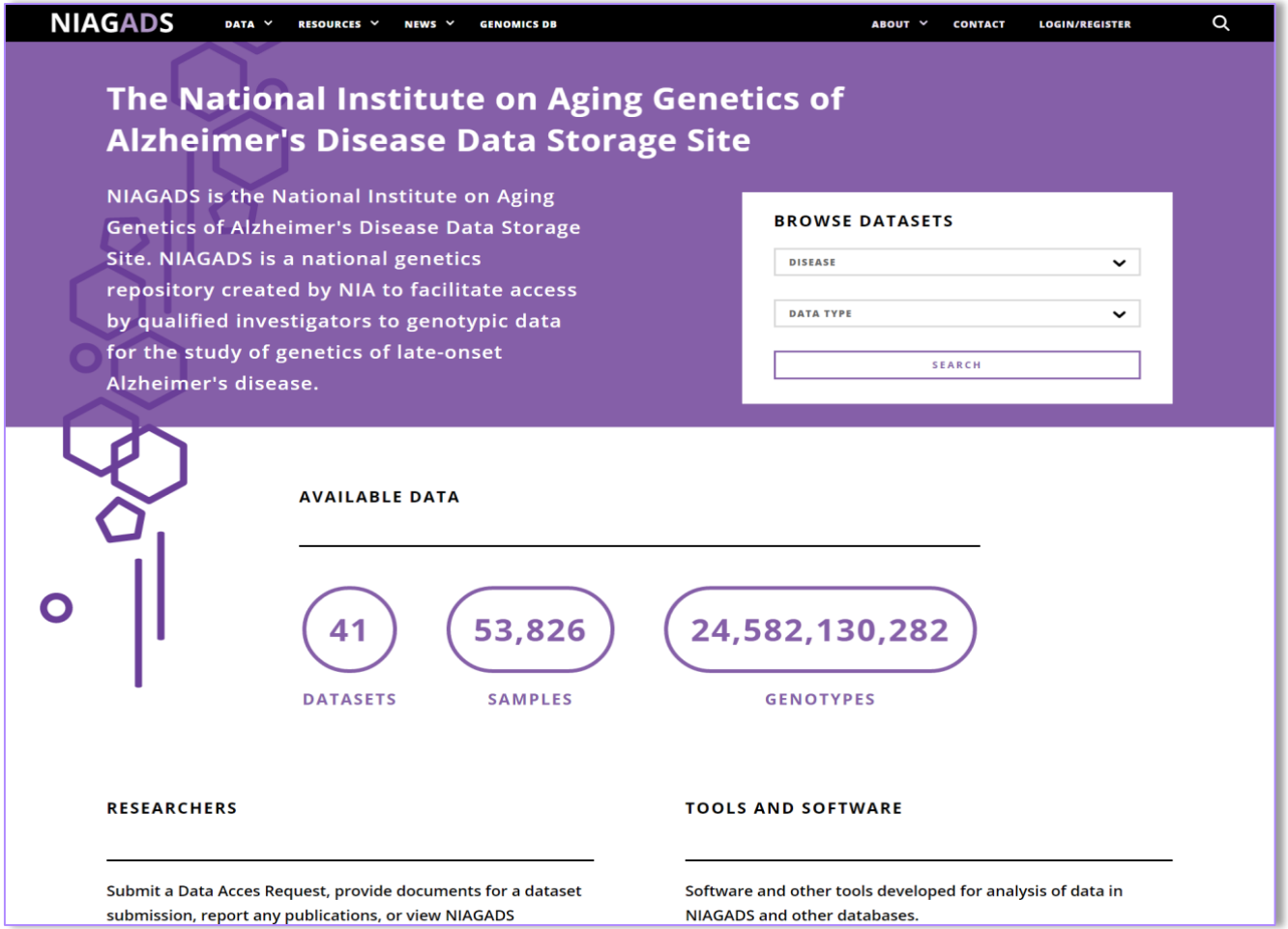
NIAGADS

ADSP Data Update

Phase	Discovery Phase		Discovery Extension Phase
Data Summary	Whole Genome Family Based	Whole Exome Case/Control	Whole Genome Family Based
Subjects	578 Subjects in 111 families	10,913 Subjects	428 Subjects
DNA-Seq (BAM)	Released Nov, 2013 & March, 2014	Released Nov 2014	Anticipate Release Fall 2016
Phenotypes	Updated Annually		Anticipate Release Fall 2016
Pedigrees	ADSP Portal	Enriched Case Families under QC	Anticipate Release Fall 2016
Genotype Calls	Released SNV VCF files April 2016 Anticipate Indel VCF files Spring 2017		Anticipate Release Spring 2017

NIAGADS

New NIAGADS Website

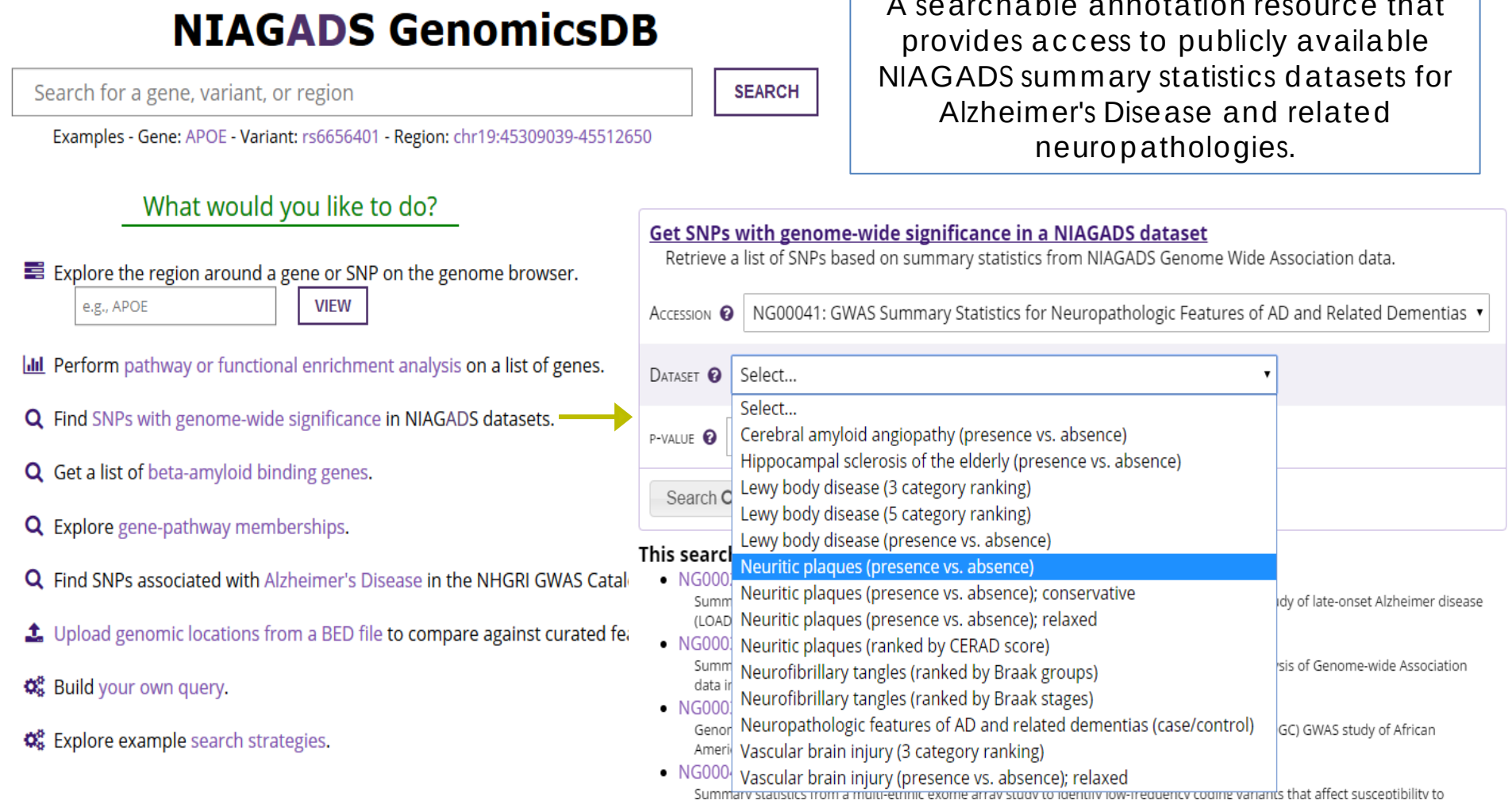


www.niagads.org

NIAGADS

NIAGADS GenomicsDB Version 2.1

A searchable annotation resource that provides access to publicly available NIAGADS summary statistics datasets for Alzheimer's Disease and related neuropathologies.



NIAGADS

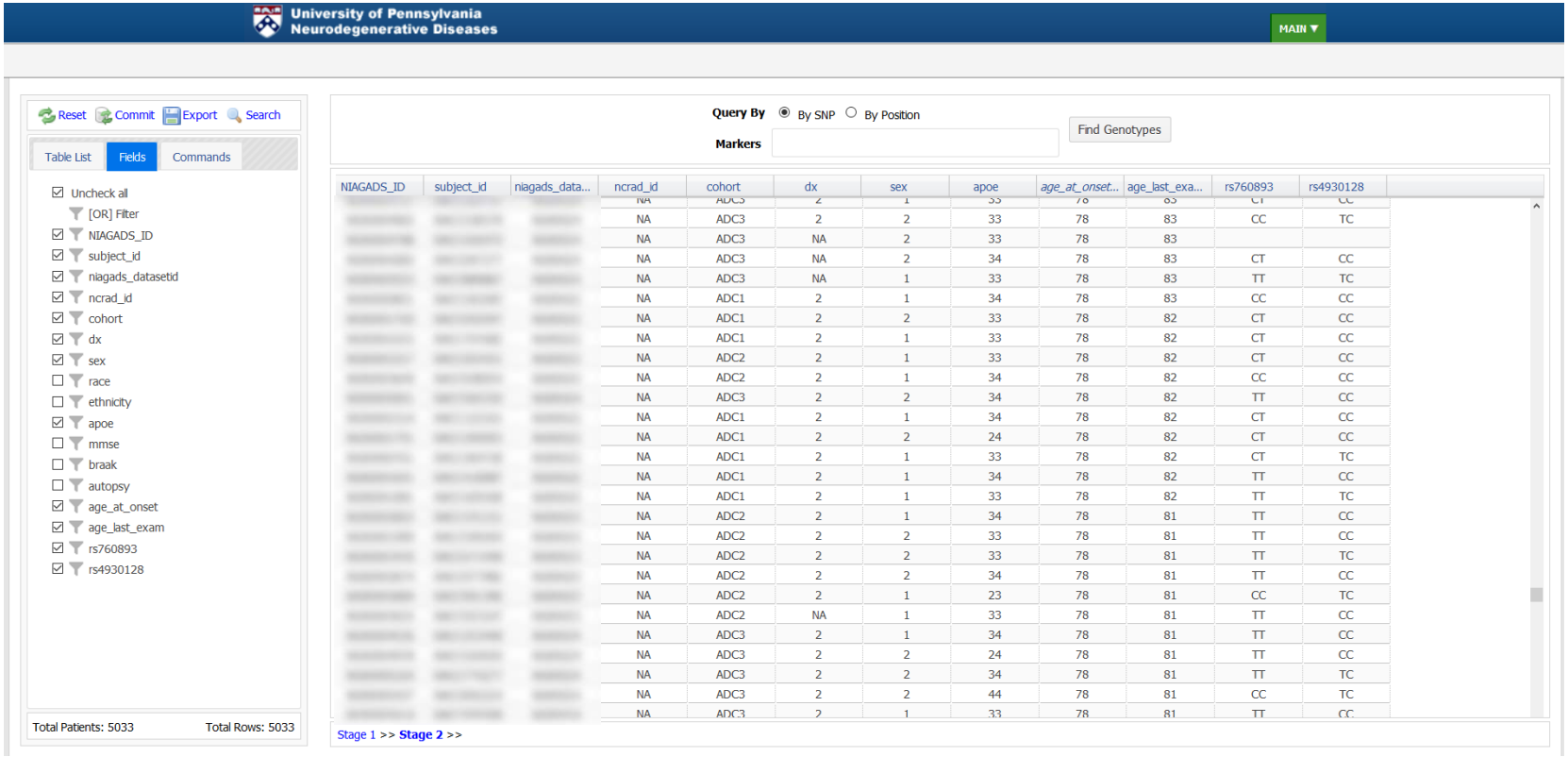
NIAGADS GenomicsDB Version 2.1

Variants, gene annotations, AD-relevant functional genomics datasets allow AD researchers to easily identify and interpret genomic regions

NIAGADS

Preview: Genotype-Phenotype Portal

- Collaboration with Penn ADC (InQuery)
- Query subjects by genotypes and basic phenotypes
- Available to approved investigators
- Planned feature: link with NCRAD sample availability
- To be released in Spring 2017



NIAGADS

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
- Chris Stoeckert
- Adam Naj
- Emily Greenfest-Allen
- Amanda Kuzma
- Otto Valladares
- John Malamon
- Fanny Leung
- Micah Childress
- Rebecca Cweibel
- Prabhakaran K.
- Han Jen Lin
- Yi Zhao
- 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