### Introduction to NIAGADS

ADC Director's Meeting Spring 2014

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Principal Investigator

NIA Genetics of Alzheimer's Disease Data Storage Site

University of Pennsylvania Perelman School of Medicine

U24-AG041689





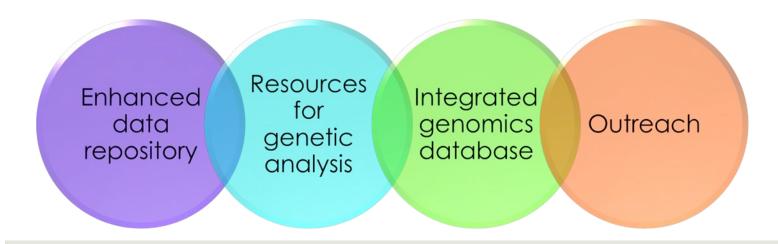




## NIAGADS Objectives

# One-step data access portal and community resource for AD genetics research

- Supported by NIA cooperative agreement U24-AG041689
- Facilitate data sharing, reanalysis, and collaboration
- Post-GWAS: integration to functional genomics knowledge and data
- Path to therapeutic discovery





#### Datasets

- We currently host 26 datasets with 42,531 subjects and 29.5 billion genotypes
- Most of ADGC1 data are available for download; cohort Pls are committed to make data available at NIAGADS
- Secondary data
  - ADGC GWAS summary statistics
  - Imputation data
  - Quality control and population structure
- Data portal for ADNI WGS data (Summer 2014). Login with your ADNI account to download VCF and BAM files

## <u>NIAGADS</u>

# AD Sequencing Project

adsp

Alzheimer's Disease
Sequencing Project

- Announced in Feb. 2012
- Participants
  - NIA, NHGRI
  - ADGC and CHARGE



Photo from http://nihrecord.od.n ih.gov/newsletters/20 12/03\_02\_2012/story5 .htm

- Large-Scale Genome Sequencing and Analysis Centers (Broad/Baylor/WashU)
- NACC (phenotype) and NCRAD (sample)
- NIAGADS (data coordinating center)
- NCBI dbGaP/SRA
- Visit ADSP website www.niagads.org/adsp to learn about study design, apply for data access, download data
- WGS data of 584 subjects available from our ADSP data portal
- Applications will be reviewed by NIH DAC and NIAGADS DUC for secondary data sharing



#### Resources

- NIAGADS Genomics Database
  - Query top SNPs and view gene and pathway annotations
  - Jbrowse genomic browser interface
- DRAW: DNA-Seq analysis on Amazon Cloud (onsite demo)
- ADGC GWAS analysis best practices and workflow (forthcoming)





# BIOINFORMATICS APPLICATIONS NOTE Vol. 29 no. 19 2013, pages 2498-2500 doi:10.1093/bioinformatics/btt422 Sequence analysis Advance Access publication August 13, 2013 DRAW+SneakPeek: Analysis workflow and quality metric management for DNA-seq experiments Chiao-Feng Lin<sup>1,2</sup>, Otto Valladares<sup>1,2</sup>, D. Micah Childress<sup>1,2</sup>, Egor Klevak<sup>3</sup>, Evan T. Geller<sup>1</sup>, Yih-Chii Hwang<sup>2,4</sup>, Ellen A. Tsai<sup>4,5</sup>, Gerard D. Schellenberg<sup>1,\*</sup> and Li-San Wang<sup>1,2,\*</sup> ¹Department of Pathology and Laboratory Medicine and ²Institute for Biomedical Informatics, University of Pennsylvania Perelman School of Medicine, Philadelphia, PA 19104, USA, ³Department of Physics, University of Washington, Seattle,

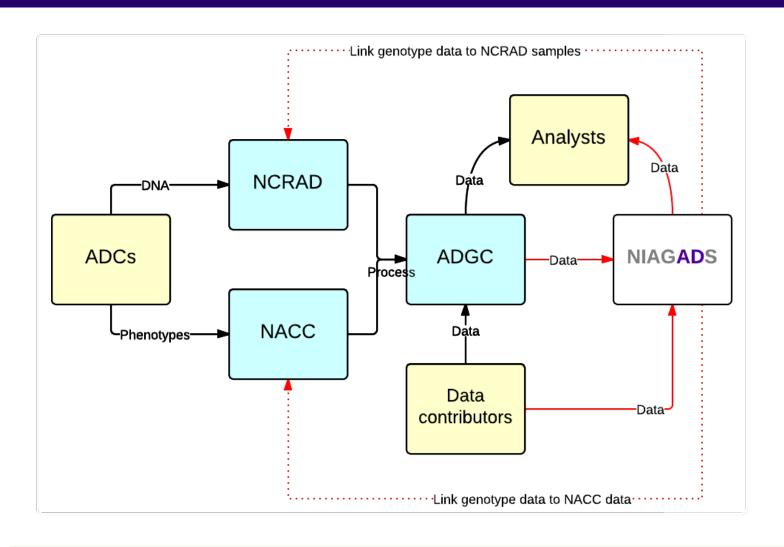
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#### NIAGADS and AD Genetics Resources





Visit our website at www.niagads.org



- We would like to visit your center to introduce NIAGADS and talk with your team to see how we can help
- Let us know what features you would like to see
- Li-San Wang Iswang@mail.med.upenn.edu