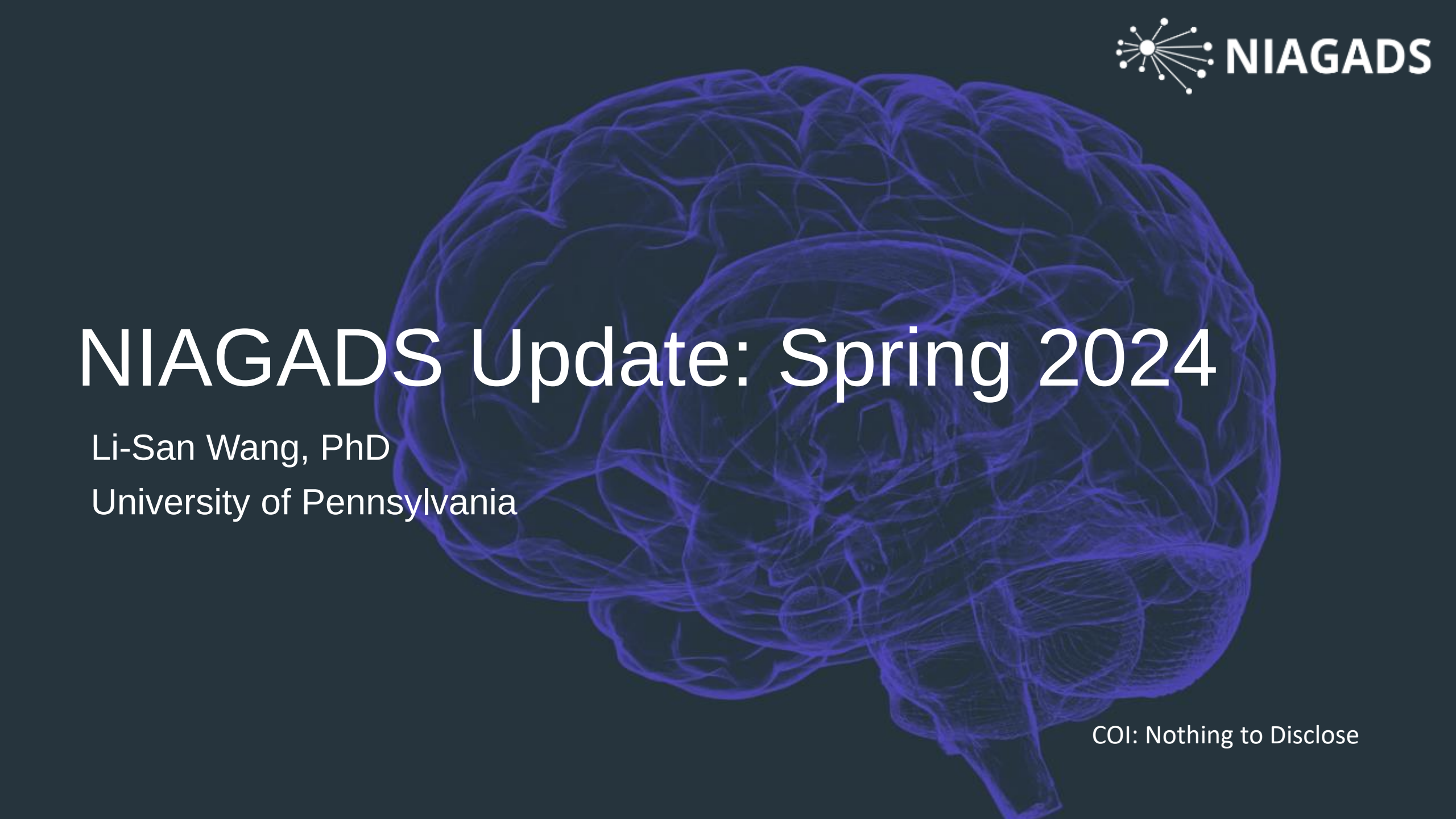


NIAGADS Update: Spring 2024

Li-San Wang, PhD
University of Pennsylvania

COI: Nothing to Disclose

NIAGADS at a Glance

NIAGADS

Data Sharing

113 datasets

>183,000 samples

497 Unique Requests

238 Institutional Certifications

Userbase

>330 articles cite U24

>4,900 verified users

ADSP

73 cohorts

23 current grant subawards

Exomes

20,503

Genomes

~350% increase from 2021 to 2024

(16,905 in 2021 to ~60,000 genomes in 2024)

ADSP Support

14 workgroups /

5 consortia

>500 members

52 institutions

NIAGADS 2023 Growth

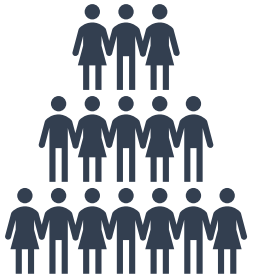
NIAGADS

+17%

datasets

+24%

articles that cite U24



+28%

verified users

ADSP WGS Releases

NIAGADS

Released in 2023

December	R4 WGS Annotation
	PHC 2023 Release

Coming Soon in 2024

Q2	SV joint genotype called pVCF
	QC'd chrX/multi-allelic pVCF
Summer	R5 WGS- ~62k genomes
	CRAMs, gVCFs, preview pVCF

ADSP 2023 Deliverables – NG00067v11
Phenotype Harmonization Consortium (PHC)

Cognition	Fluid Biomarker	Neuropathology	Vascular Risk Factors
•Harmonized composite scores (PHC_EF, PHC_MEM, PHC_LAN, PHC_VSP)	•Amyloid ß,Tau, pTau •Z-scores •A/T classification •Raw data •Platform Information	•Mapped AD Phenotypes (NIA-AA ABC score, Thal phasing, Braak staging, CERAD score) •Cerebral amyloid angiopathy, lewy bodies, vascular brain injury, TDP-43 proteinopathy and hippocampal sclerosis	•Vascular Composite (Hypertension, diabetes, heart disease [myocardial infarct, congestive heart failure, etc.], BMI, height, weight, stroke, smoking, medication, blood pressure, CVR score)

Imaging – DTI	Imaging – FLAIR	Imaging – MRI	Imaging – PET
•PreQual Processed Data (Image-Based BIDS) •Free Water Corrected Metrics •Scanner Harmonization •Multi-Site Calibration •FA and MD Scalars	•Total WMH Volume	•Preprocessed Imaging Collection •MUSE Regions of Interest •FreeSurfer Regions of Interest •SPARE-AD Scores •AD Signature (Schwartz)	•Preprocessed Imaging Collection •Global SUVRs •Centilloid Values •GMM Probabilities •Amyloid Status

Courtesy Tim Hohman / Vanderbilt: timothy.j.hohman@vumc.org

ADSP 2023 Deliverables – NG00067v11
Phenotype Harmonization Consortium (PHC)

✓ Included in ng00067.v11

Harmonization in Progress; to be included in future release

Domain data not available for harmonization

Cohorts	Autopsy	Cardiovascular Risk Factors	Cognition	Fluid Biomarkers	Imaging – DTI	Imaging – FLAIR	Imaging – PET	Imaging – T1
A4	N/A		✓	N/A	N/A	✓	✓	
ACT	✓		✓	N/A			N/A	
ADNI	N/A	✓	✓	✓	✓		✓	✓
ADFBS (NIA-LOAD)	✓	✓	✓	N/A	N/A	N/A	N/A	N/A
EFIGA		✓	✓	N/A	N/A	N/A	N/A	N/A
Knight ADRC			✓	✓				
NACC	✓	✓	✓	✓	✓	✓	✓	✓
ROS/MAP/MARS	✓	✓	✓	N/A	✓		N/A	
WHICAP			✓	N/A		✓		✓
UPenn				✓				
WHICAP		✓	✓			✓	N/A	
WRAP	N/A	✓	✓		✓		✓	✓

ADSP 2024 Deliverables
R5 WGS Release

NIAGADS

ADSP Release	Primary Ancestry/Ethnicity	Cases	Controls	Other	Totals
GCAD Release 5	Black/African American	2,646	5,608	777	9,031
	Asian	1,273	4,083	5	5,361
	Latino/Hispanic	4,488	10,248	2,777	17,513
	Non-Hispanic White	9,124	14,459	6,067	29,650
	Total	**17,531**	**34,398**	**9,626**	**61,555**

ADRC samples in R5

4,590
exomes
22% of total ADSP WES

12,629
genomes
20% of total ADSP WGS

ADRCs can get WGS/WES data from their own samples from us!
Send an email to niagads@pennmedicine.upenn.edu to initiate.

ADRC GWAS Data Release

NIAGADS

ADC 1–15 GWAS
available via DSS

- QC'd PLINK files on GRCh38
- Updated ID schemas for seamless mapping of phenotypes to genotypes
- TOPMedR2 imputed files in .bgen format
- Curated phenotypes provided by the ADSP-PHC
- ADSP to NACC ID mapping file for any ADRC participants that also have WGS, WES, or ADSP-PHC harmonized phenotypes available through the ADSP dataset (ng00067)

GWAS SNP Array Data for

29,676

ADRC Samples Passing QC and available for request.

ADC 16 & 17 GWAS
available to the ADRCs

- Raw array data available
- Total of 2,292 participants from 38 ADRCs

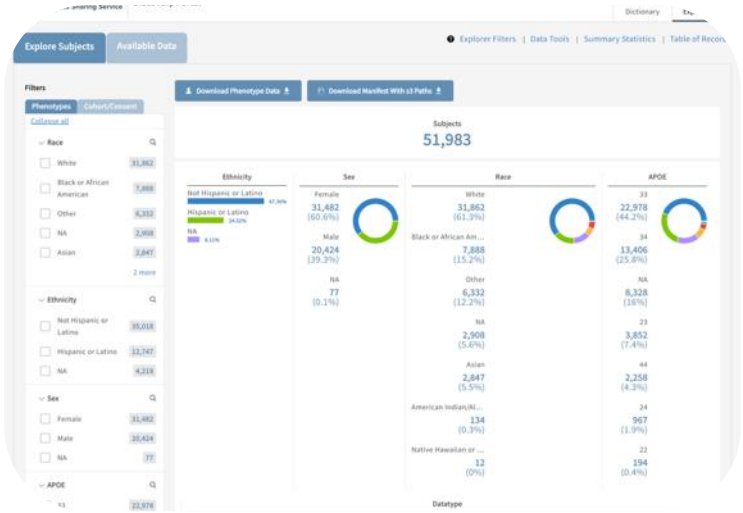
Supporting FAIR Principles

NIAGADS

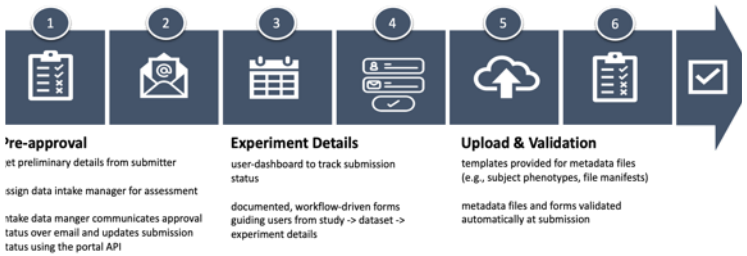
Initiatives	Findable	Accessible	Interoperable	Reusable
Gen3 Platform				
RAS Authentication				
Data Submission Portal				
DOI Implementation				
ADGC GWAS Standardization				
Outreach Initiatives				

Supporting FAIR Principles

NIAGADS



ADSP Data Discovery Platform Integration powered by **Gen3** Technology



Streamline data submission and validation using **DSS Data Submission Portal**

DataCite
FIND, ACCESS, AND REUSE DATA

Digital Object Identifiers (DOI) Enabled through **NIH DataCite Consortium** membership

NIAGADS Open Access
Data and Annotation Resources

unrestricted data and annotations (variant, gene, sequence) available for immediate access

dss NIAGADS

Open Access Portal

direct data downloads for GWAS summary statistics (p-values) and **ADSP Variant Annotations**

https://dss.niagads.org

filer [1]

75,207

standardized, searchable functional genomics tracks

https://tf.lisanwanglab.org/FILER/

advp [2]

curated AD-GWAS catalog, documenting

>18k

published associations

https://advp.niagads.org

GenomicsDB [3]

AD-genetics knowledgebase and **Genome Browser**, containing

>260M

annotated variants

https://www.niagads.org/genomics

1. Kuksa et al. 2022, *NAR Genomics and Bioinformatics,* 4(1): lqab123. 2. Kuksa et al. 2021, *Journal of Alzheimer's Disease*, 86(1): 461-477. 3. Greenfest-Allen et al. 20204, *Alzheimer's & Dementia,* 20(2):1123-1136

NIAGADS Open Access

NIAGADS API*

Lead: Emily Greenfest-Allen

advp

Alzheimer's Disease Variant Portal

filer

Functional Genomics Repository

dss NIAGADS

Open Access Portal

GenomicsDB

Alzheimer's Genomics Database

API
NIAGADS

- Programmatic access
- Bulk, ranged, and genome wide queries
- Integration into analysis pipelines
- Better support ADSP activities and AI/ML pipelines

conforms to OpenAPI standards (& is FAIR)

* under development; beta-testing version available summer 2024

Supporting Data Use

User Support

NIAGADS offers several support resources launched over the last year:

Office Hours

Held at least once a quarter

Video Tutorials

https://www.youtube.com/@niagads1814

NIAGADS Support Portal

All NIAGADS documentation in one place, new informational pages to address user questions, dedicated FAQ page

Ticket Support

Users can now submit a ticket to work on resolving technical issues with the NIAGADS team.

Awareness

Expanding NIAGADS reach through increased investigator awareness will be accomplished through informational outreach sessions at AAIC and ASHG in 2024

Exhibit Hall Booth

Visit us at booth 1329D at AAIC!

Poster Presentations

ADRC workshop at AAIC in partnership with NACC, Sage, and NCRAD

Coming in 2024

- Monthly newsletter
- More video tutorials, user support pages, blog posts

AAIC 2024 Activities

NIAGADS

AAIC Exhibit Hall Booth

Visit us at booth 1329D at AAIC

See you this July in Philadelphia!