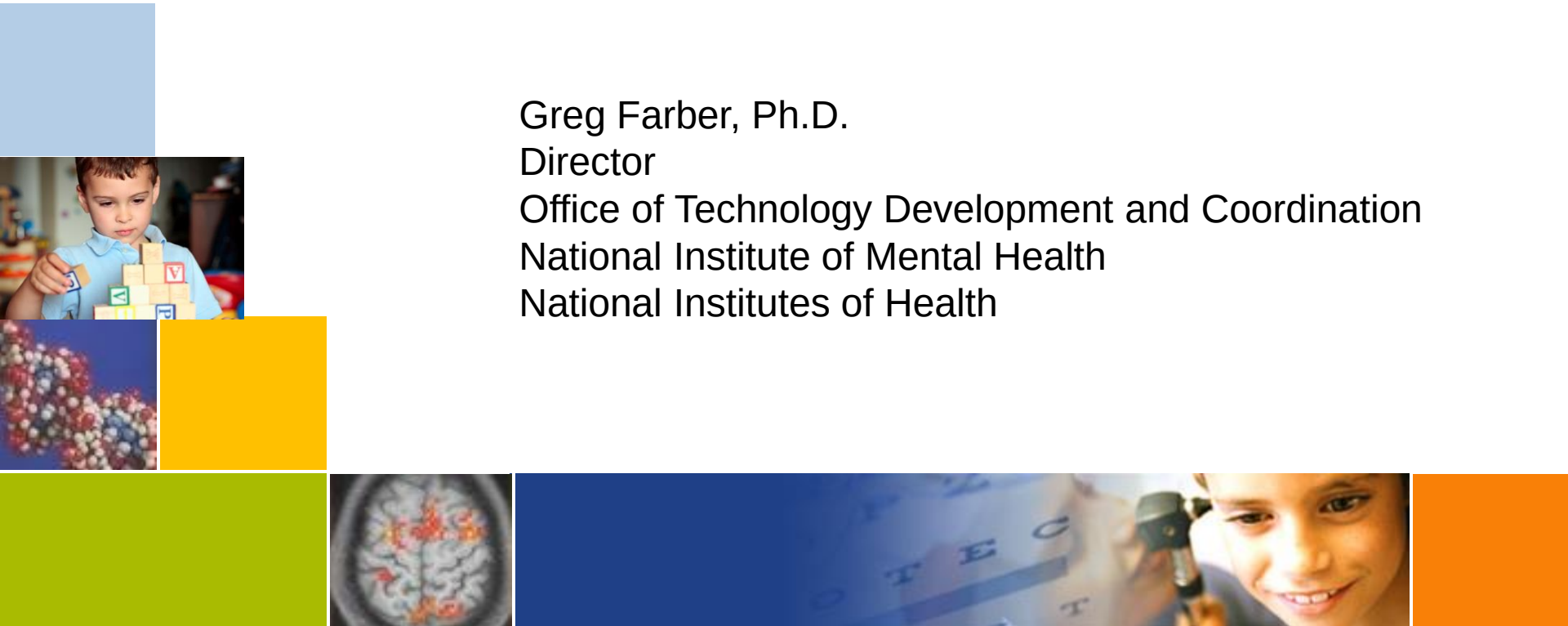


The NIMH Data Repositories

November 5, 2014

Greg Farber, Ph.D.
Director
Office of Technology Development and Coordination
National Institute of Mental Health
National Institutes of Health

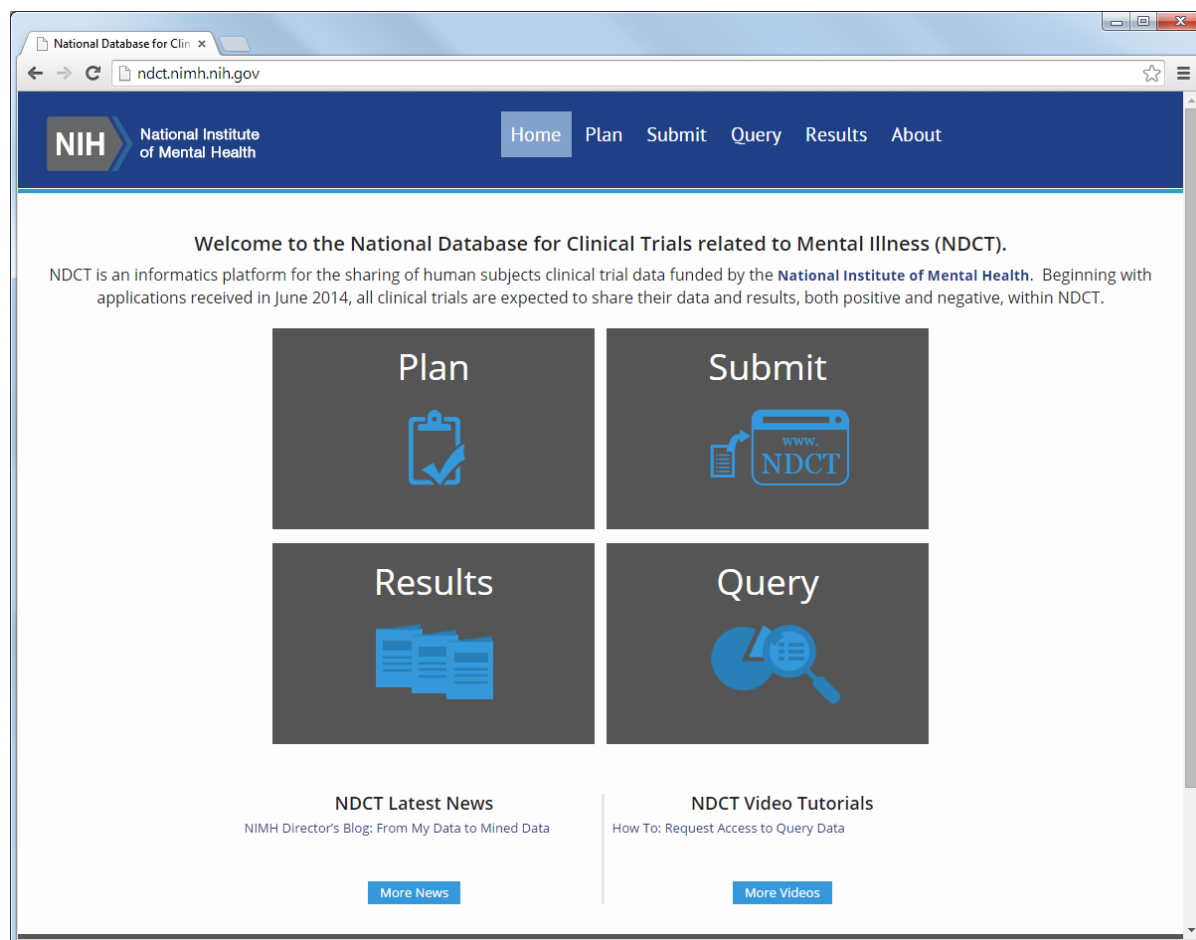


Expansion to Other Scientific Areas

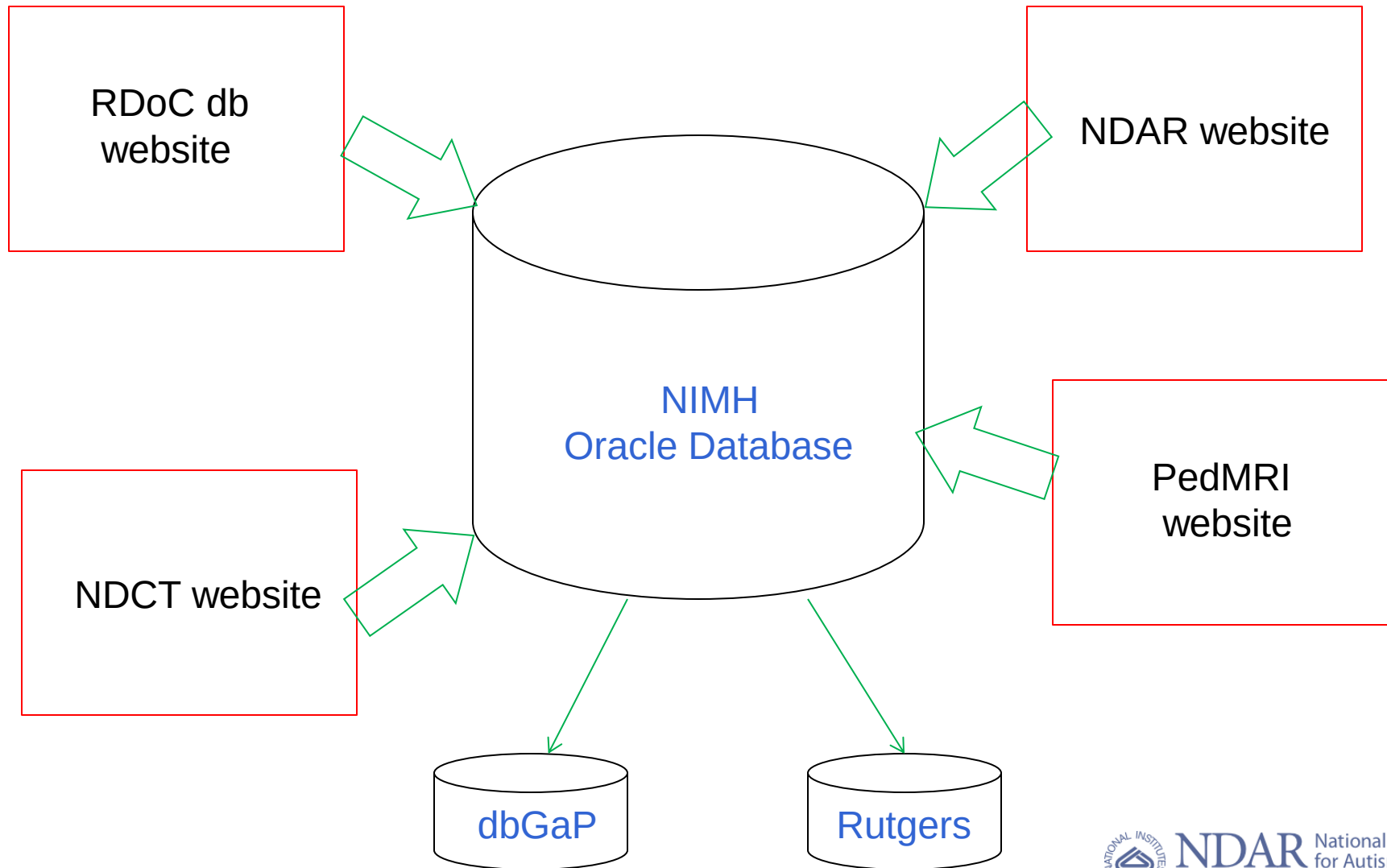
NIMH recently decided to expand NDAR to include:

- 1) Data from Clinical Trials (NOT-MH-14-005)
- 2) Data related to the Research Domain Criteria (RDoC) initiative

- Both databases now exist and data are being loaded.
- The database is shared with NDAR, so queries across all data infrastructures will be easy.



The NIMH Data Repositories



NDAR Overview

- Joint initiative supported by NIMH, NICHD, NINDS, and NIEHS
 - Federal data repository
 - Contains data from human subjects related to autism (and control subjects)
 - Data are available to the research community through a not too difficult application process
 - **Summary data are available to everyone with a browser**
- Begun in late 2006, and first data was received in 2008
- The data types include demographic data, clinical assessments, imaging data, and –omic data
- Currently has data available from over 77,000 subjects
- ~500TB of imaging and –omic data is securely stored in the cloud

NDAR Implementation

- NDAR has deep federation with the following data repositories. This federation allows NDAR to query data in those repositories and to return data to the user from multiple repositories simultaneously.
 - Autism Tissue Program
 - Autism Genetic Resource Exchange
 - Interactive Autism Network
 - Simons Foundation Autism Research Initiative
- NDAR has two key features to allow data standardization and aggregation: data dictionaries and the Global Unique Identifier (GUID)
- Generally, NIH funded investigators are expected to share their data via NDAR. Investigators with funding from other sources are welcome to deposit their data.
- Over 150 studies have registered data.

Data Dictionary – The First Building Block

- The NDAR data dictionary is one of the key building blocks for this repository. It provides a flexible and extensible framework for data definition by the research community.
- 500+ instruments, freely available to anyone
 - 50,000+ unique data elements and growing
 - A research community platform for defining the complex language characterizing autism research
 - Clinical
 - Genomics/Proteomics
 - Imaging Modalities
- Accommodates any data type and data structure
- Extended and enhanced by the ASD research community
- **Curated by NDAR**
- **Allows investigators to quickly perform quality control tests of their data without submitting data anywhere.**



Data Dictionary

Resolve Subject Identifiers

Harmonization Standards

Type:

All

Source:

All

Category:

All

TITLE	SHORT NAME	Category:	SOURCE	CATEGORY	SUBMISSION	CHANGE HISTORY
ACE Family Medical History new	ace_fammedhist01	All	ACE	Med History	Allowed	
ACE Subject Medical History new	ace_subjmedhist01	Acoustics	Common Measures V2, NDAR	Med History	Allowed	show changes
ACE Subject Physical Exam new	ace_physexam01	Behavior	ACE	Phys Exam	Allowed	
AGRE ADOS Module 1 2001	agre_ad0s1_200102	Cognitive	Common Measures V2, NDAR	Diagnostic	Not	
AGRE ADOS Module 2 2001	agre_ad0s2_200102	DTI	AGRE	Diagnostic	Not	
AGRE ADOS Module 3 2001	agre_ad0s3_200102	Demographics	AGRE	Diagnostic	Not	
AGRE ADOS Module 4 2001	agre_ad0s4_200102	Diagnostic	AGRE	Diagnostic	Not	
AGRE ADOS-G Module 1 (2000 or earlier)	agre_ad0s1_pre200002	EEG	AGRE	Diagnostic	Not	
AGRE ADOS-G Module 2	agre_ad0s2_pre200002	ERP	AGRE	Diagnostic	Not	
		Exposure	AGRE	Diagnostic	Not	
		Gen Test	AGRE	Diagnostic	Not	
		IQ	AGRE	Diagnostic	Not	
		MEG	AGRE	Diagnostic	Not	
		MRI	AGRE	Diagnostic	Not	
		Med History	AGRE	Diagnostic	Not	
		Phys Characteristics	AGRE	Diagnostic	Not	
		Phys Exam	AGRE	Diagnostic	Not	
		Questionnaire	AGRE	Diagnostic	Not	
		Resolve Identifiers	AGRE	Diagnostic	Not	
		Social Responsiveness	AGRE	Diagnostic	Not	
		Clinical Assessments	AGRE	Diagnostic	Not	

CPEA STAART ADOS G Module 4

Short Name: cs_adog_4

Version: 02

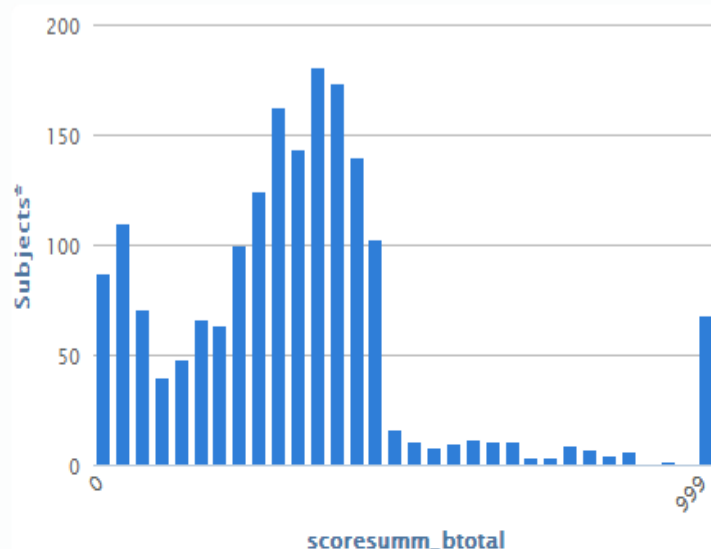
ADOS G Module 4 as defined by the CPEA STAART project

[Download Definitions](#)

ElementName	DataType	Size	Required	ElementDescription	ValueRange	Notes	Aliases
subjectkey	GUID		Required	The NDAR Global Unique Identifier (GUID) for subjects which identifies a subject in NDAR	NDAR*		
interview_date	Date		Required	Date on which the interview/genetic test/sampling/imaging was completed		Required field	ADOS_DATE_STD
cycle	Integer		Recommended	Timepoint information			
patid	String	20	Recommended	src_Subject_id		A Participant ID provided by DM-STAT must be present to process form	
network	String	20	Recommended	Network		CPEA or STAART Network	
site	String	20	Recommended	Site		Study Site	
study	String	100	Recommended	Study		CPEA or STAART Study	
ados_alg_cmsit	Integer		Recommended	Communication Social Interaction Total	0 :: 24		
ados_alg_commt	Integer		Recommended	Communication Total	0 :: 10		
ados_alg_imgcr	Integer		Recommended	Imagination Creativity	0 :: 2		
ados_alg_sbrit	Integer		Recommended	Stereotyped Behaviors and Restricted Interests Total	0 :: 6		
ados_alg_sbrits	Integer		Recommended	ADOS: Stereotyped Behaviors-Restricted Interests Severity Score			
ados_alg_sclit	Integer		Recommended	Social Interaction Total	0 :: 14		
ados_alg_sclits	Integer		Recommended	ADOS: Social Severity Score			
ados_date	Integer		Recommended	ADOS Date			
ados_dia_class	String	50	Recommended	ADOS Classification			
ados_dia_diag	String	75	Recommended	Overall Diagnosis			
ados_dx	String	15	Recommended	ADOS Diagnosis			
ados_lac_afinf	Integer		Recommended	Asks for Information		Nullled value range of: 0;1;2;3 to accept outlier data	

csa6

Distribution for Data Structure: ados1_200102 and Element: scoresumm_btotal

 to [Add Range](#)

* 112 subjects have no value provided for scoresumm_btotal

Description

Social Interaction Total

Value Range

No Restriction

Notes

None Provided

Filters

No filters currently applied. Click bar on chart to add filter.

[Return](#)

coresumm_a_a7	1_scoresumm_a7
coresumm_a_a8	ECT csa7
coresumm_atotal	1_scoresumm_a8
coresumm_a_b1	IRE csa8
coresumm_a_b3	1_scoresumm_ato
coresumm_a_b5	ATION_TOTAL CSS
coresumm_a_b9	ados_mod1_com
coresumm_a_b10	1_scoresumm_b1
coresumm_a_b11	sb1
coresumm_a_b12	1_scoresumm_b3
coresumm_btotal	PRESS_DIRECT sb3
coresumm_abtotal	1_scoresumm_b5
coresumm_a_c1	OY_INTERACT sb5
coresumm_a_c2	1_scoresumm_b9
coresumm_ctotal	e sb9
coresumm_a_d1	1_scoresumm_b10
coresumm_a_d2	JOINT_ATTN sb10
coresumm_a_d4	1_scoresumm_b11
	JOINT_ATTN RJA_r
	1_scoresumm_b12
	de SOCIAL_OVRTU
	1_scoresumm_bto
	CIAL_INTERACT_TO
	_soc_int_total ad
	1_scoresumm_abt
	_SOC_INTERACT_1
	c_ttl ados_mod1_c
	omm sabtotal
	1_scoresumm_c1
	PLAY_TOY PLAY_re
	1_scoresumm_c2
	le sc2
	1_scoresumm_ctot
	AY_TOTAL ados_m
	sctotal
	1_scoresumm_d1
	SENSE_INTERST sd
	1_scoresumm_d2
	1_scoresumm_d4

Global Unique Identifier – the Other Building Block

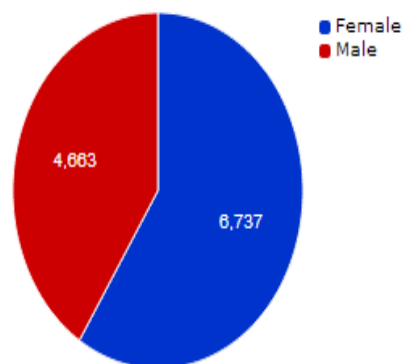
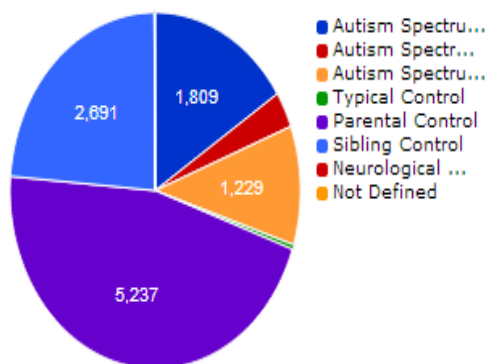
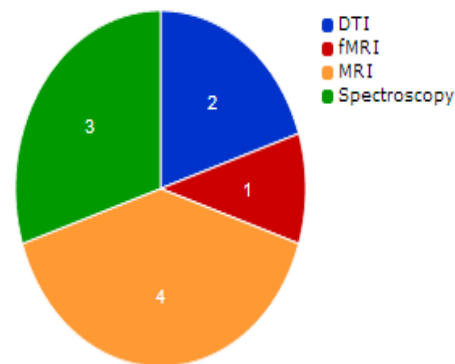
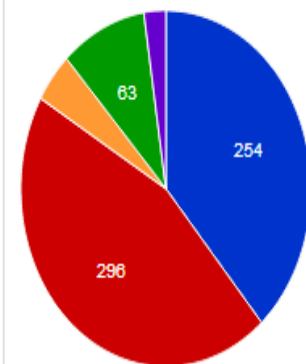
- The NDAR GUID software allows any researcher to generate a unique identifier using some information from a birth certificate.
- If the same information is entered in different laboratories, the same GUID will be generated.
- This strategy allows NDAR to aggregate data on the same subject collected in multiple laboratories without holding any of the personally identifiable information about that subject.
- The GUID is now being used in other research communities and can be made available to you. We have created a video to help with informed consent issues.

<http://www.youtube.com/watch?v=Tb6euCVoous>



**Showing all of the data
in IAN**[Home](#) [Query](#) [Harmonization Tools](#) [Cloud](#) [Contribute](#) [Request Access](#) [Policy](#) [Tutorials](#) [About](#) [FAQ](#)[Query Data](#)[Data from Labs](#)[Data from Papers](#)[Query by Data Dictionary](#)[Query by Concept](#)[Query Instructions](#)

Use "Select Data" below to query the data available in NDAR. Then, select download to create a package and download your results. Use the Data tab above to search in other ways. information on search see our [Methods](#).

Data Distribution by Gender¹**Phenotypic Data Distribution¹****Neuroimaging Data Distribution****Genomic Data Distribution**¹ Numbers reported are subjects by age[Select Data:](#) [All](#) [Basic](#) [Phenotype](#) [Neuroimaging](#) [omicSEARCH:](#) [Experiment](#) [Results](#)[Show Results](#)[Reset All](#)[Download](#)

Showing results 1 - 50 of 11,400 Results per Page: 50

Showing page 1 of 228 Jump to Page: 1

DATA SOURCES	SUBJECT ID	INTERVIEW AGE	GENDER	NDAR CATEGORY	CLINICAL DIAGNOSIS	ADI SCORE	ADOS CLINICAL DIAGN
Interactive Autism Network	8	132	MALE	SIBLING CONTROL	SIBLING CONTROL		
Interactive Autism Network	9	135	MALE	SIBLING CONTROL	SIBLING CONTROL		
Interactive Autism Network	12	384	FEMALE	PARENTAL CONTROL	PARENTAL CONTROL		
Interactive Autism Network	14	108	MALE	AUTISM SPECTRUM AFFECTED	AUTISM SPECTRUM AFFECTED		
NDAR							

- At this point, data has been received. Each subject has a GUID or a pseudo-GUID and the data have been defined in a data dictionary
- How does a user find data?

Query Data

Data from Labs

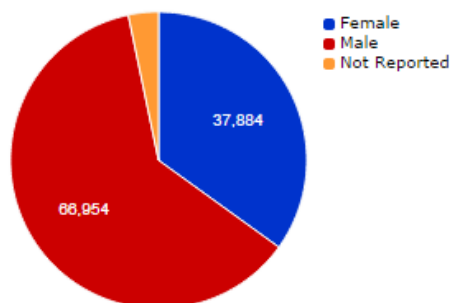
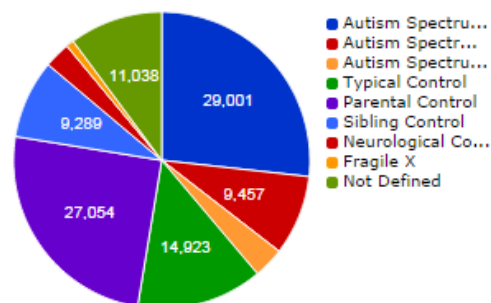
Data from Papers

Query by Data Dictionary

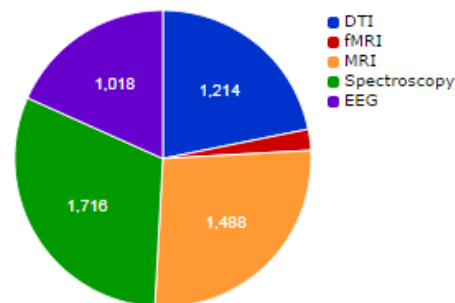
Query by Concept

Query Instructions

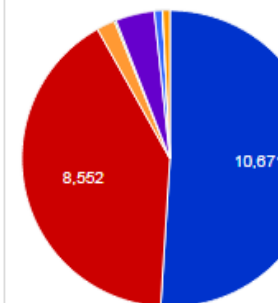
Use "Select Data" below to query the data available in NDAR. Then, select download to create a package and download your results. Use the Data tab above to search in other ways. For more information on search see our [Methods](#).

Data Distribution by Gender¹

Phenotypic Data Distribution¹


Neuroimaging Data Distribution



Genomic Data Distribution


¹ Numbers reported are subjects by age

Select Data: All Basic Phenotype Neuroimaging

omicSEARCH: Experiment Results

Show Results

Reset All

Download Data

Showing results 1 - 50 of 108,386

Results per Page: 50

Showing page 1 of 2,168

Jump to Page: 1

DATA SOURCES	SUBJECT ID	INTERVIEW AGE	GENDER	NDAR CATEGORY	CLINICAL DIAGNOSIS	ADI SCORE
NDAR	1	424	FEMALE	AUTISM SPECTRUM MILDLY AFFECTED	AUTISM SPECTRUM MILDLY AFFECTED	
NDAR	2	426	FEMALE	AUTISM SPECTRUM MILDLY AFFECTED	AUTISM SPECTRUM MILDLY AFFECTED	
NDAR SFARI	3		FEMALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR	4	30	MALE	NOT DEFINED		
NDAR SFARI	5		MALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR SFARI	6	536	MALE	PARENTAL CONTROL	PARENTAL CONTROL	
NDAR SFARI	7	537	MALE	PARENTAL CONTROL	PARENTAL CONTROL	
Interactive Autism Network	8	11	MALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR SFARI	9	136	MALE	AUTISM SPECTRUM SEVERELY AFFECTED	AUTISM SPECTRUM SEVERELY AFFECTED	NEUROLOGICAL CONTROL
NDAR SFARI	10		FEMALE	SIBLING CONTROL	SIBLING CONTROL	
Interactive Autism Network	11	32	FEMALE	PARENTAL CONTROL	PARENTAL CONTROL	
NDAR	12		FEMALE	NOT DEFINED		

An Example of Data Associated with a Particular Laboratory

National Database for Autism Research - Data - Data from Labs - Mozilla Firefox

File Edit View History Bookmarks Tools Help

National Database for Autism Research - ...

www.ndar.nih.gov/data_from_labs.html

Google

Collection Title: *Biological and Information Processing Mechanisms Underlying Autism*

Investigators: Nancy Minshew, M.D. Mark Strauss, Ph.D. Kevin Pelphrey, Ph.D. Marcel Just, Ph.D. Thomas Mitchell, Ph.D. Diane Williams, Ph.D. (Owner: Minshew, Nancy)

Collection Description: This center focuses on elucidating fundamental information processing and neurobiological mechanisms causing autism with studies of infant siblings, first-diagnosed toddlers, and groups of children, adolescents, and adults with and without autism. Project I: Development of Categorization & Facial...

Download Data

Grant Information:

Project Number	Project Title	Start Date	End Date	Organization
P50HD55748	Biological and Information Processing Mechanisms Underlying Autism	08/06/2007	07/31/2012	UNIVERSITY OF PITTSBURGH AT PITTSBURGH

Publications (Showing 3 of 39) [Show All](#)

Bishop-Fitzpatrick, Lauren; Minshew, Nancy J; Eack, Shaun M "Journal of autism and developmental disorders" *A Systematic Review of Psychosocial Interventions for Adults with Autism Spectrum Disorders.*

Dundas, Eva M; Best, Catherine A; Minshew, Nancy J; Strauss, Mark S "Journal of autism and developmental disorders" *A lack of left visual field bias when individuals with autism process faces.*

Mazefsky, Carla A; Oswald, Donald P; Day, Taylor N; Eack, Shaun M; Minshew, Nancy J; Lainhart, Janet E "Journal of clinical child and adolescent psychology : the official journal for the Society of Clinical Child and Adolescent Psychology, American Psychological Association, Division 53" *ASD, a psychiatric disorder, or both? Psychiatric diagnoses in adolescents with high-functioning ASD.*

Data Structures:

Title	Type	Number of Subjects
Autism Diagnostic Interview, Revised (ADI-R)	Clinical Assessments	212
Autism Diagnostic Observation Schedule - Module 1	Clinical Assessments	97
Autism Diagnostic Observation Schedule - Module 2	Clinical Assessments	67
Autism Diagnostic Observation Schedule - Module 3	Clinical Assessments	105
Autism Diagnostic Observation Schedule - Module 4	Clinical Assessments	200
Benton Facial Recognition Test	Clinical Assessments	341
CELF-4 Clinical Eval of Lang Fundamentals, 4th ed	Clinical Assessments	33
CHARGE Family Characteristics Questionnaire	Clinical Assessments	399
CHARGE Medical History	Clinical Assessments	312
CHARGE Physical Exam	Clinical Assessments	299

Genomics **Neuroimaging** **Phenotype**

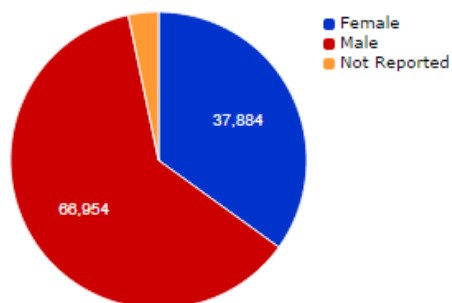
85 91 790 466

Query Data

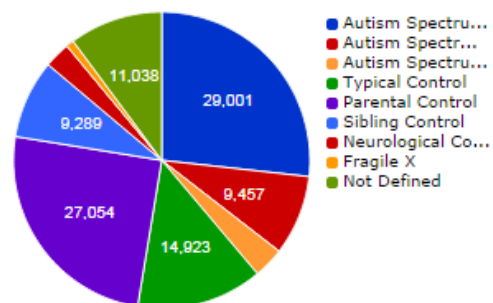
[Data from Labs](#)
[Data from Papers](#)
[Query by Data Dictionary](#)
[Query by Concept](#)
[Query Instructions](#)

Use "Select Data" below to query the data available in NDAR. Then, select download to create a package and download your results. Use the Data tab above to search in other ways. For more information on search see our [Methods](#).

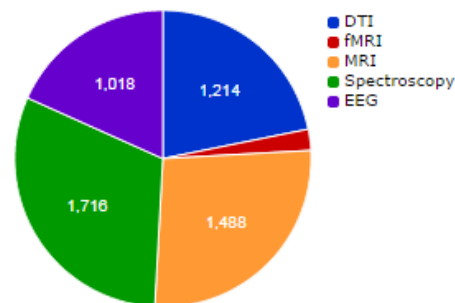
Data Distribution by Gender¹



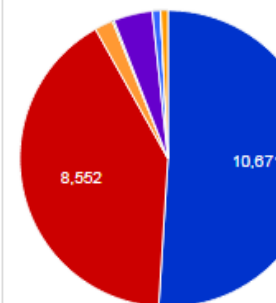
Phenotypic Data Distribution¹



Neuroimaging Data Distribution



Genomic Data Distribution



¹ Numbers reported are subjects by age

Select Data: [All](#) [Basic](#) [Phenotype](#) [Neuroimaging](#) [Genomic](#)

omicSEARCH: [Experiment](#) [Results](#)

[Show Results](#)

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Results per Page:

Showing page 1 of 2,168

Jump to Page:

DATA SOURCES	SUBJECT ID	INTERVIEW AGE	GENDER	NDAR CATEGORY	CLINICAL DIAGNOSIS	ADI SCORE
NDAR	1	424	FEMALE	AUTISM SPECTRUM MILDLY AFFECTED	AUTISM SPECTRUM MILDLY AFFECTED	
NDAR	2	426	FEMALE	AUTISM SPECTRUM MILDLY AFFECTED	AUTISM SPECTRUM MILDLY AFFECTED	
NDAR SFARI	3		FEMALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR	4	30	MALE	NOT DEFINED		
NDAR SFARI	5		MALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR SFARI	6	536	MALE	PARENTAL CONTROL	PARENTAL CONTROL	
NDAR SFARI	7	537	MALE	PARENTAL CONTROL	PARENTAL CONTROL	
Interactive Autism Network	8	11	MALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR SFARI	9	136	MALE	AUTISM SPECTRUM SEVERELY AFFECTED	AUTISM SPECTRUM SEVERELY AFFECTED	NEUROLOGICAL CONTROL
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Interactive Autism Network	11	32	FEMALE	PARENTAL CONTROL	PARENTAL CONTROL	
NDAR	12		FEMALE	NOT DEFINED		

Owner: Christopher Walsh **Owner E-mail:**
christopher.walsh@childrens.harvard.edu **State:** Shared

Using Whole-Exome Sequencing to Identify Inherited Causes of Autism #294

Summary**Cohorts (3)****Measures****Types****Data Analysis**

Investigators: Walsh CA; Yu TW, Chahrour MH, Coulter ME, Jiralerspong S, Okamura-Ikeda K, Ataman B, Schmitz-Abe K, Harmin DA, Adli M, Malik AN, D'Gama AM, Lim ET, Sanders SJ, Mochida GH, Partlow JN, Sunu CM, Felie JM, Rodriguez J, Nasir RH, Ware J, Joseph RM, Hill RS, Kwan BY, Al-Saffar M, Mukaddes NM, et al.

Abstract: Despite significant heritability of autism spectrum disorders (ASDs), their extreme genetic heterogeneity has proven challenging for gene discovery. Studies of primarily simplex families have implicated de novo copy number changes and point mutations, but are not optimally designed to identify...

Results: [Results published in Neuron, Jan 2013](#)
[Readme file for study results](#)

Documents:

Cohorts: **Control - Parents and siblings (411)**

Age: 30 to 1200 months
Gender: Both

Test - ASD (176)

Age: 17 to 1200 months
Gender: Both

Test - ASD-ID (100)

Age: 4 to 1200 months
Gender: Both

Measures: **Primary Measures (7)**

Genomics: Genomics Sample - v03

Secondary Measures (9)

Clinical Assessments:
Neurodevelopmental Phenotype - v01

Genomics: Genomics Subject - v02

Data Analysis: Genotyping/NGS

Type: SNP, CNV, Exome
Sequencing

Software: BWA, GATK

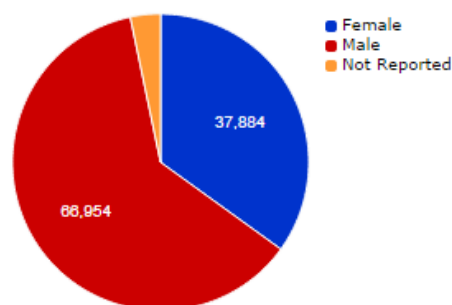
[Return](#)[Edit](#)

Query Data

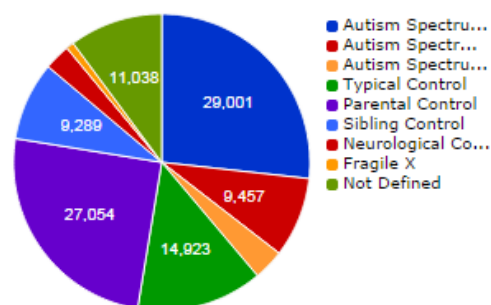
[Data from Labs](#)
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[Query by Data Dictionary](#)
[Query by Concept](#)
[Query Instructions](#)

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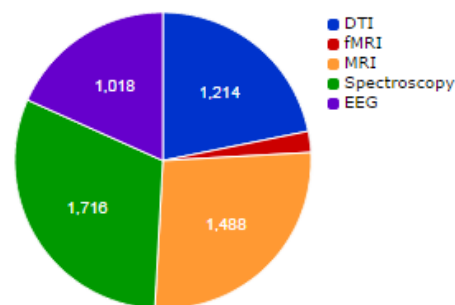
Data Distribution by Gender¹



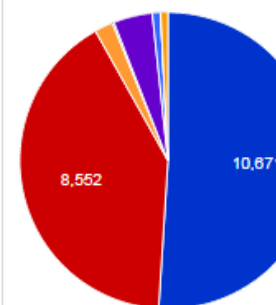
Phenotypic Data Distribution¹



Neuroimaging Data Distribution



Genomic Data Distribution



¹ Numbers reported are subjects by age

Select Data: [All](#) [Basic](#) [Phenotype](#) [Neuroimaging](#)

omicSEARCH: [Experiment](#) [Results](#)

[Show Results](#)

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Results per Page:

Showing page 1 of 2,168

Jump to Page:

DATA SOURCES	SUBJECT ID	INTERVIEW AGE	GENDER	NDAR CATEGORY	CLINICAL DIAGNOSIS	ADI SCORE
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NDAR	2	426	FEMALE	AUTISM SPECTRUM MILDLY AFFECTED	AUTISM SPECTRUM MILDLY AFFECTED	
NDAR SFARI	3		FEMALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR	4	30	MALE	NOT DEFINED		
NDAR SFARI	5		MALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR SFARI	6	536	MALE	PARENTAL CONTROL	PARENTAL CONTROL	
NDAR SFARI	7	537	MALE	PARENTAL CONTROL	PARENTAL CONTROL	
Interactive Autism Network	8	11	MALE	SIBLING CONTROL	SIBLING CONTROL	
NDAR SFARI	9	136	MALE	AUTISM SPECTRUM SEVERELY AFFECTED	AUTISM SPECTRUM SEVERELY AFFECTED	NEUROLOGICAL CONTROL
NDAR SFARI	10		FEMALE	SIBLING CONTROL	SIBLING CONTROL	
Interactive Autism Network	11	32	FEMALE	PARENTAL CONTROL	PARENTAL CONTROL	
NDAR	12		FEMALE	NOT DEFINED		

In Beta

Below are defined ontological concepts that can be used to query all NDAR and federated data. Select a concept and apply the filter to see the number of subjects available. Those that have access may then download. NDAR adopted the published ASD phenotype ontology defined in [Modeling the Autism Spectrum Disorder Phenotype](#) (McCrack et al) as an initial implementation of ontological concepts. For changes or additions to the current model, contact us at ndarhelp@mail.nih.gov.

Available Concepts (1 selected)

Clear Selections Collapse All

General Parameters

Age in Months From: 0 To: 1200

Gender Both

Personal Traits

- ☒ Cognitive Ability
- ☒ Executive Function
- ☒ Language Ability
- ☒ Motor Skills
- ☒ **Stereotyped, Restricted, and Repetitive Behavior**
 - ☒ Involuntary Behaviors
 - ☒ **Restricted and Repetitive Behavior**
 - ☒ **Adherence to Rituals and Routines**
 - ☒ Insistence on Order
 - ☒ Insistence on Routine
 - ☒ **Repetitive Actions**
 - ☒ **Excessive Repetitive Actions** ⓘ
 - ☐ No Repetitive Actions ⓘ
 - ☒ Compulsive Behavior

Apply Filters

Personal Traits > Stereotyped, Restricted, and Repetitive Behavior > Restrict...

Concept: Personal Traits > Stereotyped, Restricted, and Repetitive Behavior > Restricted and Repetitive Behavior > Adherence to Rituals and Routines > Repetitive Actions > Excessive Repetitive Actions

Rules: ⓘ cddl66 in (1;2)
ⓘ rbsr_q18 between (1::3)
ⓘ rbsr_q21 between (1::3)
ⓘ rbsr_q22 between (1::3)
ⓘ scl65 in (moderately; quite a bit; extremely;)
ⓘ rbsr_q5 between (1::3)

OK

Results in 750 subjects
being discovered

How is NDAR being used?

- With biological databases, it is not true that if you build it they will come.
- More than 270 users have been granted access to NDAR. Data access is separate from those who are depositing data.
- David Hessler and collaborators used NDAR to collect and analyze their data in a private space before publication (“Psychometric study of the aberrant behavior checklist in Fragile X syndrome and implications for targeted treatment”, J. Autism Dev. Disord. (2012), 42:1377-1392).
- David M. Richman and colleagues have published a study, “Predictors of self-injurious behavior exhibited by individuals with autism spectrum disorder” where all of the data in the paper came from NDAR (J. Intellect. Disabil. Res. (2013), 57:429-439).
- Vinod Menon and colleagues have published a paper, “Brain hyperconnectivity in children with autism and its links to social deficits” (Cell Rep. (2013), 5(3), 738-747. where some of the data is from NDAR and some is newly measures.
- Many are using data from NDAR as part of NIH grant applications.



NDAR Summary

NDAR, is a useful data archive that makes autism data:

- A) Discoverable – federation, useful queries, XML web services
- B) Useful to Others – data access, data QC, data analysis pipelines
- C) Citable – data from labs, data from papers
- D) Linked to the Literature – data link in PubMed

