Panel 5: Accelerating Improvements in IDD Data to Inform System Change Efforts

September 22, 2026

Christine Brown, Moderator

I/DD Counts Summit

Better IDD data can drive system change

- Improve how people with IDD are identified in surveys, administrative records, and health systems
- Use innovative data sources and methods to learn what existing systems miss
- Connect state, healthcare, research, and federal efforts
- How can better data improve research, policy, programs, and services?

Panelists

- **Nassira Nicola**, Massachusetts Department of Public Health
- **Mai Pham**, Institute for Exceptional Care
- **Melissa Parisi**, National Institutes of Health
- **Nik Koscielniak**, Patient-Center Outcomes Research Institute

- Moderated discussion and audience Q&A

Developing Disability Data Standards in MA

Nassira Nicola
Massachusetts Department of Public Health, Office of Health Equity and Community Engagement, Deputy Director for Access and Inclusion

IEC

Making IDD Visible – finding opportunities in Electronic Health Records

September 2026

Why Do We Need to Make IDD Visible?

IDD Remains Invisible to Healthcare Stakeholders

We can identify only a small part of the true population of people with IDD in medical claims and electronic health records (EHR) compared to other data like surveys & CDC data

- Based on analyses of Epic's Cosmos data, PCORNet EHR data, and Milliman's commercial claims
- 1-3 per 1,000 instead of 30-50 per 1,000

There is worse undercounting for certain subgroups

- Adults, females, Black/Brown, poor, rural
- Those with less visible disabilities/features

IEC

Likely causes of IDD invisibility in data

- Limited clinical training to detect IDD
- Limited resources for testing and diagnosis
- Missed co-occurring IDD conditions
- Lack of self-disclosure due to stigma
- Lost medical history during life & care transitions

e-IDD: An AI Tool To Comprehensively Identify Children & Adults With IDD

Better data is a critical first step in improving the lives of people with IDD

e-IDD can be used in daily decision-making

- For people with IDD across lifespan
- Usable by clinicians, payers, government, researchers
- Accounts for sex/racial/ethnic/geographic/lifespan diversity
- Developed on a large population

More data can improve detection

- Clinical and billing data from electronic health records
- Public socioeconomic data

IEC

e-IDD Can Support Better Outcomes For Children & Parents

Black mother of twins cannot get pediatrician to take her concerns about developmental delays in one child seriously.

- **Challenges at school:** Pediatrician diagnoses “conduct disorder”
- **Clinic manager uses e-IDD:** Child flagged with high risk of having undiagnosed IDD. Clinical leaders ask pediatrician to revisit case.
- **Formal Evaluation Confirms Autism and ADHD Diagnosis**
  - Child gets access to counselors & specialists
  - Social worker helps get individualized education plan & Medicaid coverage
- **Child Starts Receiving Services:** Less challenging behaviors; mother feels supported and less anxious

e-IDD Can help Public Payers Improve Cost/Quality Outcomes

Medicaid agency uses e-IDD to design value-based payment programs

- **Medicaid agency uses e-IDD**
  - Expands recognized population by 3-fold
- **Medicaid shares e-IDD data with health plans**
  - Plans and providers formally confirm diagnoses.
- **Health plans share e-IDD data with providers**
  - Providers see that their IDD populations are much larger than expected
- **Health plan offers better contracts to clinical groups**
  - Providers invest in tailored care strategies and reduce costs, improve quality

Our approach to Make IDD Visible?

First Focus on Autism & Intellectual Disability (ID)

- **Account for over 75% of people with IDD**

- **Harder to diagnose (especially with co-occurring conditions)**
  - Can’t be confirmed with a lab test
  - Greatest opportunity to improve detection

- **Diagnosis is uneven (biased)**
  - Opportunity to improve fairness

Use Clinical Expertise & Lived Experience To Develop "Computable Phenotypes"

- Clinicians experienced in working with autistic children and/or adults and those with ID
- Autistic adults and adults with ID
- Family members of autistic children and those with ID
- What is a **computable phenotype**?
  - Descriptions of how an autistic person or someone with ID might present clinically and/or socially, expressed in a way that allows use of data to identify cases.
  - For example – a six-year-old child who is non-speaking and anxious but has met all their physical developmental milestones.

Apply Artificial Intelligence

- Use AI approaches (some terms include **logistic regression models, decision-trees, random forest, neural networks, and deep neural networks**).
- Also use "**large language models**," like ChatGPT.
- Use many kinds of data in EHRs, such as visit notes and problem lists.
- Use information on a person's social context in the EHR.
- Use publicly available social data such as urban/rural location
- Use **agentic AI** to support better documentation and communication with patients and caregivers about the results.

Favor High Finding People Who *Might* Have ASD/ID

- The problem we're trying to solve is invisibility of IDD
- e-IDD is intended to serve as a **screening tool**
- Favor design features that **over-detect** potential autism and/or ID
- e-IDD will show how high a person's **risk** is for having undocumented autism and/or ID
- Users (clinicians, health plans, government, researchers) will decide the level of certainty they want to apply for their specific use
  - Clinicians may be willing to consider lower risk of IDD (e.g., 70%) because they can approach individual patients to confirm diagnosis
  - Researchers may focus on higher risk of IDD (e.g., 95%) depending on the question they're trying to answer, and because they cannot confirm individual cases

How Will We Make Sure e-IDD Tools are Safe and Fair?

All Team Members Receive Training In Relevant Content Areas

- **IDD Lived Experience 101:** Common life and healthcare experiences for children and adults with autism and/or ID, and for family members.
- **IDD Clinical 101:** Clinical issues common in people with autism and/or ID.
- **AI 101:** Key concepts and decision-points.
- **Algorithmic Bias 101:** Common sources of unfairness (bias) and ways to address them.
- **Ethics 101:** Ethical framework for decision-making throughout the project.

We Have an Ethics Workgroup To Maximize Benefits and Minimize Risks to People with IDD

Possible benefits

- Better access to services and supports
- People with IDD learning more about themselves
- Redefine what IDD means from the person's perspective
- Better clinical decisions
- Improve health outcomes

Possible Risks

- Stigma, negative self-image
- Feeding the narrative of autism or IDD as an "epidemic"
- Results used to deny benefits, put employment at risk, institutionalize, or do other harm
- How e-IDD works or is used not made transparent to people
- Invaded privacy

IEC

What Have We Learned So Far?

EHRs have a lot of useful “hidden” information

- **Text that relates to IDD:**
  - Diagnoses and ***misdiagnoses***
  - Descriptions of behavior and relationships
  - Clinicians’ interpretation and actions
  - Information from caregivers
- **Documents that relate to IDD:**
  - From other healthcare providers: neuropsychological testing, speech therapy reports, etc.
  - From outside healthcare: IEPs, SSI applications, etc.
- **Actions by the clinician:**
  - Referrals to specialists (OT, PT, speech therapy, neurology, etc.)
  - Orders for testing
  - Orders for medical equipment

NIH Data Systems and Programs to Improve Intellectual and Developmental Disabilities Research

Melissa A. Parisi, MD, PhD

Eunice Kennedy Shriver **National Institute of Child Health and Human Development**

I/DD Counts Summit

September 21-22, 2026

National Institutes of Health
Turning Discovery Into Health

Three NIH Tools to Accelerate Research for IDDs

01 NIH Strategic Plan for Disability Health Research

02 INCLUDE Project Down Syndrome Research

03 Autism Data Science Initiative

National Institutes of Health
Turning Discovery Into Health

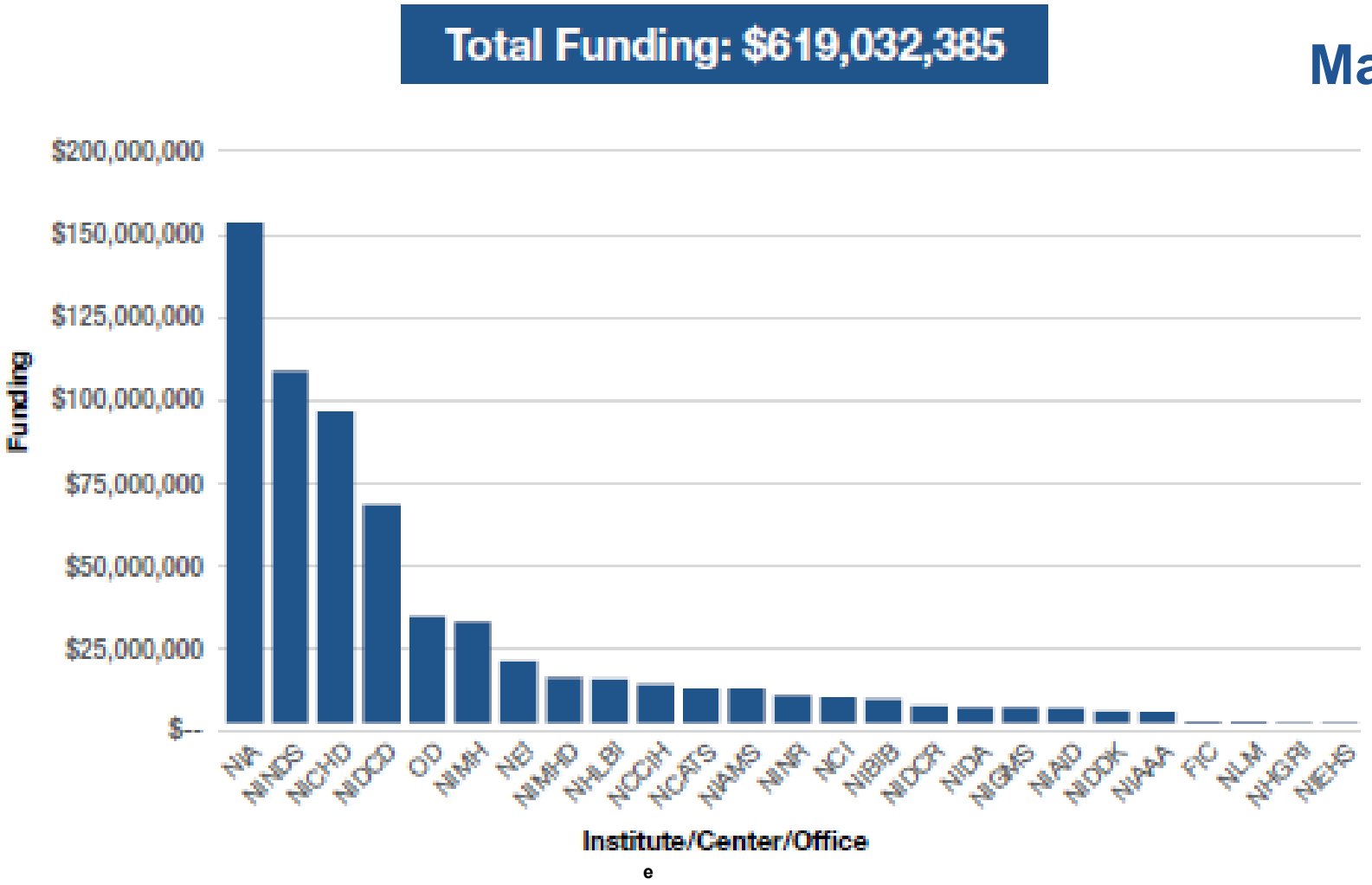
NIH Strategic Plan for Disability Health Research

FY26-FY30

Learn more and read the full plan

https://dpcpsi.nih.gov/disabilityhealthresearch/strategicplan

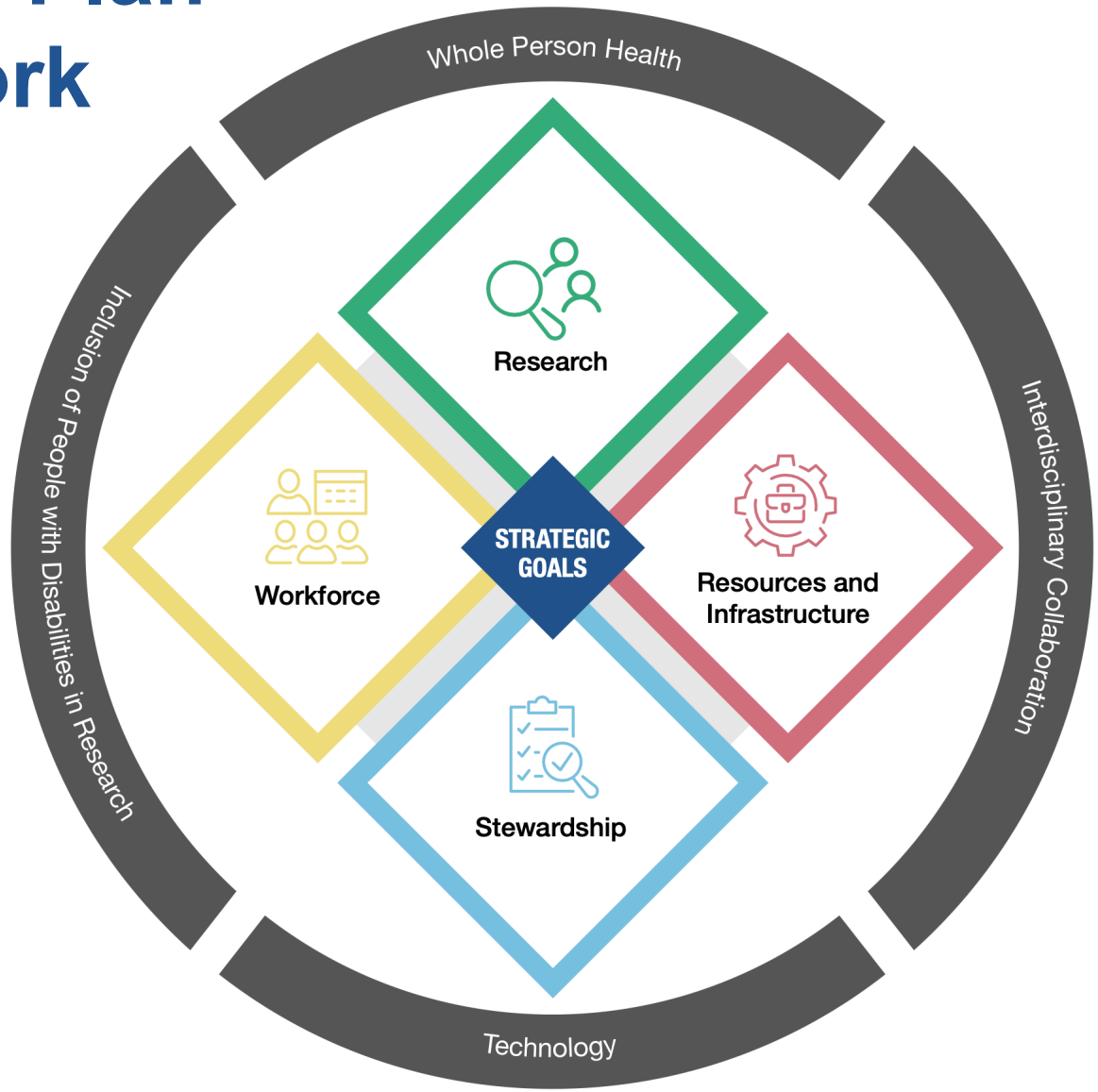
Disability Health Research Landscape at NIH in FY24



Major Developments

- **2022**: NIH Advisory Committee to the Director Issues Disabilities Report
- **2023**: People with Disabilities are given NIH Health Disparity Designation
- **2024-2025**: Disability Health Research Working Group (DHRWG)
- **2026**: Strategic Plan for Disability Health Research published

Strategic Plan Framework



Key Elements

- Research to better define disability
- Research to reduce disparities
- Health promotion and disease prevention
- Train researchers
- Maximize inclusion of people with disabilities in research
- Promote data sharing
- Ensure scientific integrity

INCLUDE Project: INvestigation of **C**o-occurring conditions across the **L**ifespan to **U**nderstand **D**own syndrom**E**

The INCLUDE Project launched in June 2018 to address a Congressional directive that called for a new **NIH-wide research initiative** on critical health and quality-of-life needs for individuals with Down syndrome (DS).

The INCLUDE Project:

- **Supports research that investigates conditions** that affect individuals with DS and the general population, such as Alzheimer's disease, autism, cataracts, celiac disease, congenital heart disease, and diabetes.
- **Aims to increase the number of investigators and trainees** studying DS.
- Works in partnership with the Down syndrome community to identify areas of need and **support work that benefits everyone.**

DS-CDP: A $32 Million Study of Down Syndrome

from birth through adulthood

INCLUDE
Collaboration for Down Syndrome Progress

GOAL
Learn about health across the lifespan

- ☑ Learn how health needs differ from person to person.
- ☑ Learn how often other health conditions occur.
- ☑ Study how life experiences and environment affect health.
- ☑ Learn which treatments may help improve health.

STUDY STATUS

1 One shared study plan
Research and clinical teams created a common protocol.

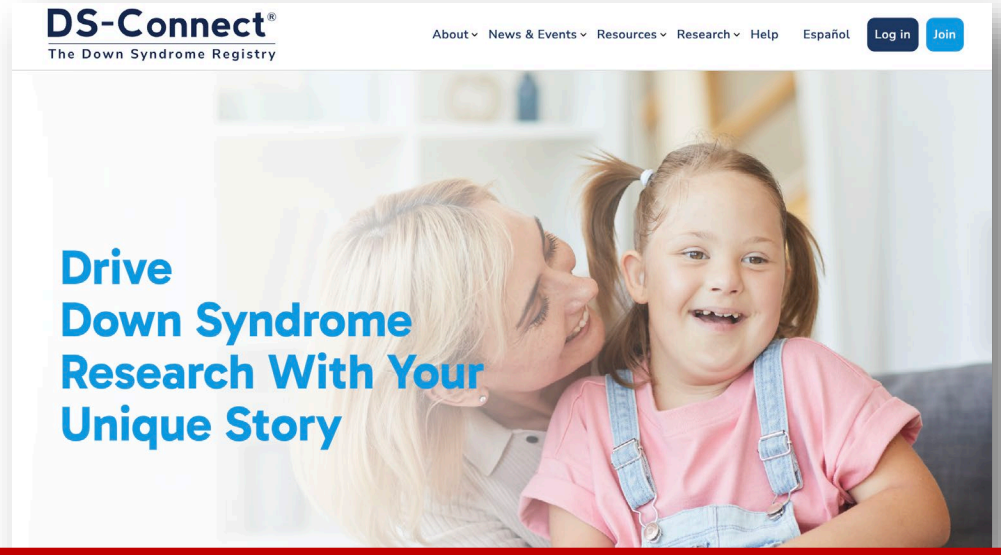
2 Enrollment began
People began joining the study in spring 2026.

DS-Connect®

The Down Syndrome Registry

A **secure, online survey-based** tool to collect basic information about people with Down syndrome

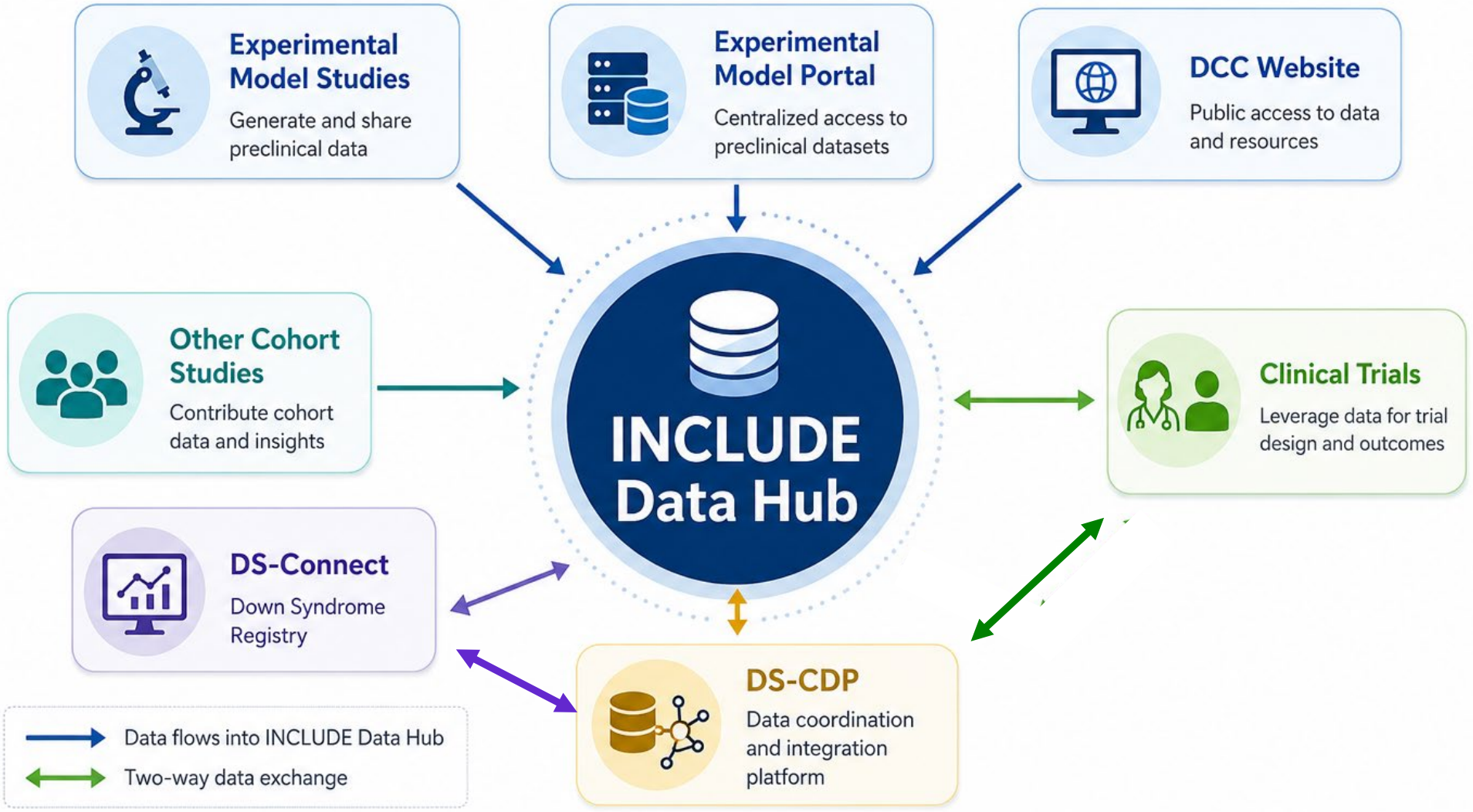
6300+ registered globally as of Aug 2026



ds-connect.org

Connecting families with researchers since2013!

INCLUDE Project: Data Ecosystem



Principles for INCLUDE Data Sharing:

- Share data without personal identifiers
- Collect data from majority of funded studies
- Encourage secondary data use
- Engage the community

Autism Data Science Initiative (ADSI)

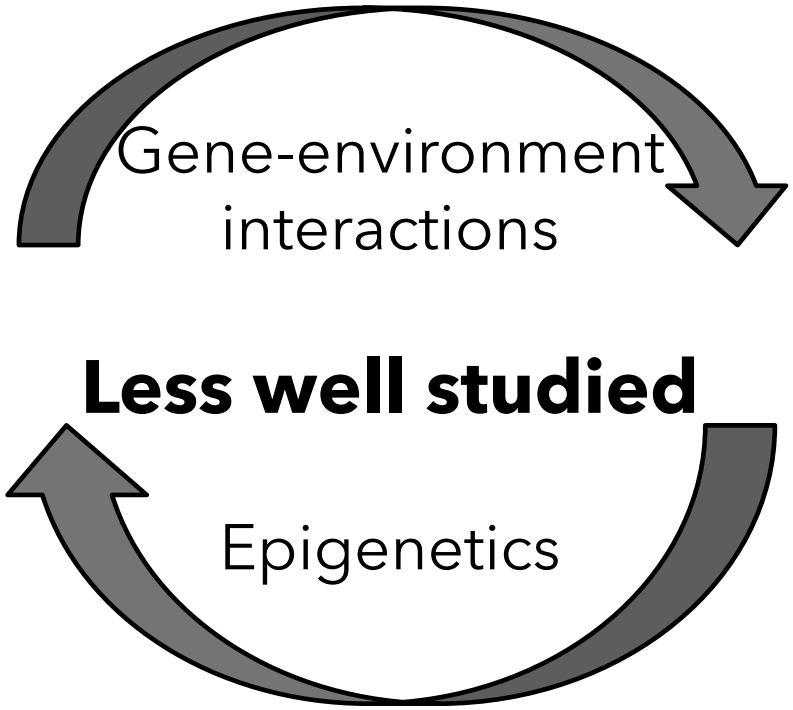
The overarching goal of the **Autism Data Science Initiative** (ADSI) is to further our understanding of autism and develop new knowledge that could be used to improve health outcomes for people on the autism spectrum.

In Sept 2025, NIH issued 13 awards with a total investment of $50 M over 24-36 months

Complex Factors that Lead to Variability in Autism

Genetic Risks

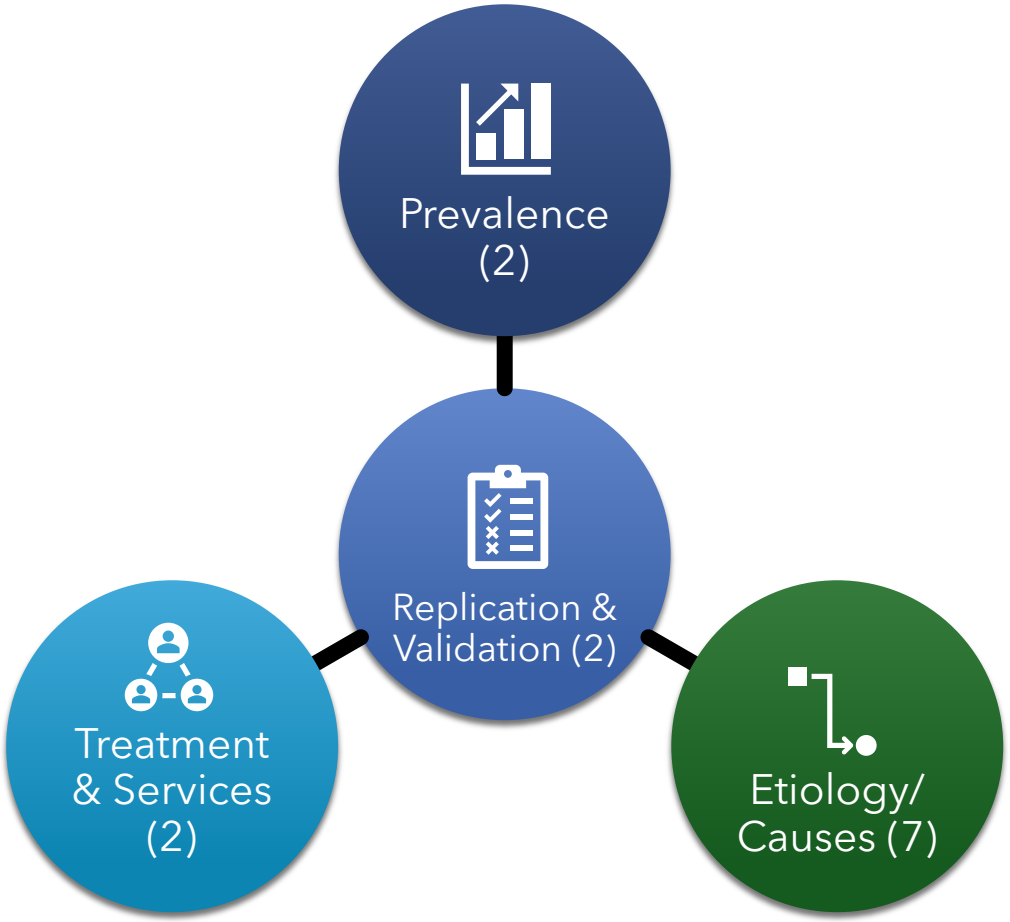
- High recurrence in families
- Complex genetics
  - Single gene syndromes
  - Aneuploidies (T21)
  - Copy number variation
  - New mutations
  - Common gene variants and polygenic risk



Nongenetic Factors

- Complicated exposures (e.g., psychosocial, nutrition/diet, medical, environmental, chemical)
- Windows of susceptibility
- Large range of exposures that could impact brain development during pregnancy

Types of Studies in ADSI: 13 total awards



Contributing Factors:

- Environmental exposures (e.g., pesticides, PFAS, air pollution, heavy metals)
- Pharmaceuticals
- Maternal diet (e.g., ultra-processed foods, folate)
- Community and Neighborhood
- Gene-Environment Interactions
- Maternal health
- Prenatal/perinatal factors

Outcomes:

- Diagnosis and autistic traits
- Longitudinal trajectories
- Mental health
- Community, employment, and educational services
- Language and communication
- Co-occurring conditions

ADSI Key Points

1. **Integrate large existing data resources** with rigorous privacy protections for analysis by autism researchers.

2. **Identify gaps in available data** and support **targeted data generation** specifically to fill those gaps.

3. Support cutting-edge data analysis to understand the contribution of genetic and nongenetic (environmental) factors to the **causes of autism** *and* identify **intervention outcomes and the use of services for autism**.

4. Support independent **validation and replication** of the Initiative's findings **while the studies are ongoing (a first for an NIH program!)**.

THANK YOU!

Melissa Parisi: parisima@mail.nih.gov

PCORnet®

Building Data Capacity for Comparative Clinical Effectiveness Research and Other Analyses on IDD

Nik Koscielniak, PhD, MPH, MSOT, OTR/L
Program Officer
Research Infrastructure and Innovation

pcori® PATIENT-CENTERED OUTCOMES RESEARCH INSTITUTE

Get To Know PCORI®

PCORI

- Nonprofit that funds patient-centered comparative clinical effectiveness research (CER)
- Widely acknowledged as a leader in driving U.S. clinical research to become more patient-centered
- Provides funding for CER, engagement in research, dissemination and implementation and research infrastructure projects

What is Patient-Centered CER?

Comparative Clinical Effectiveness Research (CER) compares benefits and harms of two or more medical treatments, clinical or care delivery strategies or other approaches to health care that are currently in use in clinical practice.

- Focuses on outcomes that are important to patients and other stakeholders
- Seeks to help patients and other stakeholders make better-informed decisions

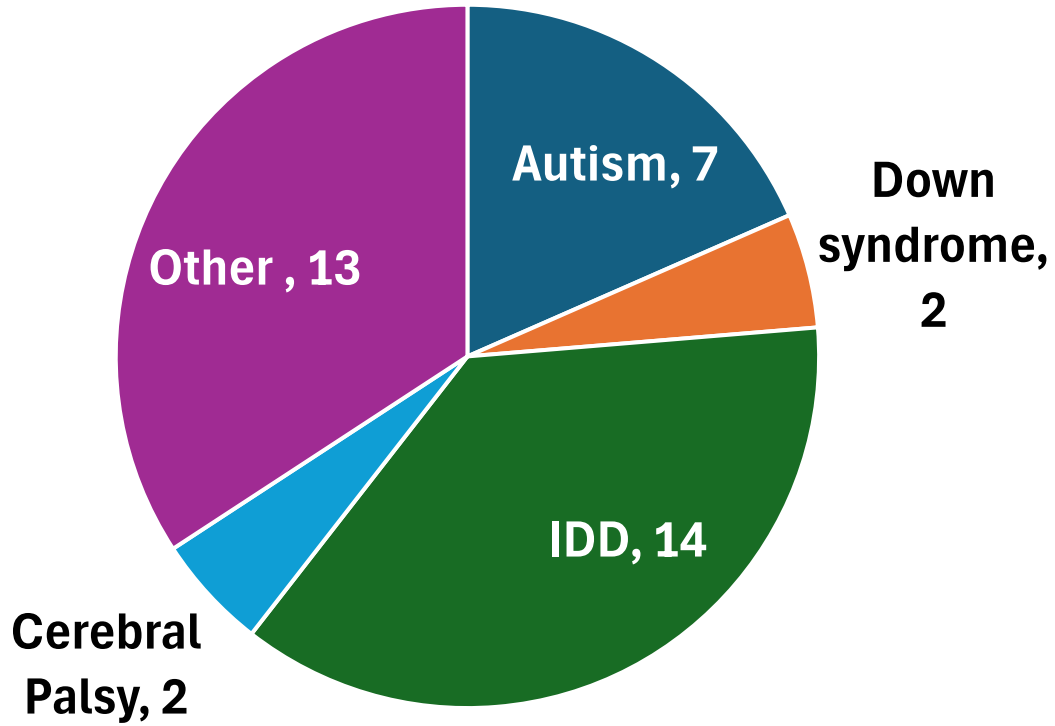
PCORI IDD Research Portfolio

- Patient-centered CER studies evaluate at least two or more interventions and/or service delivery options with evidence of efficacy or widespread use to decide which is best at meeting the needs of people with IDD.

Focus Areas

Community living
Healthcare use
Mental health
Caregiving
Symptom management/medications

38 CER Awards



As of cycle 2 2025

PCORnet®: Access to Clinical Data and Research Expertise on a National Scale

8

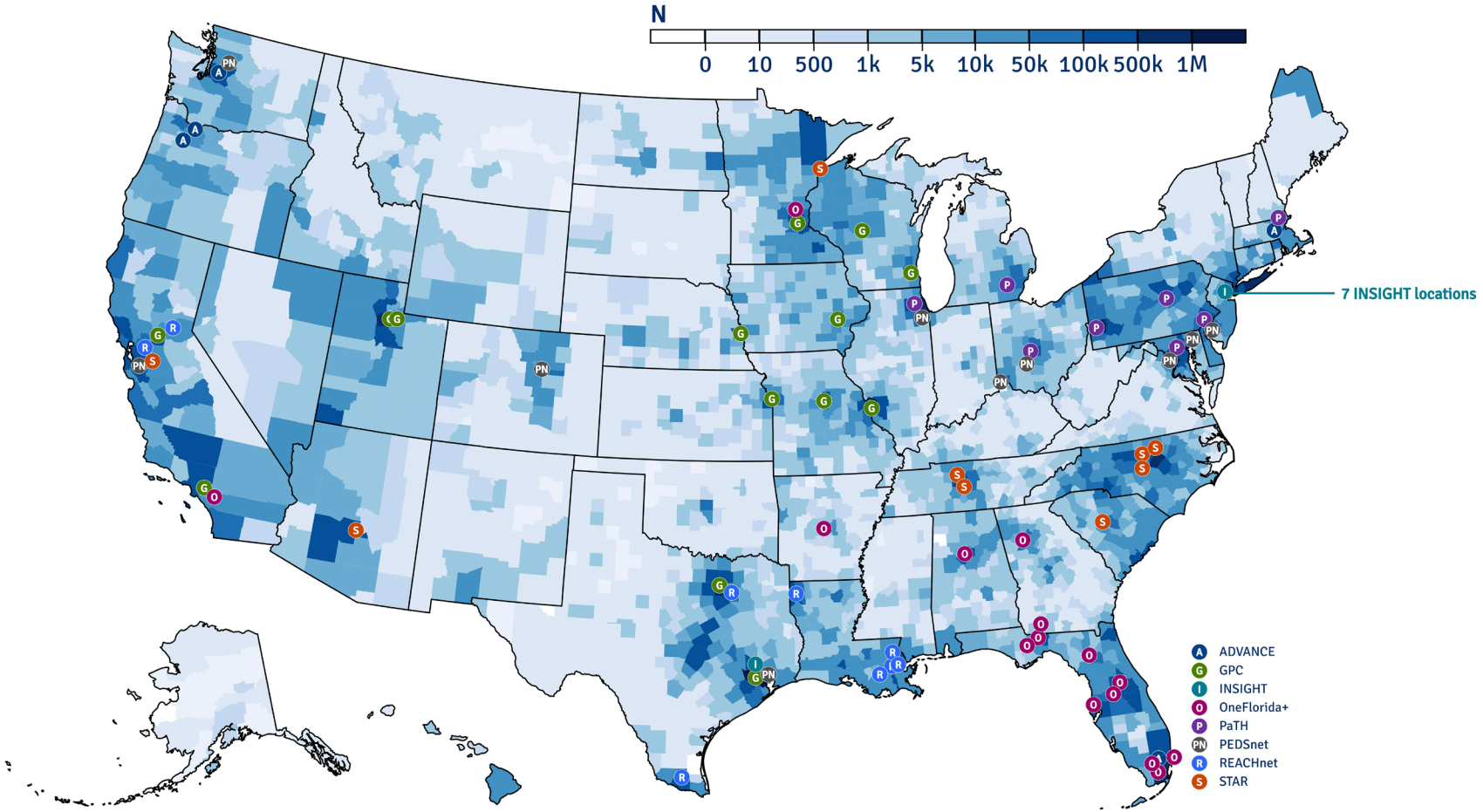
Clinical Research Networks

13K+

Clinical Care Sites

50M+

Patients With an Encounter



PCORnet®: A Vetted National Resource

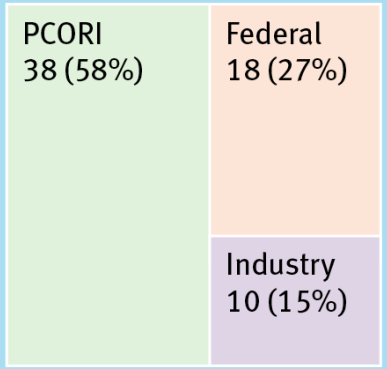
50
States
PCORnet is linked to patients in every state in the U.S.

78
PCORnet® Network Partners
8 PCORnet® Clinical Research Networks across the U.S.

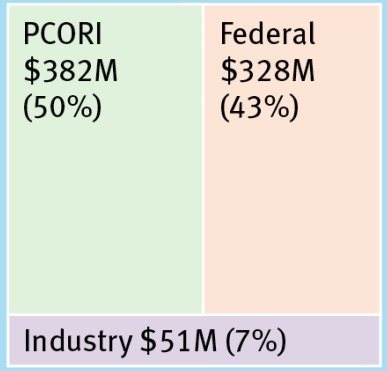
>50
Million People
PCORnet represents data from everyday healthcare encounters with >50 million people annually across the U.S.

1,110
Publications
Powered by PCORnet®

66
PCORnet® Studies
Funded by



$760
Million
PCORnet® Studies funding by source

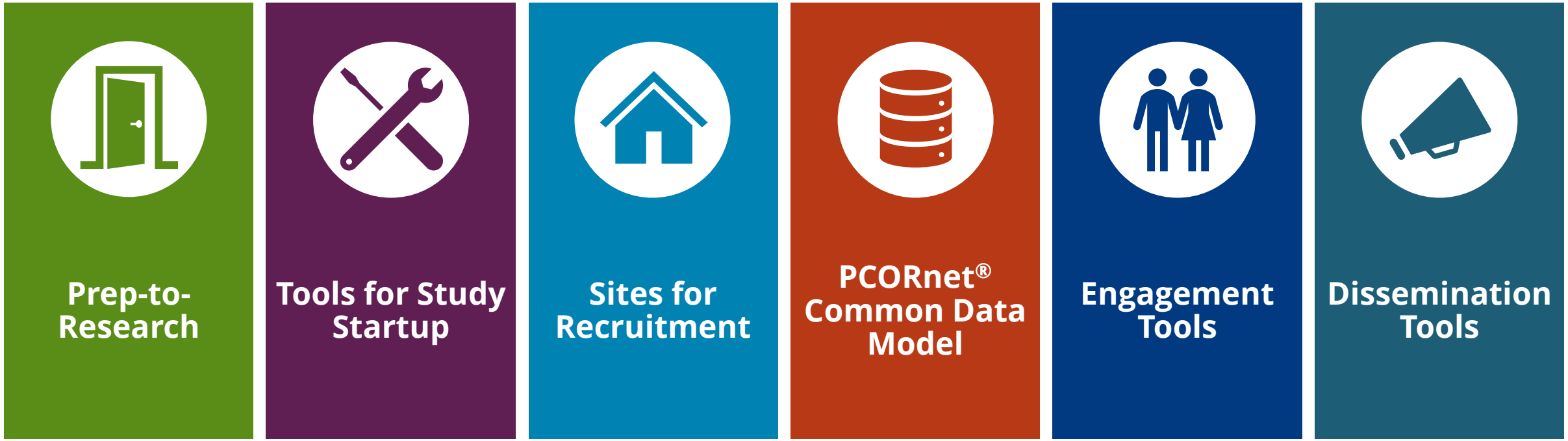


Over the last 10 years, PCORnet® Studies have already answered critical patient-centered questions on heart disease, metabolic conditions, obesity, and more, demonstrating the power of PCORnet® to improve patient care through efficient, high-quality research.

Research that leverages this network is tapping into a proven engine for discovery.

Data as of May 2026

Unique Features of PCORnet® That Investigators Can Use To Optimize Patient-Centered Research



The PCORnet® Front Door

The access point for PCORnet resources and services

A "knock" on the PCORnet® Front Door can support:

STUDY DESIGN

Preliminary data for proposals, effect sizes, and potential study power

DATA NETWORK REQUEST

- Get data insights from PCORnet® Clinical Research Networks
- Obtain aggregated results for informing research project development

CONNECTIONS TO NETWORK COLLABORATORS

- Partners to co-design research
- People with specific expertise

PCORnet® STUDY SUPPORT

Deeper partnership with PCORnet provides access to best practice sharing, patient engagement, and transparent quality improvement initiatives

Learn More About the Front Door

Ways PCORnet® Can Support IDD Research

In 2024, PCORI awarded funding to PCORnet to convene an IDD Workgroup to develop a roadmap to demonstrate a community-engaged approach to accelerate patient-centered CER using PCORnet, including:

- Identifying critical gaps and needs in knowledge, and prioritizing unanswered CER questions
- Conducting a PCORnet query to identify populations with specific conditions that are eligible for study participation
- Identifying questions that can be answered with PCORnet now as well as recommendations for infrastructure enhancements

- **Ex: What is the comparative effectiveness of clinic-based peer support vs community-based peer support for individuals with IDD on health-related quality of life, self-reported connection to community resources, and overall well-being?**

Access the IDD Workgroup Report

Franklin MS, Dolor RJ, Hendren S, Jelliffe-Pawlowski L, Wiley S, Myers SM, Quiñones A, Nowell K, Kanne SM, Kramer JM, Thompson B, Thomas E, Bello J, Pham HMH, Maslow GR. Medical Care. 2026 Feb 1;64(2S Suppl 3):S301-S313. doi: 10.1097/MLR.0000000000002259

Characterizing Patients with IDD Served by PCORnet® Network Partners

- PCORnet® Network Partners serve many patients with IDD-related conditions and co-occurring chronic conditions
- PCORI and the Coordinating Center for PCORnet® worked together to identify characteristics of the patients with IDD-related conditions and an encounter at a PCORnet site in 2025.

- PCORnet can be used to:
  - Describe use of healthcare services by patients with IDD
  - Describe experience of chronic conditions for people with IDD
  - Show how people with IDD can be identified in research data
  - Identify patients with IDD
- PCORnet supports the largest known, national-scale descriptive analysis of IDD population using EHR data
  - PCORnet can support large studies for IDD research

Learn more about PCORnet Population Insights

IDD Population Insights Report From PCORnet®

Characteristics of Patients With an Encounter in 2025

3.8M
Patients seen in 2025 with at least 1 **IDD Condition** since 2021

45.0%
Female patients

IDD CONDITIONS	N	S%	P%
ADHD	2,038,650	53.6	3.8
Learning Disability	1,170,205	30.8	2.2
Autism Spectrum Disorder	681,450	17.9	1.3
Developmental Delay	448,308	11.8	0.8
Cerebral Palsy	139,547	3.7	0.3
Congenital Malformations of Brain	138,739	3.6	0.3
Failure to thrive	121,255	3.2	0.2
Intellectual Disability ICD-10 Codes	73,378	1.9	0.1
Down Syndrome	68,861	1.8	0.1
Muscular Dystrophy	39,414	1	0.1
Additional conditions representing less than 1% each in table at end of these slides.	35,358	0.9	0.1

28.2%
Medicaid

PAYER STATUS	N	S%	P%
Private	1,527,436	40.2	40.7
Medicare	191,104	5	15.5
Medicaid	1,070,973	28.2	15
Other	162,748	4.3	3.6
Missing	851,473	22.4	25.2

Learn more about PCORnet Population Insights

53,351,854 patients with at least one face-to-face encounter and a recorded diagnosis at a PCORnet® site in 2025.

IDD Population Insights Report From PCORnet®

Characteristics and Services for of Patients Identified with IDD and Encounter in 2025

CLINICAL SERVICES	N	S%	P%
Wellness Visit	1,510,281	39.7	24.4
Audiology	441,771	11.6	4.1
PT/OT Treatments	204,680	5.4	3.9
PT Evaluation/Re-evaluation	191,322	5	3.8
Vision services	114,723	3	3.7
Psych Evaluation/Psychotherapy	314,965	8.3	2.1
Oral Exams	72,460	1.9	1.7
OT Evaluation/re-evaluation	91,727	2.4	1.7
Speech Therapy	91,149	2.4	0.6
Neuropsych Evaluation	18,353	0.5	0.1
Orthotic	6,518	0.2	0.1
Wheelchair	7,021	0.2	0.1

53,351,854 patients with at least one face-to-face encounter and a recorded diagnosis at a PCORnet® site in 2025.

26.4%
With Telehealth visits

Demographics and Geography

- 63.6% White, 14.2% Black or African American, 3.6% Asian, 3.2% multiple race
- 93.2% have address data at 5-digit ZIP
- ~4% live in small towns or rural areas

Learn more about PCORnet Population Insights

Example of a Focused Funding Award

SAE: Gastrointestinal Health

What PCORI heard:

- Gastrointestinal (GI) symptoms are prevalent in the IDD population
- Strategies addressing GI health are often applied without evidence of effectiveness

Funding opportunity:

- CER addressing GI health that consider the physical, cognitive, behavioral and communication needs of this population

FUNDED CER AWARD

Comparing Polyethylene Glycol 3350 versus Linaclotide in Autistic Children with Constipation

BRIEF REPORT

High Burden of Constipation Among Autistic Youth: A Nationwide Query Powered by PCORnet

- More than double the prevalence of constipation in autistic youths (aged 6-17) compared with non-autistic youths
- Constipation associated with greater unplanned use of healthcare such as emergency room visits or hospitalizations

Chumpitazi BP, Zelinski A, Marchesani N, Brantner CL, Tomaiuolo M, Kappelman MD. High Burden of Constipation Among Autistic Youth: A Nationwide Query Powered by PCORnet. Am J Gastroenterol. 2026 Feb 18. doi: 10.14309/ajg.0000000000003967.

Example of a Focused Funding Award
SAE: Improving GI Health for Individuals with IDD

FUNDED CER AWARD

Comparing Polyethylene Glycol 3350 versus Linaclotide in Autistic Children With Constipation

Principal Investigator – Bruno Chumpitazi (awarded 2026)

Organization – Duke University

Key Study Features

Design: Randomized controlled trial of 320 autistic children (ages 6-17) with functional constipation across 30 primary care and behavioral health clinics across the U.S.

- Polyethylene Glycol 3350
- Linaclotide

Primary Outcome: Constipation burden

Secondary Outcomes: Spontaneous bowel movements, undesirable behaviors, patient, caregiver and clinician perspectives on acceptability and barriers to implementation.

Community Partners:

- Individuals with autism, parents of autistic children, clinicians, patient advocacy organizations, payers and health policymakers.

Leveraging PCORnet®

- Learning system to assess generalizability of study findings

Comparing Polyethylene Glycol 3350 Versus Linaclotide in Autistic Children With Constipation | PCORI

Contact Information

Nik Koscielniak, Program Officer

nkoscielniak@pcori.org

www.pcori.org

/PCORI

@PCORI_ORG

PCORI

@PCORI

@PCORINews