



Promising Research at the NIH

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Global Down Syndrome Research and Medical Care Roundtable
June 25, 2020



Eunice Kennedy Shriver National Institute
of Child Health and Human Development





What if researchers studied...

- ...why some babies with Down syndrome get leukemia?
- ...the ways children with Down syndrome communicate, learn and develop?
- ...the best strategies for kids with Down syndrome to get the right eyeglass prescription?
- ...treatments for skin disorders in adults with Down syndrome?
- ...how best to measure sleep apnea in a home setting rather than going to a hospital or clinic?

They are! These and other studies are being funded through...



NEWS RELEASES

Wednesday, September 25, 2019

New NIH grant awards boost funding for research on Down syndrome

INCLUDE project aims to address conditions of Down syndrome and apply knowledge to larger population.



The National Institutes of Health has awarded approximately \$35 million in new grants in fiscal year (FY) 2019 to advance research on Down syndrome through the [Investigation of Co-occurring Conditions Across the Lifespan to Understand Down Syndrome](#) (INCLUDE) project. These awards bolster total funding for Down syndrome research in FY 2019 to an estimated \$77 million. INCLUDE is a trans-NIH effort with participation from many NIH institutes and centers. Research funded by INCLUDE will investigate critical health and quality-of-life needs for individuals with Down syndrome, while at the same time explore co-occurring conditions with individuals who do not have Down syndrome.

NIH awarded \$86 M in funding for Down syndrome research in 2019!

<https://www.nih.gov/news-events/news-releases/new-nih-grant-awards-boost-funding-research-down-syndrome>



Outline

- National Institutes of Health (NIH)
 - History of the INCLUDE Research Initiative
 - Research being funded by INCLUDE
- What you can to help
 - Sign up for DS-Connect®: The Down Syndrome Registry
 - Give us feedback: Down Syndrome Research Plan
 - Volunteer to participate in research

What is the National Institutes of Health (NIH)?

- The U.S.'s national medical research agency
- The world's largest funder of biomedical research
- Made up of 27 separate research institutes and centers
- Director since 2009: Dr. Francis Collins
- 85% of funds go to university research institutions based on peer review of grant applications



NIH leads the Down Syndrome Consortium



Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

National Cancer Institute (NCI)

National Heart, Lung and Blood Institute (NHLBI)

National Institute of Mental Health (NIMH)

National Institute of Neurological Disorders and Stroke (NINDS)

National Institute on Aging (NIA)

National Institute on Minority Health and Health Disparities (NIMHD)

National Institute of Dental and Craniofacial Research (NIDCR)

National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)

National Institute on Deafness and Other Communication Disorders (NIDCD)

National Human Genome Research Institute (NHGRI)

National Institute of Allergy and Infectious Diseases (NIAID)

National Center for Advancing Translational Sciences (NCATS)

Self-advocates



**DOWN SYNDROME
AFFILIATES IN ACTION**



alzheimer's  association®



**Down
Syndrome
International**





New Congressional Directive for DS

- In the 2018 budget legislation for NIH:
 - Develop a new **trans-NIH** initiative to study trisomy 21, with the aim of yielding scientific discoveries to improve the **health and neurodevelopment of individuals with Down syndrome** and **typical individuals** at risk for:
 - Alzheimer's disease
 - Leukemia
 - Heart defects
 - Immune system dysregulation
 - Autism
 - and other conditions
- Congress provided ~\$23 M in additional research money in 2018 for a new trans-NIH research initiative

Protected from:

- Many cancers
- Heart disease and heart attacks

[illegible]



INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrom**E**)

3 components:

- 1. Conduct targeted, high-risk, high-reward basic science studies on chromosome 21.**
- 2. Assemble a large study population of individuals with Down syndrome across the lifespan.**
- 3. Include individuals with Down syndrome in existing and future clinical trials.**



INCLUDE Funding for DS

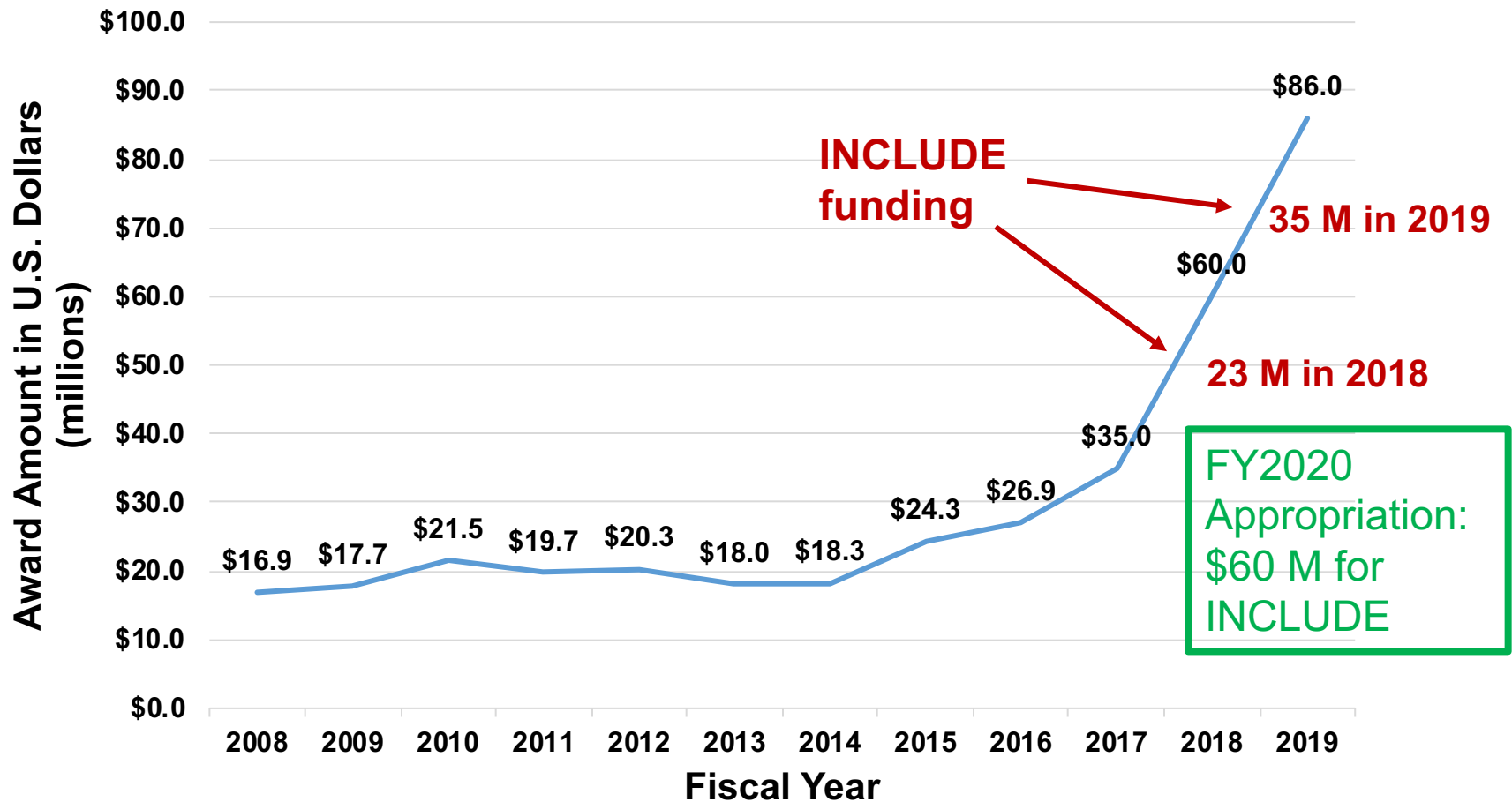
- **Basic science:**
 - Silencing genes on chromosome 21 to understand brain development
 - Studies to understand why $\sim 1/2$ of infants have a heart defect
- **Cohort development:**
 - Cognitive and developmental tests for future clinical trials
 - Effects of chemotherapy used to treat leukemia on the heart
 - Hearing in a noisy environment
 - Strategies to recruit underrepresented minorities into research
- **Clinical trials:**
 - Medications for ADHD in children with attention problems
 - Medications to treat sleep apnea in children
 - A medication for Alzheimer's disease in adults
 - Medications to treat autoimmune skin conditions
- **Training:** support for up to 18 trainees and junior researchers

URL: <https://www.nih.gov/include-project>



Total NIH Funding for Research on Down Syndrome

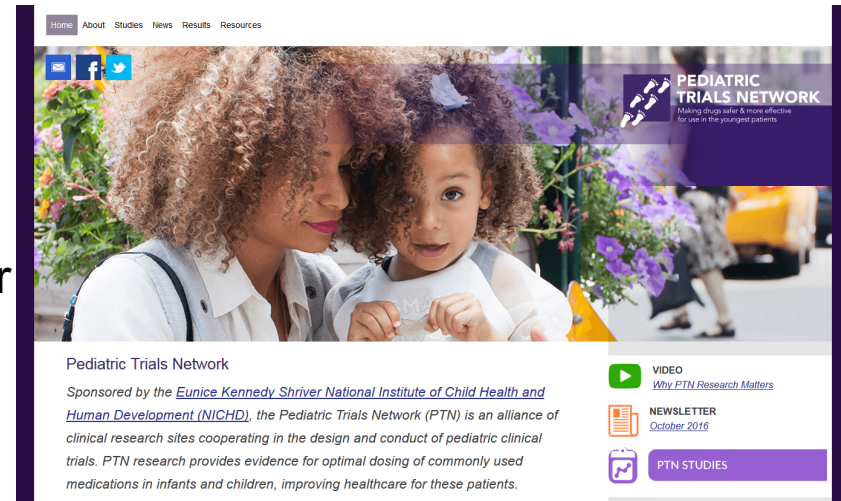
2008 – 2019





NICHD's Pediatric Trials Network (PTN)

- Studies the formulation, dosing, efficacy, and safety of drugs and medical devices used in children
- Conducts clinical trials for many different conditions
- PTN clinical research sites are located at over 100 universities and children's hospitals across the U.S.
- **The PTN will partner with INCLUDE to support drug studies in individuals with Down syndrome**
- **Additional goal is to train investigators to conduct research involving people with DS**



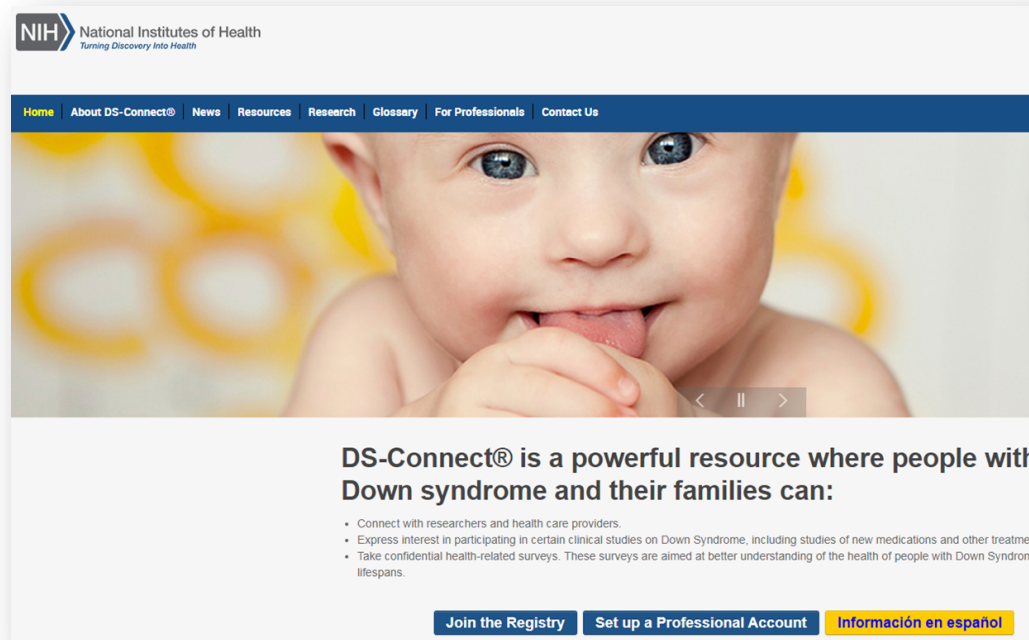
See www.Pediatrictrials.org



Building a large cohort for natural history and biomarker studies

- Several projects: Genomic predictors of heart defects, leukemia, and other features in individuals with Down syndrome
- Use DS-Connect[®]: The Down Syndrome Registry for recruitment

<http://DSConnect.nih.gov>



How you can help...



Sign up for DS-Connect® (<http://DSConnect.nih.gov>)

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

The screenshot shows the DS-Connect website homepage. At the top, there's a navigation bar with links for LOGIN, JOIN, and NEED HELP?, along with flags for the US and Spain. Below this is the NIH logo and the text 'National Institutes of Health Turning Discovery Into Health'. A secondary navigation bar contains links for Home, About DS-Connect®, News, Resources, Research, Glossary, For Professionals, and Contact Us. The main content area features a large video player showing a smiling man. Below the video, the text reads: 'DS-Connect® is a powerful resource where people with Down syndrome and their families can:'. This is followed by a bulleted list of benefits: connecting with researchers and health care providers, expressing interest in clinical studies, and taking confidential health-related surveys. At the bottom, there are three buttons: 'Join the Registry', 'Set up a Professional Account', and 'Información en español'.

NIH National Institutes of Health
Turning Discovery Into Health

LOGIN JOIN NEED HELP?  

Forgot login?
Unblock Account

Home | About DS-Connect® | News | Resources | Research | Glossary | For Professionals | Contact Us

DS-Connect® is a powerful resource where people with Down syndrome and their families can:

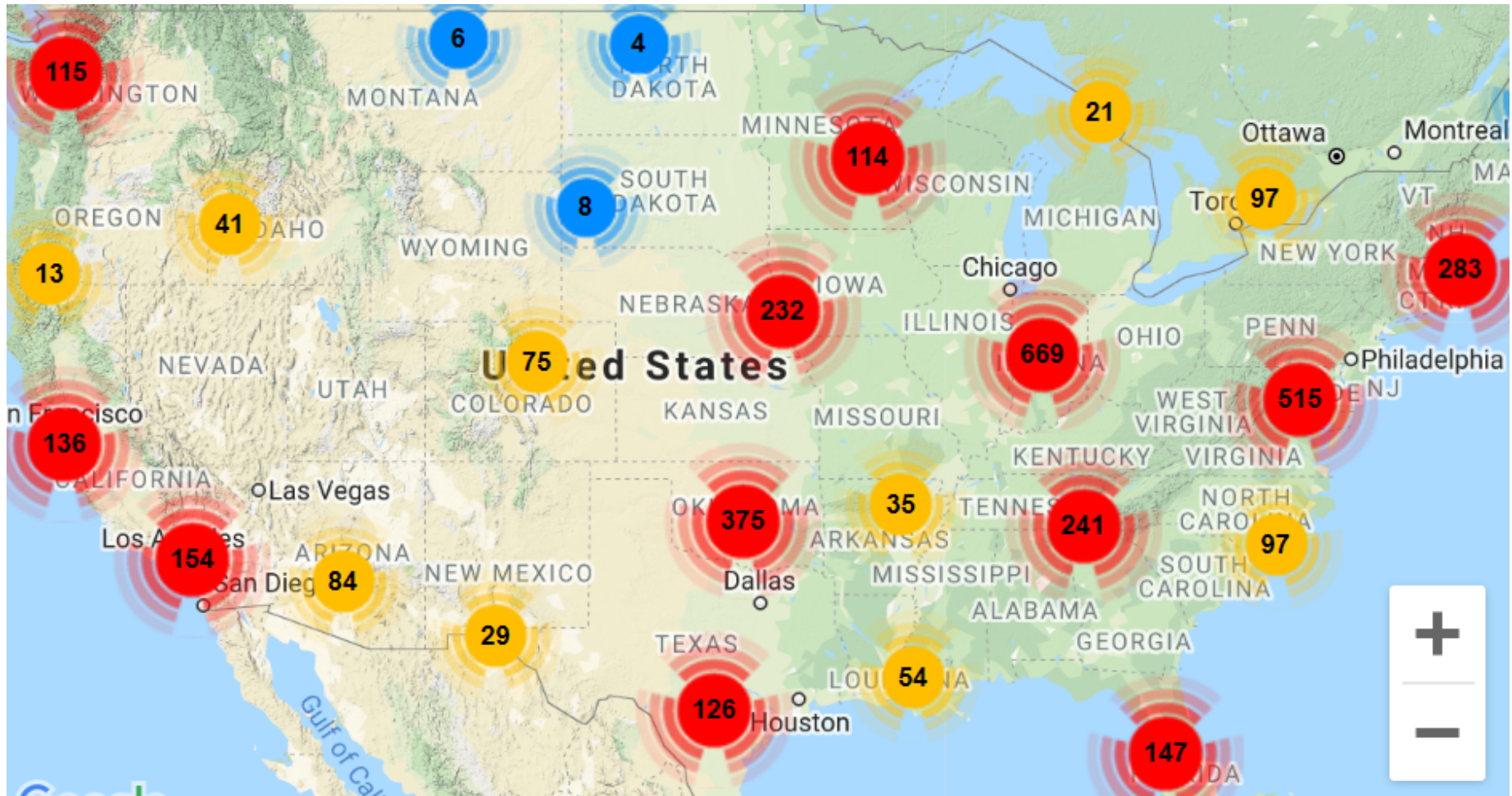
- Connect with researchers and health care providers.
- Express interest in participating in certain clinical studies on Down Syndrome, including studies of new medications and other treatments.
- Take confidential health-related surveys. These surveys are aimed at better understanding of the health of people with Down Syndrome across their lifespans.

Join the Registry Set up a Professional Account Información en español

Launched Sept. 6, 2013



DS-Connect® Registrants: North America



Over 5000 registrants globally as of June 25, 2020



A partnership with families...

➤ Access the health care provider list



[Questionnaire](#) [Healthcare Providers](#) [Growth Measurements](#) [Account Info](#) [Attachments](#) [Contacts](#) [Newsletters](#)

[Notes](#)

Search the directory for your health care provider(s) by name, facility name, city or state (2 letter abbreviation). If your health care provider is not in the directory, use the Add a New Health Care Provider link to add them in the directory. Note that you will need to search for each physician individually before the Save button will be enabled.

Please note that this list of healthcare providers does not imply endorsement or recommendation of their services.

Name/Institution

Specialty

City

State

Country



Search

Leave blank to list all

[Add a New Health Care Provider](#)

Click the  to add the healthcare provider to your account.

~764 now Listed!

Name	Specialty	Sees Adults or Youth?	Institution	City	State	Country	
John Avallone	Ophthalmology		Ophthalmology Associates	Arnold	MD	UNITED STATES	 My Health Care Provider
Mihee Bay	Developmental Pediatrics		Kennedy Krieger Institute	Baltimore	MD	UNITED STATES	 My Health Care Provider



Spread the word about DS-Connect

- **Sign up!**
- Electronic Toolkit available:
 - Video Testimonial-YouTube
 - Flyers
 - Facebook info-cards
 - Badges and Tweets
- Help us reach our goal:
 - 10,000 by Dec 31, 2020

Registration Goal

Goal: 10000
Current: 5034



**Advance science,
improve lives.**

Join DS-Connect™: The Down Syndrome Registry today.
Get DS-Connected: **DSconnect.nih.gov**

Eunice Kennedy Shriver National Institute
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<http://DSConnect.nih.gov>

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THE INCLUDE PROJECT

Events in 2020

INCLUDE Project Research Plan



- More funding opportunities
- Clinical trials support and training for investigators
- A new data coordinating center for INCLUDE
- Workshops:
 - May 2020: Clinical Trials in DS
 - Nov. 2020: Opportunities in Basic Science and cohort development in DS

URL: <https://www.nih.gov/include-project>



**HOT OFF
THE PRESS**

New Funding Opportunity for COVID-19

- Notice of Special Interest (NOSI) regarding the Availability of Urgent Competitive Revisions and Administrative Supplements for Research on Coronavirus Disease 2019 (COVID-19) in Individuals with Down Syndrome for the INCLUDE Project
- **Notice Number: NOT-OD-20-129**
- *Goal: to improve understanding and treatment of COVID-19 infection in those with DS given their increased risk of serious infection*
- **Supplements to existing grants**
- Deadlines: July 13, Nov 12 (2020); Mar 12, July 12 (2021)
- Specific areas of focus for different NIH institutes
- <https://grants.nih.gov/grants/guide/notice-files/NOT-OD-20-129.html>

NIH Research Plans on Down Syndrome

- Goal: Develop research goals and objectives for NIH research
- We are updating the 2014 plan and combining it with the INCLUDE Research Plan for 2021
- RFI (NOT-HD-20-013):
<https://grants.nih.gov/grants/guide/notice-files/NOT-HD-20-013.html>
- Seeking comments on:
 - Effectiveness of the previous plans
 - Advances made since 2014
 - Remaining research gaps
 - Suggestions concerning new future research objectives
- Respond by July 10, 2020 to DownSyndrome@mail.nih.gov

Give us your feedback!



What is missing?





Q & A

- **DS-Connect:** <http://DSConnect.nih.gov>
- **INCLUDE:** <https://www.nih.gov/include-project>
- Contact us! DSConnect@nih.gov
or parisima@mail.nih.gov



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On behalf of the entire NIH INCLUDE Team