

# 24th Meeting of the Down Syndrome (DS) Consortium: A Public-Private Partnership

January 19, 2024; 11:00-12:30 p.m. eastern time  
Virtual

## Roll Call

*Marjorie Vandy, NICHD*

Organization	Name
American Association on Intellectual and Developmental Disabilities (AAIDD)	Maggie Nygren
Alzheimer's Association	Heather Snyder
American Academy of Pediatrics (AAP)	Marilyn Bull*
Association of University Centers on Disabilities (AUCD)	John Tschida
Down Syndrome Affiliates in Action (DSAIA)	Julie Torzewski, Sarah Mulligan
Down Syndrome Diagnosis Network (DSDN)	Christy Cooper
Down Syndrome International (DSi)	Jessamy Tang
Down Syndrome Medical Interest Group (DSMIG)	Nicole Baumer
GLOBAL	Michelle Sie Whitten, Joaquin Espinosa, David Tolleson, Haley Sanders, Bryn Gelaro
International Mosaic Down Syndrome Association (IMDSA)	Colleen Jackson-Cook
LuMind IDSC Down Syndrome Foundation	Hampus Hillerstrom
National Down Syndrome Congress (NDSC)	Sean Smith
National Down Syndrome Society (NDSS)	Colleen Hatcher
National Task Group on Intellectual Disabilities and Dementia Practices (NTG)	Kathryn Pears, Marianne Barbera
Self-Advocate	Karen Gaffney, Barbara Gaffney (parent)
Self-Advocate	David Egan, Kathleen Egan (parent)
Self-Advocate	Mitchell Levitz, Jack Levitz (parent)
Special Olympics	Dimitri Christakis
Trisomy 21 Research Society (T21RS)	Roger Reeves **

Organization	Name
<b>National Institutes of Health (NIH):</b>	
<b>NHLBI</b>	Marishka Brown, Huiqing Li, Charlene Schramm
<b>NIA</b>	Erika Tarver, Courtney Wallin, Laurie Ryan
<b>NIAID</b>	Deborah Hodge
<b>NINDS</b>	Robert Riddle, Patrick Wright
<b>NICHD</b>	Melissa Parisi, Sujata Bardhan, Meghan O'Neill, Linda Garcia
<b>OD</b>	Kayla Davis

## Welcome and Overview

### *Melissa Parisi, NICHD*

- Welcomed attendees and introduced Dr. Meghan O'Neill who recently joined the [NIH INCLUDE](#) Team
- Provided an overview of the [INCLUDE Project](#)
  - INCLUDE is investigating co-occurring conditions, across the lifespan, that affect individuals with DS and the general population, such as Alzheimer's disease, autism, cataracts, celiac disease, congenital heart disease, and diabetes.
  - INCLUDE Goals:
    - Increase the number of investigators and trainees studying DS.
    - Increase the diversity of families participating in the program as well as the investigators who are part of the research teams.
  - INCLUDE components:
    - Conduct targeted, high-risk, high-reward basic science studies on chromosome 21.
    - Build a large cohort of individuals with Down syndrome for comprehensive analysis and biomarker evaluation.
    - Include individuals with Down syndrome in existing and future clinical trials.
- Since 2018, the NIH has invested nearly \$350 million in the past 6 years on 330 new projects for INCLUDE.
- Provided an update on the funding opportunities
  - Some of the funding opportunities are:

- Transformative R01s for better understanding of basic science of DS
  - R03s for the analysis and curation of data sets
  - Training grants for early career investigators
- mHealth R21 was just published
- Visit the website for the list of [funding opportunities](#) available.
- Down Syndrome Cohort Development Program (DS-CDP)
  - INCLUDE Project is still in the process of competing the DS-CDP. The DS-CDP program plans to recruit participants with Down syndrome (DS) across the lifespan; collect biospecimens; assemble phenotypic data, images, and omics for deep phenotyping; and make the data readily available to the scientific community.
  - The three funding opportunities are:
    - Cohort Research Sites: DS-CRS (U01, [NOT-OD-23-134](#))
    - Clinical Cohort Coordinating Center: DS-4C (U54, [NOT-OD-23-135](#))
    - Federated Biobanking Resource (U24, [NOT-OD-23-136](#) )
    - Awards are expected in late summer 2024.

## **INCLUDE and Diversity, Equity, Inclusion, and Accessibility (DEIA) Updates**

*Sujata Bardhan, NICHD*

- Provided an update about the activities and initiatives undertaken by the INCLUDE Project to promote inclusion and engagement with the DS community
  - Inaugural webinar focused on DEIA was launched last summer. The webinar was focused the value of diverse perspectives on DS research.
  - May 30, 2024: Second webinar will be held. The topic and registration will soon be available on the INCLUDE website.
  - Some of the NIH INCLUDE funding opportunities are focused on DEIA: mentorship, community based participatory research, research awards for undergraduate and graduate students, and pre-doctoral to post-doctoral fellow transition awards.

## **INCLUDE Community Outreach Updates**

*Linda Garcia, NICHD*

- **2024 NIH INCLUDE Investigators Meeting**

- April 15 – 16, 2024: An in-person [2024 NIH INCLUDE Investigators Meeting](#) will be held at Natcher Auditorium on NIH Campus in Bethesda, MD.
- The [registration](#) is open now.
- February 2, 2024: Deadline for [abstract submission](#) for poster session.
- Some of the upcoming events are:
  - Emory University DS Conference
  - Gatlinburg Conference on Intellectual and Developmental Disabilities
  - T21 Research Society conference
  - NDSC
  - Global Down Syndrome Medical Research Roundtable
  - DSMIG
  - DSDN
  - SACNAS
  - Annual Biomedical Research Conference for Minority Students (ABRCMS)
  - American Society of Human Genetics (ASHG)
  - New York City Buddy Walk

## **INCLUDE DCC Data Hub Updates**

*Joaquin Espinosa, GLOBAL*

- Welcomed the participants and reiterated the mission of the [INCLUDE Data Coordinating Centre \(DCC\)](#).
- The [DCC](#) is funded under a cooperative agreement to support a team from various institutions with extensive expertise in managing large-scale data. The DCC also collaborates with various investigators and NIH program officers.
- Objectives of the [DCC](#):
  - Study co-occurring conditions in people with DS to help them live longer and healthier lives.
  - Study the co-occurring conditions and make discoveries of translational potential. The DCC aims to harness the power of diversity among the DS population. There is a need for a personalized medicine approach considering the diverse manifestation of co-occurring conditions and the phenotypic variability observed in people with DS.

- To investigate the underlying causes of the phenotypic variation, an orchestrated effort that enables the testing of myriad alternative hypotheses quickly and efficiently is needed.
- DCC Data Ecosystem
  - [DCC](#) has a burgeoning data ecosystem which comprises the following components:
    - [DS-Connect](#): a study recruitment tool where families and advocates provide data and can connect with researchers and projects
    - [TrisomExplorer](#): provides intuitive data visualization about people with DS
    - [DCC Website](#): an investigator registry that has INCLUDE-related publications and grants
    - [Data Hub](#): for researchers and data scientists and has large scale data sets with registered and controlled access
    - Experimental Models Portal: Coming soon!
  - [DCC](#) has a multidimensional data set from 9,064 participants enrolled in 11 studies from all over the country. 7,463 of these participants have trisomy 21. The diverse dataset includes 4,000+ genomes, 1,700+ transcriptomes, 470+ proteomes and 400+ metabolomes.
  - If data collection is continued, it will help researchers to test hypotheses with large data sets. The following benefits are expected for the data analysis:
    - Greater sample sizes and increased statistical power
    - More diverse cohorts
    - Study of less common co-occurring conditions
    - Integrated analysis across the pediatric-adult divide
    - Reproducibility via discovery and validation cohorts
    - Data exploration before embarking on expensive experimentation
    - Identification of available biospecimens
    - Collaboration across the ecosystem
- Outreach, Engagement and Education
  - Goals:
    - To engage diverse stakeholders in research participation.
    - To promote data sharing and re-analysis.
    - To train users in the use of the Data Hub and advanced data science.
    - To disseminate research results from cohort studies in the Data Hub.
    - To promote diversity, equity, and inclusion throughout the ecosystem.
  - The [DCC](#) has established collaborations with scientific, non-scientific, and advocacy organizations and regularly attends their events.

- 51<sup>st</sup> National Down Syndrome Congress Convention:
  - DCC participated in the NDSC last summer and engaged with stakeholders at the GDSF, DSMIG, and Spanish NDSC members.
  - A panel composed of INCLUDE DCC and INCLUDE investigators presented different studies related to immune dysregulation at the DSMIG annual conference.
  - DCC presented about the INCLUDE project and advanced research studies funded by the INCLUDE project to the Spanish community. Doctors from Latin America were very interested in conducting the research studies with cohorts from their local clinics
  
- To help establish research infrastructure in Latin America, doctors from seven different countries attended a workshop and training event in Colorado from November 16-19<sup>th</sup>, 2023.
  
- Last summer, 24 domestic and international students attended the two-week Data Science for Diverse Scholars in Down Syndrome Research training course in Boulder, CO with nine instructors.
  
- Goals and milestones for next twelve months:
  - Surpass 15 studies in the Data Hub (currently at 11)
  - Surpass 750 registered users (currently at 580)
  - Surpass 10,000 unique research participants (currently at 9,064)
  - Surpass 5,000 whole genome sequences (currently at ~4,200)
  - Incorporate new data modalities: epigenomes, immune maps, images
  - Prepare for a future with a network of research sites funded by NIH

## Round Robin

### *All*

- DSMIG, Nicole Baumer, MD
  - July 25-27<sup>th</sup>, 2024: 2024 DSMIG Annual Symposium, in coordination with NDSC conference, will be held in Phoenix, Arizona.
    - May 2024: Registration likely to open
    - January 18, 2024: Abstract/poster call went out
  
  - This year, DSMIG is joining forces with NDSC to host a joint session with DSMIG and NDSC, focused on supported decision-making, addressing key issues, examples of state-wide differences, potential barriers, and strategies to educate audiences about these alternatives.

- DSMIG has expanded educational offerings beyond the Symposium to a wider audience, targeting professionals and community physicians through a webinar series and DSMIG Project ECHO.
- DSMIG is also working on increasing membership, increasing opportunity/inclusion efforts, internship opportunities for trainees and early career professionals.
- NDSC, Sean Smith
  - 4-day NDSC annual convention was held in Orlando in 2023 with a record number of attendees and included various workshops and sessions.
  - Center for Outreach & Education hosted more than 400 attendees for online webinars and one-day Educator Conferences.
  - Policy & Advocacy Team participated in more than 100 meetings on Capitol Hill with Congressional staff advocating for equal rights and opportunities for individuals with Down syndrome and their full inclusion in the workplace, in schools, and in the community.
  - National Down Syndrome Advocacy Coalition (NDAC) grew by 173 new members, including 21 new self-advocate members.
  - As part of Multi-Cultural Outreach, NDSC hosted 23 webinars in Spanish and an array of convention workshops presented by 16 international speakers.
  - National Down Syndrome Congress is committed to fostering, cultivating, and preserving a culture of diversity, equity, and inclusion where everyone is respected and valued.
  - Upcoming events:
    - February 3, 2024, in Tulsa, OK, and July 26, 2024, in Phoenix, AZ. NDSC is hosting an Educators Conference
    - February 13, 2024: Webinar on “Inclusion” as part of Ages and Stages Webinars series.
    - July 25-28, 2024, Phoenix, AZ: 52nd Annual NDSC convention and registration opens March 12, 2024
- Self-Advocate, Karen Gaffney
  - Presented at the FIRE Foundation in Denver CO in August 2023.

- Presented at the Down Syndrome Network of Oregon Buddy walk highlighting the accomplishments of people with Down Syndrome over the last 20 years in September 2023
- Presented at Pacific Northwest Employment Forum for the State of Oregon advocating for employment opportunities for people with Down Syndrome in October 2023.
- Presented at two different fund-raising galas for Gigi's Playhouse and Down Syndrome Achievement Centers in October 2023.
- Presentation about Down Syndrome to a class of medical students at the Oregon Health Sciences University in November 2023.
- Continues to work 32 hours a week at a law firm Monday thru Thursday, and dedicated Fridays to her nonprofit organization.
- Self-Advocate, Mitchell Levitz
  - Presented at New York Medical College on "Living with Down Syndrome", on December 18, 2023. 250 first-year medical students and other graduate students attended.
- Self-Advocate, David Egan
  - Spoke at Waisman Center 50<sup>th</sup> Gala Anniversary, in October 2023, and shared the impact of early intervention on his social and verbal skills Also met Chancellor of University of Wisconsin and expressed his gratitude for the research and the innovations at the center.
  - Participated in the NDSS Adult Summit in Cincinnati and spoke about importance of self-advocacy. At the summit he also met Drs. Brian Chicone and Ruth Brown and discussed the importance of creating Down Syndrome Health Centers of Excellence.
  - Gave a webinar to the Golisano Institute for Nurse Practitioners in December 2023.
  - Has been approached by a few researchers to serve on their advisory boards for their NIH projects related to Down syndrome.
  - Shared a request about the lack of reciprocity of care from one state to another and urged the consortium to advocate with Congress. David Egan also suggested establishing an international Down Syndrome Consortium; Michelle Sie Whitten will talk to him offline.
- Alzheimer's Association, Heather Snyder



- Alzheimer's Association is moving towards publishing newly proposed diagnostic criteria for Alzheimer's diagnosis and staging. One of the changes in the new criteria is the inclusion of individuals with Down syndrome. More information can be found here: [alz.org/diagnostic-criteria](https://www.alz.org/diagnostic-criteria).
- Opened a session at the Human Amyloid Imaging (HIA) meeting on February 19, 2024, with focus on advances in imaging to understand more about Alzheimer's disease in individuals with Down syndrome.
- April 11 and 12, 2024: "Genetically Determined Alzheimer Disease: Cross-fertilization between Down Syndrome & Autosomal Dominant Alzheimer's Disease" meeting will be held at Modernist Hospital of Sant Paul, Barcelona. Dr. Juan Fortea is leading this initiative, funded by Alzheimer's Association.
- Planning is underway for AAIC, with abstract deadlines at the end of January. More details can be found here, [AAIC 24](#).
- American Academy of Pediatrics, Marilyn Bull
  - AAP Publishing is partnering with Global to license the clinical report and parent pages of the AAP Down syndrome guidelines for translation into other languages. Spanish and Italian translations should be available soon.
  - Shared two publications
    - The AAP Council on Genetics published the attached AAP News article in November 2023 entitled, "[Genetics community looks for ways to improve equity, diversity, inclusion](#)."
    - The September 2023 AAP News article "[New AAP chief health equity officer sets lofty goals for generational transformation](#)."
- DSAIA, Sara Mulligan
  - Witnessed tremendous growth in its membership in 2023. DSAIA website was updated with a directory of DSAIA area points of contact. Down Syndrome affiliates can reach out to them for broader networking.
  - Reach out at [info@DSAIA.org](mailto:info@DSAIA.org) if any organization wants their events to be publicized on Down Syndrome Affiliates in Action website.
  - February 15-17, 2024: DSAIA Annual Leadership Conference in Dallas, Texas.
- DSDN, Christy Cooper
  - Working on getting the data published from the diagnosis survey, from over 880 parents of children with Down syndrome, which was conducted last summer.
  - Medical Outreach team presented "Improving the Diagnosis Experience: What We Can Learn from Families of Children with Down Syndrome," at the National Society of Genetic Counselors Annual Conference, in October 2023. They shared

results from 3 different studies including diagnosis experience, the impact of bias, and improving dissemination of information to Black and Hispanic new and expectant parents.

- A community online app is expected to be launched this year, which will move the support groups comprising of 23,000 parents, off the Facebook platform.
- Presenting a spotlight session, “How to Connect with Dads and Dads-to-Be,” as well as co-presenting “Connecting with Military Communities” at the DSAIA conference. DSDN will also be presenting about the Down Syndrome Collaborative, an effort DSDN co-founded in 2022 to support new and expectant parents.
- Retreat registration is open for the [Rockin’ Dad™ Retreat](#) in July in Nashville and [Rockin’ Mom™ Retreat](#) in September in Las Vegas.
- DSi, Jessamy Tang
  - Published a new, accessible strategy in October. Read more here: [DSi Strategy](#)
  - Awarded 5 World Down Syndrome Awards in October, with awardees chosen from over 200 nominations by a panel including people with Down syndrome. More info here: [World Down Syndrome Awards](#).
  - Training self-advocates to become consultants on inclusive organizations, including two people with Down syndrome in the UK. Read more here: [Training self-advocate consultants on inclusive organizations](#)
  - Hired a Program Officer with Down syndrome, Emma Bishop, and also appointed first ever board member with Down syndrome, Janet Charchuk.
  - Upcoming events:
    - March 21, 2024: World Down Syndrome Day Conference at the UN Headquarters in New York. This year’s campaign is around Health Equity. More info here: [World Down Syndrome Day 2024](#).
    - July 2024, 2024: World Down Syndrome Congress 2024 in Brisbane, Australia. Registrations to attend and applications to present are open at [WDSC 2024 Brisbane, Australia](#).
- GLOBAL (GDSF), Michelle Sie Witten
  - Thanked NIH INCLUDE team for their hard work.
  - Recently announced clinical trial related to study of JAK Inhibitors in children with Down Syndrome.

- New slate of authors for the Adult Guidelines has been announced and number of volunteers has been doubled. 3000 copies of Adult Guidelines have been downloaded and 4500 copies printed and delivered. Italian, Albanian and Swahili versions are coming online in March and May.
- Looking for a new medical director for the Sie Center. Recently a paper related to comorbidity referrals related to AAP guidelines was published. The paper also shows the financial viability of the Sie Centers.
- Upcoming events:
  - May 14, 2024: Building the Future Gala - Down Syndrome Alliance of the Midlands.
  - May 9<sup>th</sup>, 2024: The [AcceptAbility Gala](#) in Washington, DC.
  - November 16, 2024: Fundraising fashion show [Be Beautiful Be Yourself](#) on November 16, 2024 in Denver, CO.
  - Partnering with NDSC for [Global Research in Medical Care Roundtable](#) at the NDSC Convention.
  - Rosa DeLauro and Tom Cole Global Advancements Awards portal will be opened soon.
- Michelle has been named in the [Denver Business Journal's 40 under 40 Hall of Fame](#).
- Linda Crnic Institute, Joaquin Espinosa
  - First clinical trial for immunomodulation in Down syndrome focused on autoimmune skin conditions is schedule to complete recruitment in Spring 2024, with full dataset available by Summer 2024.
  - Clinical trial for Down Syndrome Regression Disorder is enrolling steadily ahead of schedule, with first phase involving 16 participants to be completed in Spring 2024.
  - The Human Trisome Project is now at ~1200 participants. Several publications fueled by the Human Trisome Project were published in 2023, including papers in Nature Genetics and Science Advances, with several more publications scheduled for 2024.
- IMDSA, Colleen Jackson-Cook
  - Research and Retreat Conference was held on July 26-29, 2023, at the University of California, San Diego (UCSD).
    - 5 different research teams collected data from the survey results at the conference.

- Included a session focusing on diversity and inclusion. The group also suggested topics of importance for future research and discussion at future meetings.
  - Scholarships through a conference grant from NIH were awarded to provide opportunities for people from diverse minority groups with mosaic Down syndrome and their parents to attend.
  - Videos of a subset of the information sessions presented at the conference are being uploaded on YouTube.
- Holding their annual awareness campaign in March, in coordination with a fund-raising campaign. The primary goal of this activity is to raise awareness of mosaic Down syndrome and celebrate World Down Syndrome Day.
- The Self-Advocate Leadership Team (SALT), which is for self-advocates aged 16 or older, continues to hold regular meetings.
- July 25-28, 2024: 2024 IMDSA Research and Retreat conference will be held at the Fourwinds Hotel at Lake Monroe in Bloomington, Indiana.
- LuMind IDSC, Hampus Hillerstrom
  - Completed baseline enrollment of LIFE-DSR natural history study in adults with DS over age 25.
  - Actively talking with biotech and pharma companies about clinical trials in Down syndrome-associated Alzheimer's disease (DS-AD).
  - Ongoing work on definition of early DS-AD staging for clinical practice, validation of diagnostic blood-based biomarkers for DS-AD, and prevention clinical trials in DS-AD.
  - Completed filming of videos for MRI and PET to support people with DS and their families become familiar with these procedure for clinical trials.
  - [Community petition](#) for equitable treatment access.
  - Presence and presentations at many family, affiliate, and research conferences.
  - Diversity, Equity, Inclusion, and Accessibility (DEIA) efforts:
    - Visibility in general press on equity issue for people with DS accessing Alzheimer's treatments (Boston Globe, Reuters).
    - Continuing dissemination of information on equivalencies, advisory and consensus statement for treatment access to relevant stakeholders.

- Finalizing recommendations to CMS on adaptations for the DS population of their anti-amyloid antibody registry for DS-AD.
- NDSS, Colleen Hatcher
  - Multiple new resources have been published over the past few months. These are free for the community and can be found on [NDSS website](#).
  - The 2023 NDSS Adult Summit in Cincinnati was hosted in November. 100 participants were adults with Down Syndrome.
  - Multiple webinars were held in 2023 on topics including Alzheimer's disease, regression, SSI, entrepreneurship. Recordings of these webinars can be found on the [NDSS YouTube page](#).
  - Upcoming Events:
    - March 20-21, 2024, Washington, DC: Annual Down Syndrome Advocacy Conference. NDSS has collaborated with Black Down Syndrome Association (BDSA) to ensure that a diverse community is represented at the conference. To learn more, please email [policy@ndss.org](mailto:policy@ndss.org).
    - Multiple webinars are scheduled for 2024, covering topics including employment and mental health. To learn more about future NDSS webinars, please follow NDSS on social media.
    - Fall, 2024: Adult Summit
- NTG, Marianne Barbera
  - Undertook a national “Family Needs Survey.” and the results from 530 responses showed that the top areas of interest for caregivers were the process of aging, early screening for cognitive changes, life planning for different phases of the life journey, and behavioral changes that occur with aging and dementia. Preferences for obtaining information were through online training (webinars) and one-page fact sheets.
  - Monthly virtual Family Support Group is on the NIA website and can be accessed [here](#).
  - NTG has teamed up with Lumind IDSC Foundation to sort out barriers to access issues related to newly approved anti-amyloid disease modifying therapies (primarily Leqembi). Barriers included the individual state prescribing criteria and omissions in CMS and other registries.
  - February 27th to April 23<sup>rd</sup>: 5-part Family Support Webinar Series. The series information and registration access are posted on the [NTG website](#).

- T21RS, Roger Reeves
  - June 5 – 8, 2024: T21RS biannual meeting will be in Rome.
  - There will be a half day science and society session as in previous meetings.

## **Concluding Remarks**

- Sujata Bardhan suggested that we create a space for the DEIA updates on the Down Syndrome Consortium website.
- We plan to arrange the next in-person Down Syndrome Consortium meeting at the NDSC convention in July.
- Melissa Parisi thanked all the participants for attending.

\*Not in attendance; update provided via email received on 01/15/2024

\*\* Not in attendance; update provided via email received on 01/16/2024