

23rd Meeting of the Down Syndrome (DS) Consortium: A Public-Private Partnership

July 22, 2023; 3:30-4:45 p.m. eastern time
J.W. Marriott Orlando, Florida/Virtual

Roll Call

Marjorie Vandy, NICHD

Organization	Name
National Heart, Lung, and Blood Institute (NHLBI)	Marishka Brown
NHLBI, NIH	Charlene Schramm
<i>Eunice Kennedy Shriver</i> National Institute of Child Health and Human Development (NICHD)	Linda Garcia
NICHD	Melissa Parisi
NICHD	Marjorie Vandy
National Institutes of Health Office of the Director	Nikila Natarajan
American Academy of Pediatrics (AAP)	Marilyn Bull
Alzheimer's Association	Heather Snyder*
Association of University Centers on Disabilities	John Tschida
Down Syndrome Affiliates in Action (DSAIA)	Sarah Mulligan
Down Syndrome Diagnosis Network (DSDN)	Melissa Shutwell
Down Syndrome International (DSi)	John Blascovich
Down Syndrome Medical Interest Group (DSMIG)	Nicole Baumer
Global Down Syndrome Foundation (GLOBAL)	Michelle Sie Whitten
GLOBAL	Joaquin Espinosa
GLOBAL	Bryn Gelaro
GLOBAL	David Tolleson
International Mosaic Down Syndrome Association (IMDSA)	Ruth Brown
LuMind IDSC Foundation	Hampus Hillerstrom
National Down Syndrome Congress (NDSC)	Jordan Kough
National Down Syndrome Society (NDSS)	Kandi Pickard

Organization	Name
NDSS	Colleen Hatcher
National Task Group on Intellectual Disabilities and Dementia Practices (NTG)	Matthew Janicki
NTG	Kathy Service
Special Olympics	Andrew Lincoln
Parent on behalf of Self-Advocate David Egan	Kathleen Egan
Self-Advocate	Mitchell Levitz
Self-Advocate	Karen Gaffney*

Welcome and Overview

Melissa Parisi, NICHD

- Welcomed new attendees to the DS Consortium
- Provided an overview of the INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome) Project
 - Program began in 2018 through the support of a congressional directive that called for an NIH-wide research initiative on critical health needs of persons with DS and co-occurring conditions
 - The project applies the expertise of several NIH institutes and centers (ICs) to advance this initiative across three components:
 - Conduct targeted, high-risk, high-reward basic science studies
 - Build a large cohort of individuals with DS to enable longitudinal studies that can follow conditions of interest over time
 - Conduct clinical trials research inclusive of individuals with DS
- Preparing to host an INCLUDE investigators meeting in April 2024 (rescheduled from November 2023)
 - The meeting will serve as an opportunity to receive remarks from INCLUDE leadership and perspectives from DS families and self-advocates, discuss research undertaken by INCLUDE-funded investigators, and disseminate recent study findings
 - The event will also include a demonstration of the [INCLUDE Data Coordinating Center \(DCC\) Data Hub](#) and a trainee poster session

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

Melissa Parisi, NICHD, and Linda Garcia, NICHD

- Budget and research plan
 - Since the project launched, funding for INCLUDE has progressively increased to \$75 million. While it is still early days, NIH is energized by the expansion of investments in DS research.
 - INCLUDE has funded several studies to accelerate our understanding of leukemia, congenital heart disease in infants, behavioral and developmental pathways, language acquisition, risk and resilience factors for Alzheimer's, and other related topics.
 - NIH recently finalized its [INCLUDE Down Syndrome Research Plan](#) (PDF 1.41 MB), which presents key priorities and integrates feedback from the DS community. The research plan now contains a further emphasis on training and a dedicated section to explore the long-term impacts of COVID-19 on the DS population.
 - NIH shared the funding opportunities available through INCLUDE for the next 3 years. Several opportunities will invest in a cohort study to assemble a large population of individuals with DS to understand outcomes over their lifetimes (INCLUDE Component #2).
- DS-Connect® Registry and INCLUDE DEIA Efforts
 - [DS-Connect®](#) is a registry that provides a secure, online platform to collect basic information about persons with DS. NIH encourages organizations to share information about this platform to expand the database.
 - INCLUDE continues to expand its outreach and presence globally. The team recently held discussions with DS organizations and community members in Costa Rica to identify ways to increase research participation and engagement in Latin America.
 - Held inaugural INCLUDE DEIA webinar on June 26, 2023, with more than 200 participants representing 10 countries in attendance. The event shared valuable insights on the *All of Us* program, diversity in clinical trial research, and recommendations to inform future directions. The executive summary will be available soon on the [INCLUDE Events page](#).

INCLUDE DCC Data Hub Launch

Joaquin Espinosa, GLOBAL

- The [INCLUDE DCC](#) was developed through a cooperative agreement with NIH to enable scientists and the DS community to collaborate and share data for further analysis and use.
- The DCC launched a dedicated [INCLUDE Data Hub](#) in March 2022 to store data from INCLUDE-funded grants, relevant academic studies, and other information of interest.
 - Currently, 11 INCLUDE-funded studies have been published to the hub, including 8,500 clinical DS profiles, 4,000 genomes, and thousands of -omics (transcriptomes, proteomes) studies
 - Visitors can access 55 terabytes of unpublished data
- To address the growing need to enhance data science and analytics for the large volume of data within the data hub, NIH created the Data Science for Diverse Scholars in DS Research (DS3) training program. This program will encourage students from diverse backgrounds to gain the skills necessary to import, analyze, and visualize big data.

Round Robin

All

- NTG, Matthew Janicki
 - Recently completed a national survey of families seeking information on perceptions of disability, dementia, and DS; 500 families participated, and the team is currently developing a report to share findings (expected release in fall 2023)
 - Examining equity and applicability of a new U.S. Food and Drug Administration (FDA)-approved Alzheimer's drug to the DS population
 - Created a matrix of recommendations, in collaboration with the LuMind Foundation, for the Centers for Medicare & Medicaid Services (CMS) and FDA to revise policy and reduce prescriber practice barriers across select states for persons with DS and disabilities
- NDSC, Jordan Kough
 - Continues to focus on outreach and engagement through several events and presentations, including trainings in Nashville and Tulsa in fall 2023; the events intend to engage the community and DS organizations in advocacy

- DSMIG, Nicole Baumer
 - Recently hosted 150 attendees at an in-person symposium focused on educating community providers that are not part of DSMIG or another DS-focused program
 - Expanded CME opportunities for DSMIG workgroups focused on adult health care issues and regression and autism spectrum disorder
 - Initiated Project ECHO, a monthly virtual meeting for primary care providers and trainees to collaborate and develop recommendations about the care of persons with DS
 - Revamped the organization’s website

- DSAIA, Sarah Mulligan
 - Held successful spring 2023 conference and began preparing for winter conference in Dallas next February
 - Encouraged DS Consortium members to leverage the dissemination capabilities at DSAIA to enhance connections with DS families

- Alzheimer’s Association, Heather Snyder*
 - Presented updated clinical criteria for Alzheimer’s disease to gather feedback from the community, which includes individuals with DS; criteria can be found at <https://aaic.alz.org/diagnostic-criteria.asp>

- IMDSA, Ruth Brown
 - Finalizing preparations for the “Research & Retreat” weekend and conference in San Diego, July 27-29, 2023, with support from an NICHD grant
 - Speakers will address attendees on a wide range of topics from basic science to preliminary findings from biospecimens gathered by five different research groups
 - Dr. Lori Carter-Edwards of Kaiser Permanente will lead a session on how to increase diversity across the IMDSA and encourage cross-collaboration to enhance the participation of minority populations in research
 - The conference will adopt a novel approach wherein self-advocates and their family members directly enroll in research projects or consultations of interest to them, at a setting that is most convenient for the participants

- GLOBAL, Michelle Sie Whitten
 - Working hard to increase the NIH budget while navigating a lot of challenges under the current political climate
 - DS Consortium members are supporting the U.S. National Plan to Address Alzheimer's disease (NAPA)
 - The University of Colorado's children's study continues to grow; currently working on eye and vision, expecting a white paper presenting findings from more than 500 unique ophthalmology patients
 - The team is also preparing a guideline for pregnant women on how to care for themselves if they received a DS diagnosis
 - Received a grant to launch a post-secondary program at Regis University that is now in its second year; other Jesuit colleges and universities have expressed interest in replicating the program
 - Received another grant to train adults with DS on hotel/hospitality jobs in the Denver area
 - Continues to provide support and increase awareness of projects led by the Linda Crnic Institute for Down Syndrome and NIH

- AAP, Marilyn Bull
 - Rolled out pediatric health care guidance for DS in May 2022; presentation was well received by the Council on Genetics
 - Guidelines for families are now available on [HealthyChildren.org](https://www.healthychildren.org) in both English and Spanish

- DSDN, Melissa Shutwell
 - Hosting the annual Rockin' Mom™ and Rockin' Dad™ retreats in the next few weeks for parents of a child with DS to connect and recharge
 - Expects to launch a new online community application in 2024 to move information off social media platforms, such as Facebook, which currently hosts 21,000 parents in more than 100 groups; this change will allow for better data, and easier, more effective dissemination of information, resources, research opportunities, etc.

- Medical outreach team will be attending six national medical conferences this fall, most notably, presenting and hosting a parent panel at the National Society of Genetic Counselors annual conference to discuss the diagnosis experience and what could be improved
- DSi, John Blascovich
 - Developed, translated, and published [Listen, Include, Respect](#), a set of international guidelines for inclusive participation, designed to incorporate persons with DS and disabilities; website is also currently available in [Spanish](#), with Portuguese and Arabic versions in development
 - Holding discussions with a variety of organizations to adopt these international guidelines; received commitments from the World Bank, Sight Saver, and UK DSA
 - Sponsored a [Cardiovascular Complications of Down Syndrome: Scoping Review and Expert Consensus Statement](#), published in January 2023, on congenital heart defects for the global DS population
 - Attended the annual United Nations Disability Conference with four people with DS, to advocate for easy-to-understand communication
- LuMind IDSC, Hampus Hillerstrom
 - Meeting next month with CMS to discuss two Alzheimer's drugs, which received FDA approval and demonstrate high efficacy, currently under assessment for applicability to persons with DS
 - Advocating for blood biomarkers, which are in place for the general population, to be expanded to persons with DS; the New England Health Innovation Center provided a forum to discuss equivalencies and blood biomarkers with key decision-makers and pharmaceutical companies
 - Discussing safety trials and conducting community outreach in collaboration with select organizations
- NDSS, Colleen Hatcher
 - Hosted more than 350 advocates from 42 states at the April 2023 DS Advocacy Conference (DSAC) in Washington, DC, to advocate for legislative priorities that impact the DS community

- DSAC and NDSS's ongoing advocacy enabled the inclusion of DS within the NAPA bill from the Senate HELP Committee and supported reintroduction of the Charlotte Woodward Organ Transplant Discrimination Prevention Act in the House and Senate
- Continues to update its [DS resources](#), including a regression checklist, end-of-life guide, caregiving guide, and aging guide
- Preparing for 2023 Adult Summit in Cincinnati from November 9-11; the summit will cover aging and DS through a whole-person, comprehensive approach providing attendees with information and practical resources
- T21RS, Roger Reeves
 - The report of the 2021 virtual meeting, *Innovating Therapies for Down Syndrome: An International Virtual Conference of the T21 Research Society*, is now available online (PMID: [37064334](#))
 - NIH will distribute a summary of the T21RS International Conference proceedings held in Long Beach, California
 - The next T21RS conference will be held June 5-8, 2024, in Rome, Italy
- Special Olympics, Andy Lincoln
 - Recently returned from the World Games in Berlin, where the team performed more than 15,000 health screenings and piloted a health behaviors survey
 - Currently focused on a longitudinal research study
 - Sharing cohort data in collaboration with an athlete advisory company; more information to follow
- Self-Advocate David Egan (presented on his behalf by parent Kathleen Egan)
 - Participated in the NIH RECOVER group discussion and expressed interest in identifying the incidence of long COVID-19 on the DS population; he urges the consortium to focus on physical and mental wellness for persons suffering with brain fog and other symptoms of long COVID
 - Recently visited Washington, DC, with Nick Leto (NDSS) to advocate for DS and share his book with new members of Congress
 - Received the prestigious Quincy Jones Award from GLOBAL in June

- Shared two requests, having recently learned that several states do not offer inclusive pre-school programs to citizens:
 - Further research on the impact of early childhood inclusive experiences
 - More DS-focused clinics and inclusive services across the 54 states and territories
- Self-Advocate Mitchell Levitz
 - Participating in a research study run by the Trace Research and Development Center at the University of Maryland that hopes to make health information easier to understand and more fit for people with DS
- Self-Advocate Karen Gaffney**
 - Dedicated to advocating for inclusion in families, schools, the workplace, and the community for people with intellectual disabilities
 - Presented to a parent support group as well as a Rotary Club on the importance of inclusion in the classroom and workplace settings for persons with DS
 - Expects to advocate for inclusion of students with disabilities in parish schools at an event in Denver in late August 2023
 - Will also present on employment challenges and areas for improvement at a statewide conference for providers in October 2023
 - Expects to participate in two GiGi's Playhouse fundraising events and a local Buddy Walk in Lake Oswego, Oregon

Long COVID and DS

Joaquin Espinosa, GLOBAL

Dr. Espinosa shared his current understanding based on the limited data that exist on symptoms and experiences of patients with “long COVID.” DS researchers are studying long COVID and have identified long-term sequelae in a large cohort that includes some individuals with DS. In addition, there is a DS-focused -omics study as part of RECOVER to set the stage for a larger, collective study. More research on this topic is necessary.

* Not in attendance; update provided via email received on 07/22/23

** Not in attendance; update provided via email received on 07/25/23