

22nd Meeting of the Down Syndrome Consortium: A Public-Private Partnership

June 24, 2022; 8:30-10:00 a.m. central time
National Down Syndrome Congress Annual Convention/Virtual

Roll Call

Organization	Name
American Association on Intellectual and Developmental Disabilities (AAIDD)	Maggie Nygren
Alzheimer's Association	Chris Weber
Down Syndrome Diagnosis Network (DSDN)	Jen Jacob
Down Syndrome International (DSi)	Jessamy Tang
Down Syndrome Medical Interest Group (DSMIG)	Nicole Baumer
GLOBAL Down Syndrome Foundation (GLOBAL)	Michelle Sie Whitten
GLOBAL	Joaquin Espinosa
GLOBAL	Bryn Gelaro
International Mosaic Down Syndrome Association (IMDSA)	Colleen Jackson-Cook
Jérôme Lejeune Foundation	Keith Mason
Jérome Lejeune Foundation	Julia Walsh
LuMind IDSC	Hampus Hillerstrom
National Down Syndrome Congress (NDSC)	Jordan Kough
NDSC	Tamara Pursley
National Down Syndrome Society (NDSS)	Kandi Pickard
NDSS	Colleen Hatcher
National Task Group on Intellectual Disabilities and Dementia Practices (NTG)	Matthew Janicki
Self-Advocate	David Egan
Self-Advocate	Kathleen Egan
Self-Advocate	Mitchell Levitz
Special Olympics	Andrew Lincoln
T21 Research Society (T21RS)	Roger Reeves

Organization	Name
National Heart, Lung, and Blood Institute (NHLBI)	Charlene Schramm
NHLBI	Huiqing Li
National Human Genome Research Institute	Jyoti Dayal
National Institute on Aging	Erika Tarver
<i>Eunice Kennedy Shriver</i> National Institute of Child Health and Human Development (NICHD)	Melissa Parisi
NICHD	Rachel Dows
NICHD	Linda Garcia
National Institute on Deafness and Other Communication Disorders	Elka Scordalakes-Ferrante
National Institute of Mental Health	Tara Dutka
National Institute on Minority Health and Health Disparities	Nathaniel Stinson
National Institute of Neurological Disorders and Stroke	Robert Riddle
National Center for Advancing Translational Sciences	Jamie Doyle
National Institutes of Health (NIH) Office of the Director	Cara Finley
INVITAE	Dawn Jae
LEIDOS	Adam Apostoli

Welcome, Introduction of New Members

Melissa Parisi, NICHD, and All

- Welcomed DSDN to the DS Consortium as the newest organization
- Remarks from Jen Jacob, DSDN executive director

INCLUDE Updates

Melissa Parisi, NICHD, and NIH Team

- INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome) Project was launched in 2018 under a Congressional Directive to address critical health and quality-of-life needs for those with Down syndrome.
- NIH has invested \$183 million in the past 4 years on INCLUDE on 202 new projects
- The INCLUDE team has reissued expired funding opportunity announcements (FOAs) and issued new FOAs. Updated FOAs can be found at <https://www.nih.gov/include-project/funding>.
- Planning on DS-Connect® updates, including adding more information to the DS-Connect® Professional Portal and improving mobile user access
- Community engagement updates:
 - Attended World Down Syndrome Day Conference

- Attended T21RS conference
- Attended DSMIG virtual conference
- Attending NDSC convention

Down Syndrome Diversity Initiatives

Linda Garcia, NICHD

- Upcoming workshop
 - Join us for the “Building a Diverse Community for Down Syndrome Research” virtual workshop
 - September 20-21, 2022
 - To learn more and register visit the INCLUDE events page: <https://www.nih.gov/include-project/events>
- Upcoming meetings and conferences
 - [NDSS Buddy Walk NYC](#)—September 17, 2022
 - [DSDN Rockin’ Moms Retreat](#)—Gaylord National, DC September 16-17, 2022
- DS-Connect[®]
 - The DS-Connect[®] website is a registry that collects health information from people with Down syndrome and their families through a series of online surveys. Researchers then explore the information to learn more about the health and everyday needs of people with Down syndrome. Led by NIH, the [DS-Connect website](#) provides details on how to join, what happens to the information collected, and how we protect participants’ privacy. For questions, please email us at DSConnect@nih.gov.
 - Please help us increase awareness about DS-Connect[®] by sharing our videos on your social media pages and/or websites.
 - We are planning usability updates to the DS-Connect[®] website to improve user experience and increase user numbers
- Goal of 10,000 participants by our 10th year anniversary of September 2023
 - [Access self-advocate sign-up video for DS-Connect](#)
 - [Learn one family’s take on DS-Connect](#)
 - [Learn about Down syndrome research](#)
 - [Get answers to “What is DS-Connect?”](#)



INCLUDE Data Coordinating Center (DCC) and Data Hub Updates

Joaquin Espinosa, DCC

- INCLUDE Data hub launched on March 21, 2022
- The hub includes more than 55 terabytes of unpublished data from more than 6,000 participants—of which more than half come from DS-Connect[®]
- Other half sourced from molecular data including genome sequencing; more than 2,600 genomes sequenced
- Hoping to reach 4,000 genomes in the next year

- Incorporating data from Kids First study V1.0, including data from Emory, Baylor, & Texas
- V2.0 aims for more cohorts and data types; clinical data is queued to reach portal in the next 12 months.
- Hosting DS3 training event: Data Science for Diverse Scholars in Down Syndrome this summer. Focus on training in data science for Down syndrome researchers.
- Making changes for data sharing and making data available. It's de-identified so the public can use it.
- Staying at the forefront as an example to other researchers for data sharing

Round Robin

All

- DSMIG, Niki Baumer
 - Hosted virtual DSMIG conference last week
 - Working groups formed for Down syndrome and regression, Down syndrome and autism
 - Will continue to host webinars throughout the year; plan to be in Orlando next year
- IMDSA, Brandy Hellard
 - Went to camping resorts for their retreats and participated in research
 - Activities from previous 9 months: working with families to bring awareness to mental health access
 - Want to bring attention to families in need of mental health care who are being turned away locally due to restrictions
- NTG, Matt Janicki
 - Finished report on barriers to assessing persons with dementia and an intellectual disability, published and presented to National Advisory Council
 - Developing standards with a working group on how to provide guidance for providers when assessing individuals with Down syndrome
 - Working with CDC on Healthy Brain Initiative
- T21RS, Roger Reeves
 - First live meeting in 3 years in Long Beach, California, this June
 - A lot of the info about COVID and Down syndrome was researched and developed by T21RS members
 - New animal models: large scale analysis of genomes and gene expression, learning and memory, support for clinical trials
 - Reporting large influx of young investigators to organization
 - Mark your calendars! T21RS Next Meeting: Rome 2024
- LuMind, Hampus Hillerstrom
 - Clinical trial network is now 14 sites around the United States
 - Natural history study is collecting data from second timepoint

- Training developed to support consistency across clinical trial sites and increase reliability
- More and more interest from industry to conduct clinical trials for Down syndrome
 - One drug from Biogen targeting Alzheimer's disease, but there's no data on a single research participant with Down syndrome
 - Encourage support for participants with Down syndrome to join these trials.
- Mitchell Levitz, Self-Advocate
 - Participated in public health training
- Jérôme Lejeune Foundation, Keith Mason
 - Expanding medical clinics globally: Argentina and France, identified location in Spain (working to open in November 2022)
 - Pilot program in United States to translate care, goal to expand in 2023
 - New website in French, Spanish, and English by September
- NDSC, Jordan Kough
 - NDSC convention this week here in New Orleans
 - Plenary session garnering interest among many attendees
 - Expanding programs and partnering with organizations to bring programming to different places
 - Convention has been a success!
- David Egan, Self-Advocate
 - Shout out to AAIDD: David's book earned the organization's media award
 - Involved in Reach-UP DS, You Participate: goal to bring communities of all backgrounds to participate in studies
 - Linking DS-Connect to other resources and self-advocates
 - Speaking at: self-advocate group, Source America--employing individuals with disabilities in the workplace
- GLOBAL, Michelle Sie Whitten
 - GLOBAL roundtable at NDSC, hoping to expand
 - Plans to attend DSDN Rockin' Moms convention
 - Hosted in-person AcceptAbility Gala in May in DC
 - Created fund for ambassador, Deondra Dixon, about halfway to goal
 - Dr. Fran Hickey will be retiring; recruiting for leader of largest pediatrics center in the United States
 - GLOBAL adult guidelines just published, Spanish and Japanese translations coming out later
 - November 12, 2022: Be Beautiful Be Yourself fashion show in Denver
- Crnic Institute, Joaquin Espinosa
 - On track to have 1,000 participants in Human Trisome Project donate samples

- Clinical trial of immune modulation to treat regression is upcoming; hypothesizing that regression might have an immune component
- DSI, Jessamy Tang
 - DSI is a United Kingdom-based advocacy organization committed to improving quality of life and inclusion of those with Down syndrome
 - World Down Syndrome Day conference in New York, self-advocacy conference with the United Nations
 - Soft launch of listen-include-respect guidelines collaboration with Inclusion International; feedback appreciated: www.listenincluderespect.com
 - Approached by international advertising agency to have a digital influencer: Kami
 - For info about Kami, Digital Influencer: <https://www.ds-int.org/blog/introducing-kami>
 - Next World Down Syndrome Congress in Australia, July/August 2024
 - Working with organizations on inclusion and employment, self-advocacy: Kenya, Nigeria, Rwanda, Uganda
 - Working on spreading word on COVID-19 and best practices: Brazil, Argentina, South Africa
- DSDN, Jen Jacob
 - Serve about 18,000 families online. Capturing about 22 percent of new parents (under 1 years)
 - Adding new moms every month
 - Started webinar series and online resource library
 - Hosting onsite events: Rockin' Dads (July in Indianapolis) & Rockin' Moms (September in DC) events. Parent-only events to rest, recharge, and get new information
 - Act as a "clearinghouse" to help distribute resources, research, and information
 - Jen will be resigning from her position; DSDN is looking for a new executive director and applications are being accepted through July 2022
- NDSS, Kandi Pickard
 - Three 1-day adult summits this year: Jacksonville, Indianapolis, Dallas
 - Finalizing location for fall 2024 adult summit in Ohio
 - Upcoming April 2023 advocacy conference in Washington, DC on Capitol Hill
 - Updated resources coming out in late summer
 - End-of-life and Down syndrome companion to be published this year