

21st Meeting of the Down Syndrome Consortium: A Public-Private Partnership

February 4, 2022
10:30 a.m.-Noon (eastern time)

Roll Call

Organization	Name
American Association on Intellectual and Developmental Disabilities (AAIDD)	Maggie Nygren
Alzheimer's Association	Chris Weber
American Academy of Pediatrics (AAP)	Marilyn Bull
Association of University Centers on Disabilities (AUCD)	John Tschida
Down Syndrome Affiliates in Action (DSAIA)	Lauren Camp
Down Syndrome International (DSi)	Jessamy Tang
Down Syndrome Medical Interest Group (DSMIG)	Nicole Baumer
Global Down Syndrome Foundation (GDSF)	Michelle Sie Whitten
GDSF	David Tolleson
GDSF	Bryn Gelaro
International Mosaic Down Syndrome Association (IMDSA)	Colleen Jackson-Cook
Jérôme Lejeune Foundation	Keith Mason
Jérôme Lejeune Foundation	Julia Walsh
Linda Crnic Institute	Joaquin Espinosa
Lumind IDSC	James (Jim) Hendrix
Lumind IDSC	Hampus Hillerstrom
National Down Syndrome Congress (NDSC)	Jordan Kough
NDSC	Tamara Pursley
National Down Syndrome Society (NDSS)	Colleen Hatcher
National Task Group on Intellectual Disabilities and Dementia Practices (NTG)	Matthew Janicki
NTG	Marianne Barbera
Self-Advocate	David Egan
Self-Advocate	Kathleen Egan
Self-Advocate	Mitchell Levitz

Organization	Name
Special Olympics	Andrew Lincoln
T21 Research Society (T21RS)	Roger Reeves
Invitae	Dawn Jae
Invitae	Adam Apostoli
National Cancer Institute (NCI)	Malcolm Smith
National Center for Advancing Translational Services	Jamie Doyle
National Heart, Lung, and Blood Institute (NHLBI)	Charlene Schramm
NHLBI	Huiqing Li
National Human Genome Research Institute (NHGRI)	Jyoti Dayal
<i>Eunice Kennedy Shriver</i> National Institute of Child Health and Human Development (NICHD)	Melissa Parisi
NICHD	Sujata Bardhan
NICHD	Rachel Dows
NICHD	Linda Garcia
National Institute of Mental Health	Tara Dutka
National Institute of Neurological Disorders and Stroke	Robert Riddle
National Institute on Aging	Erika Tarver
National Institute on Deafness and Other Communication Disorders	Elka Scordalakes-Ferrante
Office of the Director (OD), NIH	Erica Landis
OD	Cara Finley

Welcome, Introduction of New Members, and Discussion on Nominating New Members
Melissa Parisi, NICHD, and All

INCLUDE Updates
Melissa Parisi, NICHD, and NIH Team

- [FY21 awards posted](#) (granted \$65 million in FY2021 and funded 53 new awards)
- More than 5,400 participants registered for DS-Connect
- Support Data Coordinating Center (DCC), plan to launch DCC data portal on March 21, 2022
- Plan to reissue many expired funding opportunity announcements

Down Syndrome Diversity Initiatives

Sujata Bardhan and Linda Garcia, NICHD

- Upcoming workshops:
 - “Building Diversity Across the Down Syndrome Research Ecosystem”
 - “Hematologic Malignancies in Children, Adolescents, and Young Adults with Down Syndrome” (Led by NCI)
- Upcoming meetings and conferences:
 - **March 21:** World Down Syndrome Day (WSDS): DSI sponsoring event at United Nations (UN); theme is “Inclusion Means”
 - **June 9-12:** Fourth International T21RS Conference
 - **June 16-18:** DSMIG-USA Annual Symposium (virtual)
 - **June 23-26:** NDSC 50th Annual Convention (“Diversity and Inclusion in Down Syndrome Research: NIH and the INCLUDE Project”)

INCLUDE DCC and Launch of Data Hub

Joaquin Espinosa, DCC

- DCC has three cores: Administration and Outreach, Data Management, Data Portal
- Why a data portal? Many other medical info portals, this adheres to FAIR data-sharing principles (Findable, Accessible, Interoperable, Reusable)
- Included community outreach: “listening tour” of approximately 50 advocacy and professional organizations
- Projected launch is March 21, 2022
- More than 7,000 research participants, including:
 - More than 2,600 complete genome sequences from participants with T21
 - More than 400 research participants from Human Trisome Project
- Also developing an INCLUDE minimal common data set and common data elements as well as common neurodevelopmental test battery

Round Robin

All

- **Nicole Baumer, DSMIG**
 - DSMIG-USA will host a virtual 2022 symposium
 - Virtual event June 17-18, 2022. Many members have travel restrictions
 - New board leadership as of February 2022: Katie Frank, president; Nicole Baumer, vice president; Marilyn Bull, secretary; Angela Lombardo, treasurer
- **Jessamy Tang, DSI**
 - World Down Syndrome Congress (WDSC) held virtually in Dubai in November 2021

- Planning WDSO in Australia in 2024
- WDSO events at UN in NYC and in Geneva: What does inclusion mean?
- **Michelle Sie Whitten, GDSF**
 - David Tolleson joined GDSF
 - Collaborated with Bryn Gelaro (COVID & Down syndrome) and DSMIG on joint webinar
 - Adult health care guidelines for adults with Down syndrome: <https://www.globaldownsyndrome.org/medical-care-guidelines-for-adults/>
 - Developing metrics for adult guidelines and implementing adult rubrics in adult clinics
 - Family-friendly version of Adult Down Syndrome Health Care Guidelines coming soon
 - NDSS partner for prenatal testing pamphlet
 - AcceptAbility Gala May 24, 2022, in Washington, DC
 - Be Beautiful Be Yourself in November 12, 2022, in Denver, Colorado
- **Joaquin Espinosa, Linda Crnic Institute**
 - Safety and efficacy testing for pharmaceutical for inflammatory skin conditions
 - More than 50 percent have received prescriptions for off-label use of the drug
 - Two participants with Down syndrome regression disorder had positive response, preparing for another clinical trial based on this
- **Colleen Jackson-Cook, IMDSA**
 - Brandy Hellard is the new IMDSA executive director
 - Christina Welch is the new president
 - Not holding annual research and retreat weekend this year; planned for 2024
 - Newsletter and brochure publications
- **Keith Mason, Jérôme Lejeune Foundation**
 - Focusing on expanding medical care; clinics now in France, Argentina, and Spain
 - Completed pilot care program in United States
 - Building biodata bank
- **Jim Hendrix, LuMind IDSC**
 - CMS coverage decisions on FDA-approved Alzheimer's drug: excludes Down syndrome community
 - LuMind advocating for trials for the medication in those with Down syndrome
 - LIFE-DSR study: LuMind natural history study: 270 recruitment goal. Around 180 currently. Substudies funded with industry consortium.
 - Developed Goal Attainment Scale, caregiver assessment, launched late last year and about halfway through recruitment
 - Publishing papers on survey work:
 - [Caregivers evaluate independence in individuals with Down syndrome](#)
 - [Parental perspectives on research for Down syndrome](#)

- **Jordan Kough/Tamara Pursley, NDSC**
 - Jordan is the new NDSC executive director
 - Getting ready for convention in June 23-26, 2022, in New Orleans
 - Trying to identify areas to collaborate and support at the national and local levels

- **Colleen Hatcher, NDSS**
 - Research forum and adult summit March 4-5 in Jacksonville; visit <https://www.accelevents.com/e/2022-adult-summit-on-the-road-in-jacksonville-fl#about> for more information
 - Working on CMS Aduhelm drug issue also
 - Provided public comment to National Alzheimer's Project Act on Down syndrome and Alzheimer's link; request to form subcommittee for intellectual and developmental disabilities

- **Matthew Janicki/Seth Keller, NTG**
 - Developing virtual meeting for Florida to raise awareness for intellectual disability, Down syndrome, Alzheimer's, hoping to work with other state agencies.
 - Looking at barriers for neuro assessment in neuroatypical adults: Report of neuro-atypical conditions panel on doing cognitive assessments in adults: <https://www.the-ntg.org/screening-assessment>
 - Continuing to do trainings (virtual), including international presence

- **David Egan, Self-Advocate**
 - Presenting and speaking at different conferences, universities, and events
 - Going to Pittsburgh for data collection as part of ABC-DS project, looking to track biomarkers for Alzheimer's
 - NDSS organizing virtual gala March 17 to honor his advocacy legacy
 - Pursuing REACH-UP-DS initiative: Research, Education, Advocacy, Core Health You Participate Down syndrome

- **Mitchell Levitz, Self-Advocate**
 - Staying active in both local and larger communities with advocacy
 - Participating in national conferences on disability

- **Andrew Lincoln, Special Olympics**
 - New vice president for research and evaluation at Special Olympics
 - USA Special Olympics Games will be held in Orlando this June
 - New health screenings and fitness challenge
 - World summer games will be in Berlin 2023
 - Worldwide survey about how COVID has affected athletes

- **Roger Reeves, T21RS**
 - New leadership team in place: Andre Strydom is current president
 - Annual T21RS meeting in Long Beach, California, in June

- **Maggie Nygren, AAIDD**
 - Making plans to hold annual conference in June 2022
 - David Egan being honored at conference for his memoir, *More Alike Than Different*
 - Special issue concerned with equity for people of color in *Inclusion* journal September 2021

- **Chris Weber, Alzheimer's Association**
 - International conference being held July 31-August 4, 2022, in San Diego
 - Professional Interest Area (PIA) for Down syndrome will have meeting/abstracts
 - Dr. Jerrold Chun is leading PIA webinar. Sleep is topic in April

- **Marilyn Bull, AAP**
 - One signature away from final approval for health care guidelines revision for children and adolescents with Down syndrome
 - Major challenge in dissemination, but AAP will help with this
 - Groups here can help disseminate guidelines once published

- **John Tschida, AUCD**
 - Not available for report-back

- **Lauren Camp, DSAIA**
 - Not available for report-back

Closing Comments

- Next meeting: June 2022 at NDSC meeting in New Orleans, hopefully in person!
- **Send nominees for new Down Syndrome Consortium members to Linda Garcia, linda.garcia@nih.gov by February 18 (see attached nomination form)—being extended to February 25.**
- Discussion of sibling workshop and the sibling network: David Egan, Mitchell Levitz, Michelle Sie Whitten, Colleen Hatcher
- Need outreach and clearinghouse for messaging: amplify messages.
- What are some measurable metrics of progress for Down syndrome research?
 - Vaccine information: CDC put people with Down syndrome in high-risk group for vaccines. Worked as a team to save lives.
 - Autoimmune skin conditions—impact 25 percent or more; now dermatologist coming to meeting and treatments being tested
 - Down syndrome regression disorder—better understanding and tools to ameliorate and address it.
- May be value in coordination around messaging so that new discoveries can be amplified.