

Building a Diverse Community for Down Syndrome Research

September 2021, 2022

: September 20, 2022

Session 1: Welcome and Introduction, Scope of the Issue and Stakeholders' Perspectives

Welcome Remarks from the INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome) Team

Sujata Bardhan, M.S., Ph.D., Program Director, Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

Courtney F. Akin, Ph.D., Acting Associate Deputy Director, National Institutes of Health (NIH)

Meeting co

² A glossary of terms and acronyms used in this report can be found at the end of the document.

Accomplishing these goals will require broad thinking and innovation. INCLUDE aims to engage and amplify the voices of self-advocates and family members and to conduct community outreach to strengthen diversity, equity, inclusion, and accessibility (DEIA) plans. This workshop is one example of efforts to engage the community, listen, learn, and take action. On behalf of NIH and INCLUDE leadership, Dr. Aklin thanked the attendees and speakers, who are integral to efforts to include people with DS from diverse backgrounds in research and to advance science.

Overview of Diversity in Down Syndrome Research

Session Chair and Moderator: Sujata Bardhan, M.S., Ph.D., Program Director, NICHD

Dr. Bardhan said that NIH is the largest funder of biomedical research; 90% of funds go to university research institutions. Each year, about 6,000 babies in the United States are born with DS. They and their families face challenges that can be overcome with appropriate support and treatment. NIH invests in research that could advance our understanding of DS and help lead to treatments that could improve quality of life for people with DS.

Investment in medical research has led to a dramatic increase in the average life expectancy of people with DS, from 9 years in the 1950s to 50–60 years today. The increase occurred following the American Academy of Pediatrics' recommendation in the early 1990s that all infants with DS receive cardiac screening.

However, other health issues come with aging, and dementia is now a leading cause of death among people with DS. In addition, the median life expectancy for people with DS is much higher among White people than among people of other races, indicating a need to understand health disparities.

Dr. Bardhan asked attendees to take part in DS-Connect, a registry that collects basic health and demographic information directly from families and is available in English and Spanish. The registry helps families connect with approved researchers and take part in studies. It includes participants from across the United States, although outreach is still needed in rural communities. The goals are to make the registry helpful for families and to partner with local and national chapters to learn from underserved groups.

NIH can use DS-Connect to help INCLUDE researchers recruit for studies. However, most participants in the registry are White. Including more participants from diverse backgrounds is important because of the registry's utility for study recruitment. Failing to include a large sample population in research can lead to myths that certain types of people or phenomena do not exist. Being inclusive will help researchers understand more about DS for all.

NIH's [UNITE initiative](#) aims to address health disparities in research communities. Dr. Bardhan noted that day 2 of the workshop would include a presentation from the Chief Officer for Scientific Workforce Diversity (COSWD) about work to promote diversity in the extramural research ecosystem.

Summary of Listening Session with Families

Linda Garcia, Communication and Outreach Coordinator, INCLUDE Project and DS-Connect

Ms. Garcia presented a summary of a listening session with families that was held on May 18, 2022. NIH invited parents who had a child with DS to share their experiences.

NIH can help by ensuring that research is relevant and that families understand how it will help their loved ones who have DS. Research objectives should be clearly described, and updates should be provided through all possible channels and in families' preferred languages. Ms. Garcia noted that NIH staff regularly attend community events to connect with families, provide information, and answer questions.

Ms. Garcia invited participants to visit the [INCLUDE website](#) and to contact her at linda.garcia@nih.gov.

Summary of Listening Session with Researchers

Aruna Natarajan, M.D., Ph.D., Program Director, National Heart, Lung, and Blood Institute (NHLBI)

Dr. Natarajan shared proceedings from a listening session for DS researchers. The goal was to learn about researchers' positive experiences and their barriers and challenges, so that NIH can develop actions and initiatives to increase diverse participation.

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NIH can help in several ways:

- x Collaborating with national organizations in DS research
- x Encouraging clinicians to improve flexibility in scheduling
- x Addressing the need for study partners to accompany participants throughout studies
- x Improving compensation

Dr. Ilias presented examples of consultations:

- x The ACTIV-6 Pittsburgh site wanted to improve enrollment from populations that are underrepresented in biomedical research (UBR). CEACR put together a panel of experts that included community members from the groups the site wanted to engage. The panel provided strategies to improve enrollment, such as focusing on language inclusivity, bridging the digital divide, and reducing the burden on participants. The team also helped the site develop strategies for identifying and reaching out to trusted community members.
- x The ACTIV-6 Stakeholder Advisory Committee sought guidance on disseminating best practices to all ACTIV-6 sites. CEACR used the Pittsburgh site's consultation to showcase lessons learned and how they could be shared across sites.

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Session 2 Stakeholders' Perspectives Panel Discussion

Session Chair and Moderator: Erika Tarver, MS, Senior Project Manager, Division of Neuroscience, National Institute on Aging (NIA)

Panelists:

- x Families and Self-Advocates
 - o Yadira Carrillo, Entrepreneur
 - o Kate Dougherty, President, National Down Syndrome Congress (NDSC) Board of Directors, Rural Communities
 - o Dolores Harden, Member, Navajo Nation Advisory Council on Disabilities
 - o Taylor Murphy, High School Student, GLOBAL Self-advocate Speaker
 - o Shauntel Neal-Howe, M.B.A., Past President, NDSC Board of Directors
 - o Chidanand Rajghatta, M.S., Columnist, Washington Bureau Chief for the Times of India
 - o José Torres Rodriguez, President, Fundación Puertorriqueña Síndrome Down
- x Researchers
 - o Joaquin Espinosa, Ph.D., Professor, Department of Pharmacology, University of Colorado, Denver
 - o Andrew McCormick, M.D., FAAP, Associate Professor, Department of Pediatrics, University of Pittsburgh
 - o Esther Son, Ph.D., Assistant Professor, College of Staten Island, CUNY

families are very busy trying to meet

Dr. Espinosa added that effective communication is important. Researchers want to generate enthusiasm and excitement

DS research at Emory began in the early 1990s with the Atlanta Down Syndrome Project. In 2000, the project was expanded to become the National Down Syndrome Project (NDSP), covering six states. When the NDSP concluded, Emory began the Emory Down Syndrome Project, which was based in Atlanta but had nationwide enrollment thanks to the availability of online participation. Substudies

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Recruitment materials should be tailored to the audiences that researchers aim to reach. Researchers should make sure that participation is worth participants' time by increasing incentives and returning information to participants. Community partnerships are important and must be long-term; research teams should provide resources that extend beyond their research. Also, tts. C. Chouldte7 (s)-9 ou r tha theitreetarci(e)0

DMI is not the same as shared decision making (SDM), a concept that developed with respect to the relationship between adult patients and providers. SDM requires mutuality, shared information giving, and agreement about the decision. The concept does not necessarily apply to children, who are not equal partners in decision making.

DMI recognizes that children still need to learn and practice decision making in multiple settings. It has several benefits: It gives youth the opportunity to observe and practice decision making, enhances their self-efficacy in decision making, facilitates a positive view of the decision-making process and research, and facilitates trust and open communication between youth and the research team. Dr. Miller

disabilities. Including people with disabilities in research is critical for understanding how factors affect them differently. NIH's focus has been on identifying barriers and how to overcome them. Best practices for one

have suggested that younger children would do better with storyboards and social stories, and older children might prefer videos, but the team has not yet collected data on that.

Dr. Bardhan said that a child might assent to a portion of a research protocol, but another portion might have different procedures; that is another reason for assent to be a continuous process. She also suggested that as researchers use more storyboards and videos to explain procedures, families might become more comfortable with them, which could help reduce anxiety.

Dr. Kelly said that research on consent and assent has largely focused on ethical issues. Work on multimedia is only recently emerging, so there is an opportunity to expand the field. She believes it would have implications not only for people with DS and those with intellectual and developmental disabilities but also for children in general.

Dr. Bardhan said that many protocols in low- and middle-income countries (LMICs) use videos to explain research processes. Researchers could look to global health to help make processes easier for families. Dr. Rosser added that All of Us uses videos as an option for consent, in addition to text.

Open Discussion

Dr. Espinosa said that although much work remains to be done, INCLUDE funding and the awareness that the INCLUDE leadership is displaying will be very helpful.

Dr. Bardhan said that she was grateful to have representatives from rural communities in Session 1. With INCLUDE funding, DS-Connect can address gaps in its representation from underserved communities, including rural communities. She stressed that outreach is a constant process and that building trust with communities happens over time. She said that the workshop is a step in the right direction and noted that it will be followed by more events to keep the dialogue and momentum going.

Dr. Bardhan noted that families and researchers brought up many of the topics mentioned in the listening sessions, such as trust, transportation, and fear.

Dr. Kelly asked whether issues regarding Social Security and tax liabilities should be discussed at the NIH level. Dr. Bardhan said that NIH leads the Down Syndrome Consortium, which includes many national and international organizations. Perhaps it can expand the conversation to work with partners to address the issues. Ms. Corbett said that NIH has no control over the IRS income thresholds. However, the Office for Human Research Protections is looking into many issues regarding participant compensation, including this one. Also, the Secretary of Health and Human Services has been very engaged in participant compensation issues. NIH likely cannot make a difference on its own, but it has been elevating the problems to make sure others are aware of them.

Dr. Bardhan closed Day 1 by thanking Ms. Garcia, the workshop co

In the chat, Viviana Fernandez asked whether NIH is considering working closely with groups from other countries, such as Spain, where there are many DS research projects and findings. Dr. Bardhan reiterated that new INCLUDE funding opportunities are open to foreign applicants. She also noted that DS-Connect has been translated into Spanish, and representatives had attended the 2019 Trisomy 21 Research Society (T21RS) meeting in Barcelona, Spain. The team has been active in including researchers and families from outside the United States.

In the chat, David Egan asked about work with Down Syndrome International (DSi). Dr. Bardhan said that DSi is part of the Down Syndrome Consortium. The INCLUDE team had presented at DSi's 2022 virtual meeting on #InclusionMatters and planned to participate in its next meeting, which will be held in Australia in 2024.

Neuroscience Workforce Diversity

National Institute of Neurological Disorders and Stroke (NINDS) Strategies for Enhancing the Diversity of Neuroscience Researchers

Michelle Jones-London, Ph.D., Chief, Office of Programs to Enhance Neuroscience Workforce Diversity (OPEN), NINDS

Dr. Jones-London introduced examples of efforts to expand diversity from the field of neuroscience. The mission of [NINDS' OPEN](#) is to increase opportunities for all. The office aims to identify diverse students, recruit and prepare them by getting them on a training pathway, and provide mentors and connect them with professional networks.

OPEN uses a systemic approach, at the individual, institutional, and scientific community levels. NINDS programs span career stages from high school level to new faculty. They include fellowships and other forms of individual support, as well as institutional programs like the T32. The diversity supplement, which funds a high school, undergrad, graduate, or junior faculty trainee on an existing grant, is a program that all 27 ICs support. It provides support for the trainee's salary so that they can get pilot

Q&A

Dr. Li suggested that one benefit of increasing diversity among trainees could be an increase in the engagement of underrepresented groups in research. She asked Dr. Wu if he agreed and what strategies he used to increase engagement. Dr. Wu agreed and said his program benefitted from having students from underrepresented backgrounds. For example, Atlanta has a large Hispanic community, but until recently the lab had no one who could speak Spanish. A Spanish-sp

The team sought Dr. Gleason's advice on how best to maximize funding. Ultimately, they wanted to make sure community members were included as paid members of the outreach team and divided the funding for engagement among three team members: a parent, a self-advocate, and a liaison.

Dr. Nogueron, the mother of a young adult with DS, reflected on her experience as part of the outreach team. As someone who is active in her community, she has been building relationships with other parents for a long time. She has learned that education is an important element, especially for underserved populations who may not have had good experiences with health care. It is important to talk about historical trauma. It is also important for study staff to think about the logistics of the visit. Families may lack transportation, have to manage their child who is throwing a tantrum because they do not want to go to the doctor, or deal with worry about losing their job. Study staff should consider having materials in Spanish, having Spanish-speaking staff, and making sure people receive compensation for participating. Building relationships allows the team to think outside the box—for example, by learning about the barriers that participants face coming into the clinic and offering to meet them at home instead. It is important to show empathy for participa-

journey, I am a simple man touching people's hearts and finding my way of given what the world needs of making it better for all to be included for people that have any special needs. The best way is to work with others to learn in yourself, of listening to their stories of their lives. Coming from me, I truthly listen the brain is in the center of everything that makes life even more.

Recruitment Innovation Center (RIC)

Tiffany L. Israel, M.S.S.W., Research Services Consultant, RIC, Vanderbilt University Medical Center
Jasmine Bell, M.P.H., Research Consultant, RIC, Vanderbilt University Medical Center

Dr. Nogueron said a major issue they face is accommodating children, especially the siblings of participants in a study. Ms. Israel said being flexible has worked at their site. They allowed mothers to bring kids with them and to breastfeed as needed. The center also provided snacks.

An attendee asked in the chat if there are any recruitment practices inclusive of families at or below the poverty level. Collaborating with partners that work with those communities and finding participants using social media can be helpful. Ms. Ray emphasized the importance of flexibility—for example, by meeting at times that work with participants' work schedules and other obligations. Structuring compensation around their needs is also important. Ms. Israel added that the RIC conducts studios in spaces where people are already comfortable and where it is easy for them to navigate, such as their school or home. Dr. Gleason said that although it is not a specific focus of her team's programs, they have reached individuals in this group. She agreed that flexibility is key and added that offering culturally safe research visits is important.

In the chat, Tamara Pursely from the NDSC asked how national and local organizations serving the DS community can better promote research studies and help with recruitment. Dr. Hartley encouraged organizations to reach out to research teams in their area. She said members of the University of Wisconsin team would be happy to give a presentation or provide materials about the study and get to know the organization and the needs of the community they serve. The organization can help researchers identify opportunities to meet the members they serve and provide education.

An attendee asked in the chat if there is recruitment at events held by the Special Olympics, a consortium partner and an organization with a lot of reach in the DS community. Dr. Bardhan said that NIH has not attended their events. Ms. Whitmoyer said that ABC-DS set up a table to recruit participants at the Wisconsin Special Olympics, which was a successful experience. In the chat, another attendee mentioned a similar experience in Georgia. Ms. Nodvin said ADMH collaborates with the Special Olympics on recreational programming. Participants must have a medical release to participate, which ADMH provides.

are seven Americans with Disabilities Act (ADA) specialists. There is a 6-month wait for the vision specialist who practices in St. Louis, a 2.5-hour drive away.

It is important to include as many people as possible from the community to bring this positive pressure to bear. That includes reaching out to teachers, therapists, DS families, clinics, and more. Engagement may rely on in-person connections. Zoom may not be a viable option for providing telehealth services if the Internet connection is not working.

Ms. Dougherty emphasized the importance

Dr. Grigorenko introduced her epidemiological research on learning disabilities in rural Africa, emphasizing her eight pillars of community engagement. Although little is known about DS in Africa and there are not enough data to assess prevalence, there is no reason to believe it differs from elsewhere. The infrastructure put in place through the learning disabilities study (the Bala Bbala Project) should make it possible to conduct a DS study in the future.

The Bala Bbala Project engages 36 schools with 4,500 children (ages 7–21), who went through a long assessment battery that measured their reading ability, body mass index (BMI), IQ, and vision. Before starting the study, the researchers met with regional chiefs and local headmen and got their permission to work in their areas. Team members were recruited from the local community. The researchers used assessments that were specially developed to be child-friendly and culturally adapted and conducted the assessments in places that were convenient for the participants. The study also involved genotyping, which the research team took time to explain to participants in detail.

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support for transportation. Part of the research budget goes to materials to engage siblings who are also at the research visit.

In an effort to support families' needs comprehensively, community navigators at the center's Family Information and Navigation Desk direct families to resources. The center has a food pharmacy with food from a nearby rooftop garden. Social workers offer their services on site, and clinicians support families by attending IEP meetings for school.

State-of-the-art DS care requires additional time and resources, advocacy, and care coordination. Because this falls back on the provider to provide, funding support for the clinical program would give providers a way to enhance the care they provide with research.

Open Discussion

mentorship research awards. The funding would not have research goals attached but would support the clinical program for the purpose of diversifying participants. In addition to partnering with other government organizations, if NIH is serious about workforce development and education of the broader community, it could financially support organizations like DSMIG or professional organizations so they can offer a year of ~~tsot~~ c Tw (o) (o)no.4128m9190 .756 (h)2 -0.27 8J e -0.lin 0 Td I7.T -0.081784 31.257up Tw (o)

outcomes based on different conditions co-occurring with DS. She also shared resources and strategies used by **All of Us** to address needs. Dr. Kelly and Dr. Miller discussed how to make research easier for families. They presented examples of using storyboards and videos to make consent and assent easier and discussed how to involve children in decisions about research participation, a topic that is understudied in children with DS. Dr. Brown discussed sleep health disparities in DS, including the fact that sleep-disordered breathing, the most reported sleep disorder in DS, is exacerbated in underrepresented groups. Research gaps remain, limiting our understanding of the impact of these conditions on people with DS.

Day 2 opened with a focus on ensuring diversity in the DS scientific workforce. INCLUDE aims to fulfill a congressionally mandated directive to expand the pipeline of new DS investigators and is working to increase the diversity of trainees. FOAs emphasize diverse recruitment and prioritize diversity in investigator teams, including foreign applicants. Dr. Ley discussed the importance of recruitment and retention of diverse faculty members, and Dr. Jones-London introduced specific programs supporting diverse recruitment in the field of neuroscience. Dr. Wu gave his personal perspective on mentoring, including ways that he builds collaborative teams that rely on input from all team members.

Glossary

ABC-DS Alzheimer's Biomarkers Consortium-Down

IEP	individualized education plan
INCLUDE	INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndromE
IRB	institutional review board
LIFE-DSR	Longitudinal Investigation for the Enhancement of Down Syndrome Research
LMICs	low- and middle-income countries
NCATS	National Center for Advancing Translational Sciences
NCSDR	National Center on Sleep Disorders Research
NDSC	National Down Syndrome Congress
NDSP	National Down Syndrome Project
NDSS	National Down Syndrome Society
NHLBI	National Heart, Lung, and Blood Institute
NIA	National Institute on Aging
NICHD	Eunice Kennedy Shriver National Institute of Child Health and Human Development
NIGMS	National Institute of General Medical Sciences
NIH	National Institutes of Health
NIMHD	National Institute on Minority Health and Health Disparities
NINDS	National Institute of Neurological Disorders and Stroke
OPEN	Office of Programs to Enhance Neuroscience Workforce Diversity
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