

Building a Diverse Community for Down Syndrome Research

September 20–21, 2022
NIH-Sponsored Virtual Workshop¹

DAY 1: September 20, 2022

Session 1: Welcome and Introductions | Scope of the Issue and Stakeholders' Perspectives

Welcome Remarks from the INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndromE)² Team

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

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

Meeting co-host Dr. Bardhan opened the workshop at 12:05 p.m. ET and welcomed the participants. She reviewed the agenda and directed attendees to the [DS-Connect® website](#) and to the [INCLUDE website](#), which houses recordings of the May listening sessions and will later host the summary and videocast recordings of this workshop. Nearly 200 people registered to attend the workshop.

Dr. Bardhan thanked the Down Syndrome Consortium partners; INCLUDE leaders Lawrence Tabak, D.D.S., Ph.D., Diana Bianchi, M.D., and Gary Gibbons, M.D.; and the workshop planning committee and co-lead Linda Garcia.

Dr. Bardhan then introduced Dr. Aklin, the Acting Associate Deputy Director of NIH. Dr. Aklin advises on a broad array of complex and sensitive issues. She was previously chief of staff at the National Institute on Minority Health and Health Disparities (NIMHD).

Dr. Aklin welcomed the attendees on behalf of her fellow INCLUDE cochairs, Dr. Bianchi and Dr. Gibbons. The workshop had more than 200 registered participants, including self-advocates, family members of people with Down syndrome (DS), researchers, trainees, members of DS organizations, health care and service providers, experts in diversity and inclusion, and NIH program staff.

INCLUDE was launched in 2018 in response to a congressional directive. Since then, NIH has invested more than \$183 million in more than 200 projects and has supported numerous training awards focused on DS. INCLUDE is now completing its fifth year and is focused on expanding the inclusion of individuals with DS from diverse backgrounds to help understand and eliminate health disparities and advance health equity in underserved populations. It also aims to expand the pipeline of trainees and new investigators from diverse backgrounds who work on DS.

¹ All presentations from [Day 1](#) and [Day 2](#) can be viewed via the [NIH VideoCast](#) site.

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 in co-occurring conditions that might affect overall health.

Currently, we know that sleep health disparities occur in the general population, and poor sleep health is associated with adverse outcomes. People from racial and ethnic minority groups report shorter sleep duration, which influences many domains, including overall health.

Congress has directed INCLUDE to engage with people from diverse backgrounds who have DS and their families and to increase the number of investigators and trainees studying the condition. The NIH definition of health disparities includes not only minoritized racial and ethnic groups but also populations with less privileged socioeconomic status (SES), underserved rural communities, and sexual and gender minority groups.

INCLUDE has three components, all of which are addressed by building a diverse DS research community.

- Building diverse research teams addresses component 1, conducting basic science studies on chromosome 21.
- Assembling diverse cohorts addresses component 2, assembling a large study population of individuals with DS across the lifespan.
- Developing inclusive clinical trials addresses component 3, including people with DS in existing and future trials.

NIH has invested more than \$100 million in INCLUDE over the past 4 years, allowing the funding of 202 research projects from early and established investigators. The projects include several clinical trials. However, demographic data show that people who are of non-Hispanic ethnicity, White, and male are overrepresented in the trials. There is more work to do to achieve diverse participation.

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 diverse participants.

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, Communications 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 positive experiences with and barriers to participation in research. The goals were to engage and listen to the DS community, involve more people with DS and their families in research, and understand the needs of community members in order to engage them in research that benefits them.

Participants discussed strategies that have improved research participation of diverse communities:

- Speaking the same languages as research participants
- Educating communities about the importance of participation
- Building trust by communicating and creating positive experiences
- Making safety paramount
- Providing compensation
- Sharing research progress

Listening session participants most frequently cited time as a barrier to participating in research. Other barriers included language, trust, education, location, fear, and transportation.

Researchers can take several actions to help resolve these barriers and facilitate research participation:

- Provide educational resources
- Ensure understanding through upfront explanations and clear communication
- Build relationships within the community through organizations that caregivers trust
- Make research accessible to families
- Organize informational events in marginalized communities and offer compensation for participation
- Promote cultural competency training for research teams
- Work closely with national organizations, whose communications many potential participants receive
- Use social media effectively

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.

When asked what strategies have increased diverse participation in research, researchers focused on building trust. They cited several practices:

- Engaging and participating in communities in contexts other than research, such as health and education
- Involving communities in research design
- Building a culture that emphasized benefit to both researchers and participants
- Factoring physical features of different races into research design—for example, providing electroencephalography (EEG) leads appropriate for different kinds of hair
- Meeting participants where they are located
- Making the consent process, transportation, and logistics easier
- Having students from diverse backgrounds lead research projects
- Melding research activities and clinical support

Facilitating diverse participation requires communication. In addition to speaking participants' languages, researchers need cultural competence and behaviors that inspire trust and collaboration. Communications related to topics like developmental disabilities require nuanced language skills in participants' preferred languages. Research teams should be diverse. Also, researchers should enter participants' communities as well as using mobile and online technologies to encourage participation in registries.

Researchers must be aware of the burden that research places on participants and should balance value and burden. Practices in this area included the following:

- Building bridges between academic centers and community health care sites, where more members of diverse backgrounds may receive care
- Improving collaborative research and clinical care, with funding for both
- Reporting research outcomes back to communities, to help incentivize future participation
- Logistically coordinating research and clinical care so that families can make fewer trips
- Learning from other organizations in the community that work with diverse population groups and improving research teams' skill sets
- Using online messaging and consent forms with appropriate translation

When asked to name barriers and challenges to including diverse communities in research, researchers listed trust, language, conveying benefit, and logistics.

NIH can help in several ways:

- Collaborating with national organizations in DS research
- Encouraging clinicians to improve flexibility in scheduling
- Addressing the need for study partners to accompany participants throughout studies
- Improving compensation and logistics
- Sharing experiences among researchers
- Building relationships in the community
- Encouraging students and researchers from less-represented groups

Researchers suggested that NIH support efforts to increase diversity by funding projects that meld clinical care with research; including community support in funding mechanisms; addressing the role of medical devices that work for less-represented populations; considering diversity, community building, and technological solutions in funding decisions; and building inclusive cohorts.

[The Community Engagement Alliance Consultative Resource \(CEACR\)—Leveraging Lessons for the Down Syndrome Community](#)

Maliha Ilias, Ph.D., Program Director, Center for Translation Research and Implementation Science, Health Inequities and Global Health Branch, NHLBI

Dr. Ilias said that NIH started the Community Engagement Alliance (CEAL) Against COVID-19 Disparities in response to health disparities that became apparent during the COVID-19 pandemic. CEAL includes 21 teams across the United States with academic and community partners. As the program's resources grew, NIH realized that other researchers could use them to improve their community engagement and inclusive participation efforts. CEACR was launched in 2021 to share those resources.

The program is led by the University of Pittsburgh Clinical and Translational Science Institute. CEACR involves several partners, including the Community-Campus Partnerships for Health, which facilitates work between academic and community partners.

The CEACR team has established an asset map of the community engagement expertise and resources available within CEAL, such as communications materials and information on vaccinating children. When a consultee approaches CEACR, the team uses the asset map to curate a customized panel of experts in community engagement and experts from the communities that the consultee aims to engage. The panelists evaluate the consultee's needs and develop tailored recommendations. The team aims to complete each consultation within 30 days. After the consultation, the team checks in with consultees to obtain feedback it can use to refine the process.

Dr. Ilias displayed topics on which CEACR provides consultations, including developing community-competent outreach and communications strategies, understanding the barriers to inclusive participation in research, building an inclusive research workforce, addressing vaccine hesitancy in minoritized communities, and improving community-engaged research study designs.

The CEACR team began working in November 2021. It completed a pilot consultation with the ACTIV-6 study, refined its own process, and has since engaged in 11 additional consultations. Consultation numbers are increasing as word about the program spreads. The team has worked with multiple research programs, including major NIH-funded clinical trials such as ACTIV-6 and Researching COVID to Enhance Recovery (RECOVER). Through consultations, it is working with various community groups, including Spanish speakers, rural populations, American Indians, and older adults.

Dr. Ilias presented examples of consultations:

- 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.
- 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.

The CEACR team is learning that consultations must be specific to the research setting. CEACR encourages groups that have questions to reach out so that it can provide specific recommendations.

In addition to 30-day consultations, CEACR can provide quicker, more focused consultations. For example, it can identify speakers for workshops and conferences to provide the perspectives of patients and caregivers. It has also provided consultations to CEAL teams, including providing recommendations for considering alternate payment approaches for community-based partners.

Dr. Ilias encouraged participants to visit the [CEACR webpage](#) and to take advantage of the CEACR speaker series. Recordings from the series are available on the website; the program aims to build a virtual toolkit for researchers.

Q&A

Dr. Bardhan asked whether CEACR is available for the research community outside NIH. Dr. Ilias said that it is available only to NIH-funded researchers. Academic institutions are encouraged to have community partners, and community partners can access CEACR through their funded academic partners.

Dolores Harden asked where CEACR had met with Native American families. Dr. Ilias clarified that the program had received requests from clinical researchers who wanted to improve communication or engagement with Native American populations. CEACR had included experts who work with those populations on panels to provide targeted recommendations to the researchers.

Session 2: Stakeholders' Perspectives: Panel Discussion

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

Panelists:

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

Ms. Tarver introduced the [panelists](#). She began the discussion by asking the families and self-advocates to share positive experiences from taking part in research studies.

Ms. Dougherty said that she lives in a rural community. She found out about the hypoglossal nerve stimulator trial through the NDSC annual convention, and her son was able to take part. The trial has been life-changing, improving her son's cognition, speech and language, behavior, and quality of life. However, her family would not have known about it had they not been able to travel to access information. Also, rural communities may fear how information gained through research will be used. Ms. Dougherty said that she spreads the word about the benefits of participating in research, both for quality of life and for information gathering, but she understands that people are very protective of their family members who have DS.

Ms. Carrillo said that she has been part of the Human Trisome Project at the Crnic Institute. The staff know how to work with people who have DS, and they make her feel welcome.

Ms. Murphy said that her family participated in focus groups for the GLOBAL Adult Guidelines. Participation was all online, and her family could complete it on their own time.

Ms. Neal-Howe said that her daughter is a willing participant in research, giving blood and other samples. What works for her is that the researchers talk to her directly, rather than to her parents. Ms. Neal-Howe also said that working with trusted organizations within communities is key to engaging the communities.

Mr. Torres Rodriguez said that his participation in research has been limited, but participating is beneficial because it prompts the family to stop and think about what they can improve. Day to day,

families are very busy trying to meet their children's needs. He also said that low-income families should have the opportunity to participate in panels and in research.

Mr. Rajghatta said that his family also has limited experience participating in research and would welcome more opportunities. Mr. Rajghatta was joined by his wife, Mary Breeding, Ph.D., who said that their son's knowledge and test scores are benchmarked against early childhood test scores that are not geared toward children with special needs. As a result, she and Mr. Rajghatta receive feedback that makes them feel like research subjects.

Ms. Harden said that she had not heard of any families participating in research or of any opportunities for them to do so in her area, the northwestern region of New Mexico. She shares information with families within and around the Navajo Nation, including families in New Mexico, Arizona, and Utah. She did not believe that families living in reservation border towns or in New Mexico's 19 pueblos had participated in research, either.

Ms. Dougherty said that she agreed with Dr. Breeding's comments about inappropriate benchmarks for children. There is no good pipeline to help explain children's diagnoses to the educational system, which often takes a developmentally inappropriate perspective. Ms. Dougherty also noted that she is selective about the research opportunities that she shares through her organization. To help protect families, she does not share opportunities that do not come from a familiar, trusted source, such as NIH.

Ms. Tarver next asked the researchers to share examples of their participation to promote DEIA in their research or clinical practice.

Dr. McCormick said that academics are often siloed away from the larger community. He cited a colleague's [paper, which focused on the concept of working with, rather than on, communities](#). He also related that a Pittsburgh report had found that the city was one of the most unlivable cities for Black women. Academics were appalled, but community partners said they had been aware of and working on the problem. Those partners were angry that the academics had not sought their input for the report. The episode led to an initiative in which researchers and community scientists are paired and funded at equal levels. Dr. McCormick said that the paradigm needs to shift to include communities in identifying community needs and then working with resources from the academic community. Funding community scientists and community partners in research will help improve recruitment and bring a more inclusive perspective to research.

Dr. Espinosa said that collaborating with GLOBAL has been instrumental for recruitment and clinical trials in the Human Trisome Project. The collaboration also helps the researchers connect with community members by traveling to meetings. Dr. Espinosa said that he would be visiting the Navajo Nation in November and would be interested in meeting with Ms. Harden. Finally, partnering with the advocacy community allows researchers to gather feedback on their materials and make sure they are communicating clearly.

Dr. Spinazzi said that she was a co-principal investigator (PI) on a study that aimed to develop an assessment tool for self-management for families with special needs. The study had strict enrollment goals for various populations and found that it had enrolled "too many" African American and Spanish-speaking patients. Several steps helped the researchers achieve this enrollment. First, they considered patients' needs when designing the study. Because families are very busy, the researchers ensured that recruitment occurred during a care visit, demonstrating that they valued families' time. Also, patients feel that the medical team cares for them, so they are not surprised to be approached about research. Clinical research coordinators are skilled in using a community navigation service to help families find

resources they need. Recruiting diverse groups of participants requires supporting the clinical programs that serve the patients holistically, thereby building trust.

Dr. Tapia said that he had been part of a study that used phone interviews, which could be conducted at any time of day that was convenient for families. For the clinical trial portion of the study, the researchers reduced the burden for participants by offering different sites for families to visit. That required a lot of coordination and some travel for research coordinators. The team also conducted clinical interactions via video visits whenever possible. Dr. Tapia said that it is important to be flexible and meet families where they are.

Dr. Son said that she recruited immigrant Asian populations for her research, and translation services were critical. The populations are interested in participating in research but may not be able to take part in focus groups that are not in their own languages. For one project, Dr. Son worked with community partners who speak multiple languages and could help explain the study's purpose, research methods, and logistics. The project also recruited and interviewed families at an agency that they regularly visited, so that they did not need to make an extra trip. Families received a stipend. Most critically, the project hired a bilingual research assistant who had a racial and ethnic background similar to the participants'. On another project involving a Korean immigrant population, the research team formed a strong relationship with community partners, who then connected Dr. Son with other agencies. Dr. Son said she had learned that providing services to immigrant populations and building strong relationships with community partners have both short- and long-term benefits for research.

Ms. Tarver next asked the families and self-advocates about challenges they had faced in participating in DS research.

Ms. Murphy said that she is a high school sophomore with an active schedule, and it is difficult to miss school to participate in research. Her family tries to plan research time around her school breaks.

Ms. Carrillo said that she runs a boutique and knows about business. She feels motivated when her time is valued. For example, it helps to receive a gift card for her participation.

Ms. Dougherty said that school attendance policies that do not allow leeway for students who need to participate in clinical trials can be a barrier. Also, costs for travel and housing can be prohibitive.

Mr. Torres Rodriguez said that in Puerto Rico, the principal organization that conducts research is the Department of Health. The studies include people with all kinds of disabilities. Therefore, the results, which are used by other institutions, such as the Department of Education, apply to disabilities as a whole rather than specifically to DS. In schools, families receive some treatment and information that are not necessarily correct for people with DS. Groups look to research from the mainland United States, but that research is not representative of the Latino community or island setting of Puerto Rico. Mr. Torres Rodriguez stressed the importance of DS-specific research.

Dr. Espinosa said that representatives from the INCLUDE Data Coordinating Center would be in San Juan for the [Society for Advancement of Chicanos/Hispanics and Native Americans in Science \(SACNAS\) conference](#) in October, and Mr. Torres Rodriguez said that he would be there. Dr. Bardhan added that she would be representing INCLUDE and DS-Connect at the meeting. She would also be attending the [Annual Biomedical Research Conference for Minoritized Scientists \(ABRCMS\)](#).

Martha Ramos Fontan, M.S., said that she is from Puerto Rico and has a daughter who has DS. She asked how many families or people with DS INCLUDE is affecting in the United States, including Puerto Rico.

Dr. Bardhan said that only seven families from Puerto Rico are registered on DS-Connect. She suggested that she and Ms. Ramos Fontan connect after the SACNAS conference.

Ms. Tarver asked the researchers what can help facilitate participation in clinical research by people in underrepresented and underserved communities.

Dr. Tapia said that families with children who have DS have many competing activities. Families in underserved communities have an additional layer of commitments. For example, parents may have multiple jobs, and taking a day off may be onerous. Reimbursement must compensate for that.

Dr. Espinosa emphasized the importance of flexibility at every step of the research process, including arranging for parking and providing clear instructions for finding the research site. Also, scientists should ensure that the research team is diverse and, if possible, is bilingual. This is especially important for clinical coordinators, who work closely with families.

Dr. Spinazzi said that as a clinician researcher, she is swamped with trying to solve real problems for families. For example, she works to help families access appropriate educational services and disability benefits. It is difficult to also implement research projects. However, she has a large, diverse population of patients who would be very engaged in research. To reach those patients, researchers must support the clinical programs that support them, so that the programs can facilitate research participation.

Dr. McCormick said that clinicians base their research ideas on their own experiences and perspectives. Asking communities what problems are important to them could lead to better engagement.

Dr. Son said that participants spend time and effort to take part in research, and if they do not see clear beneficial outcomes, they will have less motivation to participate in future studies. Many researchers try to provide the best possible outcomes, but sometimes communication is unclear. Researchers should use participants' language rather than jargon and help participants understand how the outcomes will affect their communities rather than only individuals. Ms. Neal-Howe said that researchers should provide clear and timely outcomes as well as frequent status updates.

Dr. Tapia added that cab and food vouchers are important and should be included in grant budgets.

Rebecca Grzadzinski, Ph.D., said that adequate compensation for families is difficult to fit into NIH budgets. Technology will advance and produce less expensive research methods, but currently, the cost of some data collection is high, and it can be difficult to gather large enough samples with diverse representation.

Ms. Tarver asked the families and self-advocates how researchers can build trust within their communities.

Ms. Carrillo said that information and websites should be in community members' languages so that they can educate themselves about the researchers' work.

Ms. Murphy said that diversity matters. Researchers should make sure that the diversity of the DS community is represented in their recruitment materials and on their research teams.

Ms. Harden said that building trust and relationships is important in rural communities, where people do not know what to expect when new information arrives. Researchers should reach out to families and let them know what is coming up. She also agreed with Dr. Spinazzi's statement about trying to solve real problems. She said that in rural areas, services are limited. Most of the people she knows in the Navajo Nation who have DS are nonverbal, and that is because speech therapy services are so limited.

Ms. Neal-Howe said that efforts in other areas, such as breast cancer, are doing good work. Breast cancer walks seem to be very diverse and have developed champions within communities. Researchers must meet families where they are and help meet basic needs, such as the need for speech therapy. Ms. Neal-Howe said that she would like to see researchers support and partner with existing community institutions, such as pediatricians' offices, DS clinics, and churches, to build trust and engagement.

Mr. Rajghatta agreed that it is important to report outcomes to families in an easily understandable form. Families are pressed for time, and when they devote time to research, they like to know the outcome. Also, research programs should cover all legal residents, not only citizens.

Dr. Breeding said that meeting diverse socioeconomically based needs as well as language needs is critical. She and Mr. Rajghatta belong to the DS Network in Montgomery County, Maryland. It is a diverse group and has a dual-language program to help meet the needs of Hispanic community members. However, even in their well-resourced community, there are high levels of inequality in access. Accessing resources and services is challenging, and families with fewer resources who are not able to advocate for themselves may be left out. Schools and clinics need support to help them reach underserved populations. Dr. Breeding added that there is a lot of sociology and psychology literature about servicing communities, and there could be opportunities for cross-disciplinary research.

Ms. Dougherty said that researchers should understand that many people in rural and underserved communities are dealing with trauma from their experiences with educators and clinicians who lack exposure to DS. Experts who visit rural or underserved areas help build trust by letting families know that there is a larger DS community that understands best practices. It is not possible for experts to visit every underserved community, but rural communities communicate with each other.

David Chaplin, M.D., Ph.D., said that researchers must not expect to enter a community and be trusted immediately. Developing trust takes years, and researchers must have perseverance and patience. They should make sure to address research questions that the community believes are important early on, get to know community members, and include them as core members of the research team.

Mr. Torres Rodriguez said that research should be conducted in participants' native language. The objective of the research should be clear, and the results should be available to the community. Puerto Ricans are aware of past research misconduct. Participants need to understand how the research will benefit their community. Also, studies should be conducted in the same way in Puerto Rico and in other places. Finally, researchers should take into account that in Latino communities, mothers are very important in children's health and education.

Dr. McCormick said that community partners should be paid. Dr. Tapia added that researchers must understand that community partnerships are long-term and continuous.

Ms. Tarver asked the researchers how they can build trust.

Dr. Spinazzi said that researchers should show up for communities to let them know that the researchers are not there to use and abuse them. For example, researchers can provide educational opportunities, such as question-and-answer sessions. Dr. Spinazzi attends her patients' individualized education plan (IEP) meetings whenever possible, which lets families know that she is in their corner. She also speaks about institutional racism, which is a major reason for lack of trust, whenever appropriate. Acknowledging the issue is important. Also, if research finds that an intervention or service is successful, researchers should advocate for it to be available through insurance. Dr. Spinazzi said that it is very disappointing that access to hypoglossal nerve stimulation currently requires Medicaid.

Dr. Espinosa added that effective communication is important. Researchers want to generate enthusiasm and excitement but must be careful not to exaggerate or overpromise.

Dr. Son said that being a genuine friend is key to building trust. She does not have a child with a developmental disability, and communities ask her why she is interested in the topic. She shares her experience as a social worker in South Korea, where she worked with many families that had children with developmental disabilities. That experience led her to become a researcher. When she shares her personal stories with community members, they are more willing to share their own stories. She believed that sharing her stories and passion has helped community members see her as a person.

Dr. McCormick said that historically, some communities have suffered abuses through research. Researchers must remember that they have work to do to avoid repeating past mistakes.

Kathleen Egan, Ph.D., said that trust should be preceded by educating the members of a community about proven advances and remaining questions to be answered. With that understanding in place, trust can be built.

Session 3: Addressing Racial and Ethnic Health Disparities in Scientific Research

Session Chair and Moderator: *Marishka Brown, Ph.D., Director, National Center on Sleep Disorders Research (NCSDR), NHLBI*

Dr. Brown said that the session would focus on research for people living with DS, and she introduced the [speakers for the session](#).

Inclusion Across the Lifespan

Dawn Corbett, M.P.H., NIH Inclusion Policy Officer, Office of Extramural Research, NIH

Ms. Corbett said that NIH has a longstanding commitment to inclusion in clinical research, with policies in place since 1986. In 1993, a law was passed requiring NIH to include women and members of racially and ethnically diverse backgrounds. In 1998, NIH issued a policy requiring the inclusion of children. In late 2016, Congress passed the 21st Century Cures Act with new mandates for inclusion, including new requirements to report results by sex or gender, race, and ethnicity.

The 21st Century Cures Act also required NIH to convene a workshop on age grouping and age exclusion from clinical research, publish guidelines addressing consideration of age in clinical research, and publish the number of children included in NIH research by research condition or disease category. NIH began implementing these requirements in 2017. At the workshop, experts discussed barriers to including children and older adults in clinical trials. The workshop found that data about participation in research by age were lacking and that older adults and children faced similar barriers to participation.

NIH considered feedback from the workshop and the public and [announced](#) the [Inclusion Across the Lifespan policy](#) in 2017. The policy states that NIH human subjects research must include individuals of all ages unless there are scientific or ethical reasons not to do so. It also requires the submission of individual-level participant data in progress reports based on sex or gender, race, ethnicity, and age at enrollment. The policy is intended to help address gaps. For example, NIH found that the mean age of participants in clinical trials was lower than that of people who had the conditions being studied.

[NIH policy](#) requires that women and members of racially and ethnically diverse backgrounds be included in all NIH-funded clinical research studies unless there is a compelling rationale for exclusion. Cost and

convenience are not considered compelling rationales. Phase III clinical trials have some additional requirements to report analyses by sex or gender, race, and ethnicity.

Applicants for funding must provide plans for inclusion of women, members of racial and ethnic minority groups, and individuals across the lifespan. Applicants also must provide minimum and maximum age limits and an enrollment report showing the demographics of participants they will recruit.

NIH now has data on its website showing the demographics of research participants. Data are available for all condition and disease categories, including DS. Ms. Corbett displayed NIH clinical research enrollment data by age groups and by sex/gender, race, and ethnicity. In 2021, 20% of research participants were children, 58% were adults ages 18 to 64, 19% were older adults, and 3% were of unknown age. More than half of participants were female, and 11% were Hispanic or Latino. Less than 1% were American Indian/Alaska Native or Native Hawaiian or Pacific Islander, 13% were Black or African American, and 64% were White.

In 2020, NIH held a meeting to assess progress and ongoing challenges. The [meeting report](#) is available online. Themes included limiting inclusion and exclusion criteria to what is necessary, weighing the risks of exclusion versus participation, minimizing participant and caregiver burden, considering diversity within populations, assessment and adjustment of recruitment and retention, and researcher training and resources.

NIH recently funded a contract with the National Academies of Sciences, Engineering, and Medicine that produced a [report on improving representation in clinical trials and research](#). The report contains recommendations for federal agencies and the scientific community, such as reviewing reimbursement for studies, participant compensation, and NIH processes and ensuring that community engagement efforts are adequately funded.

NIH's Office of Extramural Research provides resources for the scientific community, including [blog posts](#) and [trainings](#) for researchers.

Diversity in Down Syndrome: The DS360 Project

Tracie Rosser, Ph.D., Assistant Professor, Emory University School of Medicine

Dr. Rosser said that diversity in research is necessary for adequate conclusions from the data. Individual differences are important when analyzing data, especially as we move toward an era of personalized medicine. Diversity among research participants is also important for understanding health disparities, which could be related to a variety of factors, such as race and ethnicity, social determinants of health, or environmental exposures.

Incidence of DS is higher among Hispanic families. There are also disparities by race and ethnicity in leukemia risk, congenital heart defects, some gastrointestinal defects, and life expectancy. These disparities may or may not be related to genetics.

In both the DS360 project at Emory University and NIH clinical research in general, there are more White and non-Hispanic participants than there are participants of other races or Hispanic ethnicity. U.S. Census data show that the U.S. population became more diverse between 2010 and 2020. The percentages of White, Black, and Hispanic and non-Hispanic people in the United States are comparable to the percentages among participants in clinical research, but representation of racial and ethnic minority groups in research needs to improve.

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 included the Congenital Heart Defects Study and the Down Syndrome Cognition Project.

In addition to diversity among research participants, diversity in the data types that researchers collect is important. Emory projects have collected survey data, blood and saliva samples, sequencing data, and medical record data and have conducted cognitive testing.

The NDSP found that people who are female or have Black ancestry were at increased risk for congenital heart defects, but people of Hispanic ethnicity had decreased risk. The researchers also found differences in risk for gastrointestinal defects based on sex, ancestry, and maternal age. However, the sample sizes were too small to show statistical significance. Barriers to participation in research affect researchers' ability to extend the research to find out whether the differences in risk are statistically significant.

Factors that influence participation include the time involved for participants and families; whether there are appropriate incentives, such as help with transportation; and whether participants will gain useful or interesting information. All these factors can be addressed.

Dr. Rosser said that in addition to her position as an assistant professor at Emory, she is also the Emory PI for the *All of Us* Research Program. The NIH-funded program aims to enroll one million or more people and build one of the largest, richest, and most diverse biomedical data sets of its time. One goal is to enroll people from UBR communities. Currently, about 80% of participants are from UBR communities, and about half of those identify as something other than White. Dr. Rosser stressed that UBR communities are not defined only by race and ethnicity; access to care, geography, income, and education are also factors.

All of Us hopes to enable discoveries that drive more precise approaches to care. The program engages with people and communities who have often been left out of research. *All of Us* data combine biological samples, other biological factors, and information on social determinants of health. The program expects to follow participants for at least 10 years.

All of Us uses several methods to reach and engage UBR communities. The program:

- Aims to meet participants where they are and provide them with results that they can understand.
- States in its core values that participation is open to all and that participants are partners.
- Ensures that participants have access to their information.
- Builds trust by being transparent about what it is doing and about issues in past research.
- Informs participants about how their data will be protected and the risks involved, including how receiving genetic information could affect their insurance.
- Provides education for participants.
 - For example, the program provides information about genomics, helping participants understand not only *All of Us* but why research is important overall.
- Conducts custom campaigns that aim to clearly convey why communities' participation is important.

Dr. Rosser listed some ways to increase participation and diversity. Involving participant advisory boards can help researchers obtain feedback early in their projects to find out what they might need to adjust.

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, teams should ensure that their research is relevant to the populations they aim to involve.

An Inclusive Consent/Assent Process

Andrea Kelly, M.D., M.S.C.E., Professor of Pediatrics, and Victoria Miller, Ph.D., Psychologist, CHOP

Dr. Kelly said that she and Dr. Miller would be sharing their journey toward addressing a critical methodological gap in human subjects protection, as well as their efforts to optimize the informed consent process for people with DS.

Dr. Kelly explained how she came to her current work. In the early 2000s, she was studying the intersection of sleep apnea and cardiometabolic risk in otherwise healthy people. Her mentor was a sleep expert who conducted work in DS. Later, when Dr. Kelly was pregnant with a child with DS, another mentor invited her to work on DS research. She began studying body composition and cardiometabolic risk in youth and emerging adults with DS. An experience with a participant highlighted the importance of screening for obstructive sleep apnea (OSA) and making screening more convenient for families. Now, Dr. Kelly is a co-PI with Dr. Tapia on a study on sleep apnea testing at home.

Family members participating in the study told the researchers that a previous sleep study had been traumatic for one child and that another child's doctor had said that a sleep study was not possible for the child. The comments highlighted the importance of having research team members who were sensitive to learning disabilities and sensory issues and of better education for the medical community.

Dr. Kelly works with a multidisciplinary team with expertise in DS and pediatric sleep research. The team has partnerships with parent advocates, who advise on the community's priorities and how to engage families well. As a parent of a child with DS, Dr. Kelly built trust with clinic patients and research participants that has helped with partnerships.

The team has focused on communication during the consent and assent process, using social stories and videos. One parent said they appreciated that, with videos and social stories, there were no surprises for their child. However, three people declined to participate after watching a video. Although that was disappointing, the team felt that it showed they had provided an informed study. Dr. Kelly played a clip from a video explaining what caregivers can expect in a sleep study. The videos feature people who have DS, to help community members feel more engaged.

Social stories involve placards showing various components of study procedures. A story accompanies the placards, and the team personalizes the story with the names of the child, sleep tech, and research coordinator. The coordinator lines up the cards and, if possible, asks the child to line up the cards to determine whether they understand the procedures.

A study supplement is investigating the relationship of OSA to outcomes. The team wanted to engage an older population, but because it is based at a children's hospital, the team did not have access to expertise in adult neurocognitive testing. The team sought help from Vidya Kamath, Ph.D., a neuropsychologist at Johns Hopkins. Dr. Kamath has been training the research coordinator in cognitive testing for emerging and young adults with DS.

The team originally planned to recruit 35 participants but expanded both the sample size and the age range in response to enthusiasm among the DS community. Race and ethnicity percentages are similar to those in DS research overall.

Participants were recruited through non-DS-Connect resources that the team typically uses. However, in conducting the study, the team began working with Dr. Bardhan to use DS-Connect and garnered interest from participants across the country.

Reasons that potential participants have given for declining to participate have informed the team's next steps. Limited English proficiency is one reason. The team has a Spanish interpreter, but the interpreter cannot conduct neurocognitive testing in Spanish. Also, many English-language questionnaires are not available in Spanish. Cost and travel were barriers for many people who reached out through DS-Connect.

A parent of an 18-year-old with DS said that no one had ever spoken directly to their child before in either the clinic or a research setting. This comment influenced the team's next steps, which aim to optimize the consent and assent process. The team strives to set a better standard for engaging people who have DS and their families in research.

Dr. Miller said that she would discuss what it looks like for children and adolescents with DS to have a voice in decision making about research participation and clinical care.

Assent means the agreement to participate. It is based on the moral obligation to nurture children's moral growth and their developing autonomy. Assent is not meant to be equivalent to autonomous decision making, so it does not need to mirror the elements of informed consent. Instead, the assent process gives potential participants the opportunity to choose whether to participate to the extent that they are able. The process can vary based on children's development and as their capacity matures.

Assent is required for research participation, but it can be waived if there is a possibility of direct benefit that is available only in the research context or if the child is judged incapable of assent. Local institutional review boards (IRBs) determine when and how to obtain assent, so processes vary widely.

The National Commission for the Protection of Human Subjects recommended that assent include four elements: the knowledge that procedures will be performed, the choice to freely undergo those procedures, unambiguous communication of that choice, and awareness of the option to withdraw from the research. Research on assent has largely focused on children's understanding and decision-making capabilities, assessing when children are competent to assent. Less work has focused on how parents, children, and researchers interact and communicate about research. Dr. Miller believed that it is important to consider how to involve children in decisions about research participation and their clinical care in ways that benefit them. Children can be involved in decisions without having the final say.

Decision-making involvement (DMI) refers to how adults and children or adolescents engage each other in decision making and problem solving. The concept focuses on the process and interaction rather than on who makes the decision. It also emphasizes the behaviors of parents, children, and providers or researchers. For example, parents or researchers can ask youth what questions or concerns they have and check that they understand the research procedures, and young people can share their opinions and ask questions. The concept recognizes that children and adolescents can be meaningfully involved in decision making in multiple ways and that parents and providers retain a critical role as sources of support and advice. Research has shown that many children and adolescents want their parents' input on decisions and that having a choice without the support they need can be burdensome.

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 conducted a study that found that when youth felt that that researchers involved them in discussions about research participation, they perceived more fairness, were more satisfied with the decision-making process, and felt that they had a greater voice in decision making.

Parental involvement in decision making means that that parents understand the greater context of the decision as well as the child's values and goals, they can provide information and support, and they facilitate implementation of the decision.

Little research has studied disparities in DMI. However, youth from racial and ethnic minority groups experience disparities in interactions with health care providers, and that likely includes interactions in the research context. Several factors, including negative past experiences with the health care system, racism, provider and health care system bias, and the history of research misconduct in marginalized communities, may make the assent and consent processes and DMI difficult for people from racial and ethnic minorities.

Dr. Miller listed implications of DMI for the assent process. Researchers should:

- Respect parents' role in supporting youth in decision making.
- Consider involving youth in decision making in multiple ways, including providing them with tailored, developmentally appropriate information; soliciting their opinions and questions; and checking that they understand the research procedures.
- Appreciate the ways in which the assent process can support youth development and focus on that as the goal.
- Recognize the unique barriers for youth and families from racial and ethnic minority groups. Focusing on the relational aspects of the decision-making process could help build trust and enhance patient–researcher interactions.

More research is needed to optimize the assent and decision-making process for youth and adults with intellectual disabilities. However, Dr. Miller believes that DMI is an appropriate concept for these groups, because caregiver involvement may always be relevant, and there may be a need for additional advocacy or involvement of a legally authorized representative later in adulthood.

Dr. Kelly said that the team has not formally tested the value of the storyboards or videos or obtained community input to optimize them. The team is now doing additional work to improve the process, including gathering a family advisory board to provide input on barriers to participation and experiences with consent and assent. The team will also seek feedback on the videos, the storyboards, and the utility of using a digital platform to assess understanding of study procedures, purpose, risks, and benefits. The goal is to assess the tools' utility in promoting self-efficacy in the DS community.

To address diversity, the team will recruit locally and nationally, targeting specific geographic regions to ensure representation. It will recruit from rural and urban populations and will set lower bounds for representation of people of various races and ethnicities. The team found that most people who could

not participate in its studies because of limited English proficiency spoke Spanish, so it will focus on Spanish, translating materials into Spanish and obtaining feedback on Spanish-language videos and questionnaires from people with limited English proficiency. The team always has a Spanish interpreter, and the research coordinator is also obtaining a certificate in Spanish interpretation. The team will also work with DS-Connect to optimize recruitment strategies.

The team will perform semi-structured interviews in person and through virtual platforms to include people from across the country. For people in the Philadelphia area, the team will consider in-home assessment, because the clinic environment can be stressful.

The team aims to cover travel and lodging costs in advance so that families do not have to pay for them and wait for reimbursement. The team also aims to offset costs for babysitting and wages. Dr. Kelly noted that compensation can have tax implications. Compensation of more than \$600 is taxed; this rule is based on a decision from 1990, and the cap should be updated to account for inflation. Compensation can also carry a risk of losing Social Security.

The team aims to make its educational materials easy to update for use in other studies so that they can become the standard for the consent and assent process.

Dr. Kelly listed actions for increasing inclusion of people with DS of diverse backgrounds across the lifespan, including offsetting economic burden; educating research staff about tax liability; improving training and developing best practices for consent and assent; establishing a centralized mechanism for translation and a repository for researchers; and carefully considering costs to study teams, such as costs for in-home visits.

Sleep Health Disparities: Sleep and Sleep-Disordered Breathing in Persons Living with Down Syndrome

Marishka Brown, Ph.D., Director, NCSDR, NHLBI

Dr. Brown said that sleep is required for optimal well-being. It is necessary for neurodevelopment and cognitive processes and for maintaining a healthy weight. Research has shown that deeper stages of sleep play an important role in learning by promoting consolidation of memory and that they affect behavior. Lack of good sleep undermines development and could be a burden for life.

Research has found that sleep problems are significantly greater in children with DS than in the general population. Adults with DS have a high prevalence of behavioral sleep disturbances and have less rapid eye movement (REM) sleep, which is important for learning and memory. About one-third of adults with DS have sleep problems, and the most-reported sleep disorder in people with DS is sleep-disordered breathing, or sleep apnea.

Sleep-disordered breathing encompasses a spectrum of severity, from primary snoring to OSA. It involves a partial or complete blockage of the airway during sleep. It can be difficult to diagnose; early signs may be missed, and sleep studies can be challenging in people with DS. Treatments such as adenotonsillectomy and continuous positive airway pressure (CPAP) can help, but they are not cures.

Sleep disorders increase medical risk. In addition to neurocognitive challenges, they have been linked to increases in hypertension, obesity, diabetes, stroke, and dementia. Many studies of sleep in older populations living with DS are focused on dementia or Alzheimer's disease.

Sleep is a challenge in minoritized populations. Members of racial and ethnic minority groups and people who are socioeconomically disadvantaged have a higher burden of sleep deficiencies and

disorders. Compared with their White counterparts, members of racial minority groups have shorter sleep duration, poorer sleep quality, and more severe and underdiagnosed sleep disorders.

In 2018, NIH held a workshop to define sleep health disparities, resulting in the following definition: “a difference in one or more dimensions of sleep health (regularity, quality, alertness, timing, efficiency, and duration)—on a consistent basis—that adversely affects designated disadvantaged populations.” Those populations are racial and ethnic minorities, sexual and gender minorities, socioeconomically disadvantaged populations, and underserved rural populations.

Research on sleep disparities in people with DS is lacking. Between 2012 and 2018, funding for research on sleep in DS was limited. With support from INCLUDE, funding has grown exponentially. However, research has been concentrated in the Pennsylvania and New York areas. There are many opportunities for growth in the southern and southeastern United States, where health disparities are prevalent.

Dr. Brown displayed a graph of primary topics among funded applications, including pediatrics, aging, dementia, and longitudinal studies. Health disparities are not among the most-studied topics. However, sleep research has been evolving over the past 10 years, and NIH is seeing more applications on health disparities and on sleep in people with DS.

NIH has identified some gaps and areas on which the research community could increase its focus:

- Sleep health disparities in people living with DS across the lifespan
- Sleep-related topics other than OSA in adults with DS
- Sex and/or gender differences in sleep and sleep disorders in people with DS
- Circadian rhythms in DS

Dr. Brown said that not factoring diversity, equity, and inclusion into research means that data are incomplete. The result is poor science that could affect medicine and clinical care.

Q&A

Dr. Grzadzinski asked how to work with institutions to obtain approval of “pre-imburement” procedures. Rachel Walega, M.S., said that her team uses the institution’s travel software to arrange for travel and hotel costs, and the charge goes to the team’s grant account at CHOP. Dr. Tapia added that discounted meal vouchers can also be charged to grants at CHOP.

Dr. Bardhan asked Dr. Miller and Dr. Kelly whether they see variations in the assent process when they recruit members of underrepresented groups. For example, children and families who have not participated in much research might need more information. Dr. Miller said that she had faced more challenges from a broader research design perspective. For example, a study on sickle cell disease aimed to examine the dyad of a teen and a parent, but youth often attended with an older sibling, with another family member, or on their own. The dyad was not necessarily applicable to the group.

Dr. Bardhan said that, in recruiting families to DS-Connect, she has found that conversations differ depending on whether a family has participated in research before. The concept of an online registry can be scary, so she has learned to modify the information depending on the family she is talking with.

Melissa Parisi, M.D., Ph.D., asked Ms. Corbett for an update on inclusion of individuals with disabilities in research supported by NIH. Ms. Corbett said that as NIH implements the Inclusion Across the Lifespan policy, it has found that exclusion of people with comorbidities is one reason for lower inclusion of older adults. People with disabilities are often excluded because of narrow inclusion and exclusion criteria. The pandemic had a devastating effect on people with disabilities, especially those with intellectual

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 group may work best for everyone, but that is not always the case. NIH is moving forward, but inclusion of people with disabilities in research is still not where it needs to be.

Ruth Brown, Ph.D., said that requirements to track and report inclusion of people with disabilities in NIH research would help. Dr. Parisi noted that, unfortunately, people with disabilities are not yet recognized as a population with health disparities; Dr. Bardhan said that a UNITE committee was looking into expanding the health disparity definition.

Nicole White, Ph.D., asked whether the *All of Us* Research Program includes people under age 18 and those with intellectual disabilities. Dr. Rosser said that the program is not yet enrolling children but hopes to start doing so by late 2023 or 2024. The program does not have an assent process in place, so it is not enrolling people who are unable to consent on their own, but she is pushing for a process to be added.

Beate Peter, Ph.D., asked Dr. Rosser for the source of the pie charts that she had displayed showing participant demographics in DS research. Dr. Rosser provided a link to the [NIH RCDC Inclusion Statistics Report](#).

Dr. Brown noted that Dr. Kelly had mentioned neurocognitive questionnaires and who could administer them, especially in languages other than English. She asked whether there is a dearth of people who can administer tests and whether more awareness of this issue and more training are needed. Dr. Kelly said that for neurodevelopmental work at CHOP, researchers typically work with a [Clinical and Translational Science Award \(CTSA\)](#)-funded core that includes neuropsychologists and developmental specialists who work with youth. However, the team did not feel comfortable administering tests to adults. A colleague from Johns Hopkins trained a team member to administer tests to adults, but the team member is not fluent in Spanish and could not administer the tests to Spanish speakers. That was not only because the questionnaires were not available in Spanish but also because the tests involve other interactions, such as asking participants to smell something. Dr. Kelly said that she believes there is a need to extend testing ability to other languages to better serve people with DS, as well as people with various forms of dementia or other neurocognitive issues.

Stephanie Santoro, M.D., said that instruments must be developed in diverse populations to ensure that the language used is appropriate. Dr. White added that instruments often do not apply to people who are deaf or hard of hearing.

Dr. Parisi asked Dr. Miller and Dr. Kelly whether there is a concern that a child with DS might decline to participate during the assent process discussion before they can be fully engaged in the process. She asked what happens if a family planned to have their child participate but the child says no. Dr. Miller said that that would present an opportunity to pause and explore the child's concerns and reasons for saying no. Youth may need more time for decision making than a research team has available, which points to the need for assent to be a process over time. There could be an opportunity to re-engage later to see if anything has changed. However, ultimately, even if assent is waived, researchers must respect dissent.

Dr. Kelly said that the team first has a discussion with the caregiver, who can alert the researchers to potential challenges. If a person with DS is not ready to participate, the team tells them that they are welcome to come back later. Caregivers can provide feedback on what the team could have done better, such as making different word choices. Some study participants had more anxiety after viewing a video, which is one reason the team is conducting a study to obtain feedback on videos. Also, parents

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-lead; all the Day 1 participants, session chairs, and moderators; and the Spanish interpreters for the workshop.

DAY 2: September 21, 2022

Dr. Bardhan welcomed attendees to Day 2 and introduced the agenda, which featured various programs that had achieved success diversifying research teams and participants. The session was [recorded](#). Materials from the meeting, including summaries of the May 2022 listening sessions, the videocast recording, and a summary of this meeting, would be hosted on the [INCLUDE website](#).

Remarks from the INCLUDE Team: Engaging the Community

Nathaniel Stinson, Jr., Ph.D., M.D., M.P.H., Director, Community Health and Population Science, NIMHD

Dr. Stinson gave a brief overview of the previous day's presentations, highlighting the theme of valuing the wisdom of communities. Research is unlikely to be successful otherwise.

Session 4: Ensuring Representation in the DS Scientific Workforce— Investigators and Trainees

Session Chair and Moderator: *Huiqing Li, Ph.D., Program Director, NHLBI*

Dr. Li introduced the [Session 4](#) speakers.

INCLUDE Initiatives to Promote Diversity

Sujata Bardhan, M.S., Ph.D., Program Director, NICHD, NIH

Dr. Bardhan gave an overview of what NIH, and the INCLUDE program specifically, has done to increase diversity in research teams. While stark disparities in the numbers of R01 applicants since 2010 among various racial and ethnic groups demonstrate that NIH has a lot of catching up to do, steady increases in the number of self-identified Black and Hispanic applicants in that time also show progress.

Dr. Bardhan noted that INCLUDE was established by a congressional directive that calls for an increase in the number of investigators and trainees studying DS. The 2022 Down Syndrome Research Plan, expected soon after the meeting, also underscores the importance of expanding the pipeline of new and early-stage investigators (ESIs). In the past 4 years, INCLUDE has supported 30 trainees, including ESIs. Notably, INCLUDE has introduced investigators who are experts in other topics to the DS research community. In 2020, INCLUDE launched a data coordinating center (DCC), which has established a platform to integrate and share data from various DS studies. Under Dr. Espinosa's leadership, the DCC conducted listening sessions, speaking with representatives of 13 organizations in the Down Syndrome Consortium and more than 36 community members and clinical professionals.

Dr. Bardhan introduced the following DEIA activities supported by INCLUDE:

- Organizing a breakout session on outreach and participant engagement at a 2019 virtual DS cohort planning workshop
- Submitting an INCLUDE Working Group consensus statement on outreach and engagement in DS research (in press)
- Hosting Data Science for Diverse Scholars in Down Syndrome Research, a summer course for graduate students and postdoctoral fellows from underrepresented groups
- Hosting listening sessions with families and researchers on the topic of inclusive research participation

These activities have informed activities supported by INCLUDE. In addition, new funding opportunity announcements (FOAs) issued by INCLUDE now follow NIH principles encouraging diversity among investigators and participants. Recently issued FOAs also aim to encourage diversity by prioritizing digital technologies and mobile health, broadening the geographic distribution of funding by increasing the competitiveness of Institutional Development Award (IDeA) states, and encouraging community-based participatory research. INCLUDE also funds the work of two researchers through NIH COSWD-DEIA Mentorship Administrative Supplements and supports NIH Science Education Partnership Awards to

encourage an interest in science among students from pre-K to high school and ultimately increase the diversity of trainees.

Dr. Bardhan also highlighted a series of events that NIH staff have attended to engage new research participants.

In the chat, Sujay Ghosh, Ph.D., suggested holding two additional workshops: one for parents and caregivers and one to encourage students to work on DS research in their future careers. Dr. Ghosh also asked that INCLUDE provide funding to India and other low-income countries for workshops and extension activities. Dr. Bardhan said that all new INCLUDE FOAs are open to foreign applicants, and she invited Dr. Ghosh to reach out for further discussions.

Chief Officer for Scientific Workforce Diversity (COSWD) Presentation

Christy Ley, Ph.D., Social Science Analyst, COSWD, NIH

Dr. Ley noted that DS research demands an interdisciplinary scientific approach, in addition to other forms of diversity among researchers and participants. She introduced evidence showing how diverse research groups produce better science. Science requires diverse perspectives to tackle complex problems from a variety of angles, and diversity increases innovation and the quality of science.

Many people believe heterogeneous groups have more tension due to disagreement among their members. While that is true in some cases, overall, research across a variety of contexts shows that heterogeneity helps build diverse perspectives, especially when group interactions are well designed. The group's members tend to prepare more and put forward fact-based arguments. Anticipating that others may not agree seems to lead individuals to be more rigorous. However, scientific publications data and data about the distribution of NIH grantees by race indicate that barriers remain. Underrepresented groups tend to contribute at a higher rate, but their contributions tend to be undervalued. Representation of women and other underrepresented groups among R01 grantees decreases across the career trajectory, ultimately affecting scientific advancement. The biomedical research community is not using the full talent pool.

To address disparities in the scientific workforce, the COSWD issued its new strategic plan, which was developed following an evidence-based process, in spring 2022. The plan defines three goals: building the evidence, disseminating the evidence, and acting on the evidence. Achieving these goals requires understanding what is effective and how to scale up solutions. The COSWD's cross-cutting strategies for pursuing its goals are collaboration, accountability, and evaluation.

The COSWD's activities include establishing metrics, conducting assessments in collaboration with other offices, and advising Institutes and Centers (ICs) and others in the biomedical research community on their programs' effectiveness. COSWD has piloted the use of diversity catalysts, individuals in each IC who disseminate DEIA best practices, and the 21st Century Scholars program, which aims to build mentoring support for extramural researchers.

The COSWD uses various channels to disseminate the evidence, including hosting talks on the latest scientific workforce diversity research in its Scientific Workforce Diversity Seminar Series, publishing a biweekly blog and a quarterly newsletter, and publishing in peer-reviewed journals.

The COSWD also acts on the evidence by training and deploying recruitment strategists across NIH who identify diverse candidates. This program, which has existed for 5 years, was recently expanded. The COSWD also supports DEIA mentorship supplements and sponsors a prize competition for institutional excellence in DEIA. These opportunities can support DEIA in the DS research community.

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 data to apply for their own support. These programs can be used strategically to increase diversity in a lab.

One key to growing a diverse scientific workforce is stopping leaks at career transition points. The F99/K00 award that NINDS offers through the [Brain Research Through Advancing Innovative Neurotechnologies® \(BRAIN\) Initiative](#) aims to empower trainees to see themselves as researchers by giving them support during the last part of their graduate program to find the next place to land. Another transitional award started by the National Institute of General Medical Sciences (NIGMS) that similarly empowers people to look ahead is the K99/R00 award, which provides support during the transition from postdoc to a tenure-track position.

When it comes to faculty, there is a troubling drop-off at the stage when investigators are applying for their first R01. One strategy to address this is the diversity K01, a mentored award for junior faculty that aims to provide a bridge to the R01.

Since the [Ginther paper](#) highlighting racial disparities in R01 funding was published, NIH has launched several programs, including the diversity R01 targeting new and at-risk investigators ([PAR-22-181](#)) who may struggle to stay in the research enterprise and the [NIH Faculty Institutional Recruitment for Sustainable Transformation \(FIRST\)](#) program.

Dr. Jones-London emphasized that efforts to improve diversity are not only about money but also depend on connecting people in networks and building a sense of community. This gives new

investigators a sense of belonging in their field. NINDS funds mentoring networks and brings T22 and R25 institutions together to talk about mentorship.

NINDS also offers resources, including Open Stage, to demystify the process of applying for NIH funding. Investigators can learn more by joining the NINDS listserv, listening to its podcast, or following NINDS on social media.

In the chat, Ms. Fernandez asked whether NIH partners with [Leadership Education in Neurodevelopmental and Related Disabilities \(LEND\)](#) programs around the country, and Dr. Bardhan said yes.

Dr. Ghosh asked whether people in LMICs who work on DS can join the INCLUDE community for participating in outreach programs. Dr. Bardhan said that T21RS is part of the Down Syndrome Consortium, and as part of T21RS in India, Dr. Ghosh can partner for outreach events. Because there are many virtual options now, participation in webinars and workshops will be much easier for those in LMICs. Dr. Bardhan also noted that NIH's Fogarty International Center has two programs for LMICs: [Global Brain and Nervous System Disorders Research Across the Lifespan](#) and [Mobile Health: Technology and Outcomes in Low and Middle Income Countries](#).

Building a Diverse and Inclusive Research Team

Jianhua Wu, Ph.D., Associate Professor, Georgia State University

Dr. Wu spoke on the topic of building a diverse and inclusive research team, reflecting on his experience as an award-winning mentor. In 2021, he received the Jean Landa Pytel award from the American Society of Biomechanics based on his students' nomination. It recognizes the long-term impact of mentoring on the careers of individual scientists. His remarks focused in particular on his experiences mentoring students from underrepresented groups.

Dr. Wu, who grew up in China, introduced his work and the university where he works. Georgia State University, with students from more than 167 nations and territories, is one of the most diverse in the United States. The university graduates more African American students each year than any other U.S. college. Specializing in biomechanics and pediatric rehabilitation, Dr. Wu studies how children with DS and cerebral palsy develop motor, language, cognition, and social skills. He described how doctoral, masters, and undergraduate students in his lab have come from diverse racial, ethnic, and educational backgrounds; half are women scientists or clinicians.

Dr. Wu explained his mentoring philosophy, through which he aims to promote mentees' research independence while giving them the structure and support of a team. Understanding the needs of patient populations is a key element because students can use this knowledge to improve patients' quality of life. He offers opportunities to supervise other graduate students and mentor undergraduates and encourages students to lead projects. Weekly group and individual meetings help create a fair and respectful environment. The role of the mentor is to provide support, so any topic is acceptable at these meetings, including discussing problems such as financial concerns. The training process can help students dream about what they can accomplish.

Dr. Wu said he also developed an international outreach program through which about 50 undergraduate students each year travel to China for 2 weeks.

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-speaking graduate student helped the lab reach out to this community and encourage children to participate in their study. Providing transportation and going to participants' homes to collect data helped create a welcoming environment and increase participation from these communities. Dr. Bardhan underscored how these examples illustrate what meeting attendees and INCLUDE listening session participants have shared about overcoming barriers to diverse participation. Dr. Wu's experience highlights the benefits of active mentorship and of diverse research teams.

Session 5: Enhancing Participant Diversity in Down Syndrome Clinical Research

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

Dr. Bardhan explained that the next session would feature different programs' experiences on enhancing recruitment diversity.

The Alzheimer's Disease/Down Syndrome Outreach, Recruitment, and Engagement (ADDORE) Core

Carey Gleason, Ph.D., M.S., Associate Professor, Alzheimer's Disease Research Center, University of Wisconsin–Madison

Sigan L. Hartley, Ph.D., Associate Professor, Waisman Center, University of Wisconsin–Madison

Nicole Shiree Whitmoyer, Project Manager, University of Wisconsin–Madison

Maria Nogueron, Ph.D., and Danny Chafetz, Community Outreach Specialists

Dr. Gleason, part of the ADDORE team with the Alzheimer's Biomarkers Consortium–Down Syndrome (ABC-DS) project, said that in order to succeed at inclusive research, researchers must redefine recruitment as engagement. Engagement is key at every stage and involves building relationships and investing time and energy. The research world and academia have a culture that is not always compatible with community work, so this may look different.

Research culture is described in a [paper](#) that describes the hidden hegemony of doing things in academia. One example is the research timeline. Community work may advance on a different timeline, which research can accommodate once researchers recognize the limits of their lens or culture.

Dr. Gleason underscored the importance of language, explaining preferences for “underrepresented,” “minoritized,” or “racialized” groups rather than “minorities,” a description that is imposed on groups by a dominant culture. “Underrepresented” can encompass racial/ethnic groups or those defined by SES, sexual orientation, gender identity, or geography (i.e., rural).

Even for drugs submitted for approval today, researchers are not accomplishing diverse or inclusive research. Yet, African Americans are overrepresented in research that does not require informed consent. The issue is not that underrepresented groups are not available to participate; rather, researchers often have not done the work to engage these individuals and make sure they are included.

One reason for under-enrollment by underrepresented groups in studies requiring consent is mistrust. The Tuskegee study is a prominent historical example, but researchers' bad behavior is not a thing of the past. More recent misdeeds in research or oversight include a lead abatement study that invited a lawsuit from participants concerned that researchers did not notify them about dangerous lead levels in their homes and the use of genetic data from the Havasupai tribe beyond the terms of the study for which they were originally collected; both examples are from the 1990s. In addition to concerns such as time scarcity and transportation, lack of cultural safety can be a major barrier to accessing clinical research. It is important to acknowledge that mistrust and access are limitations and to adapt research culture in a way that moves participants' experiences, not researchers' needs, to the center.

Dr. Gleason highlighted the work of many teams doing community-engaged research on [Alzheimer's disease](#), [aging](#), and [registry enrollment](#).

Dr. Gleason introduced the three-phase model adopted by the ADDORE Core to engage community members:

- Phase 1: Give first.
- Phase 2: Build advocates.
- Phase 3: Reciprocity

Examples of this model in action include the Wisconsin Alzheimer's Disease Research Center's outreach to African American and Oneida Nation communities. The center has found success in building relationships by offering exercise classes, memory screenings, and education events to the community and by establishing paid advisory boards that advocate on behalf of their community members. The center also invites trainees from the communities to participate.

Dr. Hartley gave an overview of the ADDORE Core, which is led by Annie Cohen, Ph.D., associate professor of psychiatry at the University of Pittsburgh School of Medicine. Having a core devoted to engaging with the community was a priority of ABC-DS from the outset. The ADDORE Core has three major aims:

- **Generate educational materials.** One goal is to provide education about the study, why it is important, and what it involves. The materials demystify medical terms and procedures to make research approachable.
- **Disseminate the educational materials to underserved communities, with a focus on those close to research sites.** Each of the eight ABC-DS sites was asked to consider what nearby communities they could partner with as part of their research team.
- **Increase recruitment of individuals with DS into the ABC-DS cohort.** The study aims not only to enroll participants but to make participating a good experience so that participants want to stay and build a valuable partnership over time.

To accomplish these goals, each site had funding to cover the cost of a full-time employee to engage with other cores and the core leader on these activities. In addition, the study established a Community Research Advisory Board with self-advocates and parents that helps develop research questions and outreach materials and guide the research. For example, the board helps run community-led trainings for the broader ADDORE team. Board members also gave feedback on the study flyer, pointing out that an initial version did not explain why families should participate. In addition, the board partners with the National Down Syndrome Society. Each local site identifies local partners, such as the Special Olympics in Wisconsin. The research team has also held listening sessions with the community and established working groups to work on social media, newsletters, bulletins, and information on research methods (i.e., how to provide education on brain donation, what the benefit is) and underrepresented groups.

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 participants’ circumstances. Most important to her role as a community liaison is her experience as a mother of someone with DS and her network in the community. These things empower her to talk to researchers and legislators.

Dr. Nogueron described the team’s work as shifting the research paradigm to one based on building relationships and presenting information about the study in plain language. The more appropriate model may be creating a community of practice. The outreach team moves between researchers and participants and participates in creating the final project, with its aim of improving the lives of people with disabilities. It is important to be patient and be a consistent presence in order to allow relationships and participation to build over time. Being able to bring an understanding of the participant experience to the study team means her daughter is excited to go for study visits.

Ms. Whitmoyer, a project manager who works to recruit study participants, described lessons she had learned as a liaison between community partners and the academic community. The lessons include how to set good boundaries while remaining flexible. It is important to be accessible to community members outside of regular business hours due to their working hours and to be available for weekend recruiting events. At the same time, it is important to learn not to let all boundaries go. Community partners can learn to accommodate communication preferences or limits on when study staff will respond. Another lesson is the importance of staying attuned to language, including what terminology is best in a variety of settings. Finally, study staff need to stay attuned to the needs of both researchers and community partners. Researchers place a high priority on following a consistent process, but doing so rigidly can exclude potential participants. Being willing to travel to participants requires flexibility but accommodates more people in the study. Thinking outside the box can maximize inclusion.

Mr. Chafetz, the team’s self-advocate, read an original poem about participation in ABC-DS and other studies.

Verse 1

When you have this disease the memory starts to fade away so can you promise me that any families will never be forgotten? It’s like a secret file if you afraid of losing yourself to this disease at least put your memories in the file. ABC-DS are saving lives because it matters.

Verse 2

I love being in research we find the root of communities we love. I wanna thank you for everyone in ABC-DS and a special one to Nicole and Maria. Thank you so much. From my

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

Devan Ray, Communication Sciences and Disorders, Community Engagement Studio Team, RIC

Community Advisory Board; Diversity, Equity, and Inclusion Work Group, RIC, Vanderbilt University Medical Center

Janice Nodvin, Executive Director/Clinic Director, Adult Disability Medical Home, Inc., Atlanta, Georgia

Ms. Israel, Ms. Bell, and Ms. Ray introduced the approach followed by Vanderbilt University's RIC and encouraged others to use its resources to enhance diversity in research. The RIC offers advice to overcome recruitment and retention challenges using evidence-based strategies and materials. It is part of the [Trial Innovation Network](#), a collaboration with the CTSA program.

Ms. Bell underscored the importance of acknowledging that BIPOC (Black, Indigenous, and People of Color) individuals living with DS are socially invisible. Inclusive research can help explain the lived impact of ostracizing language. BIPOC individuals with DS face more stigma and higher rates of bias and discrimination. They are more often perceived as aggressive or violent, compared with White individuals with DS. In addition, racial disparities in life expectancy among people with DS are huge.

Ms. Ray shared quotations from RIC sessions with parents reflecting on barriers they see to participating in research, including mistrust, safety concerns, logistical challenges, and implicit bias.

The team shared recommendations for enhancing diversity and resources to support these activities.

- The [Faster Together](#) course offers training on creating an anti-racist research environment with eight modules on implicit bias, community engagement, and best practices for working with diverse participants.
- Best practices in recruitment, hiring, and promotion can build a diverse staff and create a research environment that is welcoming.
- Principles of study design include best practices for reducing the burden on participants.
- A [Community Engagement Studio \(CES\)](#) toolkit helps researchers organize sessions where they invite stakeholder feedback.
- CES case studies offer additional insights on implementation, study recruitment, and messaging. One common recommendation is to embrace social media as a low-burden way to engage partners and to inform, educate, and give back to the community.

Recommendations specifically for community engagement included being mindful of building benefits for participants into the process; participation should be mutually beneficial. The research team should think creatively about who its partners are and what partnership looks like. All communities have people who are active; it is important to embrace individuals who are active in their communities but also to consider how to create spaces and opportunities that people can grow into. It is important to ensure equity and practice shared decision making. The research team should ask participants what meaningful participation and outcomes look like to them. Finally, fair compensation for partners' time, advice, and guidance is essential.

The RIC has also made considerable effort developing recruitment and retention materials. Messaging on recruitment materials should be tailored and easy to understand. These materials supplement the consent form and provide explanations of what the study is about, why it is important, what participants will be expected to do, and the potential for risk and benefit to them. The language should be clear and transparent, and the design should be sensitive to diverse sensory needs. Producing such materials does not have to be expensive. The RIC developed a [toolkit](#) for creating materials that includes templates for recruitment flyers.

Ms. Nodvin described the services and approach followed by the health care facility she oversees, Adult Disability Medical Home, Inc. (ADMH). It provides services to teens and adults with developmental disabilities and engages them to participate in research. ADMH was founded to address disparities faced by individuals with developmental disabilities, especially during the transition from pediatric to adult health care. It is one of few clinics nationwide offering both primary and specialty care in one location.

ADMH aims to look at both the individual and the family. The health of the individual encompasses their physical, psychosocial, and behavioral health. More broadly, ADMH ensures that the family has access to insurance and examines the financial aspects of the transition from pediatric to adult services. The family is then ready to share experiences through research opportunities or support groups, recreation, and residential life activities.

The transition to adult services is fragmented and difficult to navigate. Often families have been with a pediatrician and school system for many years. Until recently, there were few clinical guidelines for the care of adults with DS; their publication has helped tremendously. ADMH assembles an interdisciplinary team to address the health of the individual within the framework of a patient-centered medical home. ADMH partners with certified community-based programs. First and foremost, the individual is the central member of the team. Given that individuals may have anxiety from previous medical experiences, establishing rapport and taking cues from the individual is paramount.

Ms. Nodvin shared some statistics demonstrating the limits of access to health care for adults with disabilities. One in three adults with a disability does not have a primary care provider. One in three has an unmet health care need because of cost. One in four has not had a routine checkup in the past year. These statistics show that making health care more accessible is an urgent need.

ADMH considers transitions an ideal time to offer research opportunities to families—for example, at the onset of puberty or at the age of majority. ADMH works with families to find studies that are a good match.

Discussion

Ms. Fernandez said that addressing key social determinants of health through trusted community groups would improve participation and trust.

Dr. Santoro asked for a reference for the statistic that one in three people do not have a usual health care provider. It is a call to action. Ms. Nodvin provided a [reference from the Centers for Disease Control and Prevention](#).

Dr. Bardhan asked how the RIC community engagement studio model would work for an intramural protocol. Ms. Israel recommended submitting a request through the Trial Innovation Network, which evaluates all requests. Ms. Ray added that the presenters would be happy to discuss details individually. Ms. Israel added that nearly 40 other sites across the country have hosted a studio session, so it is worth seeing if your institution has also held one. The RIC did a demonstration studio at the National Center for Advancing Translational Sciences (NCATS).

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.

Session 6: Lessons in Community Engagement

Session Chair and Moderator: *Nathaniel Stinson, Jr., Ph.D., M.D., M.P.H., Director, Community Health and Population Science, NIMHD*

Underserved Groups

Kate Dougherty, President, NDSC Board of Directors, Rural Communities

Brian Chicoine, M.D., Medical Director, Adult Down Syndrome Center (ADSC) of Advocate Medical Group

Elena Grigorenko, Ph.D., Department of Molecular and Human Genetics, Baylor College of Medicine, and

Phil Thuma, M.D., Director, Center for Global Health, Johns Hopkins University

Noemi Spinazzi, M.D., Pediatrician, Assistant Professor, Medical Director, UCSF Down Syndrome Clinic

Dr. Stinson welcomed everyone back for the final session and introduced the presenters.

Ms. Dougherty shared a video of her son, at age 6, shoeing a horse and said she never would have anticipated when she received his diagnosis of DS that he would be able to do that one day. It is important that people in rural areas understand what is possible, and that starts with exposure to people with a disability. Although engagement may not look the same in rural and urban areas, it is possible to enact true change in rural areas by creating communities and using positive pressure. In rural areas, there is a lack of exposure to individuals with disabilities and many other demographics. There is also a limited number of specialists. For example, in the tri-state area where Ms. Dougherty lives, there

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 of language in creating community engagement. “Support group” has a negative connotation due to people’s self-reliance and sense of independence. Rural residents would be more likely to join a DS Friends and Family Group. The choice of language speaks to a different mindset. Very often, a community will not have many people with DS, but those individuals and their families are never alone. It is not realistic to expect that others will have the same level of experience with disability, but creating community by bringing along family and friends will begin to break down barriers and end individuals’ self-imposed isolation.

Making things pretty and celebrating the joy of an extra chromosome makes things attractive to other families. Making things fun creates an opening to expose people who may not have experience with disability and show them what is possible. In rural areas, people lack exposure, experience, and perspective, and misconceptions abound; introducing them to someone with DS can change how they view possibility.

The biggest barriers in rural areas are segregation, isolation, lack of exposure, and lack of inclusion. Through community, people who previously had no exposure can become the strongest advocates for children with DS. The goal is to create a culture shift. One of the most powerful ways to help families is to share their stories and end their isolation. Exposure, education, experience, and empowerment become life-altering for individuals. The possibilities are endless for children and adults with DS.

Dr. Chicoine introduced the ADSC, which has served 6,000 adults since opening in Park Ridge, Illinois, in 1992. The center provides care for people with DS ages 12 and older, including primary and specialty medical care, mental health care, hospital and residential facilities, occupational therapy, and the services of a licensed clinical social worker and a patient advocate. The facility is looking to improve research participation of all people with DS, but the priority is on physical and mental health care and health promotion. Research follows after.

Dr. Chicoine shared the estimate that less than 5% of adults with DS have access to a DS specialty clinic, and findings from a study by Dr. Santoro indicated that only a minority of specialty clinics can serve all of a patient’s health needs, which represents a stark shortcoming.

The ADSC participates in the LuMind-IDSC LIFE-DSR (Longitudinal Investigation for the Enhancement of Down Syndrome Research) study; 3% of the 2,000 patients at the center participate in the study, but this is 25% of study participants. The limited racial and ethnic diversity of the study participants is reflective of the ADSC’s patient population as a whole. To increase the diversity of the patient population, the first step is to acknowledge and address health equity issues for people with DS and across racial/ethnic groups. Efforts to enhance the diversity of the patient population and of study enrollment include reaching out to nearby urban centers, partnering with religious organizations and places of worship, and conducting Special Olympics physicals. Telehealth has been an important avenue for expanding the facility’s services. Among the study materials it provides to potential participants, the ADSC has a video to help educate participants with DS.

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.

Dr. Grigorenko introduced the team’s eight “musts” for conducting research:

1. Debrief the leadership and secure access to the participants.
2. Engage the community by recruiting local project staff (who are great advertisement agents!).
3. Accommodate the participants as much as possible.
4. Use assessments that are culturally sensitive, well calibrated, and child-friendly.
5. Appraise and respect local conditions.
6. Explain every aspect of the research, no matter how long it takes.
7. Be grateful to and reward participants.
8. Exit in such a way that a return is possible.

Dr. Spinazzi revisited the issue of trust. Clinicians and researchers must not only gain the trust of their patients and potential research participants but also actively undo the mistrust that exists. In order to build a diverse research community, investigators must acknowledge, own, and actively undo legacies of abuse and mistreatment experienced by people with disabilities. There is a traumatic history of people from this community being forced into research and sterilized without their consent.

In addition, ongoing structural racism shapes and sustains mistrust. When researchers take on the task of building trust, they must acknowledge the layers of disparity in areas where poverty, limited education, disability, and lower life expectancy may be concentrated in the same neighborhoods. Residents of these areas may be coping with issues around childcare, housing, access to fair employment, community violence, food deserts, inadequate school support, and lack of respite and are unlikely to count research among their priorities.

Dr. Spinazzi introduced the clinic where she works, which is the only pediatric-only federally qualified health center (FQHC) in the nation. It serves 10,000 unique patients, of whom 78% are covered by Medicaid. A total of 35% identify as African American, 29% as Hispanic or Latinx, and 24% as other/mixed. The center recruits heavily for race-concordant recruiters and research staff. All consent forms are reviewed by a diverse team to check for language that can trigger trauma. All materials are translated into Spanish, and video interpreters are on call to support consent in additional languages. Through strong partnerships with community organizations and with lectures and community educational sessions, researchers share results with the community.

As part of the facility’s effort to meet families where they are, clinicians strive to recruit research participants during clinic visits. Research coordinators communicate with families according to their preferences for texting versus calling or preferred time of day. The center offers flexible scheduling and

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

Anna Esbensen, Ph.D., asked how the concept of fun can be used to increase research participation in rural settings. Ms. Dougherty said that doctors and therapists have joined forces with their local community. Celebrating joy makes things attractive to others in the community. Hosting connection events can help introduce potential participants to research opportunities. Having clinicians and other members of the research team travel to rural communities is part of the process of building trust. Enrollment will be difficult without getting a representative or ambassador out to the community. Dr. Spinazzi added that researchers cannot understand a community's barriers to participation without going and seeing. It is important for researchers to go, not just to show their faces but to understand the community. Only by listening to people and making mistakes will researchers learn how to communicate effectively. Offering opportunities to meet and talk with families is an element that should be built into the budget for research studies. Dr. Bardhan added that there are similarities between LMICs and rural U.S. communities. Lessons about cultural barriers and partnering with trusted community members learned from investing in research in LMICs can be used elsewhere. Dr. Grigorenko's principle, exit in such a way that a return is possible, highlights that communication is a two-way street. Researchers need to learn from the community what the community's needs are.

In the chat, an attendee asked for input on how to get more medical professionals informed and passionate about caring for patients with DS. Dr. Spinazzi said that one thing everyone can do is to advocate for health care reform, so clinicians are not faced with the impossible expectation of providing care for such patients in a 15-minute appointment. Providers can join the [Down Syndrome Medical Interest Group \(DSMIG\)](#). Ms. Dougherty concurred; every physician her group has sent has come back with a wealth of information. Dr. Spinazzi added that she gained a lot of knowledge from NDSC symposia and seminars. She noted that the more people with DS you know, the harder it is to not get passionate about taking care of them.

Dr. Chicoine encouraged clinicians to reach out to DSMIG, which has a presentation series with a focus on local providers. He recalled his own experience as a doctor in a small town with little information about caring for DS patients and emphasized the value of information. Dr. Spinazzi added that there is no incentive for clinicians to take care of medically complex patients. Those who do so either have a personal connection or internal passion. To the advocacy and government organizations present, she asked, "How do we incentivize taking care of people with disabilities well in this country?" There needs to be clinical program funding so that programs can become models for care and pipelines for the clinical workforce.

Dr. Li commended the speakers and their dedication to the DS community. More support for clinical programs is needed. She asked attendees to give examples of how NIH can facilitate engagement of more diverse populations. Dr. Spinazzi suggested establishing a clinical pipeline award, modeled after

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 free membership or otherwise support young investigators so they can learn more about complex care. Grants could fund work with a collaboration outcome or development of a research participant pipeline as the goal rather than a research outcome. Dr. Chicoine argued that research and care have to be intertwined. Clinicians have to provide high-quality care, and then that care must be studied to establish an even higher-quality level of care. Patients will not line up for research if they do not get care. Dr. Spinazzi highlighted a conflict inherent in current models of funding, based on her experience having to turn down an invitation to participate in a study from the Pediatric Trials Network, which offered funding based on the number of patients enrolled. However, at her institution, staff have to apply for funding in advance to get a research coordinator. There was no way to make it work.

An attendee asked in the chat for information about providers in their area. The [NDSC](#) and [National Down Syndrome Society \(NDSS\)](#) websites have lists of providers knowledgeable about DS nationwide.

Next Steps, Future Activities, and Closing Remarks

Melissa A. Parisi, M.D., Ph.D., Branch Chief, Intellectual and Developmental Disabilities Branch, NICHD

Dr. Parisi summarized the meeting's highlights. Dr. Bardhan set the stage with an overview of INCLUDE and how funding can spur research in three areas: basic science, cohort development, and clinical trials. The program is in a good position to address some health disparities, especially the stark racial disparities in life expectancy among individuals with DS. Presentations on feedback from family members and researchers in listening sessions highlighted barriers to participation in research that both groups shared, including trust, time, education, language, and support for families.

Rounding out Session 1, Dr. Ilias introduced the CEAL consultative resource and what it can offer for DS research. Importantly, the consultation process for community engagement can be done in 30 to 60 days. These are valuable resources for researchers, especially in light of COVID-19–related needs, such as increasing diversity.

The panel discussion in Session 2 offered valuable perspectives from self-advocates, families, and researchers. Many participants consider research a positive experience, but lack of access is a problem. Communication is an important theme. For example, although parents often become the point of contact, it is important for researchers and staff to interact directly with participants. Researchers need to ask families what the most important factors are, instead of assuming that they already know. Making language accessible and offering translated materials are other important aspects. It is important for researchers to carefully consider logistics, including compensation, and figure out how to accommodate families' busy schedules. One underappreciated aspect of compensation is the possibility that participants will run into a compensation cap or have to handle tax or liability issues. Because mothers are often the ones providing care and bringing a family member with DS to research visits, researchers need to take particular care of mothers' preferences, needs, and concerns. Panelists also emphasized the importance of having people on the research team who look like the people the study is trying to recruit. Support for clinical programs is an essential element of funding for research. Finally, panelists emphasized the importance of translating research findings back to the community.

In the session about addressing health disparities in scientific research, Ms. Corbett discussed the importance of including underrepresented groups and people across the lifespan in NIH research. Dr. Rosser shared information from the *All of Us* Research Program illustrating differences in medical

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.

Dr. Gleason's presentation on her research team's experiences building diversity included a reminder about the importance of language and highlighted an important lesson illustrating that there is a potential for misunderstanding when values are not aligned. The participatory research framework she shared had three phases: give first, build advocates, and reciprocity. Dr. Hartley introduced the work of the ADDORE Core, which conducts community outreach. Central to this effort is the three-person outreach team of Dr. Nogueron, Mr. Chafetz, and Ms. Whitmoyer, each of whom shared their experiences and demonstrated the project's commitment to their community.

The three RIC presenters, Ms. Israel, Ms. Bell, and Ms. Ray, helped identify tools and resources that can help increase diversity in DS studies, including information about consultative studios and a toolkit for designing study materials that go beyond the standard consent forms. Ms. Nodvin talked about the adult disability medical health care model practiced by the center she runs and focused on the difficulties that families face with the transition from pediatric to adult health care.

In the session on community engagement of underserved groups, Ms. Dougherty explained some distinctive aspects of engagement in rural communities and illustrated the power of exposure. Dr. Chicoine described a model for outreach based on the experience of his own clinical center. A key takeaway from Dr. Grigorenko's presentation on epidemiological work in rural Africa was the note to leave in a way that invites being welcomed back. Dr. Spinazzi's introduction to the children at her clinic underscored the importance of embedding research in the clinical setting and of being mindful that research is not a top priority for patients or their families.

In sum, people with DS are living longer due to advances in medical care, but significant health disparities remain. New INCLUDE funding provides resources to reduce or eliminate those disparities.

The virtual INCLUDE investigators workshop will be held on October 12.

Glossary

ABC-DS	Alzheimer’s Biomarkers Consortium–Down Syndrome
ABRCMS	Annual Biomedical Research Conference for Minoritized Scientists
ACTIV	Accelerating COVID-19 Therapeutic Interventions and Vaccines
AD	Alzheimer’s disease
ADDORE	Alzheimer’s Disease/Down Syndrome Outreach, Recruitment, and Engagement
BIPOC	Black, Indigenous, and People of Color
BMI	body mass index
BRAIN	Brain Research Through Advancing Innovative Neurotechnologies®
CEACR	Community Engagement Alliance Consultative Resource
CEAL	Community Engagement Alliance Against COVID-19 Disparities
CHOP	Children’s Hospital of Philadelphia
COSWD	Chief Officer for Scientific Workforce Diversity
CPAP	continuous positive airway pressure
CTSA	Clinical and Translational Science Award
DCC	data coordinating center
DEIA	diversity, equity, inclusion, and accessibility
DMI	decision-making involvement
DS	Down syndrome
DS-Connect	DS-Connect®: The Down Syndrome Registry
DSi	Down Syndrome International
DSMIG	Down Syndrome Medical Interest Group
EEG	electroencephalography
ESIs	early-stage investigators
FOAs	funding opportunity announcements
FQHC	federally qualified health center
GLOBAL	Global Down Syndrome Foundation
ICs	Institutes and Centers
IDeA	Institutional Development Award

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	<i>Eunice Kennedy Shriver</i> 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
OSA	obstructive sleep apnea
PI	principal investigator
RECOVER	Researching COVID to Enhance Recovery
REM	rapid eye movement
RIC	Recruitment Innovation Center
SACNAS	Society for Advancement of Chicanos/Hispanics and Native Americans in Science
SDM	shared decision making
SES	socioeconomic status
T21RS	Trisomy 21 Research Society
UBR	underrepresented in biomedical research