

Building a Diverse Community for Down Syndrome Research

September 20–21, 2022
NIH-Sponsored Virtual Workshop¹

EXECUTIVE SUMMARY

On September 20–21, 2022, the National Institutes of Health (NIH) sponsored a virtual workshop of [the INCLUDE Project](#) (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndrome) titled “Building a Diverse Community for Down Syndrome Research.” Representatives from NIH, clinical researchers, clinicians, self-advocates, advocates and family members, and other members of the Down syndrome (DS) community participated in the two-day meeting and discussed the current state of diversity in DS research, participants’ and researchers’ experiences, work to address health disparities, and efforts to increase research participation among underrepresented groups as well as representation in the research workforce. The two-day meeting was co-hosted by Sujata Bardhan, M.S., Ph.D., program director at the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), and Linda Garcia, communications and outreach coordinator for the INCLUDE Project and DS-Connect[®]. Nearly 200 people registered to attend the workshop.

Day 1 opened with an overview of diversity in DS research, including summaries of listening sessions with families and researchers; continued with a panel discussion with self-advocates, family members, and researchers; and closed with presentations on addressing health disparities through research. In three sessions on Day 2, participants covered various aspects of increasing diversity in the workforce of scientists studying DS, research team outreach efforts that had been successful in enhancing diversity among research participants, and approaches to care and community engagement that supported research participation.

Overview of Diversity in DS Research

Two major goals of the INCLUDE Project are to expand the inclusion of individuals with DS from diverse backgrounds in research and to expand the pipeline of trainees and new investigators from diverse backgrounds who work on DS. Progress toward these goals could help improve understanding of health disparities and, ultimately, help advance health equity in underserved populations. The workshop was part of the project’s efforts to engage the DS community, listen, learn, and take action toward these goals.

Investment in medical research has led to a dramatic increase in the life expectancy of people with DS. However, there are disparities in life expectancy, indicating a need to better understand disparities in overall health.

NIH investment in INCLUDE has enabled hundreds of research projects, including several clinical trials. However, people who are of non-Hispanic ethnicity, White, and male are overrepresented in the trials. One way to improve participation among people from groups underrepresented in biomedical research (UBR) is to increase enrollment in DS-Connect, a DS registry that can help INCLUDE researchers with recruitment.

In May, NIH held listening sessions with parents of individuals with DS and with DS researchers. Participants discussed strategies that have improved participation in research, such as speaking the same languages as participants, prioritizing safety, providing compensation, sharing research progress, including communities in research design, making participation easier, and engaging with communities in other contexts. Families and researchers listed many of the same barriers to participation, including lack of trust, language, and logistics.

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

NIH's Community Engagement Alliance Consultative Resource (CEACR) provides consultations and resources to help researchers improve their community engagement and inclusive participation efforts. CEACR leverages assets gathered through the Community Engagement Alliance (CEAL) Against COVID-19 Disparities.

Stakeholders' Perspectives: Panel Discussion

Family members, self-advocates, and researchers discussed their experiences with research. Families had positive experiences when researchers made participation as convenient as possible, spoke directly with participants, and understood how to work with people who have DS. Clinical trials can have real benefits for participants' quality of life, but people in underserved communities may not hear about them or be able to participate. Families reiterated the barriers identified in the listening sessions, including time and costs for travel and housing. They also cited a need for DS-specific research to inform educational policies, such as benchmarks for children's test scores. Researchers can build trust by ensuring research teams are diverse, communicating clearly about research benefits and outcomes, visiting rural communities, and helping families meet basic needs.

Researchers emphasized the importance of working closely with community partners to bring a more inclusive perspective to research and connect with potential participants. Supporting clinical programs that serve patients is critical for building trust and facilitating research participation. Flexibility and considering participants' needs are key. Researchers can build trust through ongoing community involvement, advocating for successful interventions to be covered by insurance, sharing their own stories, and being aware of past research abuses.

Addressing Racial and Ethnic Health Disparities in Scientific Research

Four presentations followed the panel discussion, focusing on work to improve inclusion in DS research. Dawn Corbett, M.P.H., from the NIH Office of Extramural Research, described NIH's policies requiring that research include women, members of racial and ethnic minority groups, and people of all ages. NIH also has requirements about reporting results by sex or gender, race, and ethnicity. A [2020 meeting](#) and [2022 report](#) provided strategies to address challenges in representation, such as limiting inclusion and exclusion criteria, reviewing participant compensation practices, and ensuring that community engagement efforts are adequately funded.

Tracie Rosser, Ph.D., an assistant professor at the Emory University School of Medicine, described DS research at Emory, including the DS360 project. She noted that diversity among research participants is important for understanding health disparities. Emory research has found differences in risk for co-occurring conditions by race, ethnicity, sex, and other factors, but more research is needed. Dr. Rosser also described NIH's *All of Us* Research Program, which aims to enroll one million or more people and build a large, rich biomedical dataset. The program reaches and engages people from UBR communities by returning results to participants, informing participants about data protections and risks, and conducting custom campaigns in specific communities.

Andrea Kelly, M.D., M.S.C.E., and Victoria Miller, Ph.D., from Children's Hospital of Philadelphia, presented their work on optimizing the informed consent process for people with DS. Dr. Kelly studies sleep in children with DS, and her research team provides videos and storyboards to explain study procedures. Work is ongoing to assess the tools' utility. Dr. Miller explained that the assent process gives potential participants the opportunity to choose whether to participate to the extent that they are able. Children and adolescents can be meaningfully involved in decision making without having the final say. Although more research is needed to optimize the decision-making process for people with intellectual disabilities, Dr. Miller believed that the concept of decision-making involvement, which focuses on the decision-making process and interactions, is appropriate.

Marishka Brown, Ph.D., director of the National Center on Sleep Disorders Research at the National Heart, Lung, and Blood Institute, discussed sleep and sleep-disordered breathing in people with DS. Sleep is necessary for optimal well-being, but people with DS are more likely to have sleep problems. Members of racial and ethnic minority groups and people who are socioeconomically disadvantaged also have a higher burden of sleep deficiencies and disorders. Research on sleep disparities in people with DS is lacking. There are opportunities for growth in the southern and southeastern United States and in specific topic areas.

Ensuring Representation in the DS Scientific Workforce—Investigators and Trainees

Day 2 opened with a focus on ensuring diversity in the DS scientific workforce. Dr. Bardhan summarized INCLUDE activities to support diversity, equity, inclusion, and accessibility (DEIA). These include organizing a session on outreach and participant engagement at a DS cohort planning workshop, submitting a statement on outreach and engagement in DS research, and hosting a summer course for students from underrepresented groups. INCLUDE funding opportunities aim to promote diversity by prioritizing digital technologies, broadening the geographic distribution of funding, and encouraging community-based participatory research.

Christy Ley, Ph.D., a social science analyst in the NIH Chief Officer for Scientific Workforce Diversity (COSWD) office, reviewed COSWD activities to address disparities in the scientific workforce. To build evidence, the COSWD establishes metrics, conducts assessments, and advises NIH Institutes and Centers (ICs) and others in the biomedical research community on programs' effectiveness. The COSWD has piloted the use of diversity catalysts, individuals in each IC who disseminate DEIA best practices, and a program to build mentoring support for extramural researchers. It also trains and deploys recruitment strategists across NIH to identify diverse candidates, supports DEIA mentorship supplements, and sponsors a prize competition for DEIA excellence.

Michelle Jones-London, Ph.D., chief of the Office of Programs to Enhance Neuroscience Workforce Diversity (OPEN) at the National Institute of Neurological Disorders and Stroke (NINDS), discussed efforts to expand workforce diversity. They include fellowships, supplements, other forms of individual support, and institutional programs. One key to growing a diverse scientific workforce is stopping leaks at career transition points. Efforts to improve diversity require building networks and a sense of community, to give new investigators a sense of belonging. Finally, NINDS offers resources to demystify the process of applying for NIH funding.

Jianhua Wu, Ph.D., an associate professor at Georgia State University, spoke about building a diverse and inclusive research team, based on his experience as an award-winning mentor. He studies how children with DS and cerebral palsy develop motor, language, cognition, and social skills. Through his mentoring philosophy, Dr. Wu aims to promote mentees' research independence while giving them the structure and support of a team. Understanding patient populations' needs is key, because students can use this knowledge to improve patients' quality of life. Dr. Wu offers opportunities to supervise and mentor other students, encourages students to lead projects, and holds weekly meetings to help create a fair and respectful environment. Dr. Wu also explained how diversity among his trainees helped increase engagement of underrepresented groups in research.

Enhancing Participant Diversity in DS Clinical Research

Representatives from the Alzheimer's Biomarkers Consortium—Down Syndrome (ABC-DS) project's Alzheimer's Disease/Down Syndrome Outreach, Recruitment, and Engagement (ADDORE) Core discussed their approach and lessons learned. To conduct inclusive research, researchers must redefine recruitment as engagement. Building trust must be at the center of interactions. The ADDORE Core adopted a three-phase model to engage community members: Give first; build advocates; and reciprocity. Cores work together to generate educational materials about the study, disseminate them to underserved communities, and increase recruitment. The study also established a Community Research Advisory Board. Members of the study's paid outreach team spoke about the importance of addressing historical trauma with patients and families who may not have had good experiences with health care, showing empathy, and being flexible and setting boundaries.

Staff at the Vanderbilt University Medical Center Recruitment Innovation Center (RIC) introduced its approach and shared resources for enhancing diversity in research. The RIC offers advice to overcome recruitment and retention challenges using evidence-based strategies and materials. Recommendations for community engagement include building benefits for participants into the process, thinking creatively about partnership, practicing shared decision making, and ensuring community members receive fair compensation.

Clinic director Janice Nodvin described the services and approach followed by the health care facility she oversees, which provides services to teens and adults with developmental disabilities and engages them to

participate in research. It offers primary and specialty care in one location and aims to serve the individual and their family, addressing physical, psychosocial, and behavioral health; dealing with insurance and other financial aspects of the transition from pediatric to adult services; and helping families find studies that are a good match.

Participants discussed tactics for engaging the siblings of children who visit a study clinic. Recruitment practices inclusive of families at or below the poverty level included collaborating with partners who work with those communities, using social media to find participants, being flexible, and offering compensation that fits participants' needs. Attendees discussed Special Olympics events as one venue for recruitment.

Lessons in Community Engagement

The final session's presenters spoke about underserved groups. Kate Dougherty, an advocate and parent of a teenager with DS, spoke about the power of exposure and positive pressure to create change in rural areas. 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 strong advocates for people with DS. The goal is to create a culture shift. Using language that resonates and making things pretty and fun can create an opening to reach people who lack experience with disability and change their minds about what is possible.

Brian Chicoine, M.D., the medical director of a DS care facility, described his center's services, which include primary and specialty care for people age 12 and older who have DS, and how it has engaged patients in research. Recently the center has expanded its services to include telehealth and increased outreach in urban centers and with regional partners as part of an effort to acknowledge and address health equity issues.

Elena Grigorenko, Ph.D., an epidemiology researcher who led a study of learning disabilities in Africa, introduced eight principles she and her team follow in their research. The infrastructure put in place through their study should make it possible to conduct a DS study in the future. The principles include following the lead of the local community, engaging local leaders to secure their permission, recruiting local staff, explaining assessments and adapting them to local preferences, rewarding participants, and exiting in a way that makes a return possible.

Noemi Spinazzi, M.D., a Federally Qualified Health Center pediatrician specializing in complex care, underscored the importance of actively undoing existing mistrust. To build a diverse research community, investigators must acknowledge, own, and undo legacies of abuse and mistreatment experienced by people with disabilities, including ongoing structural racism. Dr. Spinazzi described efforts at her clinic to support families and engage them in research, including providing food, having clinicians attend individualized education plan (IEP) meetings, recruiting diverse research staff, engaging siblings who come to research visits, and sharing results. State-of-the-art DS care requires time and resources, advocacy, and care coordination from providers. Funding support for the clinical program would give providers a way to enhance the care they provide with research.

Participants discussed ways to use the concept of fun to increase research participation and the importance of visiting communities to understand their priorities and needs. Offering opportunities to meet and talk with families should be built into the budget for research studies. Also, lessons learned from research in low- and middle-income countries about cultural barriers and partnering with trusted community members can be used elsewhere. Health care reform advocacy to improve conditions for clinicians could help support providers who want to learn more about providing care to DS patients. The Down Syndrome Medical Interest Group (DSMIG) and national advocacy group events are other resources. Clinical program funding could help support programs as models for care and pipelines for the clinical workforce. NIH could establish a clinical pipeline award modeled after mentorship research awards or financially support groups like DSMIG or professional organizations. Grants could fund work with a collaboration outcome or development of a research participant pipeline as the goal rather than a research outcome. Research and care must be intertwined.